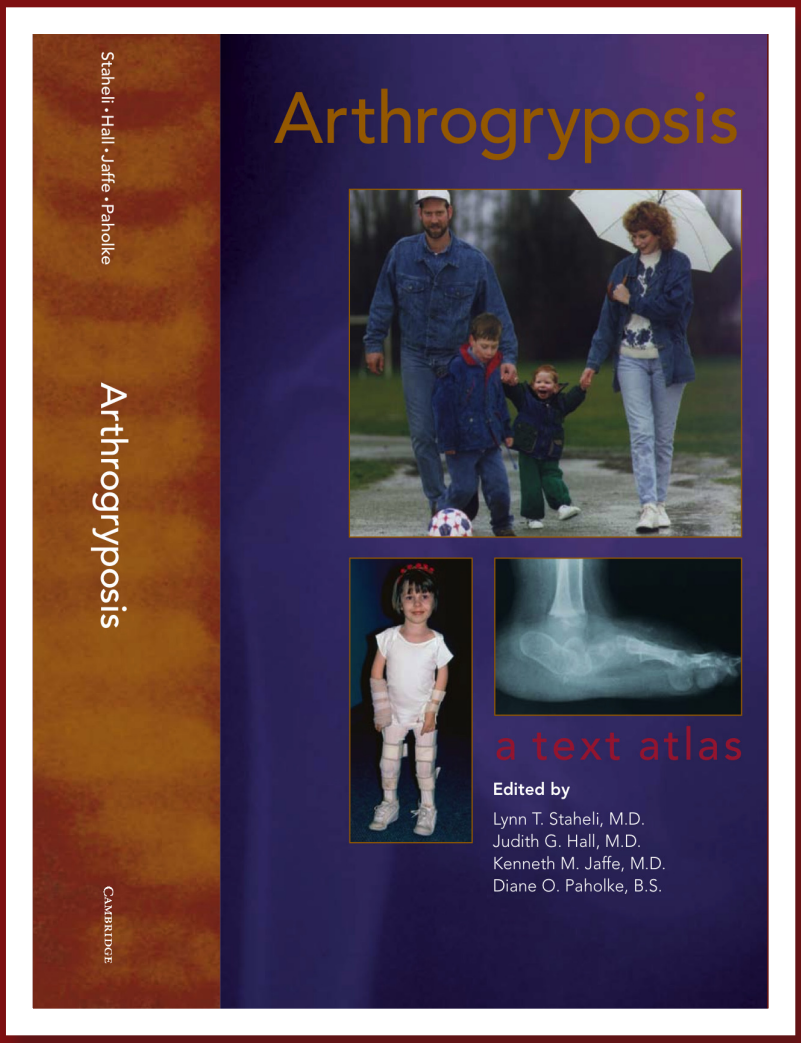
Arthrogryposis

A Text Atlas



Edited By
Lynn T. Staheli, M.D.
Judith G. Hall, M.D.
Kenneth M. Jaffe, M.D.
Diane O. Paholke, B.S.

GLOBAL HELP
Health Education using Low-cost Publications

CAMBRIDGE UNIVERSITY PRESS

Jeff McCord

A Global-HELP Republication

The Global-HELP Organization has sponsored the republication of *Arthrogryposis*, which is now available to be be download without charge from our web-site at www.global-help.org.

We are proud that this publication received a British Medical Association Medical Book Competition Award in 1999.

The authors also wish to give appreciation to:

Cambridge University Press, for releasing the copyright. This was obtained from Marc Strauss, Publishing Director of the Scientific, Technical, & Medical Division of the North American Branch of Cambridge University Press.

Jeff McCord (left), for preparing the original publication and archiving the original text and images, which made this republication possible. His professionalism and foresight in saving the material is commendable, particularly for choosing to donate his professional services for the project. Please contact him for any details at jeffmccord@freelancelot.com.

The authors support this republication and are pleased that their work will be available worldwide with the potential for even greater distribution than the original printed book.

Lynn Staheli, Editor
2008

The term arthrogryposis describes a range of congenital contractures that lead to childhood deformities. It encompasses a number of syndromes and sporadic deformities that are rare individually but collectively are not uncommon. Yet the existing medical literature on arthrogryposis is sparse and often confusing. The aim of this book is to provide health care professionals, individuals affected with arthrogryposis, and their families with a helpful guide to better understand the condition and its therapy. With this goal in mind, the editors have taken great care to ensure that the presentation of complex clinical information is at once scientifically accurate, patient–oriented, and accessible to readers without a medical background.

The book is authored primarily by members of the medical staff of the Arthrogryposis Clinic at Children's Hospital and Medical Center in Seattle, Washington, one of the leading teams in the management of the condition, and will be an invaluable resource for both health care professionals and families of affected individuals.

Arthrogryposis

a text atlas

Editors

Lynn T. Staheli, M.D.
University of Washington Medical School
Children's Hospital and Medical Center, Seattle

Judith G. Hall, M.D.
University of British Columbia

Kenneth M. Jaffe, M.D.
University of Washington Medical School
Children's Hospital and Medical Center, Seattle

Diane O. Paholke, B.S.
Parent
Redmond, Washington

PUBLISHED BY THE PRESS SYNDICATE OF THE UNIVERSITY OF CAMBRIDGE
The Pitt Building, Trumpington Street, Cambridge CB2 1RP, United Kingdom

CAMBRIDGE UNIVERSITY PRESS
The Edinburgh Building, Cambridge CB2 2RU, United Kingdom
40 West 20th Street, New York, NY 10011-4211, USA
10 Stamford Road, Oakleigh, Melbourne 3166, Australia

First published 1998

Printed in the United States of America

Typeset in ITC Giovanni and Avenir

Library of Congress Cataloging-in-Publication Data

Arthrogryposis : a text atlas / edited by Lynn T. Staheli . . . [et al.].

p. cm.

Includes bibliographical references and index.

ISBN 0-521-57106-5 (hc)

1. Arthrogyposis. 2. Arthrogryposis – Atlases. I. Staheli, Lynn T.
RG629.S53A78 1997
618.92'043 – dc20 96–28790
CIP

A catalog record for this book is available from the British Library

ISBN 0-521-57106-5 hardback

Arthrogyrposis Table of Contents

This book, *Arthrogryposis*, is a text atlas, written to meet the needs of health care professionals and families for an overview of the arthrogrypotic syndromes. The book is intended to be comprehensive, scientifically accurate, patient oriented, colorful, compact, engaging, and easily understood. A glossary is provided to help family members without a medical background. To make the book affordable, it has been computer generated. This book has been prepared as a service by the authors, with the goal of improving the understanding of the disease and providing a guide to management of children with arthrogryposis. It is authored primarily by the medical staff of the Arthrogryposis Clinic at Children's Hospital and Medical Center in Seattle, Washington, and is based on our experience in managing hundreds of children with these conditions. The honorarium generated by the book will be used to fund research on arthrogryposis.

The editors appreciate Dr. Richard Barling and his colleagues at Cambridge University Press for accepting our proposal to produce this book. We are indebted to our local contributors, whose work was indispensable:

Jeff McCord, Free-Lancelot, *design direction & prepress*
Heather Johnson, *book design*
Dayna Roberson, Dave King, *additional book production*
Dr. Charlene Butler, *project manager*
Melissa Rodriguez, *secretary*
Lynn Sapp, *Arthrogryposis clinic coordinator*
Vicki McFadden, *Orthopedic Department Manager, facilitated initial financing*
Cheryl Herndon, *illustrator*
Peter Beighton, Maureen Bocian, Bob Gorlin, Gary Greene, Barbara McGilvray, Susan Reed, and David Weaver, *Chapter One patient photographs*

All cover photographs are used by permission.
David Goetze, *family photograph (cover)*
Crane Family, *cover family*

The Club Foot (Ribera) is reproduced on the cover by permission of the Louvre Museum, Paris.

Edward E. Almquist, M.D.
Chapter Three

Clinical Professor
Department of Orthopedics
University of Washington Medical School
Seattle, Washington

Allan W. Bach, M.D.
Chapter Three

Clinical Professor
Department of Orthopedics
University of Washington Medical School
Seattle, Washington

Dawn L. Chaplin, M.S., O.T.
Chapter Six

Consultant
Division of Occupational Therapy
Department of Rehabilitation Medicine
Children's Hospital and Medical Center
Seattle, Washington

Catherine S. Graubert, P.T.
Chapter Six

Special Projects Coordinator
Division of Physical Therapy
Department of Rehabilitation Medicine
Children's Hospital and Medical Center
Seattle, Washington

Judith G. Hall, M.D.
Chapter One

Professor
Department of Pediatrics
University of British Columbia

Head
Department of Pediatrics
British Columbia's Children's Hospital
Vancouver, British Columbia

Deborah L. Hill, Ph.D.
Chapter Seven

Clinical Assistant Professor
Department of Rehabilitation Medicine
University of Washington Medical School

Clinical Psychologist and Pediatric
Neuropsychologist
Department of Rehabilitation Medicine
Children's Hospital and Medical Center
Seattle, Washington

Kenneth M. Jaffe, M.D.
Chapters Five & Six

Professor
Department of Rehabilitation Medicine
University of Washington Medical School

Adjunct Professor
Pediatrics and Neurological Surgery
University of Washington Medical School

Director
Department of Rehabilitation Medicine
Children's Hospital and Medical Center
Seattle, Washington

Michael O. La Grone, M.D.
Chapter Three

Clinical Assistant Professor
Department of Orthopedic Surgery
Texas Tech University Health
Sciences Center
Amarillo, Texas

Diane O. Paholke, B.S.E.E.
Bibliography

Computer Technologist and Designer
Parent of a Child with Arthrogryposis
Redmond, Washington

Brian Ross, M.Ed.
Chapter Eight

Director
Department of Education
Children's Hospital and Medical Center
Seattle, Washington

Lynn T. Staheli, M.D.
Chapters Two & Four

Professor Emeritus
Department of Orthopedics
University of Washington Medical School

Consulting Orthopedist
Children's Hospital and Medical Center
Seattle, Washington

Chapter Contents **Page**

J.G. Hall, M.D.

Fig 1.1 Arthrogryposis has been around for a long time.
This painting by Ribera (1642) called "The Club Foot" hangs in the Louvre Museum in Paris. The boy clearly has many congenital contractures, including the clubfoot, and may well have had the common sporadic type of arthrogryposis called amyoplasia. Reproduced by permission.

Introduction

The presence of multiple congenital contractures, or arthrogryposis multiplex congenita (AMC), has been recognized and reported in the medical literature for many years (Fig. 1.1). At first, the term arthrogryposis was used as a diagnosis for any child born with multiple congenital contractures. Over the years as different types of conditions with multiple contractures became apparent, reports in the medical literature began to use the term as a clinical sign or as a general category of disorders so that arthrogryposis became a descriptive word rather than a diagnosis. The conditions that have been called arthrogryposis range from well-known syndromes to nonspecific combinations of joint contractures (Hall, 1995). For the purpose of this book, the term arthrogryposis is used as a generic term that encompasses many different types of multiple congenital contractures.

Arthrogryposis has been considered a rare and unusual condition, and because of the many different ways the term has been used, the medical literature is often confusing. This is particularly frustrating to families with an affected individual because reports about arthrogryposis are not readily available to the lay public. When the parents of an affected child are told of a diagnosis of arthrogryposis, it is usually the first time they have heard of the condition, and reading the inconsistencies and contradictions present in the medical literature may add to their confusion.

The objective of this book is to provide individuals with arthrogryposis, their families, and the health care workers involved in their care with a helpful guide to understand the basic concepts underlying arthrogryposis and its therapy. This book is designed to answer some of the questions often asked by the affected individuals and their families, as well as to serve as a general reference for the many conditions with multiple congenital contractures.

Arthrogryposis Definition

Arthrogryposis, or arthrogryposis multiplex congenita (AMC), as mentioned previously, is a generic term used to describe the presence of multiple congenital contractures. The word arthrogryposis, *arthro*, joint, *gryp*, curved, literally means curved joint (implying that it is fixed or stuck in the curved position). Thus, arthrogryposis multiplex congenita means curved (fixed) joints in many (multiple) areas of the body, which are present at birth (congenita).

A contracture is the limitation of movement of a specific joint, in other words, a joint that does not have a full range of movement. The contractures in most forms of arthrogryposis are usually nonprogressive and involve more than one body area. The word congenital simply means that the contractures are present at birth; that is, they have occurred or been produced before birth. For the purposes of this book, arthrogryposis is defined as congenital nonprogressive limitation of movement of two or more joints in different body areas. Occasionally, there are conditions in which contractures are progressive.

Incidence of Arthrogryposis

Arthrogryposis is relatively rare. It has been estimated to occur once in every 3000 live births. However, many types of specific congenital contractures in a particular body area, such as clubfoot or dislocated hips, are much more common. At least one in every 200 infants is born with some form of congenital contracture or stiff joint (Fig. 1.2).

Type	Incidence
Clubfoot	1/500
Congenital dislocated hips	1/200-1/500
Multiple contractures	1/3000
All congenital contractures	1/100-1/250

Fig. 1.2 Occurrence of congenital contractures in the newborn.

Causes of Arthrogryposis

Studies in animals have shown that anything that prevents normal in utero or intrauterine (i.e., inside the uterus of the pregnant mother) movement of a fetus or that leads to limitation in the movement of a joint during fetal growth will lead to a contracture(s) at birth. The earlier in development and the longer the duration during which limitation in movement is present, the more severe the contracture is likely to be at birth (Moessinger, 1983; Hall, 1986a).

Arthrogryposis is not a problem in the formation of the joint or limb (the formation of organs and systems of the human body occurs in the first 8 weeks of pregnancy and is called embryogenesis), but rather it is a problem during fetal life (i.e., after 8 to 10 weeks of the pregnancy). The joint is likely to be normal, but lack of movement is associated with the development of extra connective tissue around the joint. This extra connective tissue fixes the joint in place and limits movement even more. Because the affected joint has not moved normally during fetal life, the tendons around the joint may not have stretched to their normal length, and this makes normal joint movement (and physical therapy) after birth even more difficult. Over a period of time if the joint is not used, the surfaces at the end of the bones within the joint begin to assume a different and flattened contour with more acute edges. This may lead to still further difficulty with achieving the full range of movement of that joint.

The in utero process of restricted movement leading to a contracture can be compared with a child wearing a cast as therapy to limit the movement of a broken bone while it is healing. When the cast is removed, the joint that has been held in place is usually very stiff, and there is limitation of the full movement of the joint. In the situation of casting for a broken bone, the

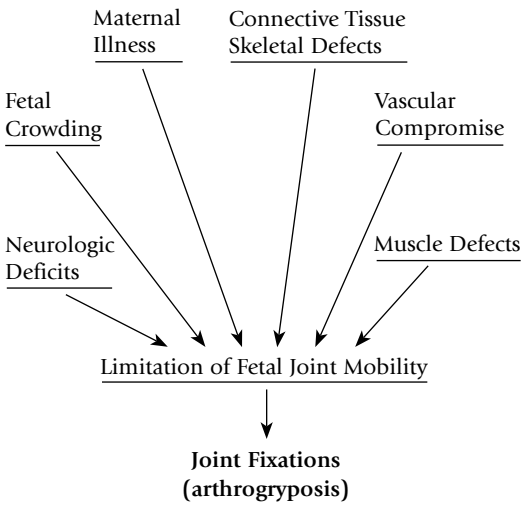


Fig. 1.3 Causes of arthrogryposis.
Anything that causes decreased fetal movement or prevents normal fetal movement may lead to contractures in the newborn. Any limitation of movement of the fetal joints, even for relatively short periods of time, such as a few days, may result in fixation of the joint.

vlimitation of joint movement is only temporary (usually 6 to 8 weeks), and with physical therapy the joint range of movement is usually regained completely. However, when there is limitation of movement during a pregnancy there is also limitation of growth of the limb, which seems to compound the severity of the contracture even more. Also the period of limited movement during the pregnancy is usually several months.

In general, there are six major categories of problems leading to limitation of movement in an embryo or fetus (Fig. 1.3).

Abnormalities of the Muscle Structure or Function

These are called myopathic processes. In these individuals, muscles form abnormally or develop normally but do not function properly. In most cases, the cause of this lack of muscle development or abnormality in muscle function is not known. Some suspected causes include muscle disease such as congenital muscular dystrophies, mitochondrial disturbances such that the muscles do not have enough energy to function normally, and abnormalities of the biochemistry of the muscle.

Abnormalities of the Nerves That Connect to the Muscles

These are called neuropathic processes. There may have been a failure of the nerves to form, failure to mature, or failure to function properly. The problem can be in the brain, in the spinal cord, or in the peripheral nerves and their connection to the muscle. When the central nervous system and spinal cord are malformed, as in individuals with neural tube defects (defects in the closing of the spine), there may be very severe lack of movement. Failure of neurons to mature or myelinate (formation of the insulation of the nerves) properly can also lead to lack of normal movement. Arthrogryposis due to abnormalities in the development and function of the central nervous system is often accompanied by structural abnormalities that can be seen on imaging studies or if nerve tissue is examined carefully at autopsy.

Abnormalities of Connective Tissue

In this type of problem, the tendons, bones, joint, or joint lining develop abnormally in such a way that normal movement cannot occur during fetal development and contractures are present at birth. Examples of abnormal connective tissue are seen in diastrophic dysplasia (a dwarfing condition with clubbed feet and hands) or when there are abnormal tendon attachments. The tendons may have developed appropriately but may not have attached to the proper place around the joint or on the bone. If this happens, normal movement of the joint may not occur during fetal life, leading to contractures at birth. This is the case in some forms of distal arthrogryposis (Hall et al., 1982a).

Limitation of Space or Restriction of Movement Within the Uterus

In certain situations there is limited room within the uterus. In multiple births, such as in twin pregnancies, there is less room for the fetuses to move around. Twins are more prone to develop contractures than singletons. In other cases there may be a lack of the normal amount of amniotic fluid (i.e., amniotic leakage leading to less room to move). The mother may have structural abnormalities of the uterus that do not allow the fetus to move freely. Any force that causes compression within the uterus may cause limitation of movement and secondary contractures.

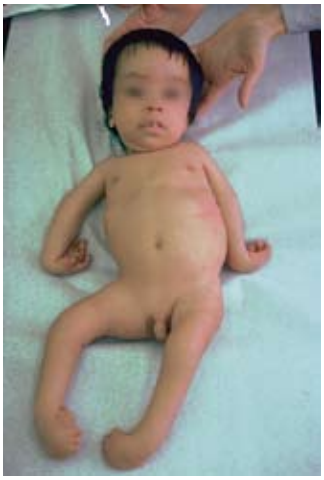
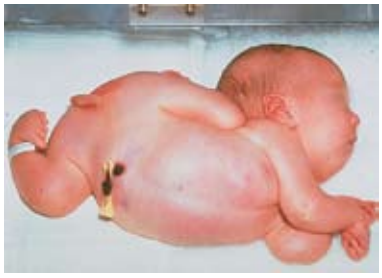


Fig. 1.4 Infant with typical amyoplasia.
Amyoplasia is a specific form of arthrogryposis. It is characterized by typical positioning of the limbs. The involvement is usually, but not always, symmetric. When the arms are involved, the shoulders are usually internally rotated and fixed in extension and the wrists are flexed. When the legs are involved, the feet are usually clubbed in equinovarus.



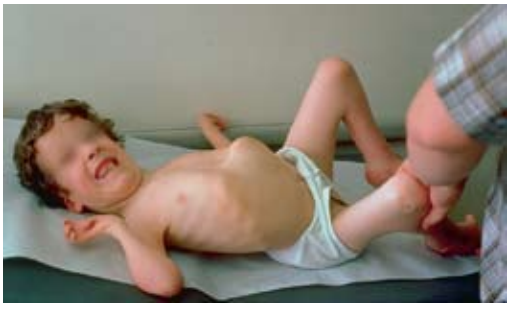


Fig. 1.5 Severely affected individual with amyoplasia.
(Top) This newborn is very severely involved with scoliosis and fixed flexion of his elbows and knees with practically no muscle in his limbs. The muscle has been replaced by fat and fibrous tissue. (Bottom) The same boy, bright and clever, has figured out how to do many things in spite of very little strength and limited range of movement. Here he is receiving physical therapy to prevent recurrence of the contractures.

Vascular Compromise Leading to Loss of Neurons

In this type of problem, the contractures are the result of the lack of blood circulating normally to nourish the nerves that lead to the muscles or to the bones that make up the joint. There have been several reports of individuals who were born with multiple congenital contractures after severe bleeding during the pregnancy or after a failed attempt at termination of the pregnancy (Hall, 1996).

Maternal Illness Leading to Contractures

A number of maternal metabolic disorders and maternal illnesses, such as multiple sclerosis (Livingstone and Sack, 1984), diabetes mellitus, and myasthenia gravis (Moutard-Codou et al., 1987), have been associated with the presence of multiple congenital contractures in the newborn. Maternal hyperthermia during the first trimester that raises the mother's core temperature for a certain period of time (high fever, prolonged hot baths, jacuzzis, or hot tubs) can be associated with congenital contractures in the newborn (Reid et al., 1986; Edwards, 1986). Maternal antibodies against fetal neurotransmitters can also lead to arthrogryposis.

In a given individual or specific entity, several processes may be taking place at the same time during pregnancy, which may accentuate the deformities.

Common Types of Arthrogryposis

At least 150 specific entities have been recognized that have multiple congenital contractures (arthrogryposis). It is important to make an accurate diagnosis in each individual with arthrogryposis and make use of all the diagnostic tools available. A specific diagnosis will provide information about the natural history, the prognosis, the recurrence risk, and the best therapies.

The easiest way to approach the differential diagnosis of different types of arthrogryposis is to separate and classify a specific case into one of three groups: (1) disorders with mainly limb involvement, (2) disorders with limb involvement plus involvement of some other body area(s), and (3) disorders with limb involvement and central nervous system dysfunction.

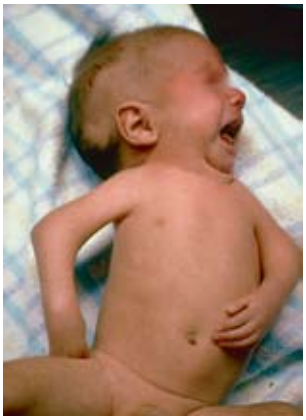


Fig. 1.6 Infant with amyoplasia.
This baby has very little muscle mass in his limbs, but he likes to move his trunk and head. His jaw is just a little small, and he has mild flexion at the elbows. He has a mild birthmark over his nose and forehead.

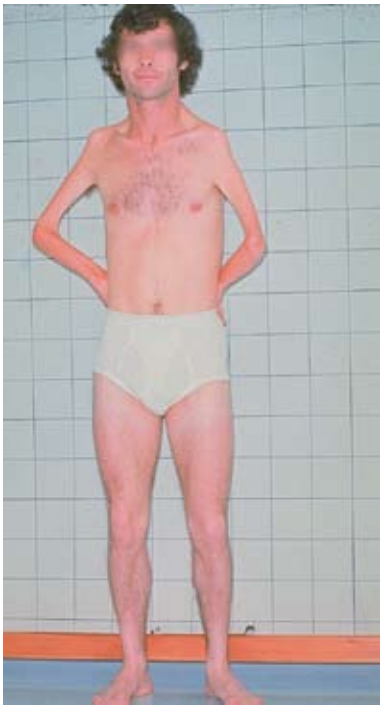


Fig. 1.7 Man with amyoplasia.
This man with amyoplasia has involvement only of his arms, where he has markedly decreased muscle mass and internal rotation. As a young child, his elbows were much straighter (more extended), but as the bones in his arms grew, the fibrous bands that had replaced most of his muscle did not grow as much, leading to flexion of the elbows. The bones in affected limbs do not grow as much as normal.

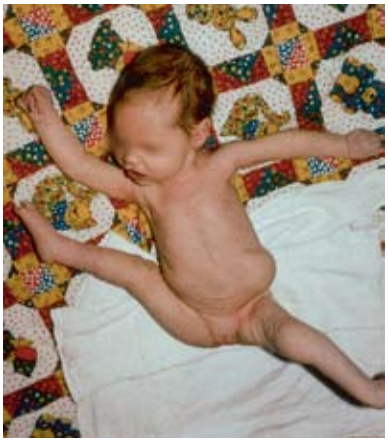


Fig. 1.8 Infant with amyoplasia and abdominal wall muscle defect.
10% of all individuals with amyoplasia have some type of abnormality of intestine or abdominal wall, which appears to be due to intrauterine vascular accidents.

Mainly Limb Involvement

Some of the most common disorders involving mainly limbs are discussed in the following section.

Amyoplasia

Amyoplasia is the most common type of arthrogryposis. In the older medical literature, it is called "classical arthrogryposis." The term means, *a*, no, *myo*, muscular, and *plasia*, growth. There are other types of arthrogryposis in which there is very little muscle growth, but this is a very specific condition. Amyoplasia has an incidence of 1 in every 10,000 live borns. It represents one third of all cases of liveborns with arthrogryposis. Amyoplasia is characterized by typical symmetric positioning of the limbs (Fig. 1.4) with severe equinovarus feet and extended elbows, absent muscle tissue with fibrotic replacement, midfacial haemangioma, and normal intelligence (Figs. 1.5 and 1.6). It has surprisingly good response to early physical therapy (Sells et al., 1996). Some individuals have only legs involved and more rarely only arms involved (Fig. 1.7). Most affected individuals have all four limbs involved but the trunk spared. About 10% of the individuals with amyoplasia have abdominal structural anomalies (Hall et al., 1983a) (Fig. 1.8). Amyoplasia also appears to be increased in one of identical twins (Hall et al., 1983b) (Fig. 1.9). Amyoplasia is considered a sporadic disorder and has not been observed to recur in siblings or in offspring.

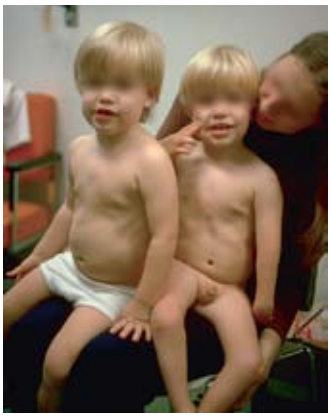


Fig. 1.9 Monozygotic twins, one with amyoplasia.
(Top left) Amyoplasia appears to be increased in one of monozygotic (identical) twins. Many cases have been reported where only one twin has contractures. Obviously, they do not seem identical, but they have come from one fertilized egg. (Bottom left) These twins are a little older, and the affected twin is smaller. Bones that are not used do not grow quite as much as normal. The lack of normal muscle in amyoplasia also makes the arms look smaller around. (Right) The twin on the right hides his affected arms by wearing long sleeves, but his cast gives away his arm involvement. This twin has only arm involvement.

Fig. 1.10 Typical hands in distal arthrogryposis type I.
There are characteristic changes in the hands. (Top) In the newborn, the hand is clenched and the fingers overlap. (Middle) With physical therapy and use, the fingers usually open up and are quite functional. There may be some residual contractures and abnormal creasing. (Bottom) Occasionally, ulnar deviation and contractures persist and may look like arthritic changes. However, there is no pain or progression.

Distal Arthrogryposis

Another type of arthrogryposis with only limb involvement is distal arthrogryposis type I. This disorder has a characteristic position of the hands (Fig. 1.10) [medially overlapping fingers, clenched fists, ulnar deviation of fingers, and contractures of the fingers (camptodactyly)] together with foot contractures. Contractures at other joints are variable. In addition to the contractures of the hands and feet, usually only knees and hips are involved and usually fairly mildly (Fig. 1.11 and Fig. 1.12). There are no associated visceral anomalies, and intelligence is normal. Distal arthrogryposis type I has a relatively good response to physical therapy (Hall et al., 1982a; Hall, 1995). It is inherited as an autosomal dominant trait, and the gene has been mapped to chromosome 9, specifically 9p22-q22.3 (Bamshad et al., 1994).

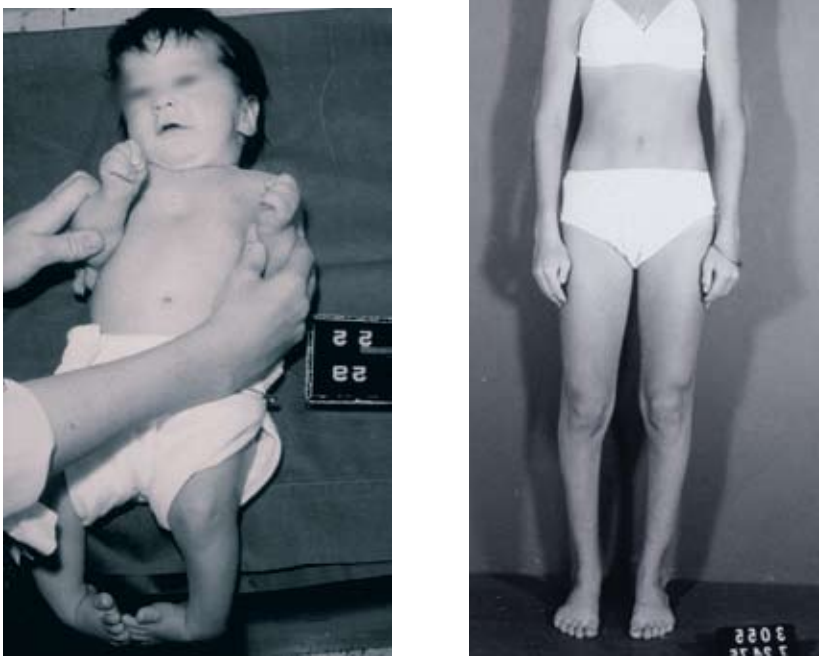


Fig. 1.11 Distal arthrogryposis Type I.
The hands and feet are usually the most severely involved, and the trunk and head are spared. (Left) The typical clenched fist with overlapping fingers can be seen in the infant. The feet may be deformed in many different ways. (Right) At an older age, a very good and functional result has been achieved.

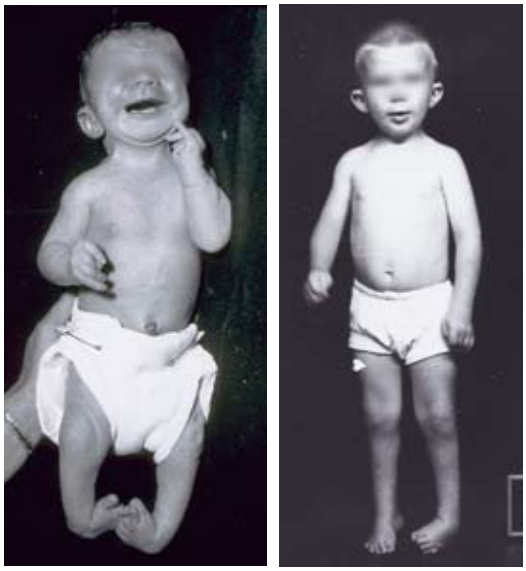


Fig. 1.12 Distal arthrogryposis Type I.
(Left) Typical changes primarily involving the hands and feet are seen in this baby. The hands are beginning to open. (Right) After a few years, he has become much more functional. Intelligence is normal, and usually there is no involvement other than contractures. Within a family, the amount of involvement can be quite variable.

Bony Abnormalities Confused with Arthrogryposis

There are many bony anomalies where bones are fused to each other that can be confused with arthrogryposis. These include symphalangism or fusion of the phalanges (bones of the fingers) (Fig. 1.13) (Matthews et al., 1987), coalition or fusion of the carpals (bones of the wrist) and tarsals (bones of the ankle), and synostosis or fusion of other bones, such as the elbow or ear bones (ossicles) (Fig. 1.14). Some of these types of bony fusions can run in families. Others occur sporadically. There are many specific entities that cause limitation of movement.

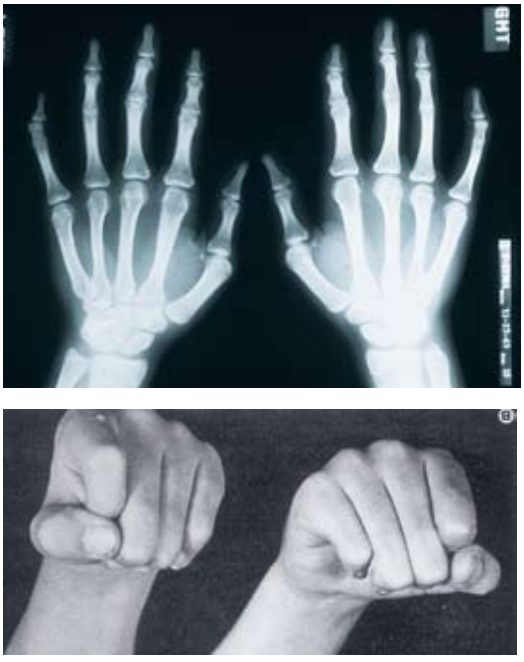


Fig. 1.13 Symphalangism.
When bones are fused together, the joint will not move. This can be confused with arthrogryposis where the joint is held by connective tissue. (Top) An x-ray helps to identify bone fusion. (Bottom) The bones of the fingers are fused and will not flex. Many other types of bones can be fused at birth and mimic arthrogryposis.

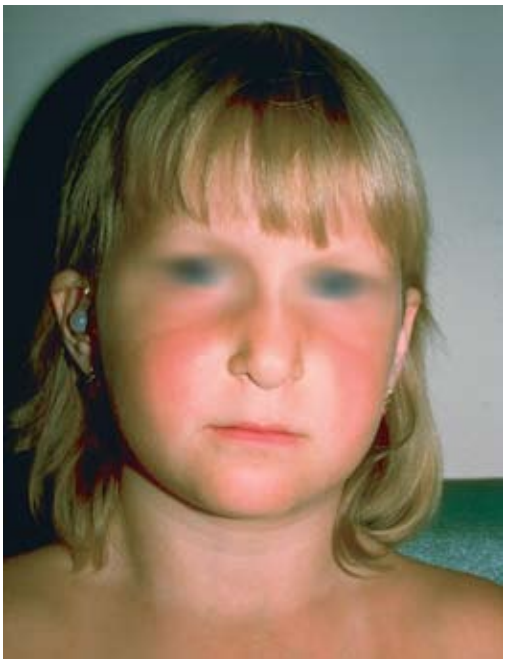


Fig. 1.14 Multiple synostoses.
This girl has fusion of many bones in her body. Multiple synostoses may even involve bones of the ear, leading to deafness. The bones of the nose are typically broad in the midnose.

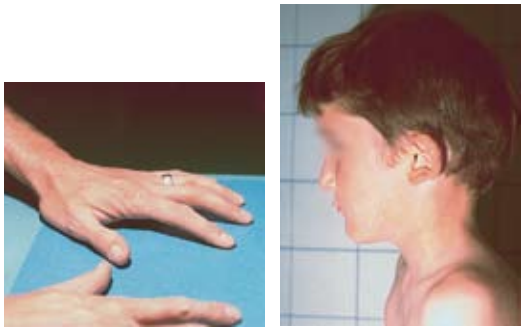


Fig. 1.15 Contractural arachnodactyly.
Multiple contractures together with long thin fingers and toes suggest contractural arachnodactyly.

Contractural Arachnodactyly

Contractural arachnodactyly is often referred to as Beals syndrome (Beals and Hecht, 1971). Individuals with contractural arachnodactyly are usually very long and thin and have a characteristic crumpled ear in addition to contractures of their joints (Ramos-Arroyo et al., 1985) (Fig. 1.15). This disorder is inherited as an autosomal dominant trait and has been associated with the fibrillin gene located on chromosome 5q23-q31 (Lee et al., 1991). The differential diagnosis of contractural arachnodactyly includes disorders with loose joints, such as Marfan syndrome. However, the cardiovascular and ocular problems seen in Marfan syndrome do not occur in individuals with contractural arachnodactyly (Viljoen, 1994).

Limbs Plus Other Body Areas

There are many specific syndromes with contractures and involvement of other areas of the body.

Multiple Pterygium Syndromes

The best examples of arthrogryposis that involves the limbs plus other body areas are the various types of multiple pterygium syndromes (Fig. 1.16). There are many different types of pterygium syndromes. A pterygium is a winglike structure, web, or triangular membrane that forms across a body joint (Fig. 1.17). The different pterygium syndromes have different forms of inheritance and characteristic features (Figs. 1.18 through 1.22). Individuals with pterygium syndromes often have webs of skin at their neck, knees, and elbows, as well as multiple congenital contractures (Hall et al., 1982b; Hall, 1984a).

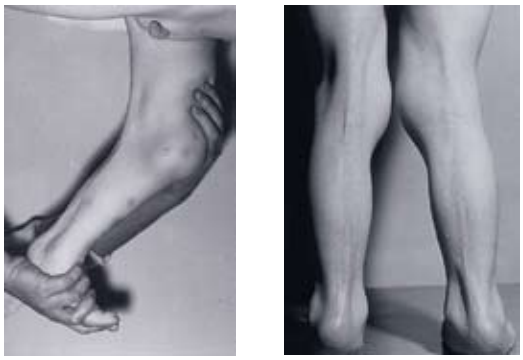


Fig. 1.17 Pterygium (web) across knee joint.
Sometimes, the edge of the web is thickened and fibrous and may even contain a nerve or a blood vessel or both.

Type	Inheritance	Distinguishing Features
Popliteal pterygium	AD	Clefts, lip pits, normal hands, abnormal nails
Antecubial pterygium	AD	Only elbows, abnormal elbow joint
Multiple pterygia *(Escobar type)*	AR	Cervical vertebral anomalies, hands involved, chin-sternum ptergyia, dysmorphic facies
Multiple pterygia	AD	With and without mental retardation
Lethal multiple pterygium	AR	Extensive contractures, hypertelorism, chin-sternum ptergyia, small chest
Lethal popliteal pterygium	AR	Facial cleft, syndactyly (hands and feet), genital skin anomaly
Pterygium and malignant hyperthermia	AR	General congenital contractures, cleft palate, torticollis, malignant hyperthermia

Fig. 1.16 Pterygium syndromes.

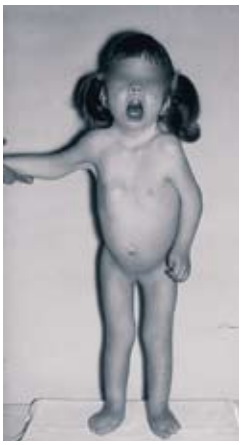
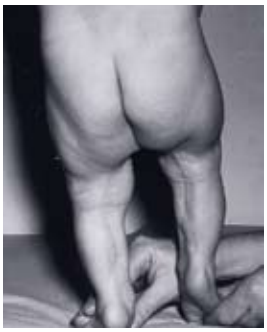


Fig. 1.18 Multiple pterygium syndrome.
Also called Escobar syndrome. (Top) In the newborn period, the webs are often not striking. (Bottom left) They become more obvious with age. (Bottom right) By adolescence, there is often increased lumbar lordosis and involvement of the spine.



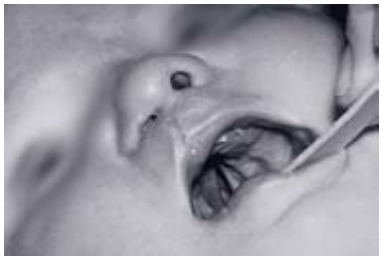
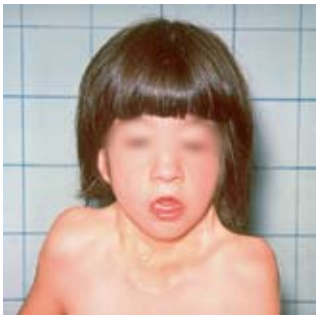


Fig. 1.19 Popliteal pterygium syndrome.
This child has popliteal pterygium syndrome in which cleft palate, cleft lip, webs in the mouth, and unusual nails are seen. Typical popliteal webs are seen at the knees. Marked variability within an affected family is often present.



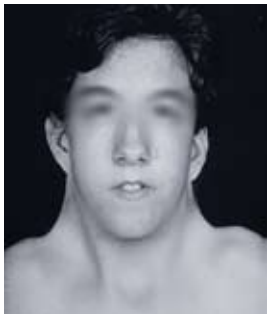


Fig. 1.20 Multiple pterygium syndrome (Escobar type).
Webbing of the neck may increase with age, and the face may seem to have decreased movement.

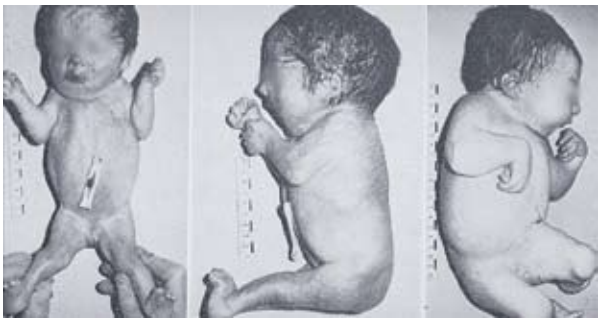


Fig. 1.21 Lethal multiple pterygium syndrome.
Marked webbing of multiple joints is seen at birth. These children usually have underdeveloped lungs and do not survive. The involvement tends to be consistent within a family. As an autosomal recessive disorder, there is a 25% recurrence risk for additional children to be affected.

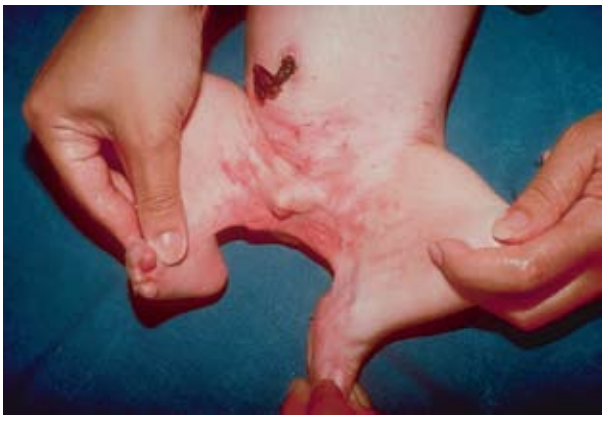


Fig. 1.22 Lethal popliteal pterygium syndrome.
This baby has lethal popliteal pterygium syndrome (Bartsokas-Papas syndrome) in which there are severe webs present in the newborn period across the knee. There is also facial clefting and fused digits at birth. These children usually do not survive. It is an autosomal recessive disorder, with 25% recurrence risk after one affected child has been born to a couple.

Fig. 1.23 Freeman-Sheldon syndrome.
Note contractures of feet and hands with limited movement of the face in this father and daughter. Dad hides the facial involvement with his handsome beard.

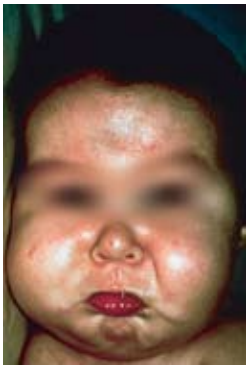


Fig. 1.24 Freeman-Sheldon syndrome.
Infant with contractures of facial muscles giving a puckered appearance. Her lips look like she is trying to whistle. Hence, this syndrome is sometimes called whistling face syndrome.

Fig. 1.25 Freeman-Sheldon syndrome.
This Northwest Indian mask has many features suggestive of Freeman-Sheldon syndrome. The legends about the totem Zunoqua contain many features that would be seen in Freeman-Sheldon syndrome.

Freeman-Sheldon Syndrome (Whistling Face Syndrome)

This disorder was first described by Freeman and Sheldon in 1938. It is an autosomal dominant disorder, although there are some families reported as having autosomal recessive inheritance (Fitzsimmons et al., 1984). Individuals with whistling face syndrome have a full forehead and masklike faces with a small mouth giving a whistling face appearance, deepset eyes, broad nasal bridge, epicanthic folds, strabismus, small nose, high arched palate, small tongue, an H-shaped cutaneous dimpling on the chin, flexion of fingers, equinovarus feet with contracted toes, kyphosis, and scoliosis (Figs. 1.23 through 1.27). Other abnormalities include postnatal growth deficiency, inguinal hernias, and incomplete descent of the testes.

Osteochondrodysplasia

Many osteochondrodysplasias, or dwarfing conditions, also have contractures and thus have a combination of disproportionate short stature and arthrogryposis. Most dwarfing conditions have abnormalities of the connective tissue and bones. Metatropic dysplasia, Kniest syndrome, camptomelic dysplasia, osteogenesis imperfecta, parastrammatic, Jansen metaphyseal dysplasia, Saul Wilson syndrome, geleophysic syndrome, synspondylism, spondyloepiphiseal dysplasia, otospondylometaphyseal dysplasia, and diastrophic dysplasia are some of the osteochondrodysplasias known to have congenital contractures.

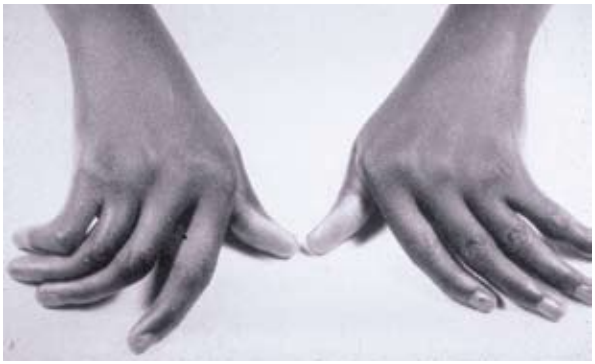


Fig. 1.26 Hands in Freeman-Sheldon syndrome.
When the finger contractures and clenched hands open out, they often have some ulnar deviation.

Fig. 1.27 Feet of father and daughter with Freeman-Sheldon syndrome.
Both had a good response to therapy.

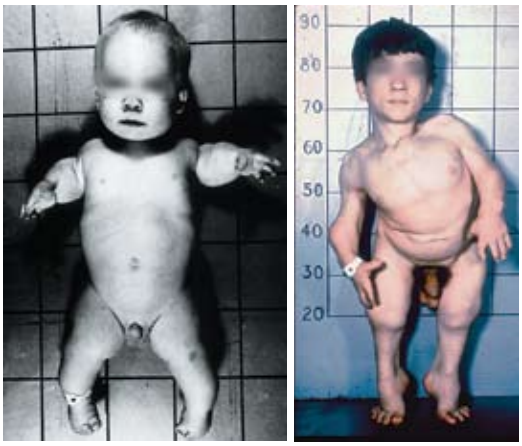


Fig. 1.28 Diastrophic dysplasia.
Many joints are involved and usually get worse with age. Bones and other connective tissue are also involved. The basic defect is related to lack of an enzyme in connective tissue.

Fig. 1.29 Mother and daughter with distal arthrogryposis Type IIB.
Note the lack of movement in the face and residual contractures in the mother's hands.

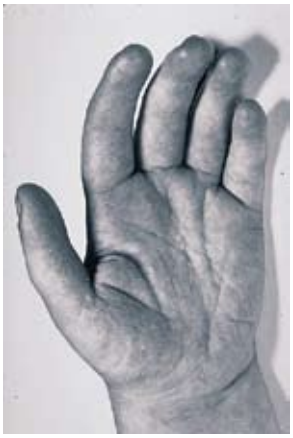


Fig. 1.30 Hand in distal arthrogryposis type IIB.
Note the thickened skin and abnormal flex in creases.

Diastrophic Dysplasia

Fig. 1.28 is a type of dwarfism with autosomal recessive inheritance, char-acterized by short stature, short extremities, multiple joint contractures, clubfeet, proximally placed hypermobile thumbs, and progressive kyphoscoliosis. The contractures involve the shoulders, elbows, interphalangeal joints, and hips. Other features include cystic masses of the external ear usually appearing between the first and twelfth week of life, and cleft palate in 10% of cases.

The major radiographic findings are shortening and metaphyseal widening of the tubular bones, flattening of the epiphyses, irregular deformity and shortening of the metacarpals, metatarsals, and phalanges, pes equinovarus, and kyphoscoliosis.

Infants with diastrophic dysplasia have a high mortality rate, but after infancy their life expectancy is normal. However, in some cases, severe kyphoscoliosis may compromise cardiac and pulmonary function. They have moderate to severe restriction of movement and normal intelligence.

Distal Arthrogryposis Type IIB

One type of distal arthrogryposis that involves the muscles has firm-feeling muscles, decreased eye movements, and thickened skin (Figs. 1.29 and 1.30). It may be inherited as an autosomal dominant condition. Some affected individuals have abnormalities of the mitrochondria (small structures in the cells involved in providing energy).

Distal Arthrogryposis Type IIE

Among cases of arthrogryposis that primarily involve the distal parts of the limbs is a relatively common, sporadic (does not run in families) condition with limited jaw opening (trismus) (Fig. 1.31) and an unusual contracture of the hand in which the wrist is flexed but the metacarpalphalangeal joint (palm to finger) is extended.

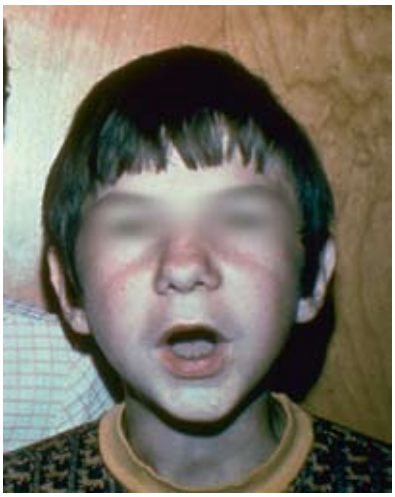


Fig. 1.31 Limited opening of the jaw (trismus).
This is seen in type IIE distal arthrogryposis and in trismus pseudocamptodactyly.

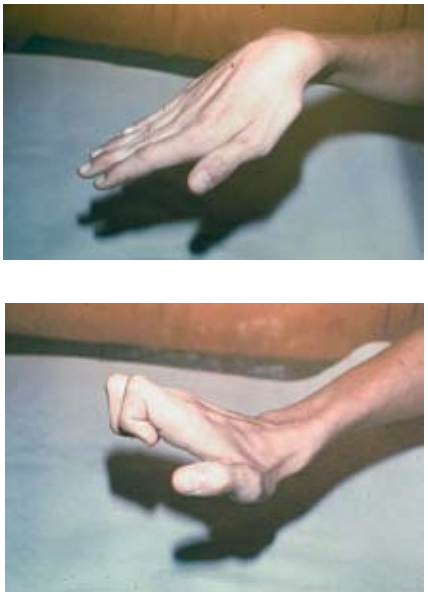


Fig. 1.33 Hands in trismus pseudocamptodactyly.
When the wrist is flexed, the fingers can open (top), but when the wrist is extended, the tendons are too short to allow the fingers to open and can look like or lead to contractures (bottom). Limited jaw opening (trismus) is inherited together as an autosomal dominant trait.

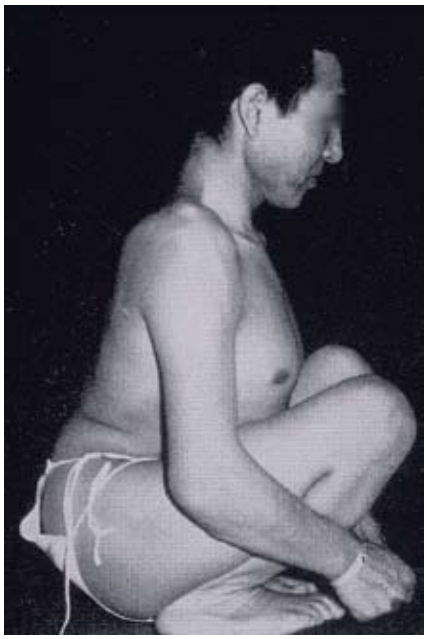


Fig. 1.34 Kuskowin syndrome.
A type of congenital contractures seen among Alaskan aboriginal peoples.

Fig. 1.35 Infant with tuberous sclerosis and arthrogryposis.
A rare occurrence probably reflecting CNS involvement with tuberous sclerosis.

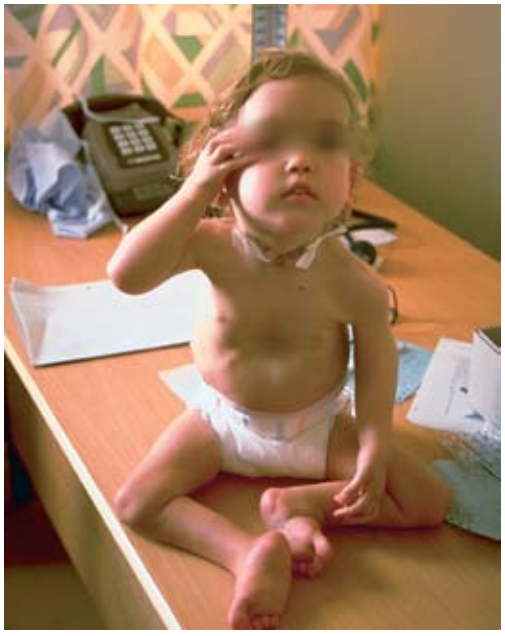
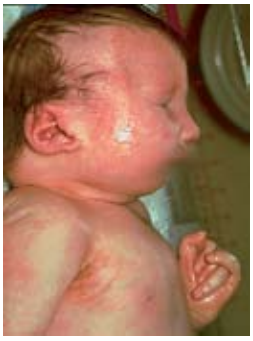


Fig. 1.36 Larsen syndrome.
Dislocations together with contractures and flat "dished-out" face are seen in this syndrome.



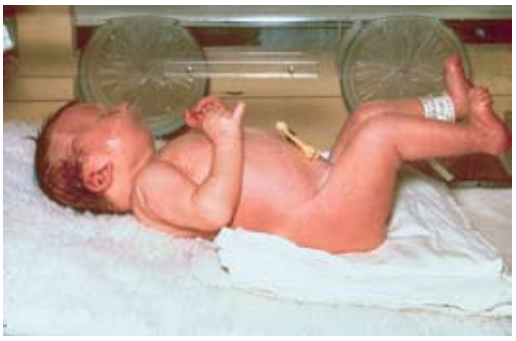


Fig. 1.37 Newborn with cerebrooculofacioskeletal (COFS) syndrome.
Note the small eyes. There are cataracts present. The contractures are obviously quite fixed. Note the unusual position of the fingers with overlapping fifth finger and unusually shaped ear.

Fig. 1.38 Hand contractures in child with chromosomal mosaism.
Note the tight fist and elevated "tea drinker's" fifth finger, often seen with severe CNS involvement.

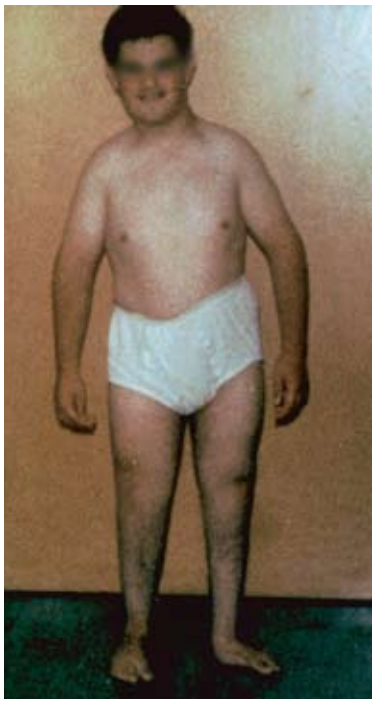


Fig. 1.39 Chromosomal mosaism and arthrogryposis.
This child has developmental delay, arthrogryposis, and streaky pigment in the skin. Chromosome studies show some normal cells and some with an extra chromosome.

Other Syndromes

Syndromes that also affect the limbs and other areas of the body include trismus pseudocamptodactyly (Fig. 1.33), Kuskowin syndrome (Fig. 1.34), tuberous sclerosis (Fig. 1.35), and Larsen syndrome (Fig. 1.36).

Limbs Plus Central Nervous System Dysfunction

Cerebrooculofacioskeletal (COFS) Syndrome

COFS is an autosomal recessive disorder with intrauterine growth retardation, microcephaly (small head), structural abnormalities of the brain, eye abnormalities such as microphthalmia (small eyes) and cataracts, micrognathia (small jaw), abnormal ears, hypotonia (floppiness), and congenital contractures (Fig. 1.37). The congenital contractures include overlapping flexed fingers and flexed hips and knees. Infants with COFS are usually very unresponsive and do not interact with their environment. COFS is a degenerative disorder with progressive failure of proper maturation of the nerve cells. It is lethal, although the natural history of this disorder varies from family to family. In some cases, the contractures are present at birth, whereas in others the contractures develop later in life (Winter et al., 1981).

Restrictive Dermopathy

There are several reports of children born with contractures as a result of failure of the fetal skin to grow normally. The skin is so tight that it restricts movement during development of the fetus and leads to contractures. Other structures are normal. This disorder is lethal and usually familial (Happle et al., 1992).

Congenital Contractures and Chromosomal Abnormalities

It is important to do chromosome studies in individuals without a specific diagnosis who have multiple congenital contractures and mental retardation (Figs. 1.38 and 1.39). It has been found that the presence of contractures in these individuals may be due to chromosomal abnormalities. To rule out chromosomal mosaicism (some normal and some abnormal cells), chromosome studies in fibroblasts (skin) must be done (Fig. 1.40).

Among arthrogryposis patients studied: *80/350 (23%) were mentally retarded*

Among mentally retarded patients with nonspecific multiple congenital contractures: *13/80 (16%) had an abnormal karyotype*

Among mentally retarded/multiple congenital contractures patients with normal karyotype: *2/13 (15%) had familial chromosomal rearrangements*

Among mentally retarded/multiple congenital contractures patients with de novo abnormal karyotype: *5/11 (45%) had mosaicism with some normal cells*

Among mentally retarded/multiple congenital contractures patients with mosaicism: *3/5 (60%) had tissue mosaicism*

Among mentally retarded/multiple congenital contractures patients with mosaicism: *2/5 (40%) had normal lymphocytes and abnormal fibroblasts*

Fig. 1.40 Chromosomal studies in arthrogryposis.

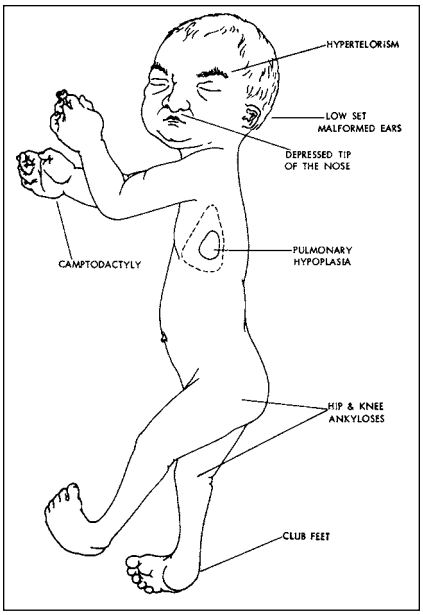


Fig. 1.41 Diagram of fetal akinesia. *Abnormalities seen when the human fetus lacks normal movement.*

Fetal Akinesia

Fetal akinesia refers to features seen when a fetus does not move (*a*, no, *kinesis*, movement) during the pregnancy (Fig. 1.41). Use (i.e., movement) of muscles is essential for normal development of the structures of the fetus. The features of a fetus who has not moved in utero were first described as Pena-Shokeir syndrome (Pena and Shokeir, 1970). These features are noted to occur when there was absent or very little movement of the fetus during the pregnancy. This lack of movement in utero leads to congenital contractures, and the degree of congenital contractures depends on the time of onset of the akinesia during the pregnancy.

Decreased movement during the pregnancy leads to a whole series of abnormalities, including intrauterine growth retardation, congenital contractures of the limbs, craniofacial abnormalities with micrognathia with or without cleft palate, small mouth, and a distinctive nose with a very high bridge and depressed tip. Pena-Shokeir syndrome is lethal because without movement during intrauterine development, the lungs of the fetus do not develop properly (i.e., the lungs are hypoplastic), which leads to respiratory failure and death after the infant is born. Other anomalies seen with fetal akinesia (Pena-Shokeir syndrome) are cleft palate and small jaw (Davis and Kalousek, 1988). The pregnancies often have excessive amniotic fluid (polyhydramnios) because the fetus does not swallow (Moessinger, 1983). The intestines of the fetus need stimulation by the swallowed amniotic fluid in order to mature. The lack of swallowing interferes with development of the ability of the intestines to function. This leads to failure to thrive and malabsorption in the newborn.

Diagnostic Approach

As mentioned previously, the most practical approach to define a specific type of arthrogryposis is to establish what part(s) of the body is involved. Using this method, arthrogryposis can be divided into the three main groups: (1) disorders where mainly the limbs are affected, and (2) disorders where there are affected limbs and other abnormalities, and (3) disorders where there are affected limbs with central nervous system dysfunction.

To distinguish between different types of arthrogryposis several things need to be done. The family history is essential, especially in regard to consanguinity (marriages between cousins or close relatives), previously affected children, and paternal and maternal age. A careful prenatal history should include exposure to teratogens (drugs, alcohol, medications that may cause birth defects), and maternal illness or fever, and fetal movement must be documented. The birth history should include time and length of the delivery and perinatal outcome. The newborn examination is the most important part of the study of a patient with arthrogryposis. Documentation of the exact position and range of motion of the contracture(s), as well as any other abnormality, is crucial. Photographs of an individual born with arthrogryposis *must* be taken, and should be considered to be a laboratory test essential for accurate diagnosis, prognosis, and management of arthrogryposis (Hall, 1981).

Family History

a. Other affected family members
b. Marked variability of contractures within family
c. Natural history in other affected family members, i.e., degenerate versus improve with time
d. Increased incidence of congenital contractures in second and third degree relatives
e. Hyperextensibility or hypotonia present in other family members
f. Rule out myotonic dystrophy, myasthenia gravis in parents, particularly mother
g. Consanguinity
h. Advanced parental age
i. Increased previous stillbirths or miscarriages

Fig. 1.42 Table of family history.

Pregnancy History

a. Maternal illness, chronic or acute diabetes, myasthenia gravis, myotonic dystrophy
b. Infections, e.g., rubella, rubeola, coxsackie virus, enterovirus, Akabane virus
c. Fever above 39°C (determine timing in gestation)
d. Nausea, e.g., viral illness or encephalitis
e. Drugs, e.g., curare, robaxin, alcohol, dilantin, addictive drugs
f. Unusual fetal movement, e.g., polyhydramnios, fetal kicking in one place, rolling decreased
g. Oligohydramnios, amniotic leakage
h. Uterine anomaly
i. Trauma during pregnancy, e.g., blow to the abdomen, attempted termination
j. Other complications during pregnancy, e.g., bleeding, abnormal lie, threatened abortion

Fig. 1.43 Table of pregnancy history.

Delivery History

a. Unusual presentation, e.g., breech, transverse
b. Length of gestation
c. Initiation of labor
d. Intrauterine mass, e.g., twin, fibroid
e. Abnormal uterine structure or shape
f. Abnormal placenta or membranes
g. Time of year
h. Geographical location
i. Multiple birth, or evidence of a twin

Fig. 1.44 Table of delivery history.

Evaluation of a Child with Multiple Congenital Contractures

Family History

An extensive family history is a crucial part of the evaluation of a child with arthrogryposis (Fig. 1.42). The physician evaluating the child will ask all the appropriate questions. However, a parent may want to clearly point out any consanguinity (marriages between cousins or close family members) in the family or if there are any other family members with contractures and whether their contractures are similar or different, since there may be marked variability within families with arthrogryposis. It is possible that some relatives have a milder form of the same disorder. This will become very important when trying to establish the inheritance and recurrence risk of the disorder.

Pregnancy History

It is important to remember that anything that leads to decreased movement in utero may lead to contractures in the fetus, so any information or suspicions a mother may have regarding this would be useful for the physician (Fig. 1.43). Any unusual fetal movements, such as movement in only one place, rolling movement, and decreased movement, will be helpful in establishing the position of the fetus in utero or in providing a clue to what led to the contractures. Any trauma or injury as well as surgical procedures or accident during pregnancy must also be recorded. Infections or suspicions of a probable infection during pregnancy, such as persistent nausea, must be noted. Amniotic fluid leakage may cause space restriction. Rupture of the amniotic sac may be associated with amniotic bands. Any drug or medications taken during pregnancy must be carefully documented (curare, methocarbamol, and alcohol, for example, are known to affect fetal movement and may lead to contractures). It is important to make a careful reconstruction of the timing of these unusual events during the pregnancy.

Delivery

It is important for both the physician and the parent to note the length of the pregnancy (i.e., week of delivery), length of labor, the duration of the delivery, and the position of the child at birth (photographs) (Fig. 1.44). This information may come in handy if the child is evaluated by other doctors. Pictures of the child at birth and pictures of the child at different ages with range of motion of joint may later be important documents that provide very valuable information, as they will allow both parent and physician to evaluate the changes in the contracture(s) of the child.

Newborn Examination

This is an important crucial step that will be conducted entirely by the physician. Photographs, as mentioned previously, must be considered important documents and must be taken at this point by the physician for the medical record. Of course, the parents may wish to take pictures also. This newborn evaluation may provide the best and most useful information to differentiate among different types of arthrogryposis and give an accurate diagnosis.
The newborn examination should include careful evaluation and description of the position of the child at rest, the limbs and joints involved in the contractures and their range of motion, whether the contractures are in flexion or

extension, and the amount of muscle and connective tissue mass of the limbs (Figs. 1.45 through 1.63). Measurements of the limbs are another important part of the newborn examination. Any other abnormalities, such as birth marks, dimples, scoliosis, amniotic bands, webbing, abnormal genitalia, malformations of the nails, eyes, palate, or skull, as well as characteristic facial features, should be noted.

There should also be documentation of the neurologic status of the child. Strength, receptiveness, and presence or absence of reflex are important in assessing the possibility of central nervous system involvement. The neurologic examination may be difficult at birth, and evaluation and response to therapy during the first 2 years are important.

Description of Contractures

a. Which limbs and joints
b. Proximal versus distal
c. Flexion versus extension
d. Amount of limitation (fixed versus passive versus active movement)
e. Characteristic position at rest
f. Severity (firm versus some give)
g. Complete fusion or ankylosis versus soft tissue contrature
h. PHOTOGRAPHS!!!

Fig.1.45 Table of description of contractures.

Fig. 1.47 Newborn examination.
This infant is weak and hypotonic as well as having contractures.

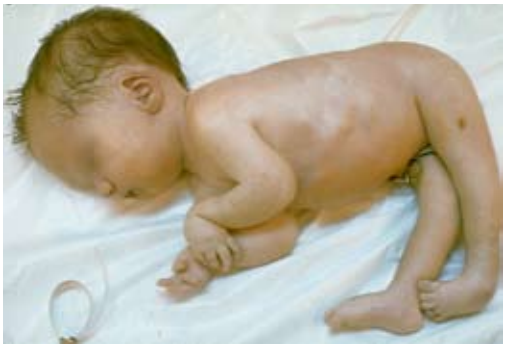


Fig. 1.46 Limb position in newborn.
The exact position of contractures is important to describe, since it helps to identify the specific type of arthrogryposis. Photographs are an important part of the record.

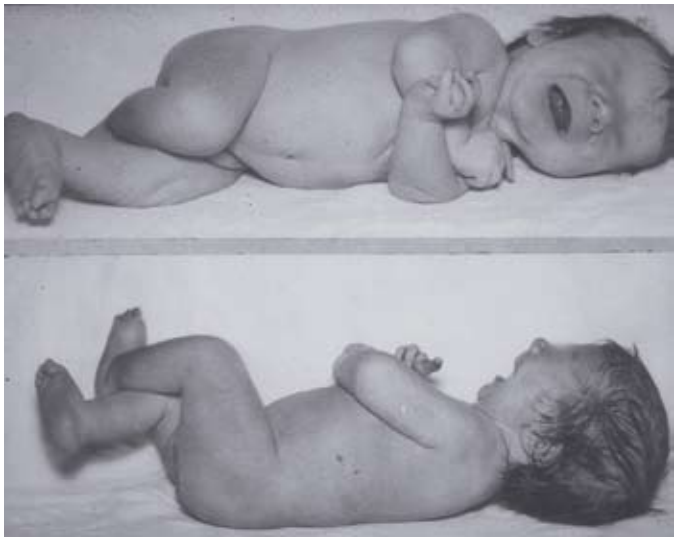


Fig. 1.48 Newborn examination.
In addition to the position of the contractures, asymmetry and other anomalies should be described. This infant has asymmetry of the face. Only one side moves with crying.

Deformations	Malformations	Other Features
Genitalia (cryptorchid, lack of labia, microphallus)	**Eyes** (small, corneal opacities, malformed, ptosis, strabismus)	**Neurologic examination**
Limbs (pterygium, shortening, webs, cord wrapping, absent patella, dislocated radial head, dimples)	**CNS** (structural malformation, seizures, mental retardation)	Vigorous vs. lethargic
Jaw (micrognathia, trismus)	**Palate** (high, cleft, submucous)	Deep tendon reflexes (present vs. absent, slow vs. fast)
Facies (asymmetry, flat bridge of nose, hemangioma)	**Limb** (deletion anomalies, radioulnar synostosis)	Sensory (intact or not)
Scoliosis	**GU** (structural anomalies of kidneys, ureters, and bladder	**Muscle**
Dermatoglyphics (absent, distorted, crease abnormalities)	**Skull** (craniosynostosis, asymmetry, microcephaly)	Mass (normal vs. decreased)
Hernias (inguinal, umbilical)	**Heart** (congenital anomalies versus cardiomyopathy)	Texture (soft vs. firm)
Other features of fetal akinesia sequence	**Lungs** (hypoplasia versus weak muscles or hypoplastic diaphragm)	Fibrous bands
intrauterine growth retardation	**Tracheal and laryngeal clefts and stenosis**	Normal tendon attachments or not
pulmonary hypoplasia	**Vascular** (changes in vascular struture, hemangiomas, cutis marmorata, blue cold distal limbs)	Change with time
functional short gut with feeding problems	**Visceral** anomalies	**Connective tissue**
craniofacial anomalies (hypertelorism, cleft palate, depressed tip of nose, high bridge of nose)		Skin (soft, doughy, thick, extensible)
		Subcutaneous (decreased fat, increased fat)
		Hernias (inguinal, umbilical, diaphragmatic)
		Joints (thickness, symphalangism)
		Tendon attachment, length

Fig. 1.49 Table of other anomalies to watch for in arthrogryposis.

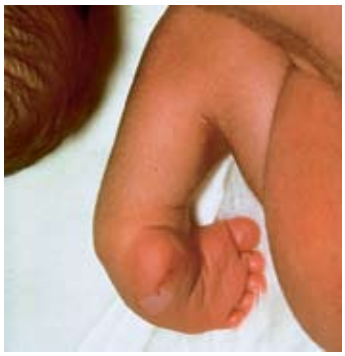


Fig. 1.50 Severe equinovarus deformity of the foot.
Deep creases are present at the hip and ankle. The usual creases are not present on the sole of the foot.

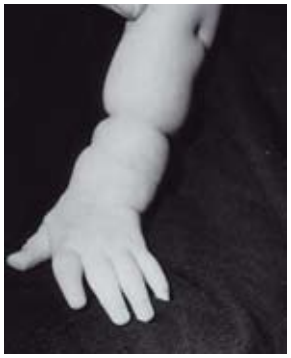


Fig. 1.51 Dimples and bands.
Deep grooves, tight bands, and dimples are often seen on the limbs in arthrogryposis.

Fig. 1.52 Finger position.
When contractures of the fingers are present, the position may help to identify the specific condition. When the fifth finger is cocked up high, it is a poor prognostic sign.

Fig. 1.53 Syndactyly and smudged digits.
Decreased growth of fingers or toes or webbing (syndactyly) is often seen in arthrogryposis.

Fig. 1.54 Webbing or syndactyly.
Lack of complete growth or failure to completely separate digits is often seen.

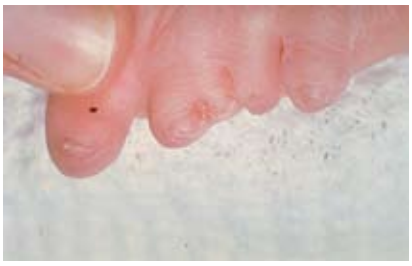


Fig. 1.55 Loss of the end of the digit.
Loss of the nail or underdevelopment of the end of a digit is a frequent finding in both fingers and toes.

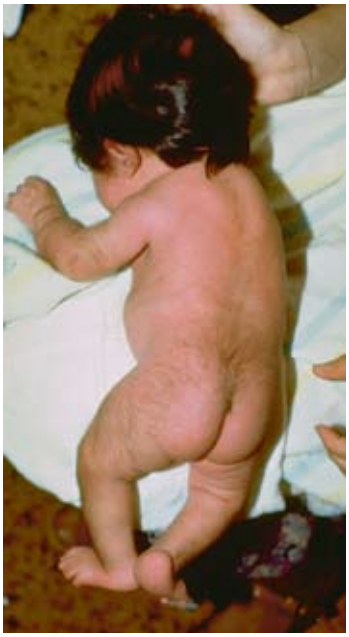


Fig. 1.56 Hirsutism.
Extra hair, or long dark hair, is often seen in areas where activity has been low. This baby with arthrogryposis has extra hair all over his back.

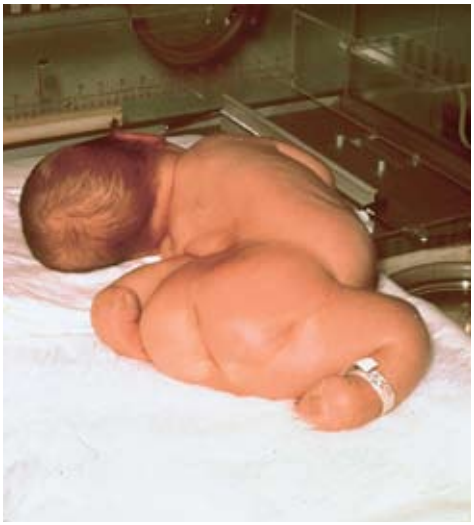


Fig. 1.57 Dimples.
Deep dimples (where skin connects to underlying tissue) are often seen over joints in arthrogryposis. They suggest decreased movement in fetal life.

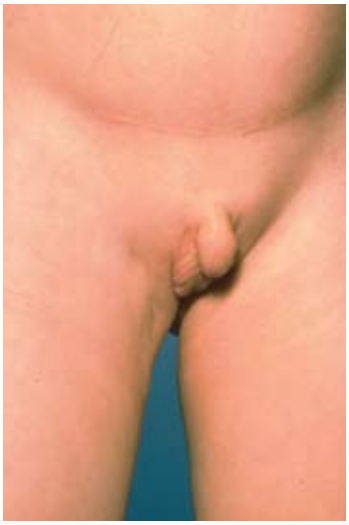


Fig. 1.58 Cryptorchidism.
Undescended testicles are a common finding in boys with arthrogryposis.

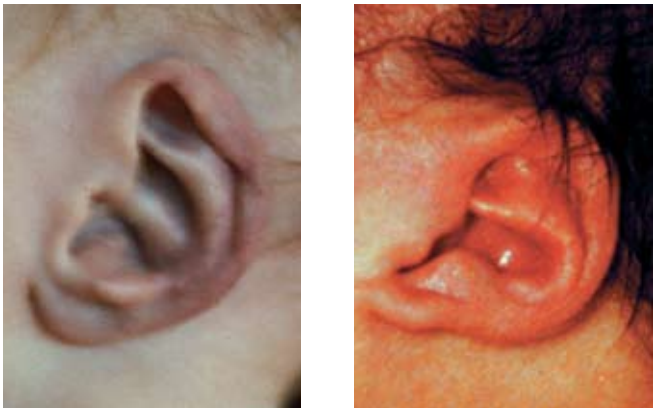


Fig. 1.59 Ear shape and folding.
It is important to describe any unusual features seen at the ears. Often, they are unusual in shape or folded in an unusual way. Sometimes they stand out from the head.

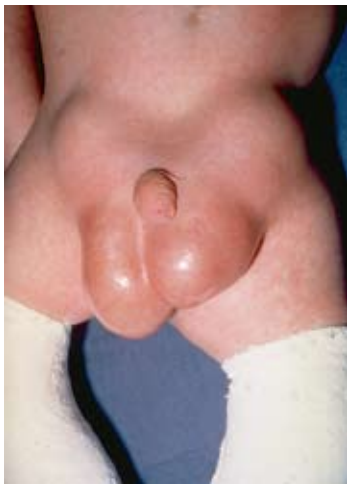
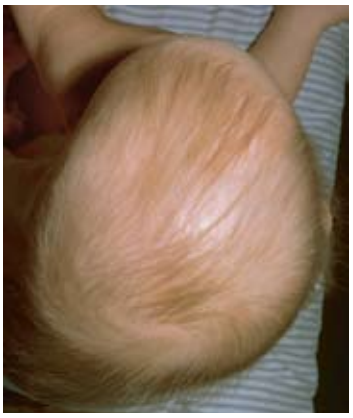


Fig. 1.60 Hernias and hydroceles.
Both hernias and hydroceles (extra fluid around the testicle) are seen with increased frequency in arthrogryposis.



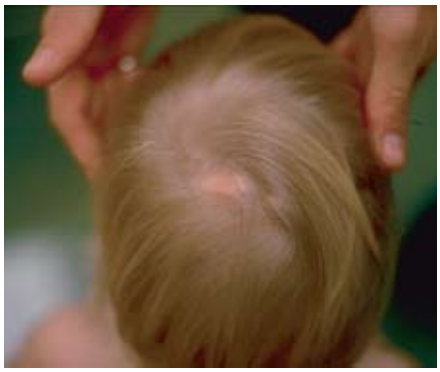


Fig. 1.61 Shape of head and defects in scalp or hair pattern.
Scalp defects imply a vascular accident. Abnormal hair patterns imply unusual growth of the brain. Unusual head shape is often a deformation because of unusual muscle pull or prolonged position of the head in one place.

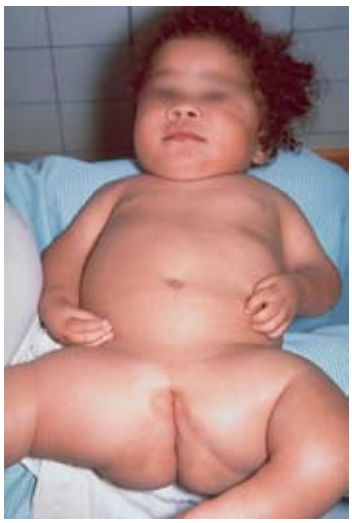


Fig. 1.62 Undeveloped labia.
Girls with arthrogryposis who have widely open hips often fail to develop normal labia. However, internal female organs are usually normal.

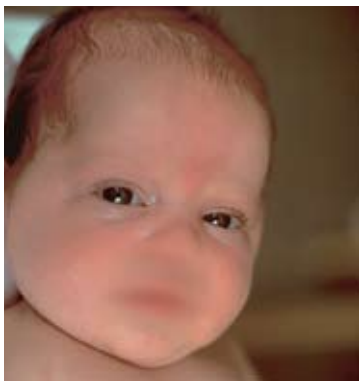


Fig. 1.63 Facial features.
Midline facial hemangiomas or birthmarks are frequent, particularly in amyoplasia. Droopy eyelid (ptosis) is seen in some types of arthrgryposis. It can be asymmetric, as in this child.

Occurrence of Arthrogryposis

Type	Occurrence
All live births	1 in 3000
Amyoplasia	1/3
CNS/newborn lethal	1/3
Hetergeneous groups of disorders	1/3

Fig. 1.64 Occurrence risk for arthrogryposis.

Gene Mapping

Name	Chromosome
Contractural arachnodactyly	5q23-31
Diastrophic dysplasia	5q31
Distal arthrogryposis type I	9q21.2
Lethal spinal muscular atrophy	5q13.3
Lethal X-linked arthrogryposis	Xp11.3-q11.2
Ophthalmoplegia	12p11.2-q12
Symphalangism	9q

Fig. 1.65 Mapping of the genes for various types of arthrogryposis.

Fig. 1.66 Family in which the mother and one child have distal arthrogryposis type I.

Genetics of Arthrogryposis

To address the inheritance of particular forms of arthrogryposis, it is important to make a specific diagnosis if this is possible (Fig. 1.64). If a specific diagnosis is made, the mode of inheritance can be established (autosomal dominant, autosomal recessive, X-linked, or sporadic), and the parents can be counseled with a specific recurrence risk. If a specific diagnosis cannot be made, the parents may be counseled with an empiric recurrence risk usually in the range of 5%.

There are a number of ways in which different types of arthrogryposis can be inherited (Fig. 1.65). Sometimes, the disorder is caused by a single gene, that is, a single abnormality in the genetic information the parent passed on to the child. In this case, the disorders can fall into any of three groups: autosomal dominant, autosomal recessive, or X-linked. Mitochondrial inheritance and maternal effects are also seen.

The risk for having another affected child in a couple in which one parent is affected with an autosomal dominant type of arthrogryposis is 50%. This means that future pregnancies are at a 50% risk of being affected (Fig. 1.66). If the disorder is an autosomal recessive disorder, the parents are carriers of the gene for that specific disorder and the couple's next pregnancy is at 25% risk of being affected. Rarely, there are disorders where there may be a much higher risk of recurrence, such as mitochondrial disorders.

Specific types of arthrogryposis may be caused by the presence of chromosomal abnormalities. This means that there may be a piece of genetic information (piece of a chromosome or DNA) that is missing (deletion) or in excess (duplication). It may be that all the chromosomes are present but they have rearranged; this is called a translocation. Most of the chromosomal abnormalities seen in arthrogryposis appear for the first time in the affected child (de novo), but they may also be inherited from a carrier parent. Chromosomal abnormalities are particularly important in cases of multiple congenital contractures associated with mental retardation. If the blood chromosomes are normal, it may be necessary to do chromosome studies in skin in order to find a specific abnormality.

Finally, there are the sporadic types of arthrogryposis, such as amyoplasia, and the nongenetic or environmental cases in which there is a known environmental cause or event leading to the congenital contractures (Fig. 1.67). Individuals born to mothers with diabetes mellitus, multiple sclerosis, or myasthenia gravis, those born with contractures associated with teratogens that interfere with the development of the limbs, or individuals born from a multiple birth pregnancy that led to compression of one twin are all examples of nongenetic or environmental causes of arthrogryposis. Some of these may have a risk of recurrence.

Prenatal Diagnosis

Once the specific diagnosis and the form of inheritance of a disorder have been established, it may be possible to offer prenatal diagnosis to those parents with a higher risk of having another affected child. Most parents with a previously affected child with arthrogryposis will want reassurance in future pregnancies. The most useful tool for prenatal diagnosis of arthrogryposis is ultrasound. If a couple is known to be at risk or if the mother notices a decrease in the fetal movement, a real-time ultrasound may be performed at 16, 20, and 24 weeks of pregnancy and then again prior to birth (Hall, 1985; Hogge et al., 1985; Bendon et al., 1987). These studies can give not only diagnostic information but also information to help manage the pregnancy.

Mainly Limbs

Absence of dermal ridges
Absence of DIP creases
Amyoplasia
Antecubital webbing
Camptodactyly
Coalitions
Congenital clasped thumbs
Contractural arachnodactyly
Distal arthrogryposis type I
Guadalajara camptodactyly
Humeroradial synostosis
Impaired pronation/supination of the forearm (familial)
Liebenberg syndrome
Nievergelt-Pearlman
Poland anomaly
Radioulnar synostosis
Symphalangism
Symphalangism/brachydactyly
Tel-Hashomer camptodactyly
Trismus pesudocamptodactyly
X-linked resolving

Limbs and Other Body Areas

Antley Bixler syndrome
Camptomelic dysplasia
Chondrodysplasia punctata
Diastrophic dysplasia
Distal arthrogryposis type IIB
Distal arthrogryposis type IIE
Freeman-Sheldon (whistling face) syndrome
G syndrome
Gordon syndrome (distal IIA)
Hand muscle wasting and sensorineural deafness
Holt-Oram syndrome
Kuskowin syndrome
Larsen dysplasia
Leprechaunism
Megalocornea with multiple skeletal anomalies
Metaphyseal dysplasia
Metatropic dysplasia
Moebius syndrome
Moore-Federman syndrome
Multiple pterygium syndrome(s) (See Figure 1.18)
Multiple synostosis syndrome
Myopathy
  Central core
  Congenital fiber disproportion
  Nemaline
Nail-patella syndrome
Neurofibromatosis
Oculodentodigital syndrome
Ophthalmomandibulomelic dysplasia
Orocraniodigital syndrome
Osteogenesis imperfecta II
Otopalatodigital syndrome
Pffeifer syndrome
Popliteal pterygium syndrome(s)
Robert syndrome
Puretic syndrome
Sacral agenesis
Schwartz-Jampel syndrome
SED congenita
Sturge-Weber syndrome
Tuberous sclerosis
VATER association
Weaver syndrome
Winchester syndrome

Limbs and CN, Possibly Lethal

Adducted thumbs
Arthrogryposis with liver and kidney
Bowen-Conradi syndrome
C syndrome
Cerebrooculofacioskeletal (COFS) syndrome
Cloudy cornea, diaphragmatic defects, distal limb deformities
Craniofacial/brain anomalies (intrauterine growth retardation, IUGR)
Cryptorchidism, chest deformity, contractures
Faciocardiomelic syndrome
Fetal alcohol syndrome
FG syndrome
Lethal multiple pterygium
Maternal multiple sclerosis
Maternal autoantibodies
Marden-Walker syndrome
Meckel syndrome
Meningomyelocele
Mietens syndrome
Miller-Diecker syndrome
Myotonic dystrophy (congenital)
Myasthenia gravis (congenital)
Neu Laxova syndrome
Pena-Shokeir syndrome
Popliteal pterygium with facial clefts
Pseudotrisomy 18
Restrictive dermopathy
Spinal muscular atrophy
Toriello-Bauserman syndrome
X-linked lethal arthrogryposis
Zellweger syndrome
Chromosomal abnormalities
  46XXY/48XXXY
  49XXXXX
  49XXXXY
  4p trisomy
  Trisomy 8
  Trisomy 8 mosaicism
  Trisomy 9
  Trisomy 9q
  Trisomy 10q
  Trisomy 13
  Partial trisomy 14
  Trisomy 15
  Trisomy 18
  Trisomy 21
  Turner syndrome

Fig. 1.67 Disorders with multiple congenital contractures.

Regular examination
Special examination
- Nervous system:
  - Brain: Structural anomalies
  - Spinal cord: Multiple levels
    - Number, type of anterior horn cells
  - Myelinization
  - Peripheral nerves
  - Neuromuscular connections
- Muscles: Multiple sites
  - Affected and unaffected
  - Voluntary, smooth, cardiac, ophthalmic
  - EM and special stains
  - Mitochondrial studies
- Eye: Nerve connections
  - Muscle structure
- Joints: Thickened capsule
  - Muscle attachments, tendons
  - Secondary deformities of bone or joint

Careful evaluation of any other organ anomalies
Photograph, preserve unusual tissue, consider special studies such as DNA, chromosomes

Fig. 1.68 Special aspects of autopsy.

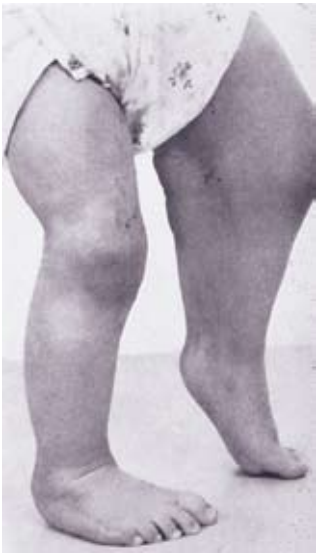


Fig. 1.69 Pterygium has abnormal anatomic structures.
Webs, such as this pterygium, seen in some cases of arthrogryposis may have the blood vessel and nerve running along the edge of the web. Obviously, care must be given not to destroy such important structures.

Lack of movement of the fetus in utero plays an important role in the formation of contractures. This has led to the possibility of in utero physical therapy. If contractures are seen on ultrasound or if the pregnancy is known to be at risk for multiple congenital contractures, the mother is encouraged to do some exercise. This has been shown to increase the movement of the child in utero. Other types of medication may be considered.

After birth, physical therapy is used to improve muscle strength and range of motion of the joint. Casting and splinting are used to improve foot position and range of motion. Surgery can be used as a supportive measure after other forms of treatment have achieved their maximum results.

Autopsy

Doing an autopsy in lethal cases of multiple congenital contractures is important, since it may give a definitive diagnosis and lead to a better understanding of why the death occurred. Just as in the newborn evaluation, an autopsy should document the location, position, and situation (flexion or extension) of the contractures. Photographs should be taken. The central nervous system, spinal cord, and peripheral nerves must be carefully examined, as well as muscle attachment and muscle pathology. A careful and accurate autopsy will provide a definitive diagnosis that will be useful when giving a recurrence risk.

Although we would wish that no children with arthrogryposis died, approximately 20-30% do die during the first year. These are usually the severely affected children, and it usually is obvious in the first few months that survival will be difficult. When an individual with arthrogryposis dies, an autopsy will help to understand why. An autopsy may also help to determine whether the congenital contractures have a genetic basis and could recur in other children in the future. Sometimes, unexpected results at the autopsy demonstrate some preventable cause or complication that can help in another affected individual or family member.

An autopsy involves careful examination after death by a trained pathologist. Areas that deserve particular attention are listed in Figure 1.68. The family of the deceased individual will expect to receive a full report and explanation of the findings. However, certain parts of the examination, particularly nerve tissues, will take several months to analyze properly.

Sometimes when a child with arthrogryposis dies, a family may feel the child had already suffered and they do not wish it to suffer further. Of course, an autopsy does not cause suffering in the person who has died. Sometimes for religious or personal beliefs, a family does not wish to have an autopsy, and this should be respected. However, very frequently at some much later time, information garnered from the autopsy will help in family planning, treatment of other affected individuals, or prevention of complications.

Preventable Complications

Over the years, a great deal has been learned about how to treat various types of arthrogryposis and various complications seen in arthrogryposis. Some of these are covered elsewhere in this book, but this list will serve as a summary of preventable complications.

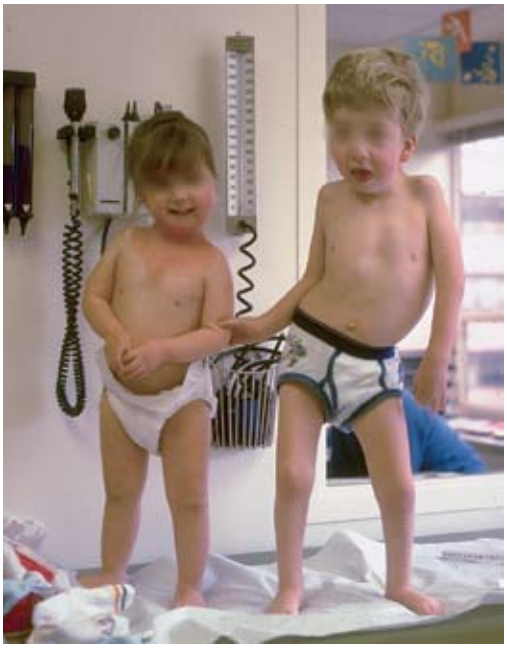


Fig. 1.70 Arthrogryposis and malignant hyperthermia.
A brother and sister who have this combination. Several other families have been reported.

Fig. 1.71 Kyphoscoliosis in a boy with generalized contractures whose back curve has progressed.

Surgery

The normal position of muscles, tendons, blood vessels, and nerves may be altered in individuals with arthrogryposis (Fig. 1.69). These alterations may be part of the deforming process of the contractures or part of the underlying disorder. However, they are common.

Anesthesia

There are major concerns when an individual with arthrogryposis needs anesthesia for surgery or manipulations.

Hypotonia Associated with Cervical Vertebrae Instability

Many children with arthrogryposis have decreased muscle mass and weakness. Occasionally, they also have underdevelopment of the first and second cervical vertebrae such that these two vertebrae may slip on each other, compromising or even damaging the spinal cord. When such an individual is put to sleep with anethesia, damage can be done to the spinal cord. It is possible to evaluate for this type of slippage before surgery to avoid such a complication.

Multiple Congenital Contractures with Malignant Hyperthermia

Multiple congenital contractures and malignant hyperthermia have been described in families with osteogenesis imperfecta (breakable bones) and muscular dystrophy (degeneration of muscle). Malignant hyperthermia is a condition in which there is an abnormal response to anesthesia, leading to high fever that may cause severe damage and in some cases lead to death. Individuals with congenital contractures and malignant hyperthermia may also have cleft palate, torticollis (stiff neck), progressive scoliosis (curvature of the spine), and low serum CPK (blood chemistry) (Fig. 1.70). It is important to be aware of this possible complication of surgery and anesthesia, since it can be treated and prevented.

Aspiration During or After Surgery

Because children with arthrogryposis do not have normal movement and often do not have normal respiratory muscles, they are more prone to aspirate fluids during and after surgery. Attention to this possible complication should help to avoid it.

Scoliosis and Kyphosis

Because of weak trunk muscles and occasionally vertebral structural anomalies, abnormal spinal curves are seen in about one-third of all individuals with arthrogryposis (Fig. 1.71). These curves can be progressive and lead to compromise of respiration and visceral function. It is important to treat them early and aggressively so as to avoid secondary complications due to compromised respiration.

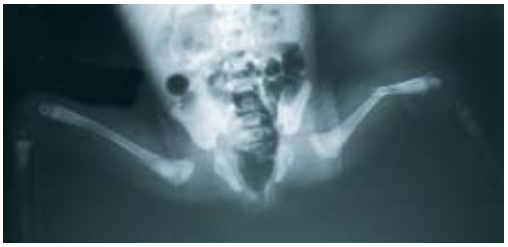


Fig. 1.72 Broken femur in newborn.
This bone was broken at birth during a difficult delivery.

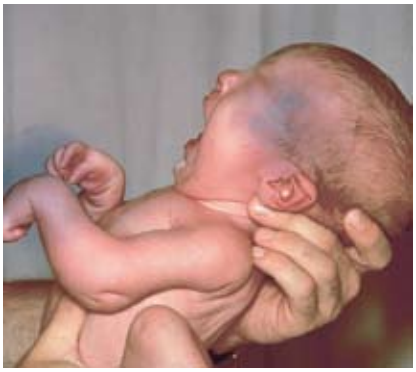


Fig. 1.73 Bruises from difficult birth.
This baby with severe amyoplasia had bruises of the head and face related to a very difficult delivery.

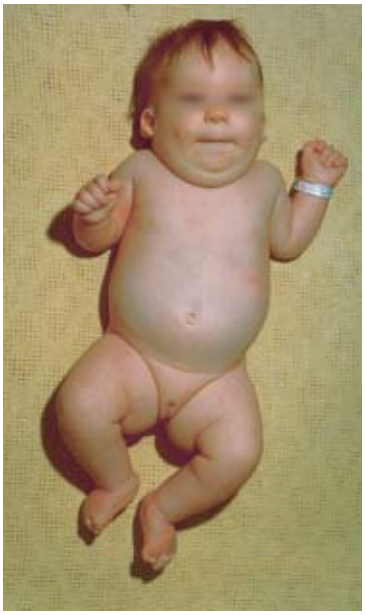


Fig. 1.74 Infant with excess fat.
Individuals with arthrogryposis have a risk of becoming overweight for their age and size because they get less exercise and usually have less muscle bulk.

Arthritis

Wear and tear arthritis seems to develop with aging in joints that have been affected by congenital contractures. It is not clear whether this is related to the presence of an abnormal surface to the joint (which may develop because of lack of movement during fetal life) or because of the vigorous physical therapy needed to mobilize joints with contractures. Whichever the case, adults with arthrogryposis seem to have an increased occurrence of wear and tear or degenerative arthritis. This can be treated like any other degenerative arthritis.

Fractures

Many infants with arthrogryposis are born with fractures of long bones (Fig. 1.72). This seems to be related to the abnormal position of the limbs, which are stiff and make birth difficult. The bones that have not been used seem to have less calcium deposited and to be long and thin (gracile). This makes them more likely to fracture (break) with less trauma or pressure. The bones heal normally and should be treated like other broken bones. However, care should be taken in handling newborns and doing physical therapy in those who have thin bones (osteoporosis).

Difficult Delivery

The process of being born may be difficult (Fig. 1.73). If the presence of arthrogryposis is known before birth, ceasarean section should be considered. Many babies end up with bruises and broken bones. The contractures are not due to the difficult birth, since it usually takes at least 3 weeks of lack of movement by the fetus to develop contractures. However, if the birth process is very difficult, central nervous system damage can occur. This is surprisingly rare.

Obesity

Obesity is seen with arthrogryposis because infants, children, and adults eat too much (Fig. 1.74). This overfeeding is often at the urging of well-meaning, concerned relatives and health professionals. Affected individuals usually weigh less than normal for age or height because they have less muscle mass. They may also appear thin because of lack of muscle bulk on their limbs. However, the excess fat and weight make it more difficult to move the limbs because there is often little strength and the extra weight of the fat tissue requires extra strength. Not uncommonly, there are feeding difficulties in early infancy, so at first weight gain is considered a success. Obesity should be avoided if possible, and skin thickness rather than limb size should be used to judge proper weight.

Growth Curves

Very little information is available for the common types of arthrogryposis on the expected height and weight. However, most adults will end up 4-8 inches shorter than their families and weigh 10-20 pounds less than other people their age (Hall, 1985b) (Fig. 1.75).

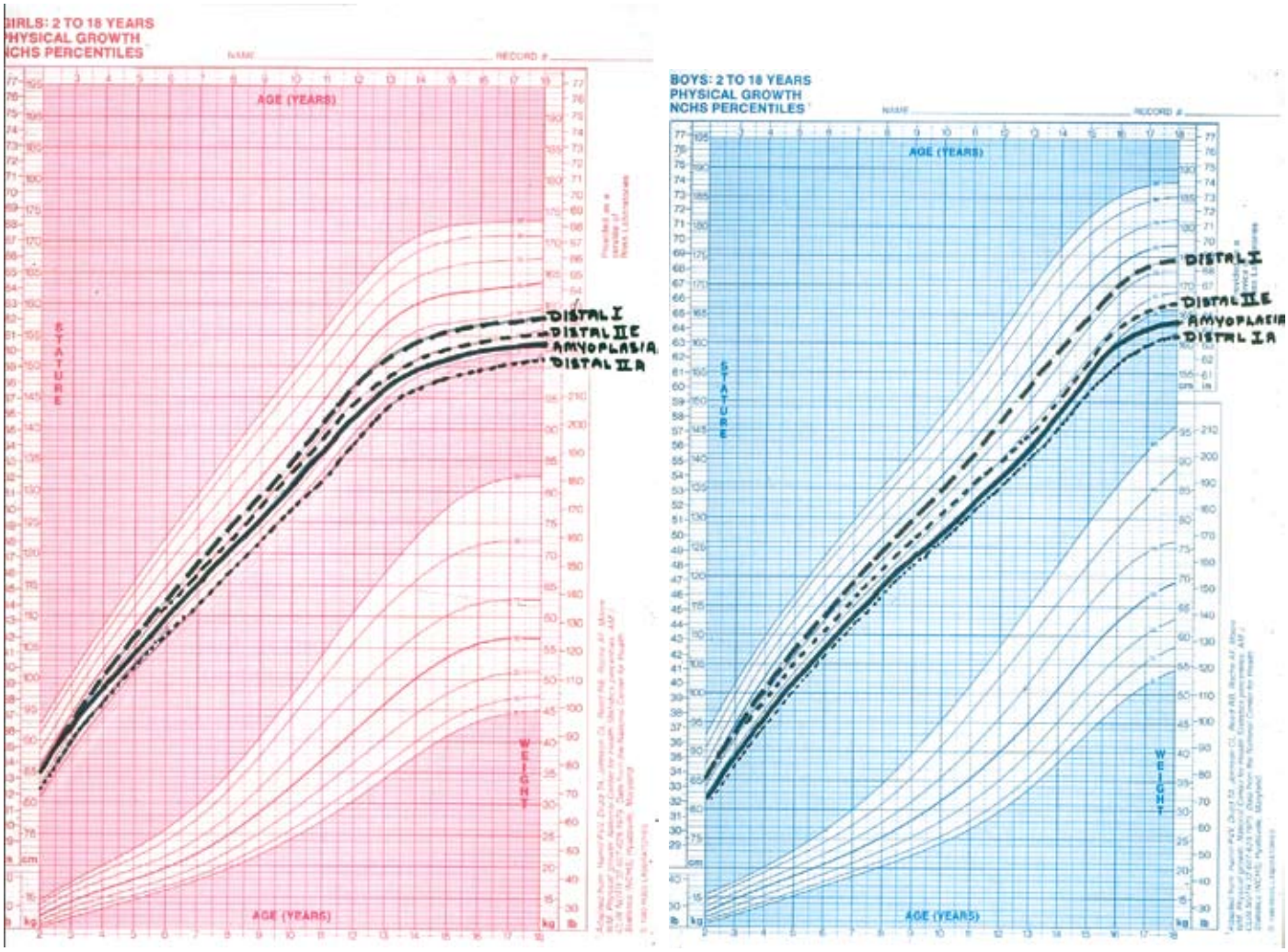


Fig. 1.75 Growth curves in amyoplasia.
(Left) Amyoplasia growth curves for girls. (Right) Amyoplasia growth curves for boys.

Chapter Contents **Page**

L.T. Staheli, M.D.

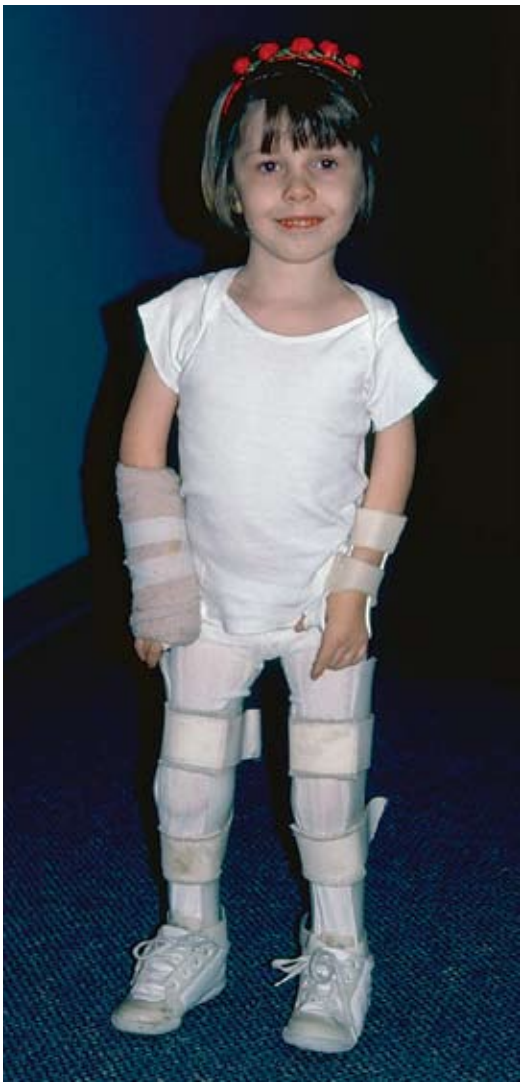


Fig. 2.1 Happy childhood.
Most children with amyoplasia can expect to have a happy childhood.

Introduction

In this chapter we direct our attention to the management of the musculoskeletal deformities present in arthrogryposis. This chapter deals with the principles that are the foundations on which specific treatment is based. These general principles usually apply to all parts of the body when dealing with musculoskeletal problems. There are, however, some unique differences in the way deformity is managed in different regions of the body. For example, fine motor function is the major objective of upper extremity management. In contrast, in the lower limbs, our primary objectives are symmetry and stability.

Although our primary focus is on amyoplasia, these basic principles usually apply to all forms of congenital contractures. Other types of arthrogryposis that commonly require orthopedic treatment are briefly covered at the end of this chapter.

The primary objective of orthopedic management is to improve function by correcting deformity. Secondary objectives include improvement in appearance, facilitating care and control of discomfort, and reducing the risk of pain in adult life. Plan management of the child is based on a lifetime perspective.

Our goal is to help each child reach its potential. Approach treatment with optimism, as most children with amyoplasia have the potential of living a satisfying and productive life (Fig. 2.1). Unlike many children with other neuromuscular disorders, the amyoplastic child looks most deformed at birth. Time is the infant's friend. With advancing age, deformities improve

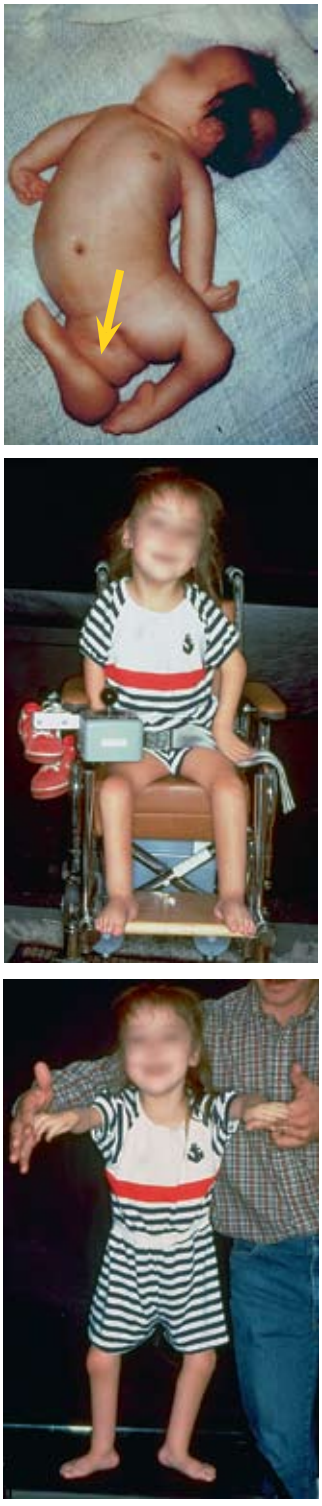


Fig. 2.2 Effectiveness of time and treatment.
This infant with amyoplasia had severe deformities at birth, with a leg rotated 180° (arrow), clubfeet, and knee contractures. By age 7, she was independent. She is independently mobile in an electric wheelchair and can stand with assistance and transfer easily. She is an outstanding student.

and the child's healthy and happy personality emerges (Fig. 2.2), making management of these children a gratifying experience. Function in these children can be significantly improved by treatment. As these children possess normal intelligence, sensation, and perceptive mechanisms, their potential is only mildly limited (Fig. 2.3).

Develop a realistic management plan. Each treatment has its costs: discomfort; interference with play, socialization, and schooling; risks to health; and stress and disruption for the family. Poorly planned, overzealous treatment may provide little benefit to the child, exhaust the family, and deprive the individual of childhood experiences. Planning must be thorough and focused and use proved, effective treatment methods.

This book was written with the belief that childhood has intrinsic value; it is not merely a period of preparation for adult life. Childhood should be valued and savored. Normal childhood experiences are an important foundation for a healthy adult life. The arthrogrypotic child's needs are the same as those of other children. Plan management that interferes least with childhood and integrate the management plan. Tailor the plan for the specific needs of the child and family. Avoid overemphasizing any one method of management to the exclusion of the rest. Work as a team, employing management that is most effective and efficient. The approach to management varies from one center to another. Be concerned if the recommended management becomes too focused on only one type of treatment.

We have found that an integrated management program involves all modalities of treatment. In general, deformity is corrected by casting and surgery, recurrent deformity is best prevented by night splinting, and function and independence are enhanced by physical and occupational therapy.

Fig. 2.3 Productive, independent adult life.
This man with amyoplasia is independent, has a successful business, and lives a nearly normal life.

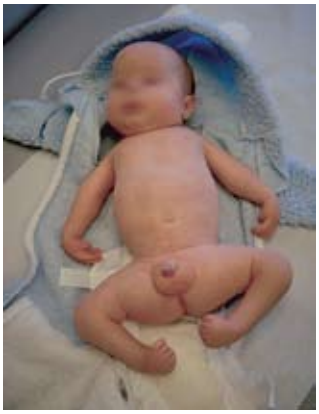


Fig. 2.4 Primary deformity.
These congenital contractures occurred early in intrauterine life, producing rigid deformity that requires operative correction.

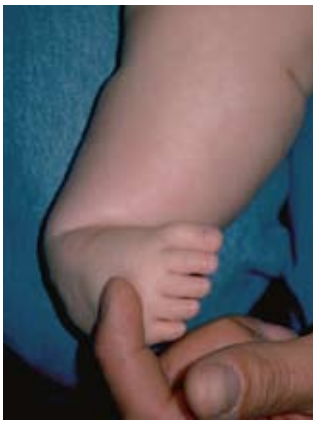


Fig. 2.5 Positional deformity.
This foot is flexible, and the deformity can be nearly corrected with pressure from a finger.

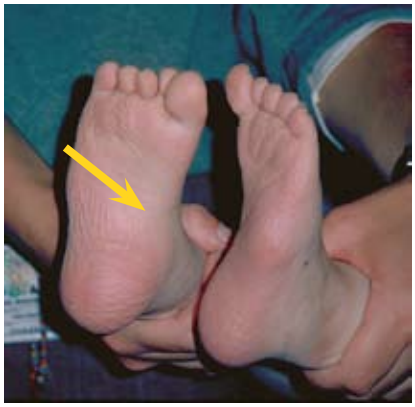


Fig. 2.6 Recurrent deformity.
Recurrence of clubfoot deformity is very common. Often the foot develops a varus deformity with pressure over the base of the fifth metatarsal (arrow).

Types of Deformity

Deformity may be classified into three basic types based on time of onset.

Primary Deformity

During fetal life, pathogens damage nerve (Clarren and Hall 1983; Brown et al., 1980) or muscle cells. This damage causes reduced fetal movement, which in turn causes various deformities. The spectrum of pathologic findings is broad (Banker, 1986). Immobilization provokes a collagenic response (Swinyard, 1982; Swinyard and Bleck, 1985) with increased collagen synthesis (Ianasescu et al., 1970). This in turn causes contractures, deformed articular contour, thinning and shortening of capsules, and fibrosis and hypoplasia of muscle. Fetal akinesia results in a loss of normal skin creases and dimpling over bony prominences. This intrinsic, primary deformity produces stiffness so severe that only surgical correction is effective (Fig. 2.4).

Positional Deformity

Positional deformity occurs late in fetal life secondary to akinesia together with abnormal intrauterine position. These positional deformities are usually mild and tend to improve as the infant moves freely and the joints are gently ranged or stretched (Fig. 2.5) during therapy.

Recurrent Deformity

Unlike positional deformity, primary deformities tend to recur after correction. Recurrent clubfeet (Fig. 2.6) and knee flexion contractures are typical examples. Recurrence occurs more rapidly and is likely to be more rigid if the primary deformity was severe. Recurrence develops most rapidly during the months following correction and during infancy and early childhood but often continues insidiously throughout the remaining growth period.

Prevent recurrent deformity by night splinting. If deformity does develop, correct the recurrence by serial casting. Untreated recurrent deformity becomes more fixed and severe with time, causing secondary cartilage and bony changes. Fixed, rigid recurrent deformity may require operative correction.

Evaluation

The evaluation necessary to establish a diagnosis was discussed in Chapter 1. In this section, the evaluation necessary for management is detailed. In most cases, a thorough musculoskeletal examination provides most of the information necessary to plan treatment.

Screening Examination

In addition to evaluation of specific deformities, a screening examination should be a routine part of the evaluation. Look at the whole child. Perform a forward bending test to assess the spine for scoliosis. This may be performed with the child sitting or standing (Chapter 3). Avoid focusing only on the deformity that is currently the major problem.

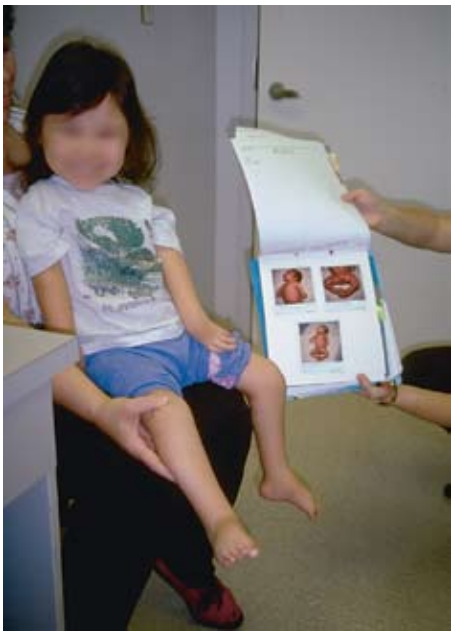


Fig. 2.7 Note the effect of time on deformity.
Make serial measurements or record the degree of deformity by photographs.

Fig. 2.8 Bonding.
Promote close physical contact between the parents and infant.

Sequential Evaluation

Assess the effect of time (Fig. 2.7), growth, and treatment on joint motion. Record sequential measurements. These measurements are most reliable if made by the same person. Positional deformity improves during early infancy. This improvement often plateaus once positional deformity is corrected and the primary deformity is encountered. This primary deformity is more rigid and may require operative correction.

Sequential measurements are especially important following operative correction of deformity. Be concerned if correction is lost. This suggests that the deformity is recurring, and the effectiveness of the splinting program needs to be assessed. If the loss of correction is significant, regain the correction by serial casting.

Imaging

In most infants, a radiograph of the pelvis should be made to assess the status of the hip joint, as this is difficult by physical examination alone. A lateral radiograph of the foot may be necessary to confirm the diagnosis of a vertical talus. Although ultrasound and magnetic resonance imaging (MRI) studies may in the future be useful in assessing muscle status, their value in establishing prognosis and planning management is yet to be determined.

Priorities

Plan management based on age-related priorities. Apply each method of treatment at the age when that treatment is most effective. Avoid overwhelming the child and family with too many different treatments at one time. Usually the order of employing various treatment methods is roughly the same, but the exact timing varies from child to child.

Early Infancy

As bonding between infant and parents occurs during the first months, be certain that treatment does not interfere with this vital process. Be certain that the mother is comfortable holding and playing with the infant even if clubfoot casts have been applied (Fig. 2.8). Emphasize the importance of physical contact between family and infant.

Infancy

Correct most deformities during this period. The positional deformity component will improve with time and gentle ranging of the joints. This is best performed by the family following instructions taught by a therapist. Splint in the best position obtained by gentle stretching. Correct rigid deformity by surgery. Most operative correction is performed during this period.

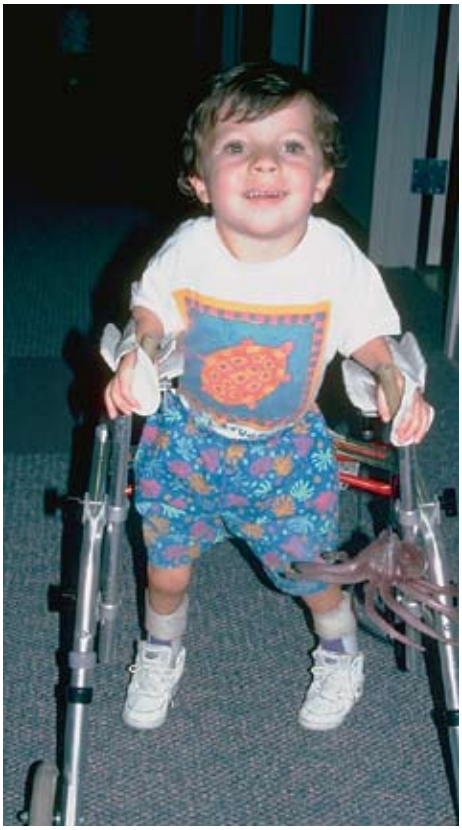


Fig. 2.9 Establish effective mobility.
During early childhood, the child needs time and help to establish an effective method of mobility. Walkers are helpful in making the transition to independent walking. The role of the therapist is especially important during this period.

Fig. 2.10 Importance of family.
The most important factor in the success of child is the health of the family.

Early Childhood

Encourage play and independence during the day and prevent recurrent deformity by splinting during the night. Physical and occupational therapy is most important during this period. Correct upper limb deformities that interfere with function. Provide effective mobility (Fig. 2.9). Most children become walkers between ages 2 and 5 years.

Childhood

The focus is on education and the development of special talents. The parent (Fig. 2.10) and teacher should explore the child's special abilities in art, science, or other areas on which to focus. Minimal intervention by therapists and physicians is best. Usually, walking ability peaks during late childhood.

Adolescence

Focus on education, vocational planning, socialization, independence, and preparation for adult life. Address psychologic problems. Correct the disability that is producing the deformities by surgery. Avoid prolonged periods of immobility, as recovery may be slow or incomplete. Promote good nutrition to minimize any tendency for obesity. This is a period of maintaining walking and mobility skills and preparing for independent living as an adult. Most patients function well, and the degree of independence depends primarily on personality, education, and coping skills rather than physical disability (Carlson et al., 1985).

Nonsurgical Treatment Methods

Optimum treatment requires an integrated plan that employs the most effective and efficient methods of achieving the desired outcome. Select the method of treatment that is both effective and necessary. Focus on correction of deformities that are most disabling. Employ only treatment methods of proven effectiveness.

Fig. 2.11 Cast application.
Allow the parent to comfort the infant while the cast is applied. Hold the limb in the corrected position throughout the period during which the cast is rolled on.

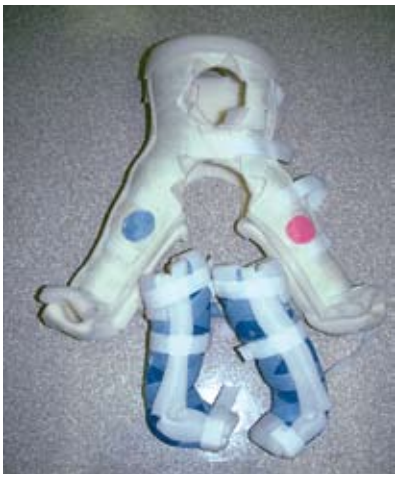


Fig. 2.12 Night splints.
These are typical night splints made of cast material and converted to removable splints.

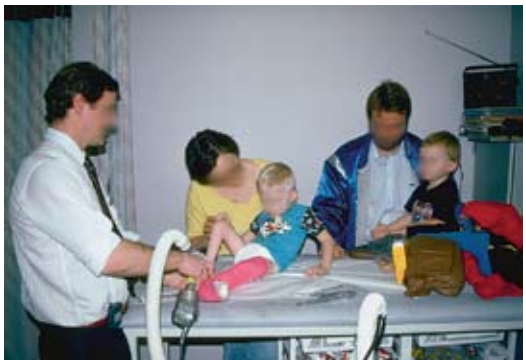


Fig. 2.13 Unthreatening cast applications.
By involving the family, the child's experience in the cast room is much less traumatic.

Cast Correction

Casting has a variety of uses. Apply casts if rapid correction of positional deformity is necessary (Fig. 2.11). Apply serial stretching casts to stretch soft tissues prior to surgical correction of primary deformity. Bivalved casts are useful night splints (Fig. 2.12) to prevent recurrent deformity. Should recurrent deformity occur, correct with serial casts before the deformity becomes fixed. As immobility is an underlying cause of congenital contractures (Smit and Barthm 1980; Jago, 1970; Drachman and Coulombre, 1962), avoid excessive periods of immobilization. The infant needs movement and freedom for optimal development. Use cast treatment prudently and for short periods. Before applying a cast, gently range the joint to achieve maximum correction. Be certain that the cast is well padded and include enough of the limb to be both comfortable and effective.

A common mistake is to apply a short leg cast for correction of a clubfoot deformity. The long leg cast is much more effective in correcting equinus and medial rotation of the foot. Apply the long leg cast segment with the knee flexed, the ankle dorsiflexed, and the foot laterally rotated and positioned in valgus. Extend the cast above the flexed knee. Flexion of the knee stabilizes the upper portion of the cast so effective correction can be applied to the cast applied to the foot. The thigh-foot angle in the cast should be laterally rotated.

Select the cast material based on the situation. Use fiberglass for spica casts, as the material is light in weight and radiolucent. Use fiberglass for making night splints, as the material is strong and light. Use plaster casts for clubfoot correction, as the plaster is more easily molded and the cast may be removed at home. Teach the parents to soak off the plaster cast at home just before they leave for the clinic visit. This avoids the terror from the noise and reduces the risk of skin lacerations from the cast saw. This often makes the clinic quieter and more efficient for the staff and less stressful for the child and family (Fig. 2.13).

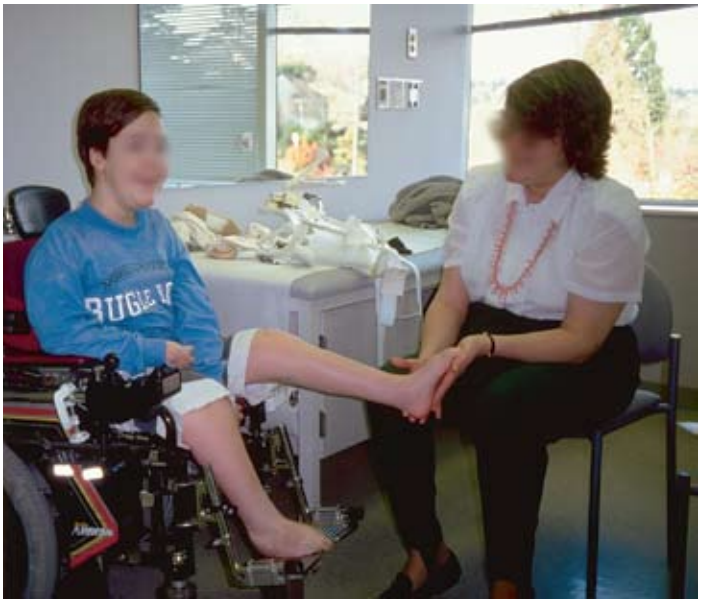


Fig. 2.14 Range of motion exercises.
The family often provides the exercise program. This is effective, convenient, and inexpensive.

Fig. 2.15 Effective mobility options.
These children come together to play by different methods of mobility. The child above rolls across the floor to play with the other children.

Fig. 2.16 Electric powered wheel chair.
This method of mobility allows this severely affected child effective mobility. This is facilitated by a supportive family.

Physical and Occupational Therapy

Physical and occupational therapy are vital components in a well-integrated management plan. The role of the therapist is very broad and includes education, emotional support, monitoring for problems, and traditional methods of treatment.

Range of Motion

Gentle stretching of the joints is useful in overcoming positional deformity. This is of special importance in the upper limb. The stretching should not be painful but should stretch to the edge of the arc of motion and be held in the position of maximum correction for about 10 seconds. This ranging should be continued for 20-30 minutes four times daily by the family (Palmer et al., 1985). The therapist teaches and monitors. Ranging should be a positive experience for both the infant and parent and should never be painful (Fig. 2.14).

Facilitation of Bonding

The therapist can help parents become comfortable holding and playing with the infant. This is especially necessary if clubfoot casts have been applied.

Making Splints

In many centers, splints, especially hand splints, are made by the therapist. New products, such as the silicone rubber material (Bell and Graham, 1995), allow better molding and correction. Splinting should be integrated with the range of motion exercise program.

Monitoring

Monitoring is an important diagnostic role to detect recurrent deformity or other problems in management.

Mobility Training

Providing effective mobility is an essential part of managing the young child. Effective mobility includes rolling, crawling, scooting, and other ways of getting about (Fig. 2.15). As the child becomes proficient at a level of mobility, the next level should be introduced – a standing device, a walker, crutches, all in order. A short trial period with the next level of mobility will usually determine if the child is ready for the next higher level. If the child is not ready, avoid pushing the child into a level that is not effective. This causes only frustration. Allow the child to function at the next lower level that provides an effective means of getting around.

Some children are unable to walk. Provide these children with an electric powered wheelchair (Fig. 2.16). A common misconception is that the early use of a wheelchair will habituate the child to this means of mobility. The child has an innate desire to be as independently mobile as possible and will walk if physically able, regardless of any prior use of a wheelchair or other mobility aid.

Fig. 2.17 Gait training.
Walking may progress slowly and may require bracing and training.

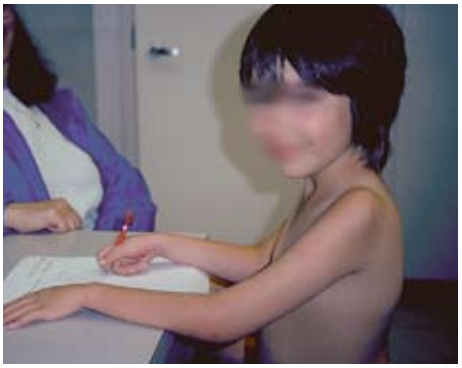


Fig. 2.18 Handwriting skills.
Upper extremity function is limited by deformity but facilitated by good sensation and an intelligent and motivated child.

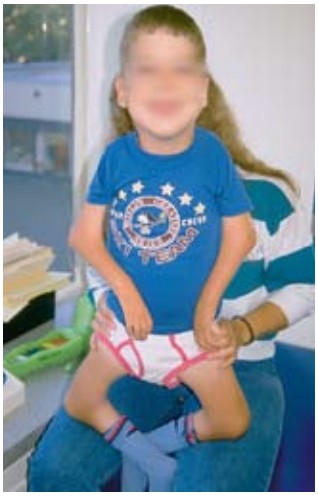


Fig. 2.19 Knee standing.
The ability to knee stand demonstrates good trunk balance and the potential for walking once the knee contractures are corrected.

Walking

Most children with congenital contractures become independent walkers (Gibson and Urs, 1970; Hoffer et al., 1983). Acquisition of walking is often slow (Fig. 2.17), and most children will need braces, a walker, or crutches in the beginning. Integrate the use of training, walking aids, and braces. Tailor management since each child is unique. If the upper extremities are involved, forearm platforms may be required for walkers or other aids.

Self-Care Skills

Self-care skills are usually taught along with mobility training. Optimal upper limb function requires careful assessment and the use of adaptive equipment or special modifications of the child's clothing. In about a third of children with upper extremity involvement, operative correction is useful in improving function. Plan operative correction carefully. Communication and cooperation between the therapist and surgeon are essential.

Communication

Handwriting (Fig. 2.18) and computer skills may be improved by training, adaptive devices, and optimum seating programs.

Home Assessment

It may be helpful to evaluate the home situation to determine what aids can best facilitate the child's independence.

Presurgical Assessment

Preoperative assessment by the therapist to identify problems in self-care is important in planning upper extremity surgery. In the lower limb, determining the ability of the child to knee stand is often helpful in predicting the effect on walking following correction of knee flexion contractures (Fig. 2.19).

Night Splinting

Night splinting is the most effective method of preventing recurrent deformity. As sleep accounts for a third to a half of the child's life and the period of time correction is applied is a critical factor in preventing deformity, this treatment is very important. Nighttime splinting costs the child little, if it does not interfere with play or socialization as do devices worn during the day. Night splinting is of special importance following correction of clubfeet and knee flexion contractures, as these deformities are most likely to recur.

Material

We have found night splints made of fiberglass and lined with foam to be the most efficient design. Adequate splints may be made of plaster casts or plastics, and the material is not critical. If the splint can be made during a clinic when the physician is available to monitor the positioning and final product, the fabrication is most efficient and convenient for the family.

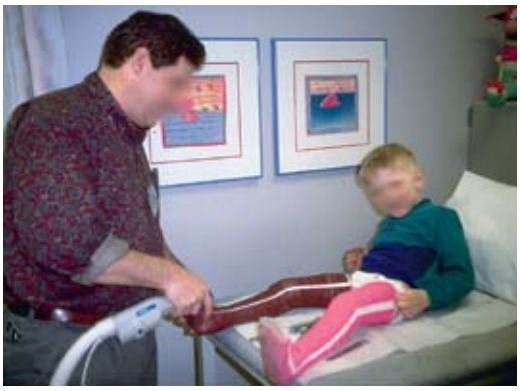


Fig. 2.20 Making of night splints.
Night splints are most valuable for maintaining correction of knee flexion contractures and clubfeet. The splints are made with the knees maximally extended and the feet in as much dorsiflexion and lateral rotation as possible.

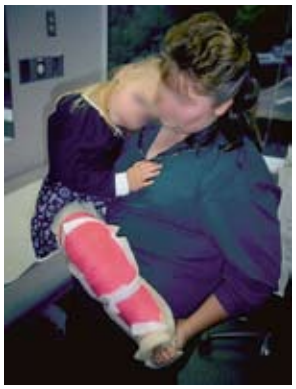


Fig. 2.21 Adapting the child to night splinting.
This child adapts to splinting readily.

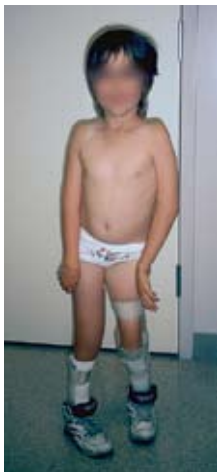


***Fig. 2.22 Long and short leg braces** (left).*
These nonarticulated braces are light, durable, and easily removed.

***Fig. 2.23 Conventional braces** (right).*
These braces allow knee joints while making sitting easier if the brace is used throughout the day.

Position

Place the limb in the best position to resist the tendency for the original deformity to recur. For example, to prevent recurrence of a clubfoot deformity, position the foot in lateral rotation, dorsiflexion, and eversion with abduction of the forefoot (Fig. 2.20).

Comfort

The night splints must be comfortable. Pressure areas will prevent the child and thus the whole family from sleeping, and the program will fail. Instruct the family to watch the skin for persisting redness. Signs of irritation that persist for an hour after removing the splint indicate that the splint should be remade.

Effective Life of a Night Splint

Night splints usually last about 3 months during early infancy, about 4 months during the second year, and about 6 months thereafter.

Duration of Night Splinting

Continue night splinting as long as there is a significant tendency for the deformity to recur. Most clubfeet should be splinted until at least age 5 years. Splint after correction of severe deformity for the longest period. The duration of splinting may be affected by the compliance and attitude of the family.

Adapting the Child to Night Splinting

Infants and children adapt quickly to night splinting if the splints are comfortable and the family deals with the initial reaction of the child appropriately (Fig. 2.21). Apply the splints for only 3-4 hours the first night. Remove and observe the skin. If it is not irritated, the splint can be left on throughout the second night. Advise the family to avoid the mistake of removing the splint if the child fusses. The child soon associates splint removal with crying.

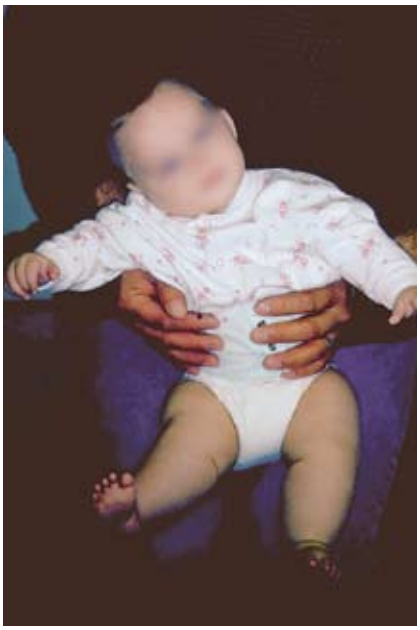
Bracing

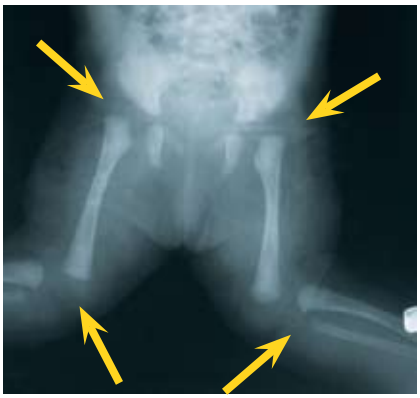
Bracing is used during the day to facilitate function. Use braces only if they enhance effective mobility. Long leg braces (knee, ankle, foot orthosis, or KAFO) often are needed as the child first starts to walk. Later, short leg braces (AFO) may be adequate (Fig. 2.22). Finally, the child often graduates from bracing altogether.

Brace Design

Braces should be lightweight, durable, easily applied and removed, comfortable, and affordable. The use of plastic and aluminum reduces weight.

The first brace may be made without a knee joint (Fig. 2.23). This reduces weight and cost and is also useful if knee motion is limited.





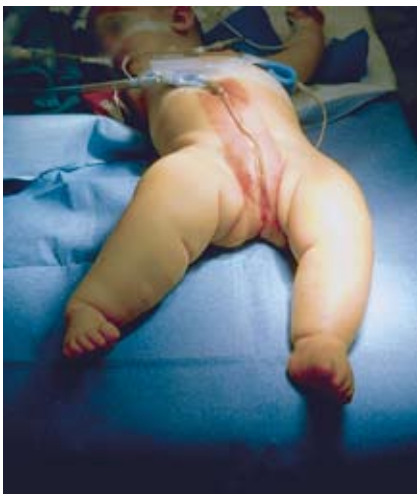


Fig. 2.24 Combining procedures.
This infant with amyoplasia had a dislocated hip (top arrows), hyerextended knees (bottom arrows), and a right clubfoot. Operative correction was undertaken.

Accommodating Foot Deformity

The presence of a foot deformity requires molding of the foot portion of the brace. If the foot is stiff and deformed, the skin over bony prominences may become irritated. This requires relieving the pressure area by molding the orthosis. This molding must be exaggerated to be effective.

Shoes

The child's shoes should be comfortable, ample in size, flexible, and of acceptable appearance. Usually, inexpensive sneaker types of shoes are adequate.

Surgery

Operative correction should be employed early, briefly, aggressively, and later sparingly and only as absolutely necessary. Surgery is usually necessary to correct primary deformity. It is effective but carries significant risks and costs. At best, the surgery is decisive and correction is permanent. At worst, surgery achieves inadequate correction and the deformity recurs.

Timing of Surgery

Most infants with arthrogryposis require surgery. Correct most lower extremity deformities during the first year. Correct upper extremity deformities in early childhood when deformities that limit function are isolated. The recommended age for operative correction varies from center to center. Williams (1978) recommends correcting feet at about 4 weeks, knees at 4 months, and hips at 6-8 months. Lloyd-Roberts and Lettin (1970) recommend that all deformities be corrected before the walking age of 18 months.

Combining Procedures

A basic objective of operative treatment is to correct all of the limiting deformities with the least number of procedures and the shortest possible period of immobilization. This objective is best achieved by combining procedures. The number of procedures that can be performed during one operative session is somewhat dependent on the skill and experience of the surgeon and

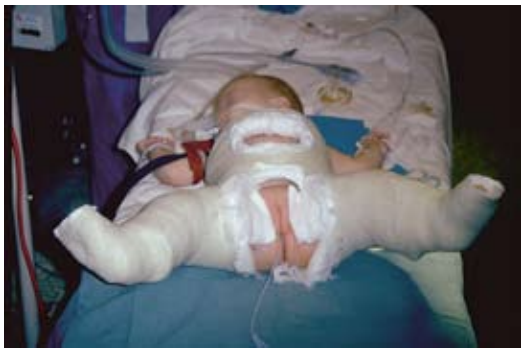


Fig. 2.25 Combining procedures.
The procedures were performed and a spica cast was applied (top). A night splint was used to maintain correction of the clubfoot (middle). The child at age 19 months (bottom).

anesthesiologist. How many procedures can be performed during one session? We have corrected both clubfeet, reduced both hips, and lengthened the quadriceps tendon during one session of 4-5 hours. The infants tolerate these multiple procedures well and greatly benefit by a reduction in the periods of hospitalization and immobilization and the added risks of multiple procedures. In addition, the family benefits by reduced cost and interruption of family routines (Figs. 2.24 and 2.25).

An alternative method of combination convalescence involves spacing the operations about 2 weeks apart so the infant can convalesce from both procedures during the same period of immobilization.

Avoid stringing out procedures to occupy much of infancy and childhood. This unnecessarily prolongs the period of immobilization, leads to greater stiffness, delays development, and robs the patient of normal childhood experiences.

Incision

Plan the incision carefully. The Cincinnati incision is useful in correcting clubfeet and vertical tali. An anterior vertical midline incision is best for lengthening of the quadriceps. Correction of knee flexion contracture may be approached through a transverse incision, a lazy S, or, if severe, a single large Z-plasty. The oblique medial incision is useful for the medial approach in reducing a dislocated hip. Closure with subcuticular dissolving sutures is ideal, but incisions that are subjected to postoperative stretching require interrupted suture closures.

Types of Operations

The type of operation depends on the age of the patient and the nature of the deformity. Correct deformities in infants using soft tissue procedures. Bone procedures are necessary for the older child or adolescent or for more severe deformities.

Soft Tissue Procedures

Most primary deformity can be corrected during infancy using soft tissue procedures, which are preceded and followed by corrective casts and night splinting. Consider each soft tissue element in the correction.

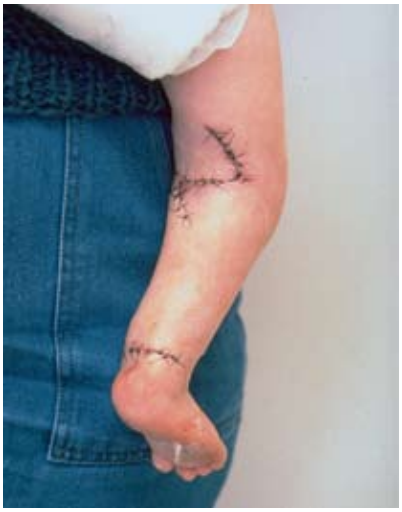


Fig. 2.26 Z-plasty.
This single Z incision behind the knee provided excellent exposure and immediate lengthening of the skin to improve correction.

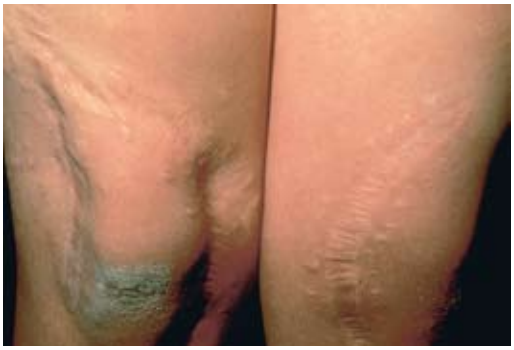


Fig. 2.27 Ugly scar.
This scar over the front of the knee posed a significant cosmetic problem for this girl. The position and use of interrupted sutures make the scar unacceptable.

Skin

The skin is usually contracted because of the intrauterine immobility and deformity. Contracted skin often limits the initial correction. This contracture may be overcome by Z-plasty (Fig. 2.26). For established scars that are con tracted, a series of small Zs may be used to lengthen or break up the scar. If the skin is under tension at the time of closure, interrupted sutures are necessary. For most incisions, subcuticular closures are best. Scarring is least and suture removal is avoided (Fig. 2.27).

The contracted skin may be corrected by postoperative serial casting (Fig. 2.28). The deformity is released, but the initial operative cast is placed with the extremity in a position of only partial correction. After the skin is healed (2-3 weeks), casts are applied weekly to gradually stretch the skin to achieve the level of correction obtained interoperatively (before skin closure).

The use of soft tissue expanders has been tried, but outcomes have varied and the reported complications rate has been high.

Tendon and Muscle

In amyoplasia, muscles may be normal, hypoplastic, or completely absent. Most often, muscles are hypoplastic and partly replaced by scar tissue. This causes weakness and limited muscle excursion. The status of the muscle can usually be determined at the time of operation. If the muscle is absent, the exposed tendon is hypoplastic and inelastic. With traction, the tendon will not elongate and represents only a deforming band and usually is best simply released unless the tendon is maintaining the contour of the limb segment. An example is the fibrotic heel cord. To retain the shape of the ankle, it is preferable to lengthen rather than section this tendon.

Lengthening of muscle tendon units may be achieved by one of several methods (Fig. 2.29).

1. Z-plasty or step-cut lengthening is the most commonly used method. This technique preserves the function of the muscle. The amount of lengthening is important. Lengthen generously to allow the joint to be functionally positioned. Overlengthening is seldom a problem in amyoplasia. Problems are nearly always due to undercorrection and recurrence.

2. Simple division may be necessary if the muscle is absent and only a tethering band remains.

3. Aponeurotomy is lengthening achieved by dividing the fibrous envelope encasing the muscle. This technique is used for muscles that attach to bone with little or no tendon. Examples include the semimembranosis and biceps muscles. The amount of lengthening depends on the degree of contracture of the envelope and the number of divisions made in the envelope.

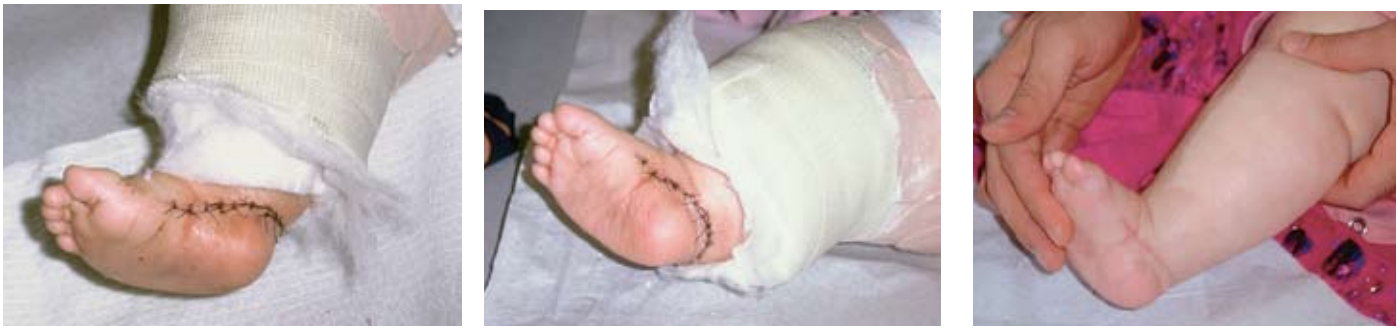


Fig. 2.28 Postoperative serial cast correction.
Full correction may not be possible at the time of surgery. The cast is removed 2-3 weeks following surgery (left), and gradual correction is achieved by weekly cast changes. The appearance at the second change (middle) and following correction (right).

4. Direct origin release. Simple release from the bone allows lengthening at either end of the muscle. The muscle reattaches to bone in an elongated position. Release of the gastrocnemius from the distal femur to correct knee flexion contractures is an example of this type of lengthening.

Tendon Transfers

Muscle-tendon transfers are performed to transfer the power of a functioning muscle to a location of greater functional value. Transfers in amyoplasia are seldom used in the lower limb. Transfers in the upper limb are sometimes useful if the procedure improves the functional position of the hand. To be effective, the transferred muscle must be strong and have a suitable excursion, and the loss of its original function must be acceptable. Deformities should be corrected and mobility achieved prior to the transfer. As these conditions are seldom met, transfers are rarely performed.

Capsule

Capsules are fibrous envelopes that enclose joints. In most congenital contractures, capsules are thick and contracted and pose a significantly limited joint movement. Release (capsulotomy) is nearly always necessary. Divide the capsule completely, and be certain that satisfactory joint motion has been achieved. Do not expect to achieve greater motion postoperatively than was possible with the joint open.

Ligaments

Ligaments are soft tissue connections between bones. They provide stability. In congenital contractures, they may be shortened and prevent repositioning the bony elements in a functional position. In such cases, the ligaments must be released. An example is the interosseous ligaments between the talus and calcaneus in severe clubfeet.

Nerves and Arteries

These structures cannot be surgically lengthened but may be elongated by gradual stretching. Although some elongation is possible at the time of surgery, most correction must be achieved by gradual postoperative stretching with traction, serial stretching casts, or an external fixator. If the deformity is severe, bone shortening may be necessary to achieve correction to avoid overstretching these structures. An example is a severe knee flexion contracture. Femoral shortening allows full immediate correction without excessive stretching of the popliteal nerve and artery.

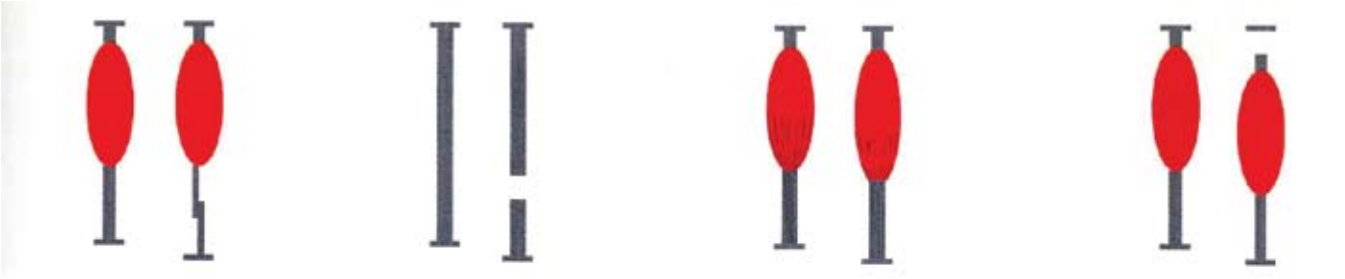


Fig. 2.29 Types of muscle-tendon lengthening procedures.
Lengthening muscles may be accomplished by a variety of techniques.

Bone

Bone procedures are numerous and include a variety of osteotomy types (Fig. 2.30). Rotational osteotomy changes the alignment in the transverse plane. Wedge osteotomies are of several types: Removal of a wedge of bones is called a *closing wedge osteotomy*. If a wedge of bone is added, it is described as an *opening wedge osteotomy*. A segment of bone may be removed and this is described as *shortening osteotomy*. A bone may be removed entirely, such as talectomy or astragalectomy, to correct the clubfoot deformity. If only the center of the bone is removed, it is termed a *decancellation*.

Fixation methods are either internal or external. Internal fixation is applied directly to bone as part of the operation (Fig. 2.30, right).

Joints

Most contractures result in deformities through joints. The joint may be fixed in a functional or nonfunctional position. This fixation is described in comparison with the anatomic position (Fig. 2.31). When a dislocated joint is corrected, it is said to be reduced. Most surgical procedures in arthrogryposis move the arc of motion into a more functional plane. The actual range of motion often remains about the same.

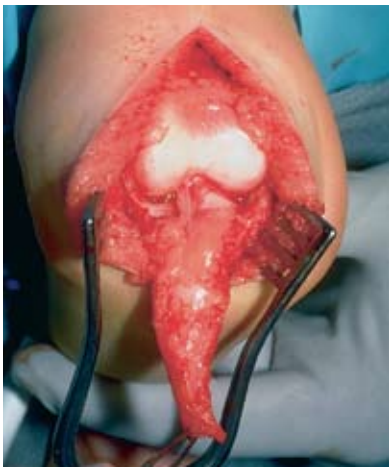


Fig. 2.31 Joint surgery.
This shows the knee joint during a lengthening of the quadriceps for an extension deformity of the knee.

Internal Fixation

1. Pins across the osteotomy site and supplemented with a cast are a common combination. Pins may be smooth or threaded. Smooth pins may be left protruding through the skin and removed in clinic without anesthesia. Threaded pins do not migrate and may be removed after the bone is healed or left in place. Removal of threaded pins usually requires an anesthetic.

2. Plates applied with screws are a common method of fixing osteotomies. Plates are often removed, as they affect the elasticity of bone, and fractures may occur through the end screw holes.

3. Removal of hardware. The need to remove metallic fixation devices is controversial. As the long-term effect of metallic implants appears to be benign, removal is becoming less commonly performed.

External Fixation

Several methods of external fixation are useful.

1. Cast immobilization. Plaster or fiberglass casts are commonly used. Try to limit immobilization to 6 weeks or less.

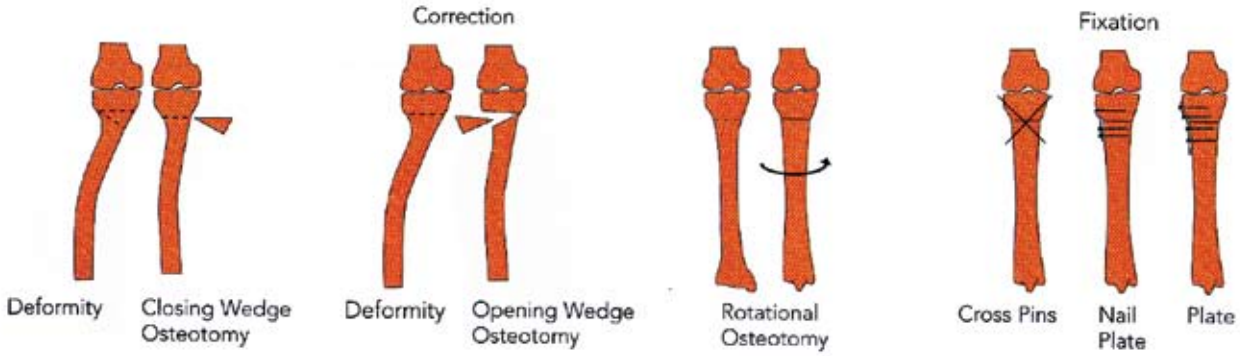


Fig. 2.30 Osteotomies and fixation.
Various types of osteotomies are designed to correct specific deformities. Fixation is required to hold position until healing is completed.

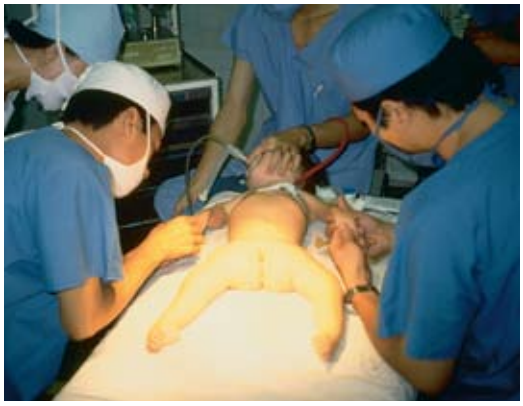


Fig. 2.32 Anesthesia.
Anesthesia has become much safer with modern techniques. Providing a good airway and IV connection is essential.

2. Serial cast immobilization. Serial casting may be started after 2 weeks when the skin is healed. Change casts weekly until the desired correction has been achieved.

3. External fixators are of two types: ring and cantilevered. External fixation allows convenient inspection of the skin and circulatory status and, most importantly, an effective means of achieving gradual correction of deformity at a precisely controllable rate. The disadvantages include risks from pins (i.e., infection), nerve or vascular damage, pain, psychological problems, and cost. These techniques allow unparalleled flexibility in correcting deformity. The role of external fixators in managing congenital contractures is in the process of being determined.

Risks and Complications

The risks of surgery for arthrogryposis include the usual risks of infants or children undergoing orthopedic procedures: anesthetic complications, wound infections, and so on. Fortunately, arthrogrypotic patients have fewer complications than those with such conditions as cerebral palsy or spina bifida, as sensation is intact, communication skills are excellent, and muscle tone is normal. This results in fewer pressure sores, pathologic fractures, and overcorrection. Patients with congenital contractures face special risks and problems. In some forms of arthrogryposis, problems with ventilation (Fig. 2.32) and malignant hyperthermia may be present. Discuss these risks with the family openly. Identify special problems in advance and provide special care to avoid the problems.

The greatest operative risk is incomplete correction or recurrent deformity. Operative releases improve the deformity but cannot address the underlying pathology. Certain deformities, such as clubfeet and knee flexion contractures, tend to recur. The family should be made aware of these problems before the procedure. A common misconception is that an operation is definitive and permanent. The operation is but one step in management. Continued follow-up is necessary through the period of growth.

Fractures

Most fractures occur during delivery (Friedlander et al., 1968) and during the perinatal period (Diamond and Alegado, 1981; Simonian and Staheli, 1995) (Fig. 2.33).

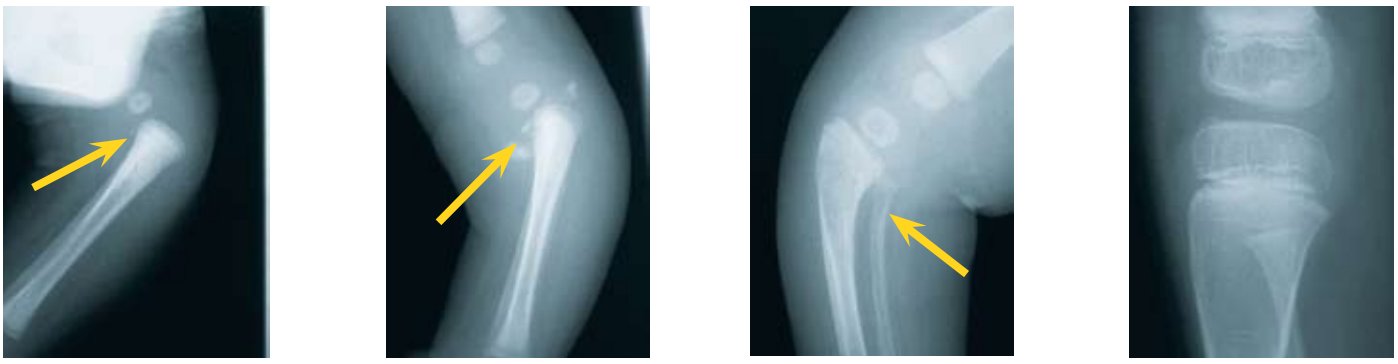


Fig. 2.33 Neonatal iatrogenic fracture.
This infant was born with hyperextended knees and dislocated hips. In an attempt to correct the knee extension deformity, the tibia was fractured (left arrow). This healed (middle arrow) and gradually remodeled. Remaining deformity present at 12 months (right arrow) and full correction by remodeling at 3 years (right).

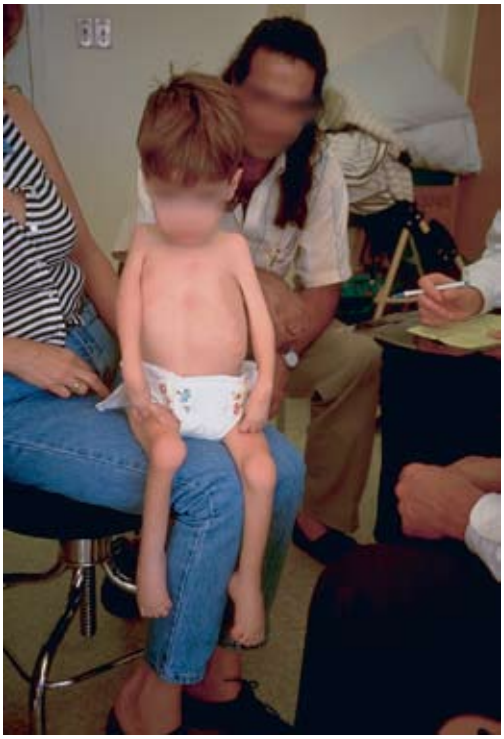


Fig. 2.34 Atypical forms sometimes cannot be categorized .
If the infant is hypotonic, consider delaying operative correction until the respiratory status is optimum.

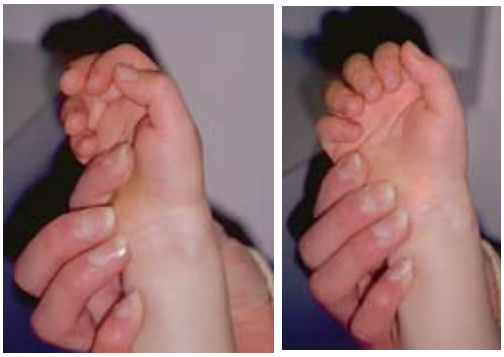


Fig. 2.35 Distal arthrogryposis.
Flexed overlapping fingers are common in distal arthrogryposis.

Other Forms of Arthrogryposis

Most congenital contractures are due to the classic form of arthrogryposis, amyoplasia. These infants have multiple contractures usually in the upper and lower limbs, with loss of skin creases about joints, muscle hypoplasia, loss of motor function, dimpling over bony prominences, and multiple deformities. Clubfeet, flexed or extended knees, dislocated hips, extended fingers, flexed wrist, elbow extension, and shoulder hypoplasia often are present. Sensation is intact, and intelligence is normal, and the occurrence is sporadic. If the findings are atypical, consider one of the less common forms (Fig. 2.34). I have included some of the more common forms that often require orthopedic management because of deformity.

Distal Arthrogryposis

Distal arthrogryposis was described by Hall et al. in 1982 (Hall et al., 1982a). The disorder shows heterogeneity. Type I is most common. It is autosomal dominant, the fist is clenched at birth, and fingers overlap (Fig. 2.35) and are ulnar deviated. The foot may show a vertical talus or equinovarus deformity. The IQ is normal. Types II a through e are extremely varied. Patients have cleft palate, cleft lip, small tongue, trismus, ptosis, short stature, scoliosis, and dull normal IQ.

Pterygium Syndromes

Webbing across joints is present in a number of syndromes. Classic locations include the neck in Klippel-Feil, Noonan's, and Turner's syndrome and the elbows in the nail-patella syndrome. Some multiple pterygium syndromes are lethal.

Multiple Pterygium

Escobar syndrome (Fig. 2.36) is a rare autosomal recessive disorder characterized by short stature and multiple deformities, often including scoliosis, vertical tali, finger deformities, facial dysmorphia, and genital abnormalities. Webbing occurs most commonly on the lateral neck, knee, shoulder, elbow, fingers, and anterior chin (Escobar et al., 1978).

Popliteal Pterygium

This is an autosomal dominant disorder characterized by popliteal webbing that is usually bilateral. Cleft palate or lip and genital abnormalities are common. The popliteal web includes a fibrous band that extends from the ischium to the calcaneus. The popliteal nerve lies immediately below the band. The vessels are deep. Neurovascular structures may be identified by an MRI. Correction of the knee flexion contracture is usually appropriate. The band may be released, skin Z-plasty performed, and the hamstring tendons lengthened. Femoral shortening is required if the contracture exceeds about 45°.

Diastrophic Dysplasia

Diastrophic dwarfism is a rare autosomal recessive disorder characterized by short-limbed dwarfism, multiple contractures, hitch-hiker's thumb, deformed pinnae, cleft palate, normal IQ, and varied spine and foot abnormalities. The most common foot abnormalities (Ryoppy et al., 1992) include hindfoot valgus and metatarsus adductus, equinovarus, and metatarsus adductus.

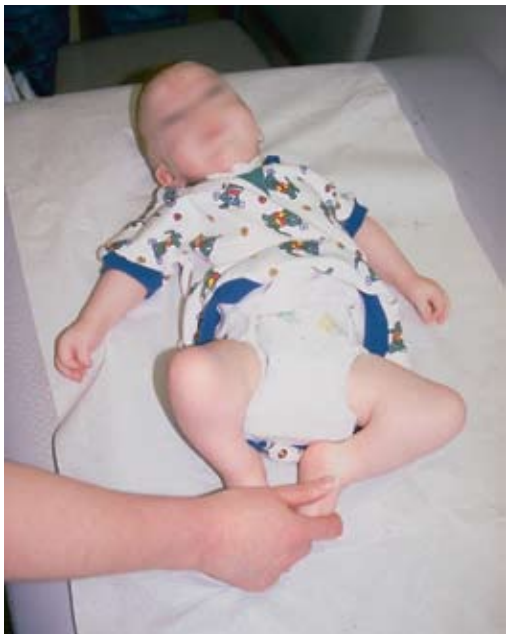


Fig. 2.36 Multiple pterygium syndrome.
Webbing of the knees is most severe.

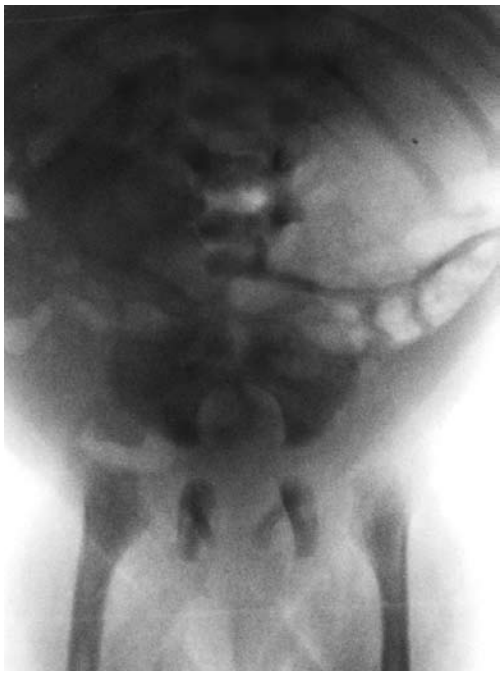


Fig. 2.37 Sacral agenesis.
These infants often show a variety of deformities that are difficult to manage.

The common spine lesions (Poussa et al., 1991) include cervical kyphosis, scoliosis, and spinal stenosis in older individuals. Evaluate the stability of the upper cervical spine before administering an anesthetic (Richards, 1991).

Lumbosacral Agenesis

Sacral agenesis with caudal regression (Fig. 2.37) is a rare disorder often occurring with diabetic mothers and characterized by a variety of lumbosacral abnormalities and lower limb anomalies. These include hip dislocation, neurologic impairment, spine instability, and lower limb contractures. The goal of treatment is to have the patient standing or sitting depending on the severity (Guidera et al., 1991) and degree of neurologic impairment. In a long-term study, the best results were obtained by knee disarticulation and prosthetic fitting. Spine-pelvic instability and dislocated hips are not a problem (Phillips et al., 1982). The spine pelvis dissociation was managed aggressively by fusion using autogenous bone from knee disarticulations (Winter, 1991). Knee contractures may be corrected by soft tissue release and femoral shortening.

Larsen's Syndrome

Larsen's syndrome is an autosomal dominant or recessive heterogeneous disorder characterized by multiple joint dislocations and characteristic facial defects. Differentiate from amyoplasia by multiple joint dislocations, more spine involvement, and a binuclear os calcis. Significant early morbidity may be attributed to cardiopulmonary problems. Reduced elastic fibers in larynx, trachea, and bronchi causes tracheomalacia (Ronningen and Bjerkreim, 1978) and may be associated with problems in wound healing (Lutter, 1990) following orthopedic surgery. Employ conservative methods for correcting hip dislocations, clubfeet, and genu recurvatum. Delay operative correction until the general health is stable (Laville et al., 1994).

Freeman-Sheldon Syndrome

This is also called cranio-carpo-tarsal dysplasia, or whistling face syndrome. It is a rare, autosomal dominant disorder with classic facial features of a pursed mouth, deep-set eyes, and a small nose. Intelligence is normal. The infant is often seen because of foot and hand deformities, including flexed, ulnar deviation of fingers, clubfeet, or vertical tali. There may also be dislocated hips and scoliosis and small stature. Operative correction is usually necessary. Recurrent deformity is common. Anesthetic complications of airway difficulties, malignant hyperthermia, and muscle rigidity following halothane use have been reported.

Contracture Arachnodactyly

This variant of Marfan's syndrome is autosomal dominant and characterized by spidery hands and feet and multiple contractures. The contractures usually involve the knees, elbows, toes, and fingers. The contractures tend to improve with growth and nonoperative management. Scoliosis may require surgery. Knee flexion contractures may be most disabling and may require operative correction (Langenskiöld, 1985).

These varied forms of arthrogryposis are usually readily differentiated if a careful evaluation is made. The management principles are, however, very similar to those for amyoplasia. In Chapter 3, we deal with management of lower extremity deformity.

Upper Limb

A. Bach, M.D.
E. Almquist, M.D.

Spine

M. La Grone, M.D.

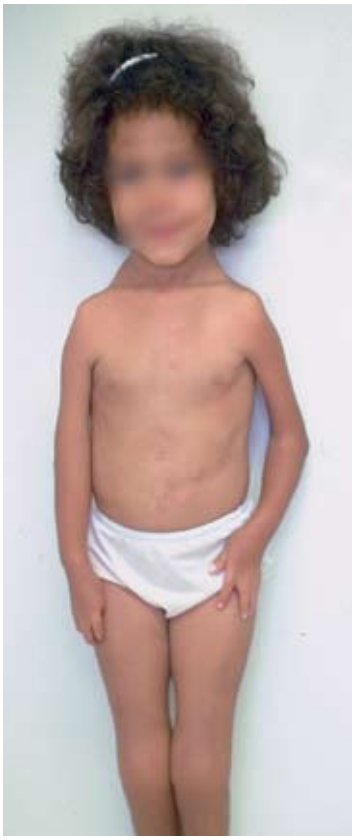


Fig. 3.1 Upper involvement in amyoplasia. *This child shows the typical upper and lower extremity involvement. Note the shoulder muscle hypoplasia.*

Upper Limb

Introduction

Children begin to explore their surroundings with their hands soon after birth. This employs not only the motor function of grasp and hold but also the sensory information received from touch. These functions develop throughout childhood from the most simple grasping motions to the sophisticated manipulation of a musical instrument. Upper limb involvement in arthrogryposis impairs hand function through both weakness and lack of joint mobility while leaving sensation completely normal. The goals of treatment of the upper limb are twofold: first, to maximize hand prehension and grasp, and second, to mobilize the shoulder, elbow, and wrist to maximize the placement of the hand in space. The upper limb may also be called on to provide support during ambulating via a cane, crutch, or walker.

With over 150 specific causes, arthrogryposis has extreme variability in limb involvement (Hall, 1985). In the upper limb, the majority of patients treated will have amyoplasia as a diagnosis. Most amyoplasia patients will have rather symmetric involvement of their limbs. Another group will have distal arthrogryposis, a heritable disorder primarily involving the hand.

Care of the upper limb in arthrogryposis combines the skills of therapists, nurses, and physicians. Tools available to this team are range of motion treatment, splinting and casting, occupational therapy, and surgical treatment.

Patterns of Involvement

Shoulder

Significant loss of shoulder function is seen in most patients with amyoplasia (Fig. 3.1) and is typical for other forms of arthrogryposis. The changes about the shoulder are marked in many cases, but these changes have been described

Fig. 3.2 Lack of elbow extension.
The lack of active elbow flexion can be a severe but not insurmountable handicap. Here, the child uses counterpressure from the tabletop to bring his hands to the facial area.

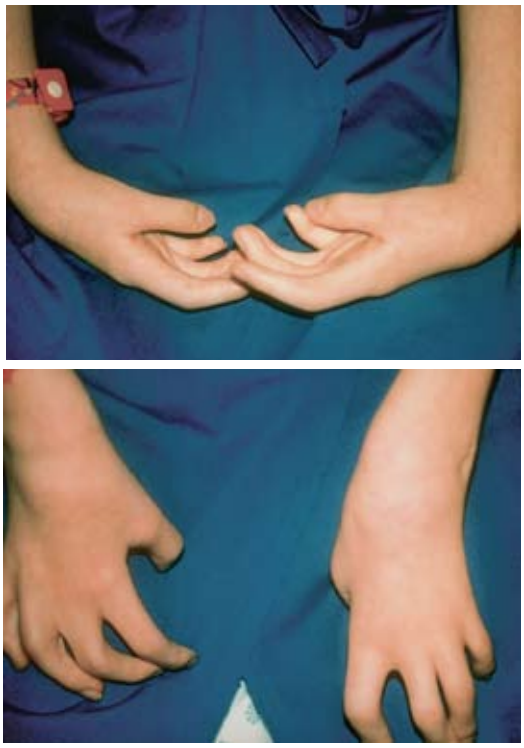


Fig. 3.3 Wrist flexion deformity.
Uncorrected wrist flexion and thumb-in-palm deformities in a 9-year-old child. The decision to surgically correct the position involves a thorough assessment of the child's current function and desired functional goals.

as having little impact on the patient's overall disability (Williams, 1985). However, ankylosis of the shoulder in the best functional position still is considered a 40% impairment of the upper extremity. This low emphasis on the shoulder in arthrogryposis may reflect the limited treatment options.

Limitation of shoulder abduction and external rotation is noted from the neonatal period. Muscle weakness of the deltoid and external rotators accompanies these contractures. Pectoralis function is often present even in severe cases and applies an unopposed internal rotation force on the humerus.

Elbow

Even with an elbow ankylosed in a functional position, nearly half of the potential for upper extremity function is lost. The stiffness seen in the arthrogrypotic limb is highly variable and may be in flexion or extension. Passive range of motion at birth may be limited to just a toggle. The joint capsule, muscle, tendons, and skin are all affected. Early joint changes have been found with flattening of the articular surfaces before age 1. The elbow will often be in extension. Triceps function will be present, but biceps and brachialis are nonfunctional or extremely weak (Fig. 3.2). When the elbow is flexed, biceps function will be better but is limited by the stiffness of the elbow.

It is important to consider the lower extremity function before planning treatment for a stiff elbow. The usual goal for elbow treatment is to allow the hand to at least passively reach the face. However, if crutches or other assistive devices are needed for ambulation, an elbow release may not be wise or may be deferred until lower extremity function is improved.

Along with the extension deformity of the elbow, the forearm will often be in pronation.

Wrist

A flexion and ulnar deviation deformity is most often present at birth in children with significant upper extremity involvement (Fig. 3.3). Occasionally, the wrist may be in an extended position, and the forearm muscle development will show some flexor power of the wrist even in the most severe cases. The volar wrist capsule will be tight, and intraarticular adhesions have been demonstrated during wrist releases. X-rays of the wrist may show intercarpal fusions. The changes seen in patients with distal arthrogryposis are usually milder than those seen with amyoplasia.

Hand

The hand position will depend on the specific cause and severity of the disease. There is wide variation in the deformities of the hand. With distal arthrogryposis, the fingers are flexed and often overlapping. The metacarpal phalangeal joint will be in ulnar deviation, as seen in Freeman-Sheldon syndrome, and will respond very quickly to splinting. In amyoplasia, the fingers will be in a position of intrinsic contracture, and a thumb-in-palm deformity is present. Lack of digital skin creases is variable and reflects the severity of the problem. The interphalangeal joints are slightly flexed, and the interdigital spaces may be severely webbed.

Because of the marked variation, it is difficult to categorize hand deformities, but usually the hand deformity can be classified in one or more of three groups. The most common is thumb-in-palm deformity, where the MCP joint is flexed in 90°, the metacarpal is adducted, and the interphalangeal (IP) joint can either be flexed or stiff in extension. The thumb occupies the palm and therefore limits finger grasping. The second group is flexion deformities

of the fingers. This usually involves the PIP joints while the MCP joints are in relative extension. This deformity prevents flattening of the hand, which is often quite functional. This deformity allows the limited muscle excursion to move the fingers through a functional range for grasping and prehension. The deformity actually enhances the limited power. The third general group is aplasia, with limited action movement and varying stiffness from an extended position. The MCP joints are usually in ulnar deviation, and the PIP joints lie in extension and may be stiff in extension or have considerable passive flexion. The distal interphalangeal (DIP) joints are usually stiff in extension. If this hand has a mobile thumb, this deformity at least allows prehension, if not grasp.

Treatment

There are three general treatment goals for the arthrogrypotic upper extremity: gaining and maintaining a functional range of motion of the upper extremity joints, first passively and then actively, if possible; increasing functional abilities, particularly the activities of daily living of eating, dressing, and toileting with occupational therapy, adaptive devices, and surgery; and maximizing educational and vocational potential, which often involves using computer keyboards.

Early institution of splinting and range of motion treatment has been a universal element of our treatment program. It is our recommendation that corrective splints for the elbow and hand be applied within a few days at birth. Although the end results of splinting in arthrogryposis remain controversial, we have found that early application of these splints enhances their effectiveness. Serial casting and the application of thermoplastic splints are both useful techniques. Our choice has been to use thermoplastic splints in the upper extremity, which allow skin care and functional use and are easily adjusted to increasing corrections. However, a very skilled occupational therapist is necessary for correct application. Range of motion treatment is encouraged and monitored by the therapist but is primarily done by the parents.

The majority of children with upper extremity manifestations of arthrogryposis will not need surgical treatment (Fig. 3.4). At Children's Hospital and Medical Center in Seattle, 70% of the children seen at the arthrogryposis clinic did not undergo surgery on the upper extremity. The 30% who did undergo surgery were mostly patients with amyoplasia.

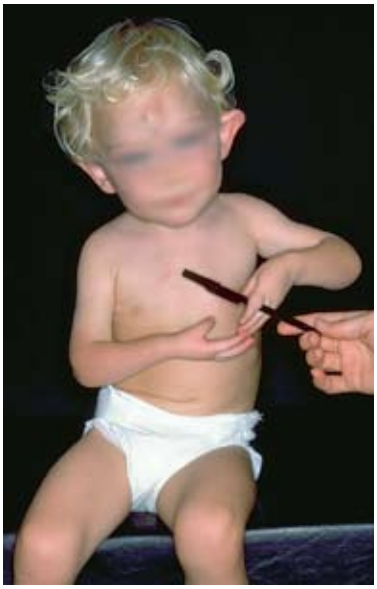


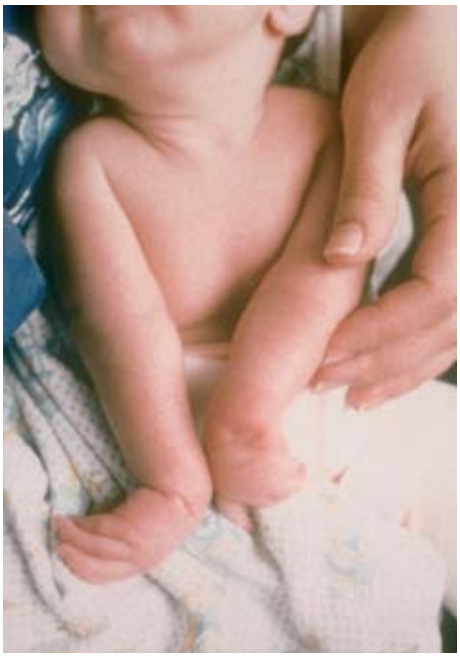
Fig. 3.4 Adaptive movement.
This child has adapted well in function. With voluntary control and good sensation, hand function is remarkably good even without surgical intervention.

Shoulder

Early institution of passive range of motion is the mainstay of treatment for the shoulder. Most of the children with amyoplasia will have poor active shoulder abduction and internal rotation deformities of the shoulder. No splints have been used. No releases have been done. All improvements in active range of motion have come with ROM therapy in our clinic, and no muscle transfers have been done. This management has allowed us to avoid performing humeral rotational osteotomies in nearly all cases (Bennett et al., 1985). The primary indication for this procedure has been to facilitate computer keyboard use.

Elbow

Our goal has been to obtain flexion to 90° in elbows that are initially extended. Thermoplastic splints are applied within a few days of birth. The orthopedist monitors progress every 4-6 months and continues treatment until no improvement is seen. We have not usually made decisions on the



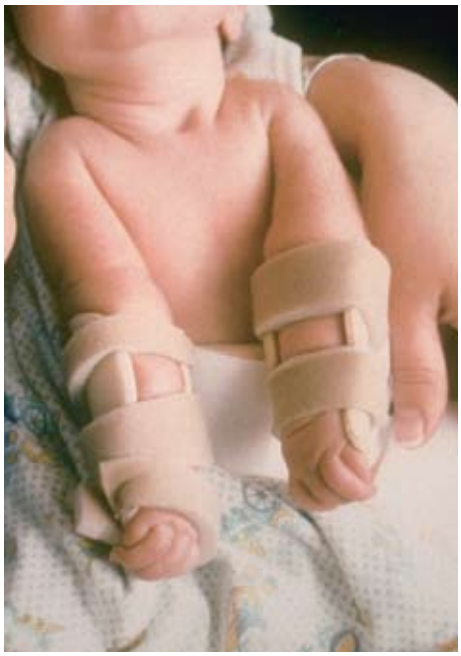


Fig. 3.5 Hand splinting in the newborn.
(Top) Newborn infant with amyoplasia. The upper extremities show typical deformities of shoulder internal rotation, elbow extension, forearm pronation, and wrist, finger, and thumb flexion deformities. (Bottom) With proper fitting, wrist splints can be applied within a few days of birth, and passive correction of the wrist flexion deformities can begin.

necessity of elbow release procedures until the child has reached approximately 8 months of age. If both elbows are not required to be in extension for ambulation, then a posterior elbow release is considered to allow one hand to reach the face for feeding and self-care. This is often done early.

During posterior capsulotomy of the elbow, the triceps tendon is lengthened by a long oblique tenotomy, and the posterior capsule is released. Even aggressive releases rarely result in more than 100° of passive flexion (Williams, 1973). Splinting continues for 8-12 weeks after surgery.

A few children will have bilateral flexion deformities of the elbow that may not respond to splinting. However, anterior elbow release is indicated only in severe contractures, and this is rare. Even with the elbow flexed at 90°, the functional level is high.

Restoration of active elbow flexion in the arthrogrypotic child is often a consideration, since some active flexion power will greatly improve feeding, facial care, and carrying. However, all the muscle transfers available involve some cost. Available donor muscles include the latissimus dorsi, pectoralis major (Bennett et al., 1985; Doyle et al., 1980), triceps, sternocleidomastoid (Carroll, 1962), and the common forearm flexors (Steindler, 1949). The latissimus dorsi offers a large donor muscle with little loss of function if it is transferred. Unfortunately, in most cases of arthrogryposis, the latissimus muscle does not develop to a point that a transfer is possible. The pectoralis major is most often a fairly strong muscle. Transfer is possible, but the cosmetic appearance of the donor site is often unacceptable. The patient will lose ability to forward flex the shoulder in most cases. Triceps transfer can be done simply but will result in loss of active elbow extension, and this procedure should only be performed unilaterally (Bennett et al., 1985; Carroll and Hill, 1970; Williams, 1973, 1985). Progressive flexion contracture of the elbow has been noted after triceps transfer but may be purely a natural progression of the arthrogryposis and not due directly to the transfer. Many children with amyoplasia will have little strength of finger and wrist flexion, and a Steindler flexorplasty cannot be considered. The Steindler procedure ideally can be combined with posterior elbow release and triceps lengthening. It can be performed with little extension loss but gives active flexion to 40°-50°. The strength depends on the muscle power available. Often, both the forearm extensor and flexor muscles are advanced proximally.

Rarely, supracondylar extension or flexion osteotomy will be indicated for the stiff elbow. This should be performed for specific functional demands.

Wrist

Maintaining a functional position of the wrist is an ongoing battle in many arthrogrypotic children. Passive range of motion exercises can improve wrist motion about 50%, but splinting is necessary to avoid recurrent deformity (Palmer et al., 1985). As in the elbow, position splints are applied very early, and this is when the most correction can be achieved. The wrist will be in a flexed and ulnarly deviated position. In most cases, we try to restore a neutral position to the wrist and then use resting splints to maintain position (Fig. 3.5).

When is surgical treatment for the wrist indicated? Several factors must be considered to answer this question. The optimal functional position for the patient should be identified. This will vary with the patient's finger flexor power, digital extension power, and specific functional requirements. The splinting program should have an adequate trial. Wrist surgery is rarely performed early.

Surgical correction involves release of the volar wrist capsule. The release must be complete and may involve taking down intraarticular adhesions.

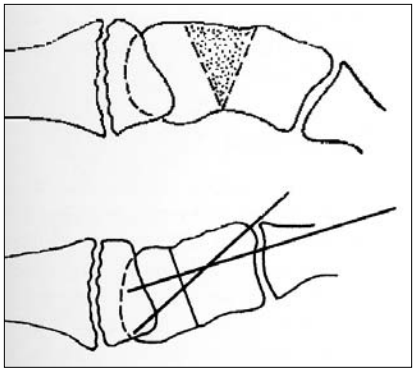


Fig. 3.6 Correction of wrist flexion deformity.
Occasionally wrist flexion deformities involve an intracarpal fusion. Here, the lunate and capitate are fused in a flexed or humpback position. A dorsal closing wedge osteotomy is necessary for adequate correction. With the wedge defect closed and held with K-wires, a position of slight dorsiflexion of the wrist can be maintained.

Fig. 3.7 Computer game use.
Being able to position the hands to use a computer is an important objective in upper extremity management.

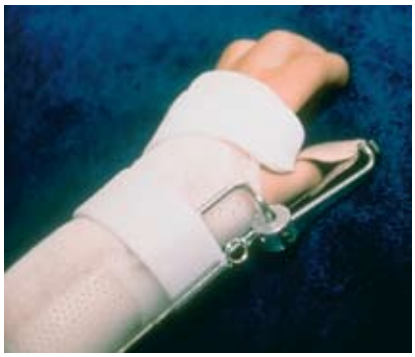


Fig 3.8 Functional wrist splint.
Functional dynamic brace for a thumb-in-palm deformity when active thumb extension is absent. Passive correction is achieved with static splinting techniques.

Maintaining the correction postrelease remains problematic. Some authors believe this is not possible and recommend capsulotomy or wrist fusion when the child is near skeletal maturity (Bennett et al., 1985). Our preference has been to do earlier volar capsular release and employ a flexor carpi ulnaris (FCU) to extensor carpi radialis brevis (ECRB) tendon transfer to maintain position (Palmer et al., 1985). Osteotomy of the distal radius has been used, but recurrent deformity has been seen (Lloyd-Roberts and Lettin, 1970). Intracarpal extension osteotomy is useful when a natural intracarpal fusion exists, but this should be combined with a palmar capsular release. Intercarpal fusion or marked deformity of the carpal bones is seen with flexion contractures over 60° in older children. Correction then must involve intracarpal osteotomies. This often involves a wedge resection osteotomy through the midcarpal area (Fig. 3.6). These fusions can be seen in patients as young as 10 years of age, particularly in children who use the flexed wrist for ambulation transfers, often developing a callous on the back of their wrist.

Hand

Since the degree of observed joint stiffness, strength, and active range of motion varies greatly in the digits of arthrogrypotic patients, the treatment programs must also vary, and the goals of treatment must be made on an individual basis (Fig. 3.7). The primary deformities are the thumb-in-palm and finger flexion contractures. Total lack of skin flexion creases, no active motion, and extremely stiff joints are bad prognostic signs, but as with other joints, the initial appearance at birth may give a falsely pessimistic impression of potential function. Although the efficacy is debated, we think early application of corrective splints is helpful in improving digital deformities, and this is our first step in treatment. The splints are useful only if they are properly molded and applied. A splinting program for the digits may continue for years using resting and night splints. Functional splinting, particularly to abduct the thumb while writing, may be useful (Fig. 3.8).

The thumb-in-palm deformity in arthrogryposis is a combination of metacarpal adduction and metacarpal phalangeal joint flexion contracture. This blocks effective grasp and eliminates the opposability of the thumb. When splints have not been successful and the hand is believed to have functional potential, surgical correction for the thumb-in-palm deformity is considered. Surgical options include first web skin release, adductor pollicus release, sublimis transfer, first metacarpal osteotomy, and first MCP joint fusion. Bennett et al. (1985) reported poor results with skin and adductor release only and recommend MCP joint fusion. Bayne (1985) employs metacarpal osteotomy along with soft tissue release. If possible, we believe that the best addition to release procedures is the addition of an active thumb extensor, if absent, to balance the first ray. Often, the brachioradialis is available.

The interphalangeal joint flexion contractures found in arthrogryposis are particularly difficult to treat. Reviews of this problem have suggested that soft tissue releases of the PIP joints do not give lasting correction and that fusion may be indicated in severe cases (Bayne, 1985; Bennett et al., 1985; Lloyd-Roberts and Lettin, 1970). The interdigital webbing seen at the base of the fingers, however, can be released and allow the patient increased function, especially when using an interlacing grip. In our practice, we have found splinting to be effective in correcting metacarpalphalangeal (MP) joint position. Fusions of the PIP joints should wait until growth is complete. Occasionally, a PIP joint release is indicated if the contracture is severe and

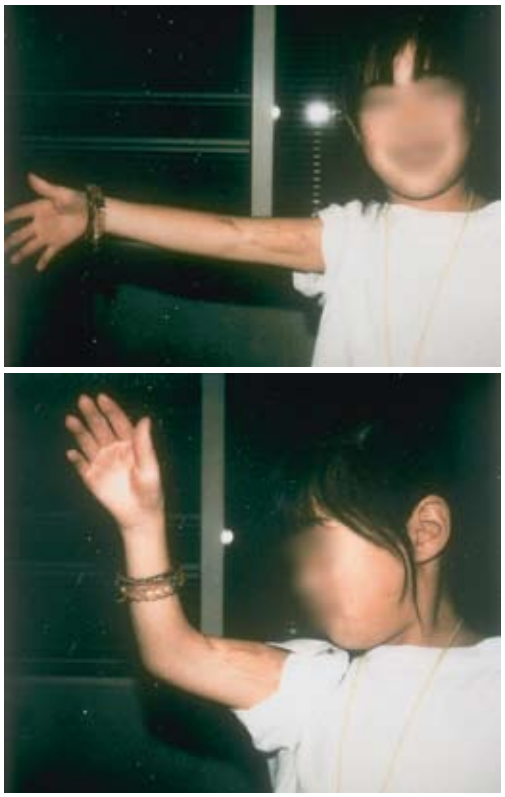


Fig. 3.9 Latissimus dorsi transfer.
A 7-year-old child with lack of active elbow flexion and excellent hand function. Note the active shoulder abduction (top). Following transfer of the latissimus dorsi (bottom) the patient has nearly 90° of strong active elbow flexion without significant change in shoulder function.

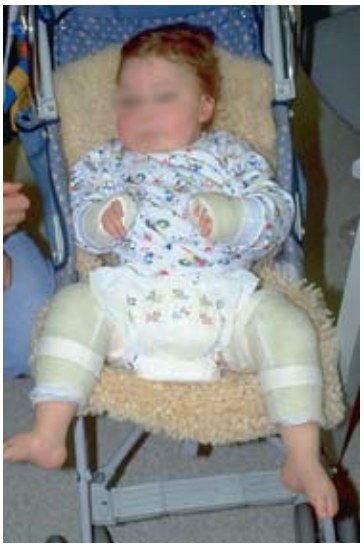


Fig. 3.10 Individualization of management.
This child underwent upper and lower extremity surgeries in a planned sequence to allow convalescence concurrently.

hindering the function of the rest of the hand. Tendon transfers are rarely performed for finger deformities. There is usually too little active muscle excursion to properly balance a transferred finger motor.

Results of Surgical Treatment

From 1970 to 1989, 25 patients underwent surgical procedures for treatment of upper extremity arthrogrypotic deformities. This represents about 30% of our clinic population. Since 1989, an additional 12 patients have undergone surgery. Sixty percent of these patients carry the diagnosis of amyoplasia.

Above the elbow level, the only procedure performed was humeral rotational osteotomy in two patients. Correction in both cases allowed use of a computer keyboard. Nine patients underwent posterior elbow releases with an average range of motion of 41° to 96° of flexion. The average improvement in the arc of motion was 35°.

We have found that even some augmentation of elbow active flexion can be helpful to the patients. The results of our Steindler flexorplasties show that the patient can actively initiate elbow flexion and further flexion can be done passively. No patient developed a more significant wrist flexion contracture following Steindler procedures. The results of triceps to biceps transfer in two patients have been excellent, with active flexion from 40° to 110°. One latissimus transfer has been performed with active flexion to 80° (Fig. 3.9).

For wrist flexion deformities, we have chosen to do palmar capsular releases and FCU to ECRB tendon transfers in younger children rather than do late wrist fusions. Without exception, wrist position of no more than 10° short of neutral was achieved and maintained. The tendon transfers worked as check reins rather than achieving much active dorsiflexion. Carpectomies and fusions were reserved for persistent or untreated deformities in older children and have predictable success. The ability to improve the wrist flexion contracture allows keyboarding, an important function for these relatively immobile people.

In the hand, the best results came from treatment of thumb-in-palm deformities. In 16 cases, we had good results with combinations of soft tissue release and tendon transfer or MP fusion. The 2 cases which were treated by simple skin release resulted in recurrent deformity. We also found that soft tissue release of the PIP joints was not predictable, whereas web space release gave improved function but not range of motion.

Summary

The primary goal of treatment for the upper extremity in the arthrogrypotic child is to maintain and maximize functional capabilities. We believe that early institution of splinting and range of motion treatment offers distinct advantages over delayed treatment and serial casting. A team approach to treatment is necessary.

Surgical treatment in this group of patients is difficult to standardize because of the extreme variability of the disease. Difficulty in achieving treatment objectives of surgery is reflected in the literature by varied experience, lack of uniformity of opinion, and inconsistency of results. Therefore, each patient requires a thoughtful and individualized approach (Fig. 3.10).

Patients with arthrogryposis demonstrate remarkable adaptability with their deformities, and functional evaluation is very important. Our treatment should not interfere with those positive adaptations.

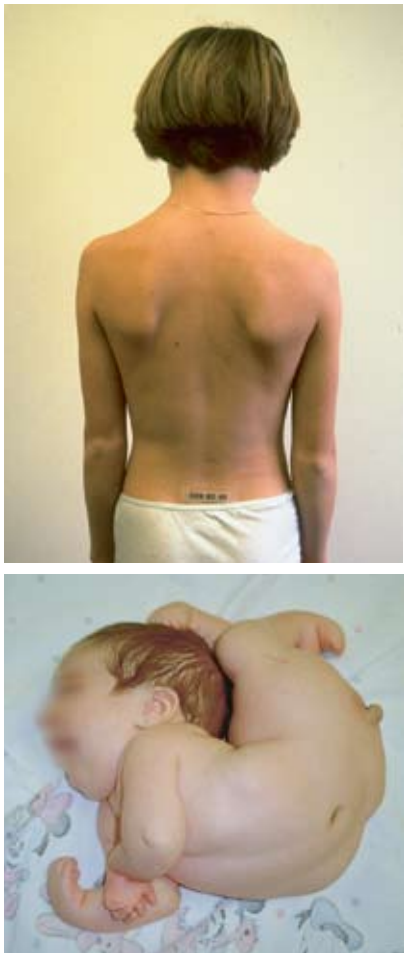


Fig. 3.11 Varied severity of spinal deformity. *The child at the top has a minimal deformity. In contrast, the infant pictured below has a severe hyperextension deformity of the spine. Fortunately, most curves are mild to moderate in severity.*

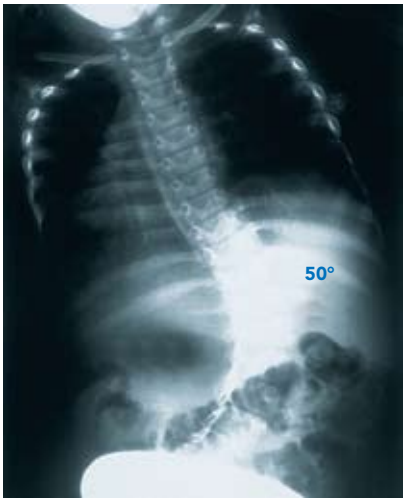


Fig. 3.12 Prominent curve in young child. *This 3-year-old girl has a 50° lumbar curve and pelvic obliquity.*

Spine

Introduction

In the early literature on arthrogryposis, little mention is given to the problem of scoliosis (Friedlander et al., 1968). Stern's original description of the syndrome (1923) does not mention spinal involvement. In more recent years, it has become increasingly evident that the spine is involved frequently in this condition (Drummond and MacKenzie, 1978; Gibson and Urs, 1970; Herron et al., 1978; Sarwark et al., 1990; Spencer et al., 1977; Thompson and Bilenker, 1985).

Incidence

The reported incidence of scoliosis in children with arthrogryposis multiplex congenita varies depending on the group of patients studied. Drummond and MacKenzie (1978) reported on 50 patients with arthrogryposis multiplex congenita. All patients had rigid contractures present at birth that involved at least two extremities. Scoliosis was noted in 14 of 50 patients for an incidence of 28%. There were 8 girls and 6 boys, with ages ranging from 1 month to 6 years at the time of diagnosis. Eight of the 14 patients were reported to have curves greater than 40°. Herron et al. (1978) found significant scoliosis in 20% of the 88 patients they reviewed with arthrogryposis multiplex congenita. Spencer et al. (1977) reviewed 112 patients with arthrogryposis multiplex congenita and reported a 31% incidence of scoliosis..

It would appear that the incidence of scoliosis in patients with arthrogryposis multiplex congenita is between 20% and 30% on average. The variance in reported incidence is due to the wide spectrum of clinical syndromes included in some studies. If only patients with amyoplasia are included, the numbers are more consistent. In the review by Sarwark et al. (1990) a 35% incidence of scoliosis in patients with amyoplasia was reported.

Curve Types

Spinal deformity varies greatly from minimal to severe curves (Fig. 3.11). There is no single typical curve type in patients with arthrogryposis. Three different curve types have been described: congenital, paralytic, and idiopathic-like. The studies by Drummond and MacKenzie (1978) and by Spencer et al. (1977) include a significant number of patients with congenital spinal anomalies (14% and 7%, respectively). On the other hand, Sarwark et al. (1990) have pointed out that patients with congenital scoliosis usually have other specific syndromes, and patients with amyoplasia typically do not have congenital vertebral anomalies.

Paralytic or collapsing-type curves appear to be the most common patterns seen in amyoplasia, particularly in severely involved nonambulatory patients (Fig. 3.12). Sarwark et al. (1990) suggested that the paralytic pattern of most curves supports the theory that amyoplasia is due to an anterior horn cell defect occurring in utero. With respect to curve location, lumbar and thoracolumbar curves are most common (Fig. 3.12 and 3.13), although double thoracic and lumbar curves as well as single thoracic curves can be seen. Lumbar and thoracolumbar curves are frequently associated with pelvic obliquity and can lead to seating imbalance. Pelvic obliquity can also be caused by soft tissue contractures about the trunk or hips.

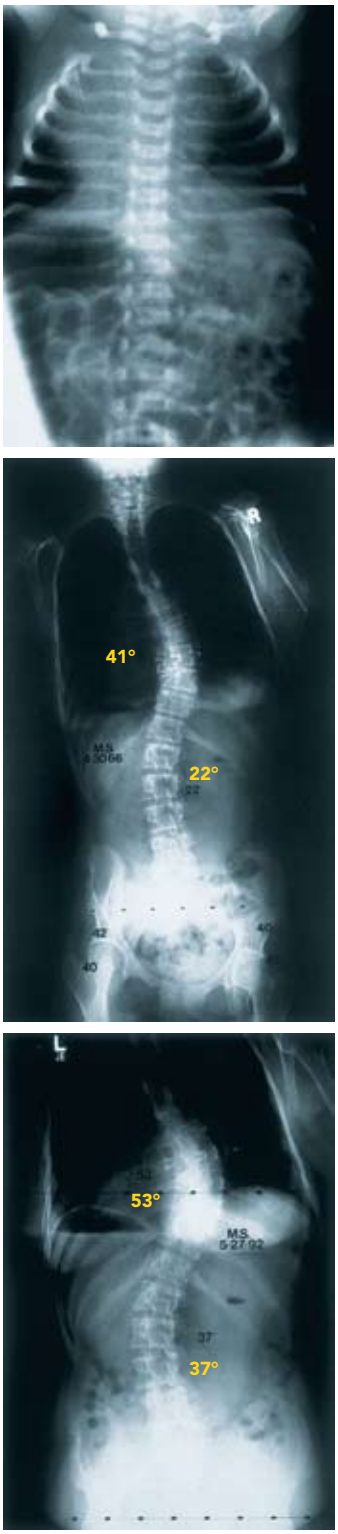


Fig. 3.13 Natural history of scoliosis.
(Top) Eight-day-old girl with amyoplasia and no evidence of spinal deformity. (Middle) By age 13 years, the girl had developed a right thoracic curve of 41° and a left lumbar curve of 22°. (Bottom) By age 18, the thoracic and lumbar curves measure 53° and 37°, respectively.

Natural History

Unlike involvement of the extremities, spinal involvement is not typically present at birth, but it is usually detected within the first few years of life (Fig. 3.13). Drummond and MacKenzie (1978) found that all of their patients with paralytic C-shaped curves had their scoliosis detected within the first year of life and demonstrated a relentless progression of their scoliosis. Herron et al. (1978) stated that most of the patients in their series had curves that were progressive and became rigidly fixed at an early age. They noted that if scoliosis was not present within the first few years of life, it was unlikely to become severe. Therefore, poor prognostic signs for curve progression and subsequent development of severe spinal deformity are early onset, a paralytic curve pattern, and pelvic obliquity.

Evaluation

Every infant or child with arthrogryposis should have a back examination as part of the general screening evaluation. Furthermore, with each interval examination, the back should be assessed, as sometimes curves can be rapidly progressive.

With the child's clothing removed, observe the general appearance from the back and side views. Note asymmetry or deformity. Scoliosis is most readily apparent on the forward bending test (Fig. 3.14). This may be done with the child standing or sitting. Look at each level of the spine for evidence of asymmetry. As the scoliotic deformity includes a rotational component, even a few degrees of scoliosis are detectable by this test.

For measuring the severity of the curve in degrees, radiographs are necessary. These should be taken with the child sitting or standing whenever possible. For older children and adolescents, a 36-inch cassette is very helpful to include the entire spine on one film. Measure the curve by identifying the upper and lower involved vertebrae. Mark the end plates and construct a right angle line from the end plates. The angle enclosed by the intersection of these lines is the degree of scoliosis (Fig. 3.15). As the measures are subject to

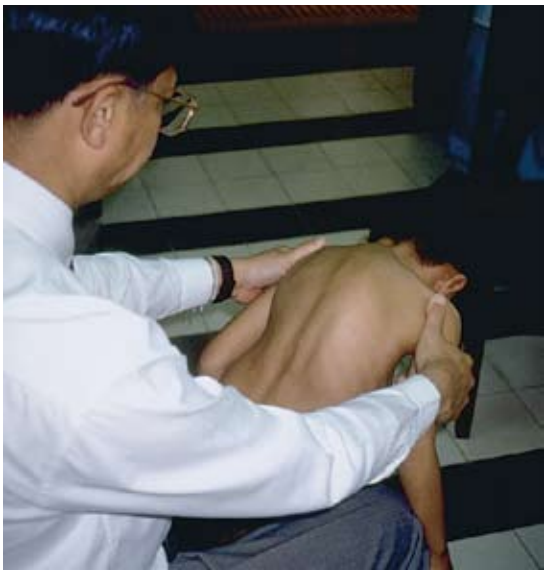


Fig. 3.14 Forward bending test.
Minor degrees of scoliosis can be detected by the forward bending test. Look for asymmetry.

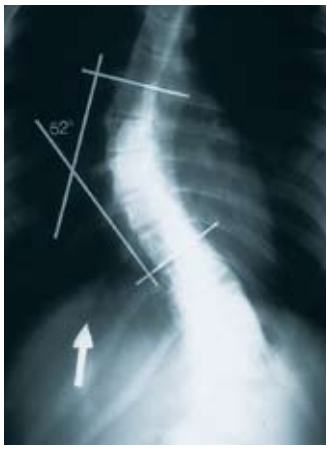


Fig. 3.15 Measurement by radiography. *Measure the severity of the curve by constructing a right angle line from the top of the upper vertebra and the bottom of the lower vertebra in the curve. The developed angle indicates the severity of the curve.*

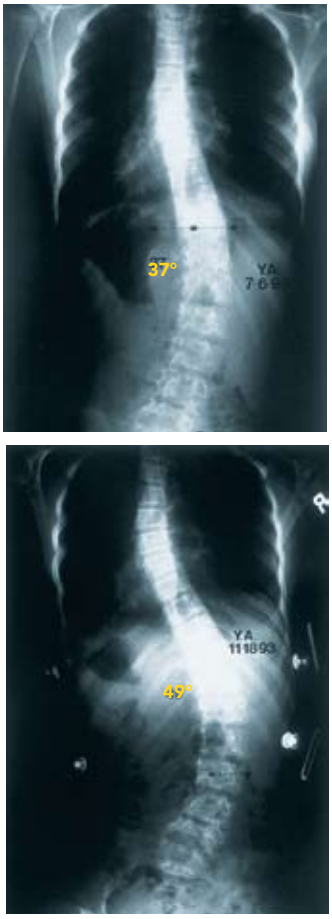


Fig. 3.16 Rapidly progressive scoliosis. *(Top) A 5-year-old girl with amyoplasia and a 37° thoracolumbar curve. (Bottom) In only 4 months, the curve has progressed to 49°. This rapidity of progression is unusual. It does demonstrate that these curves require continual monitoring.*

variations in position of the patient at the time the radiograph was made and differences in marking the film, the accuracy of these measurements is subject to 5°-10° of error. As curves are sometimes rapidly progressive, follow-up studies are essential (Fig. 3.16).

In children with hip and spine deformity, it is often wise to order radiographs that include the pelvis and spine on the same film. This allows an assessment of both problems in one study. This combined study is also useful in assessing pelvic obliquity.

Treatment

As with idiopathic scoliosis, treatment options in patients with arthrogryposis and scoliosis include observation, bracing, and surgery. In contrast to patients with idiopathic scoliosis, brace treatment is rarely successful in patients with arthrogryposis and should be used only in patients with small, flexible curves (Herron et al., 1978; Sarwark et al., 1990). If a curve is between 25° and 40° and flexible, bracing can be attempted. Most studies suggest that bracing will only delay surgical treatment. If bracing is to be successful, early detection is imperative.

Most progressive curves will require surgical treatment. Surgery is generally recommended for curves measuring 50° or greater (Herron et al., 1978; Sarwark et al., 1990; Siebolt et al., 1974). Untreated progressive scoliosis in these patients may cause severe and debilitating spinal deformity. Previous experience suggests that there is no place for expectant management of progressive scoliosis in patients with arthrogryposis.

In patients with thoracic curves and idiopathic-type curve patterns, selection of fusion levels and instrumentation technique is the same as that used for patients with idiopathic scoliosis. These patients are typically ambulatory with less severe involvement. In patients with paralytic-type curves, fusion should usually include the sacrum. In patients with pelvic obliquity, fusion to the sacrum is required. Patients with severe, rigid deformities (curves that cannot be passively corrected to 40° or less or pelvic obliquity that cannot be passively corrected to within 15° of neutral) should be considered for anterior release and fusion, followed by posterior segmental instrumentation and fusion to the sacrum.

The results of surgical treatment are generally good (Daher et al., 1985; Herron et al., 1978; Siebolt et al., 1974). Operative correction is less than that obtained in patients with idiopathic scoliosis. This is likely because the deformities are more rigid. On the other hand, loss of correction is typically low. Daher et al. (1985) reported an average loss of correction of only 5° following surgical correction of scoliosis.

Summary

Scoliosis is seen frequently in patients with arthrogryposis multiplex congenita. It occurs in approximately one third of patients with amyoplasia. Paralytic curve patterns are most frequent, although there is no typical curve pattern in arthrogryposis. If the scoliosis is to become severe, it is typically present within the first few years of life.

Brace treatment is generally ineffective for halting the progression of scoliosis. Most patients with significant scoliosis will ultimately require surgical treatment. One should not allow other orthopedic problems to delay early management of progressive scoliosis. All patients with arthrogryposis should be evaluated at an early age for the possibility of scoliosis in order to avoid a severely debilitating deformity that is difficult to treat.

L.T. Staheli, M.D.

Fig. 4.1 Effective mobility.
Mobility must be practical. A wheelchair may be a more practical means of getting around than difficult walking.

Introduction

The objective of management of lower limb contractures is to help the child become as independent as possible by overcoming the disability and developing an efficient and practical means of mobility. The information presented in this chapter is based on our experience in managing 95 children with amyoplasia and a review of the literature.

Definitions

Effective Mobility

For optimum development, a child requires a means of mobility that is self-directed, practical, and efficient. The means of getting around is not as important as that it occurs at the appropriate developmental age (Fig. 4.1). The capacity for effective mobility is necessary for normal social, psychologic, and intellectual development. A common misconception is that aided mobility, such as using a wheelchair, delays the acquisition of independent walking skills. The child will walk when walking becomes practical.

Levels of Ambulation

Hoffer et al. (1983) have classified walking ability into four levels: (1) community ambulators can walk without aids outside the home, (2) household ambulators walk about home with aids and use wheelchairs in the community, (3) nonfunctional ambulators can only walk with support and aids, such as parallel bars or walkers, and (4) nonambulators are unable to walk in any situation (Fig. 4.2). The ability of functional ambulation depends on many factors, including the severity of lower limb deformity, muscle strength, and degree of upper limb involvement.

Classification	Ability
Community Ambulator	Able to walk about the community
Household Ambulator	Able to walk short distances at home
Nonfunctional Ambulator	Can only walk with assistance
Nonambulator	Requires a wheelchair

Fig.4.2 Levels of ambulation.
This is a commonly used classification of walking ability (Hoffer et al., 1983).

Our Patients with Amyoplasia

Our 95 children with amyoplasia included 55 girls and 40 boys.

Birth History

A birth history was available in 49 patients (Fig. 4.3). The infants were often delivered breech. Some had Streeter's bands, and many had dimpling over bony prominences. Because of the contractures, delivery was often traumatic, and birth fractures were common.

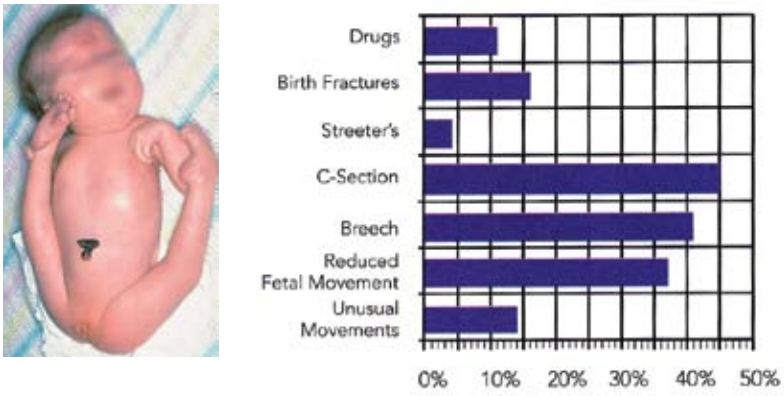


Fig. 4.3 Birth history.
These data were obtained from 49 patients in our series. Breech deliveries were common.

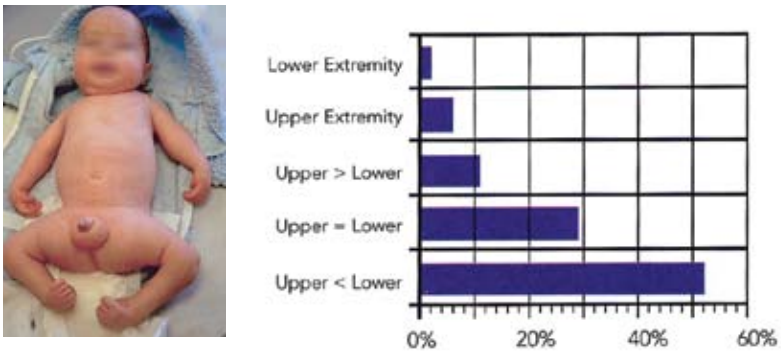


Fig. 4.4 Limb involvement in amyoplasia.
Most infants with amyoplasia have multiple contractures involving both the upper and lower limbs.

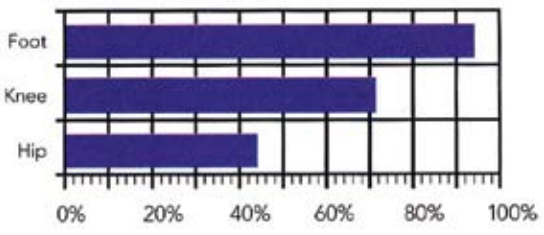


Fig. 4.5 Level of involvement of lower limb.
In our patients with amyoplasia, deformity was more common distal in the limb.

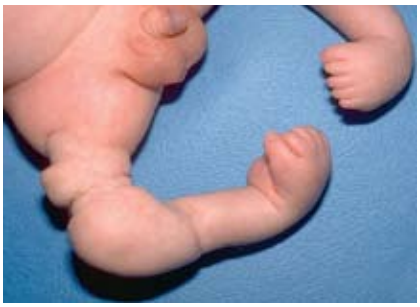


Fig. 4.6 Streeter's ring contractures.
This infant demonstrates multiple Streeter's ring contractures. These were surgically released.

Limb Involvement

Most infants show multiple limb involvement (Fig. 4.4). This extensive degree of involvement emphasizes the need for a comprehensive management plan. Most major lower limb deformities should be corrected during the first year.

Most infants showed foot, knee, and hip deformities (Fig. 4.5). Often the femora were laterally rotated.

Steeter's Ring Contractures

These contractures (Fig. 4.6) are caused by uterine amniotic bands. Such banding may be minimal or so severe as to jeopardize circulation of the limb. Deep bands require operative release. Releases may be performed in a single or a staged procedure.

Dimpling

Deep dimpling is a common feature of amyoplasia (Fig. 4.7). The primary pathology of dimples is a loss of subcutaneous fat. They may result from pressure caused by intrauterine constraint and immobility. Dimples may be excised to improve appearance. They do not alter function.

Fractures

Fractures may occur during delivery (Fig. 4.8) or as a result of manipulation either during examination or more commonly when a joint is ranged. As the joints are contracted and stiff, the bone may fail before any additional joint motion is gained.

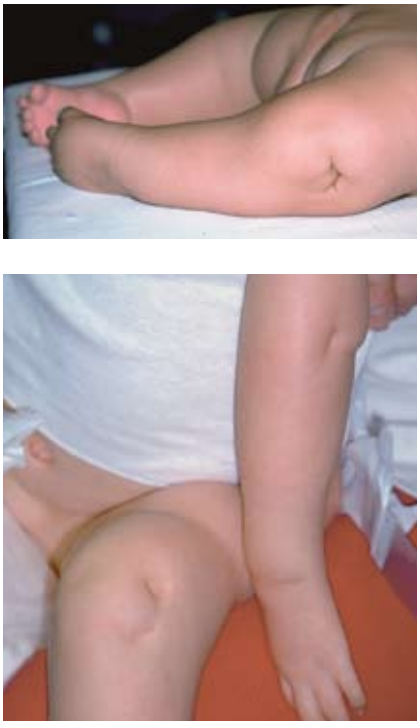


Fig. 4.7 Dimpling.
Dimpling was common and seen most frequently over the elbows and knees.

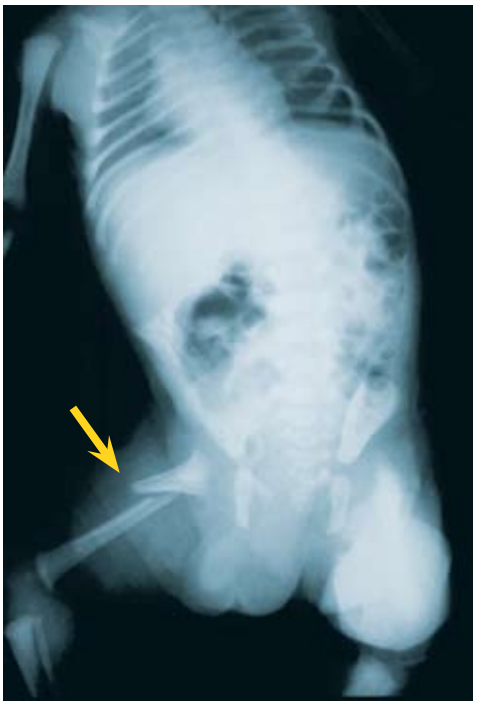


Fig. 4.8 Birth fractures.
This femoral fracture occurred during delivery. Because of the congenital contractures, birth fractures are relatively common in arthrogryposis.

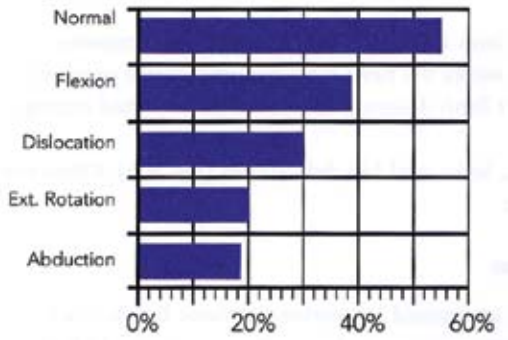


Fig. 4.9 Limb involvement in amyoplasia.
In our series of 95 children with amyoplasia, most had all extremities involved.

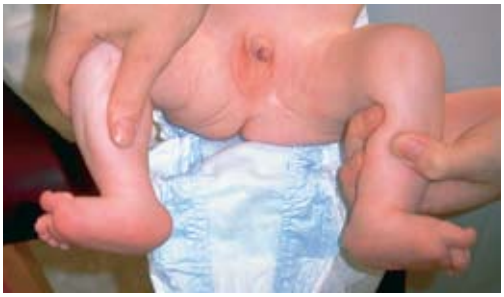


Fig. 4.10 Hip flexion contractures.
This infant has a combination of lower limb contractures which include hip flexion deformity. These deformities resolved without operation.

Fig. 4.11 External rotation deformity.
This child at age 8 shows the typical lateral rotation pattern of the lower limbs. Rarely is the deformity serious enought to require correction. A femoral rotational osteotomy would be required to correct the deformity.

Hip

Various hip deformities occur in nearly half of infants with amyoplasia (Fig. 4.9). Classically the hips are flexed, externally rotated, and abducted. Variable patterns are common.

Flexion Contractures

Hip flexion contracture (Fig. 4.10) is often compensated by increased lumbar lordosis. This compensation requires a mobile lumbar spine. Hip flexion contractures may limit walking (Hoffer et al., 1983), and severe hip flexion contractures may prevent walking. Moderate deformity may make walking difficult or tiring. Delay release of a flexion contracture until it is clear that the child will walk and that the contracture is restricting ambulation. Contractures above 30° may be significant. Contractures exceeding 45° usually require release.

Operative Release

Drape the lower limbs free so a Thomas test can be performed intraoperatively to assess the completeness of the release. Make an oblique incision parallel to the inguinal crease over the sartorius. With care to avoid injury to the lateral femoral cutaneous nerve and femoral vessels and nerves, release the sartorius, rectus femoris, and anterior hip capsule as necessary. Monitor correction by the Thomas test. Continue the release until the contracture has been reduced to at least 10°-20°. Release bilateral contractures during the same anesthesia. As the procedure is relatively minor, correction of other deformities during the same anesthesia may be appropriate. A spica cast may be applied for a period of 2 weeks to allow soft tissue healing. Convert the cast into a night splint and use for an additional 6-8 weeks.

External Rotation Deformity

External rotation contractures (Fig. 4.11) are common and are due to femoral retrotorsion. The arc of hip rotation is rotated laterally with little or no medial rotation. The combination lateral rotation of the hip and medial rotation of the clubfoot compensate one another so the foot faces forward. This places the knee in a laterally rotated position and often results in ranging the knee in the wrong arc, stretching the collateral ligaments.

During growth, the lateral hip rotation gradually becomes less pronounced. I have not found operative correction necessary. A rotational femoral osteotomy would be required for correction.

Abduction Contracture

Abduction deformity is common. The inguinal crease is often displaced to a midthigh level (Fig. 4.12). The hip abductors may be contracted, and radiographs often show a reduction in the neck shaft angle. In rare instances, excision of the extra tissue on the medial aspect of the thigh may be useful to improve adduction and appearance. Should the hip be dislocated, open reduction and this soft tissue excision may be combined.

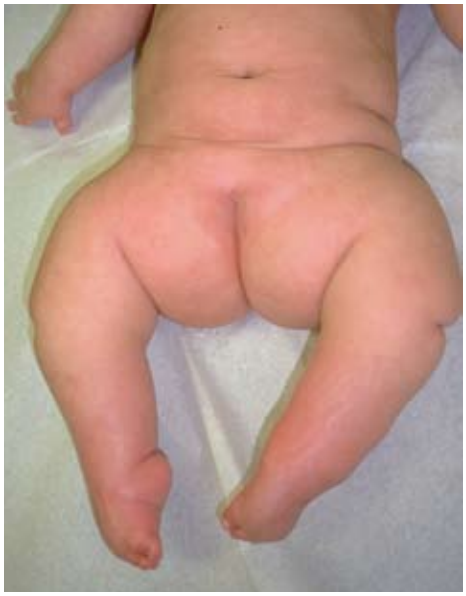


Fig. 4.12 Abduction contracture.
Abduction contractures are common and frequently associated with excessive soft tissue in the groin region.

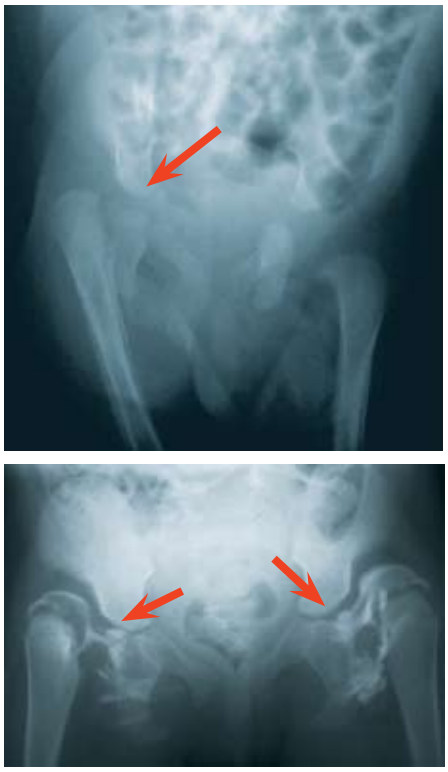


Fig. 4.13 Hip dislocations in amyoplasia.
Hip dislocations occurred in about a third of our patients. The position of the hip sockets is shown by the red arrows. The unilateral dislocations shown in the top picture produce more pelvic asymmetry than bilateral dislocations, shown in the bottom picture.

Hip Dislocations

Hip dislocations (Fig. 4.13) occur in about a third of the children with amyoplasia. Dislocations are congenital and teratologic and can seldom be reduced without surgery. The iliopsoas tendon is severely shortened, and the joint capsule is contracted.

Unilateral Dislocation

Unilateral dislocation causes pelvic and truncal asymmetry, and the need for reduction is not controversial.

Bilateral Dislocations

The appropriatness of reducing bilateral dislocations is controversial. As the children with dislocated hips can walk and have reasonable mobility and little pain, proponents of accepting dislocations believe reduction is unnecessary. In contrast, I believe that reduction improves the quality of gait in both function and appearance. The hips are more stable, and the gait is more efficient.

The poor outcomes following open reduction of dislocated hips were reported using outdated treatment methods. The majority of authors recommend open reduction and femoral shortening osteotomy (St. Clair and Zimbler, 1985; Gruel et al., 1986; Grill 1990). In our experience (Staheli et al., 1987; Szoke et al., 1996), good results can be achieved by a medial approach open reduction if performed during infancy. We adopted the medial approach open reduction based on our experience with the procedure in infants with developmental dysplasia (Mankey et al., 1993). As the approach involves little dissection, can be performed easily bilaterally, and is readily combined with other procedures, we have found it useful in amyoplasia (Staheli et al., 1987). These good results were confirmed by our more recent, larger study (Szoke et al., 1996).

Open Reduction: Ludloff Technique

Of the four approaches to open reduction, the Ludloff approach between the adductor longus and pectineus has several advantages. This approach offers a direct access to the major obstacles to reduction and is still well medial to the femoral vessels and nerve, blood loss is minimal, and the approach is entirely between muscle planes requiring minimal dissection. As the procedure can be performed quickly, bilateral dislocations may be reduced during the same anesthesia.

An arthogram will demonstrate the presence of an acetabulum (Fig. 4.14), but this procedure is usually not necessary.

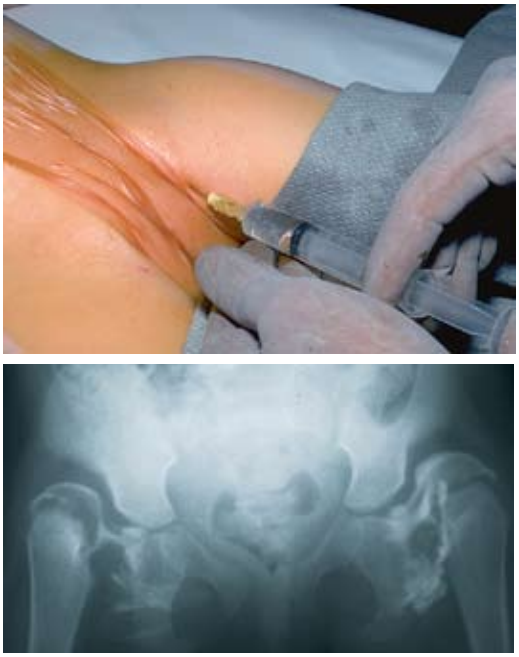


Fig. 4.14 Arthrography.
Arthrogram is performed through a medial approach. The study demonstrates the heads to be high but the acetabulum is present medial to the dye pool.

I have reduced 25 hips in infants with amyoplasia by this approach (Fig. 4.15). Redislocation occurred in 1. This hip was rereduced through an open reduction using the lateral approach. Four hips (16%) developed avascular necrosis. Overall, 80% were considered good, 12% were fair, and 8% were poor. These results are very good when compared with other series of open reduction for teratologic dislocations.

Reductions are best performed early in the first year. Reduction may be effectively achieved by the Ludloff approach until about 24 months of age. Reduction of dislocations is often combined with other procedures. If procedures are combined, perform the hip reduction last, as maintaining the reduction until securely stabilized in a cast is essential.

Technique

The infant is placed on a folded towel to elevate the pelvis. Adhesive plastic is placed to protect the genitalia. We prepare the skin with a 1% solution of iodine in alcohol.

The incision is centered over the lateral margin of the adductor tendon (Fig. 4.16). A 3-cm oblique incision is made that parallels the inguinal ligament. Avoid the long saphenous vein lateral in the incision. Expose the tendon of the adductor longus. Divide the fascia to expose the interval between the adductor longus and pectineus.

Release of the Iliopsoas Tendon

Use finger dissection to find the lesser trochanter. This dissection is easier if the thigh is flexed and laterally rotated. Place retractors to expose the lesser trochanter. Place a small right angle clamp under the tendon (Fig. 4.17), divide the tendon, and allow it to retract. This will allow the capsule to be exposed.

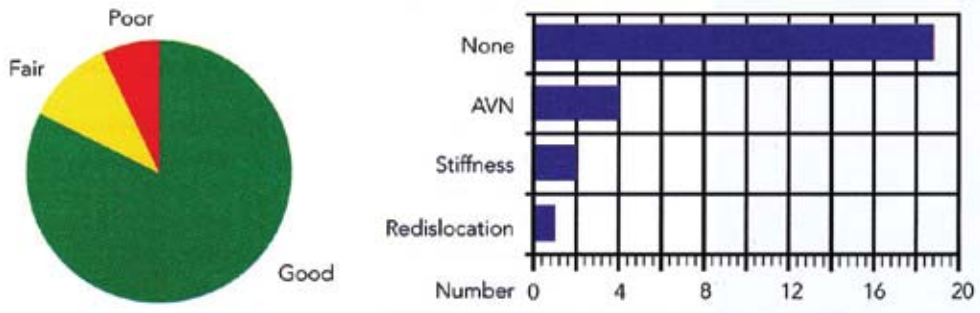


Fig. 4.15 Results of open reduction via the medial approach.
Good results may be obtained by medial approach open reduction. The risks of avascular necrosis, stiffness, and redislocation are acceptable.

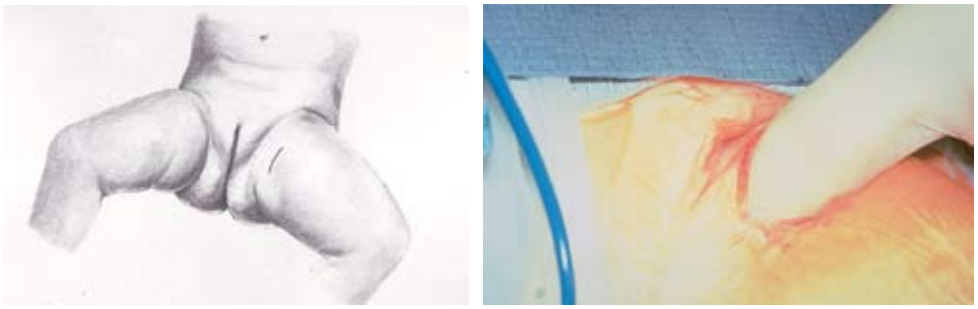


Fig. 4.16 Ludloff exposure.
Through the oblique incision and finger dissection, the interval anterior to the adductor longus is developed to expose the lesser trochanter.

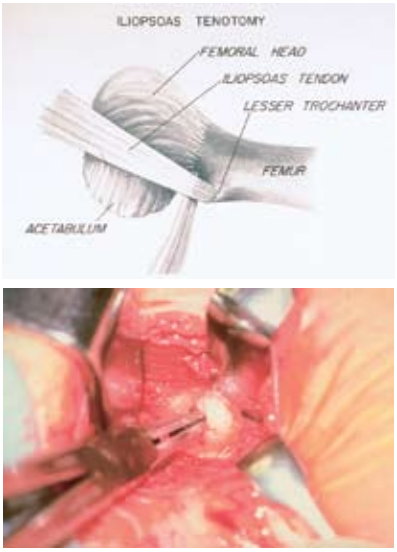


Fig. 4.17 Exposure of iliopsoas tendon.
The iliopsoas tendon is isolated with a curved clamp and divided. It retracts to expose the joint capsule. The tendon reconstitutes itself with time.

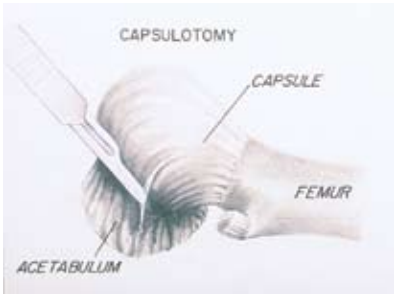


Fig. 4.18 Capsulotomy.
The capsule is divided to release the hourglass constriction. The release is extended medially to include division of the transverse acetabular ligament.

Expose Joint Capsule

Remove the retractors and expose the joint capsule by finger dissection. This is aided by applying some traction on the limb. Replace the retractors and apply traction again to the limb. Further identify and expose the capsule using a Kitner dissector. Make certain the capsule is well exposed at this time.

Capsulotomy

Perform an anterior capsulotomy (Fig. 4.18) and extend it medially to include section of the transverse acetabular ligament. Use a small hook to be certain that the ligament has been completely released. Section the ligamentum teres from the femoral head. Place a clamp on the ligament and follow it to its acetabular attachment. Divide the acetabular attachment to remove the ligament. This steps ensures that the base of the acetabulum has been identified. Remove acetabular fat with a small rongeur.

Reduction

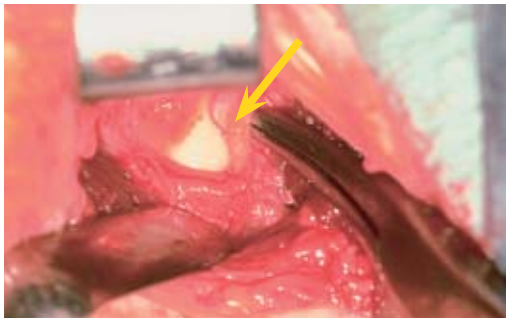
The hip will now reduce (Fig. 4.19). Determine the position of greatest stability. Avoid excessive abduction or forced positioning. Make a radiograph to confirm the reduction and to provide a baseline for comparison with postoperative radiographs to ensure that the reduction is concentric.

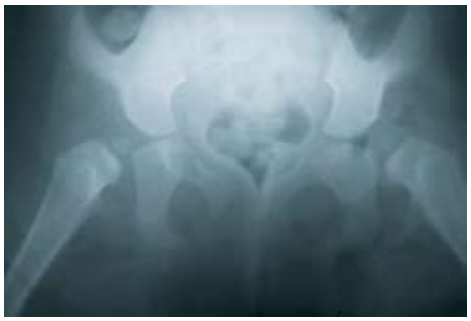
Closure

While maintaining the reduction, the assistant closes the skin with absorbable subcuticular sutures. If the other hip is dislocated, the wound is packed, and the other hip is reduced. Both hips are then reduced and positioned in the safe, stable position by the surgeon while both wounds are closed.

Cast Immobilization

A spica cast is applied while maintaining the reduction (Fig. 4.20). This usually includes the feet. While the infant is still sleep, a radiograph of the pelvis is made and compared with the previous intraoperative radiograph to be certain the hip remains reduced. Further confirmation may be made by a CT scan should the reduction be tenuous. The cast is left in place for 5-6 weeks.





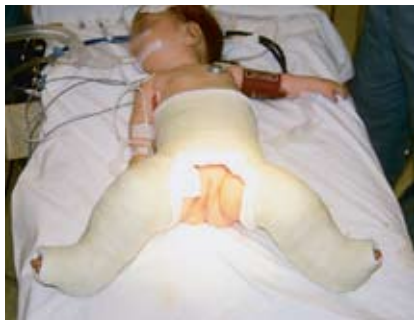


Fig. 4.19 Hip reduction *(left & center).*
Once the hip is reduced, the femoral head is seen (arrow) through the gaping capsulotomy. With the hips oriented in the position of greatest stability, an AP radiograph is made to confirm reduction.

Fig. 4.20 Spica cast immobilization *(right).*
The hips are immobilized in the position of greatest stability with a spica cast. A radiograph is made in the cast and compared with the previous film to be certain the reduction has been maintained.

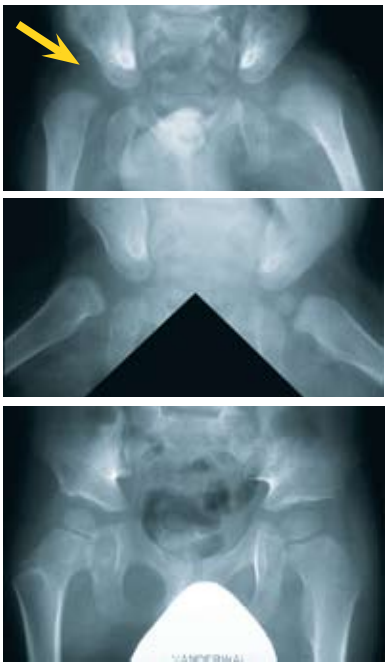


Fig. 4.21 Dysplasia improving with time.
This sequence shows gradual improvement of dysplasia with time. The prereduction radiograph shows dislocation of the right hip (arrow) at 12 months of age. Following reduction, the hip is reduced, but acetabular dysplasia is severe. Four years later, the joint is well formed.

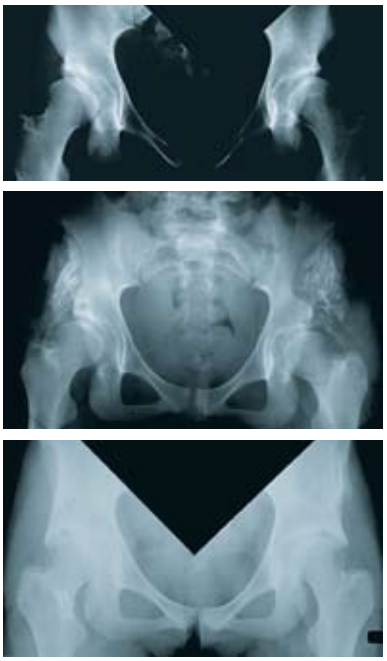


Fig. 4.22 Bilateral acetabular augmentation procedure.
This 15-year-old boy had acetabular dysplasia that was corrected by bilateral acetabular augmentation procedures performed during one operative session.

After Treatment

If clubfoot correction has been performed concurrently, start serial cast correction about 2 weeks after the operation. Remove the foot portion of the cast, manipulate to improve correction, and replace the foot portion of the cast. Continue weekly until correction is satisfactory. At about 5-6 weeks following surgery, remove the spica cast and allow free mobility of the hip. Long leg night splints are made for the feet. Make radiographs at 3, 6, and then at 12-month intervals for the first 3 years. Afterwards, radiographs at 3-year intervals are adequate.

Acetabular dysplasia may be present. This dysplasia often improves with time. My threshold for performing acetabular reconstruction is higher for amyoplasia than for simple developmental dysplasia. The least reasonable intervention is judicious to avoid stiffness.

Open Reduction: Femoral Shortening with or without Pelvic Osteotomy

Femoral shortening combined with anterior open reduction is a standard method of managing dislocated hips in otherwise normal children over about 2 years of age. The femoral shortening relaxes the muscles about the joint and allows reduction with reduced joint compression and less stiffness. This principle has been applied to arthrogryposis. We have found it unnecessary in the young infant, although others have recommended it as a method of reducing most arthrogrypotic hips (St. Clair and Zimbler, 1985; Gruel et al., 1986).

Residual Hip Dysplasia

Often, acetabular dysplasia resolves with growth (Fig. 4.21). In others, the dysplasia persists. Most dysplastic hips remain stable throughout childhood. I recommend that if the hips are stable, wait until puberty before correcting residual dysplasia. For residual dysplasia, determine the site of the major deformity. In most hips, the acetabulum is more abnormal than the proximal femur and is the best site for correction. Correct with a procedure unlikely to produce stiffness. In my experience, acetabular augmentation meets this criterion. As the augmentation is totally extraarticular and does not alter joint pressure, stiffness has not been a problem. If the dysplasia is bilateral, both hips can be corrected during one operative session (Fig. 4.22). Cast immobilization is limited to 6 weeks.

Numerous other methods for correcting residual dysplasia are available, including a variety of pelvic and femoral osteotomies. Each is designed to provide greater hip stability and less deformity.

Knee Deformity

Most amyoplastic infants have knee involvement (Fig. 4.23). In our series, the most common deformity was a flexion contracture (Fig. 4.24). Without normal motion, the knee joint becomes deformed. The femoral condyles flatten in the arc in contact with the upper tibia. Fat and fibrous tissue replace the normal synovial membrane. The joint capsule becomes thickened and contracted. The suprapatellar pouch may be absent. The quadriceps muscles are often hypoplastic or absent, and the muscle is completely or partially replaced with fibrous tissue. This fibrosis reduces the arc of motion of the knee. Knee deformity seriously affects walking ability. Contractures above 20° make walking difficult (Hoffer et al., 1983). A fixed extended knee allows stable standing but makes sitting difficult. If the knee is stiff, a position of about 15° of flexion is the best compromise for both standing and sitting.

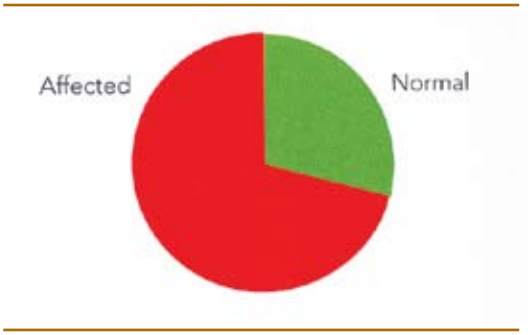


Fig. 4.23 Knee deformity.
Most children with amyoplasia have some type of knee deformity.

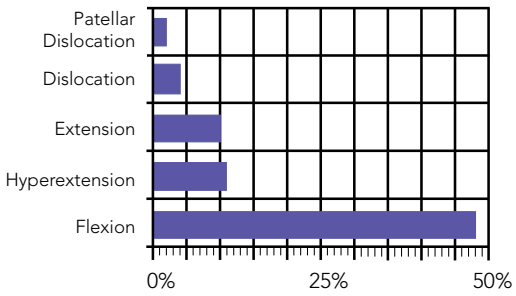


Fig. 4.24 Knee deformities in amyoplasia.
Flexion and extension deformities are common.

Severity	Degrees	Management
Very Mild	0 - 20	No Treatment Required
Mild	20 - 40	Simple Lengthening
Moderate	40 - 60	Stretch Post Op
Severe	60 - 80	Femoral Shortening
Very Severe	80+	External Fixator

Fig. 4.25 Classification of severity.
The severity of the contracture strongly influences the method of correction.

Usually the knee contracture is due to both primary and positional deformities (Chapter 2). Positional deformities improve with time and stretching. Stretching alone, without bracing or casting, is often not successful (Thomas et al., 1985). In most cases, the primary deformity is severe enough to require operative correction, which is most successful in extension deformities (Sodergard and Ryoppy, 1990). Flexion contractures are most disabling and have the greatest tendency to recur.

Flexion Contracture

Flexion contractures were present in nearly half of our infants (Fig. 4.24). These may be classified based on severity (Fig. 4.25). Except for very mild deformity, operative correction is usually necessary. The functional improvement following operative correction is often dramatic. The child, for the first time, becomes ambulatory – an exciting event.

Principles of Correction

Unless the potential for ambulation is uncertain, correct flexion contractures early. Correct the positional component of the deformity by gentle manipulations of the knee into maximal extension with each diaper change. With time, improvement of 10° to 20° often occurs. Correction then may plateau, and further correction requires surgery. Usually, correction is best performed late in the first year. Avoid attempting to correct the flexed knee and clubfoot at the same time. Correction and maintenance of correction of the clubfoot are difficult if the knee is positioned in extension.

The objective of surgery is to place the arc of motion in the most functional position. Because the arc of motion is determined by the fibrosis of the muscles, the arc is not greatly increased by surgery. Stiffness is still common. The family should be prepared for this outcome; otherwise they may be disappointed.

Make certain that the child is standing or has the potential for standing before undertaking correction. If the child can knee stand, the child will benefit from correction (Fig. 4.26).

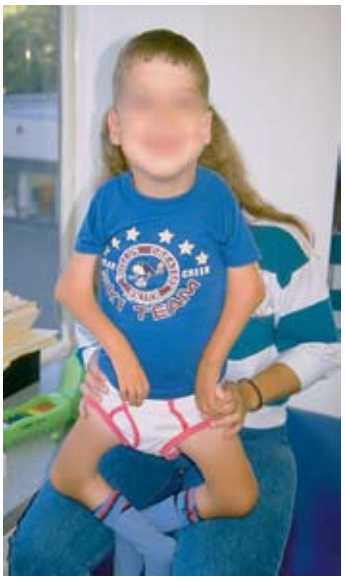


Fig. 4.26 Severe knee flexion contracture.
This 7-year-old boy was unable to walk because of severe contractures. He became ambulatory following correction of these contractures.

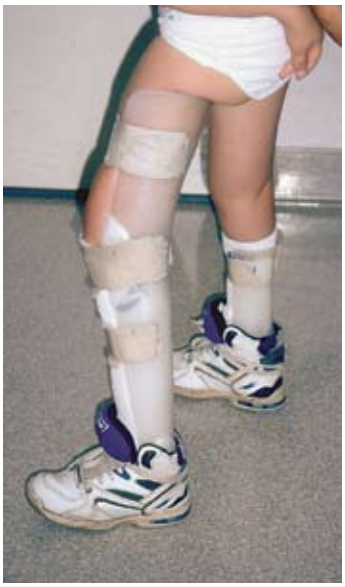
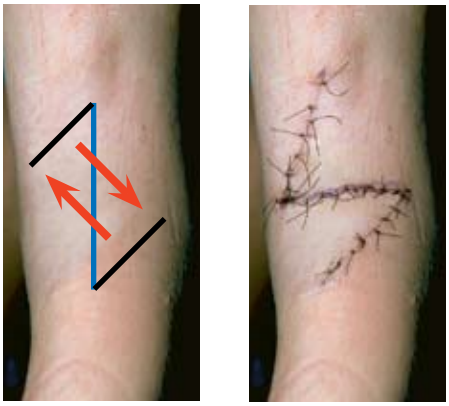


Fig. 4.27 Mild knee flexion contracture.
This child has a contracture of 25°. With a brace, walking is possible.



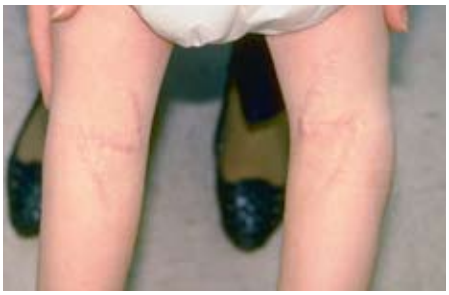


Fig. 4.28 Z-plasty.
The incision is first made with the midsegment of the Z in line with the contracture (blue). The other Z segments are incised at 45°-60°. Make thick flaps. Excellent exposure is obtained. The flaps are reversed (red arrows) for closure. This wound was closed with interrupted sutures.

Operative correction requires lengthening of contracted muscles or simple excision of fibrotic bands. The posterior capsule must be completely opened. Usually after these releases or lengthening, the skin and neurovascular structures are tight and prevent extension of the knee. These structures are best gradually stretched after the skin is healed. This may be accomplished with traction, but serial cast correction is usually more practical. Start serial cast correction about 2-3 weeks following surgery. Change the cast weekly. Once correction is complete, the cast is left in place until soft tissue healing is complete. Employ night splinting to prevent recurrence.

Severity

The details of management are based on the severity of the deformity. Difficulty and complications increase with increasing degrees of contracture.

Very Mild

Very mild flexion (10°-20°) allows the infant to stand and walk. Contractures less than 10° allow nearly normal gait; those between 10° and 20° cause increased energy expenditure for standing. This is usually acceptable.

Mild

Mild contractures (20°-30°) often make walking very fatiguing unless a brace is used (Fig. 4.27). Operative correction is usually appropriate to allow brace-free walking.

Correct during the first year. Usually, correction of both knees is done during the same operative session. Avoid combining correction of knee flexion contractures and clubfoot during the same procedure since clubfoot correction requires postoperative immobilization with the knee flexed.

Place the infant in the prone position. A transverse incision is made across the popliteal region. This may be extended proximally or distally, converting the incision into an S if additional exposure is required. Avoid the saphenous vein and posterior sural nerve. Expose the neurovascular structures. Lengthen the gracilis and semitendinosus by Z-plasty. Lengthen the semimembranosus and biceps by aponeurotomy. Sometimes, the origin of the gastrocnemius requires release. Expose the posterior joint capsule on both sides of the neurovascular bundle. Determine the level of the joint by palpation while flexing the knee. Divide the capsule transversely. The knee should then freely extend, limited only by the popliteal nerve and artery. Note the degree of extension that is just short of making these structures excessively tight. This will be the initial position of immobilization in the cast. Following closure, apply a long leg cast in maximal safe extension. If the knee is not fully extended, achieve the final correction by weekly cast changes starting 2 weeks following surgery. The infant may stand and walk in the casts. After 6 weeks, remove the cast, make night splints, and allow free movement during the day. A nonarticulated bracing may be necessary to stabilize the knee for walking.

Moderate

Correct moderate deformity (40°-50°) with the same approach except for the skin incision. A single large Z incision (Fig. 4.28) provides excellent exposure and allows immediate lengthening of the skin. The disadvantage is the appearance of the scar. As the scar is behind the knee, it is not very noticeable (Fig. 4.28 bottom).

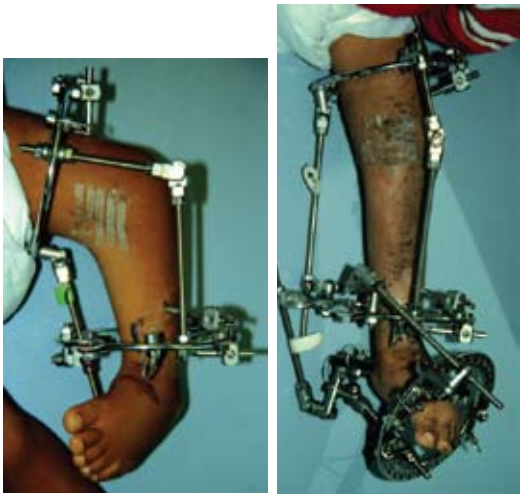


Fig. 4.29 External fixator correction.
This 4-year-old boy with amyoplasia has 80° knee flexion contracture and a recurrent clubfoot deformity. (Left) Before correction with external fixator. (Right) Following correction of both the knee and foot deformity. Note that the fixator has been lengthened to include the foot. Courtesy of Dr. Vincent Mosca.

Severe

Correction of severe contractures (50°-80°), in addition to the release as described for moderate deformity, usually requires gradual correction with an external fixator (Fig. 4.29) or femoral shortening (Fig. 4.30). Correction makes walking possible, whereas, before correction, even with bracing, walking is not possible.

Very Severe

Correction to full extension may not be possible for some very severe deformity (80+°). This is in part due to the severe intra-articular deformity. Correct by releasing the contractions and applying an external fixator. Correction is then achieved gradually over a period of weeks.

Most children require bracing following correction. Order a nonarticulated, lightweight plastic orthosis.

Correction by Osteotomy in the Growing Child

Correction of deformities by osteotomy in the growing child is usually followed by recurrence. Just as fractures remodel, osteotomies that change bone alignment tend to recur with time and growth. This recurrence occurs at the rate of about 1° per month (DelBello and Watts, 1996). If the child is severely disabled by deformity and soft tissue correction is considered unsafe due to extensive scarring or unlikely to be effective because of severity, osteotomy may be accepted as the only method of restoring function. The family must be prepared for recurrence and reoperation. The decision to correct must weigh the risks of the osteotomy against the functional improvement for a limited period of time.

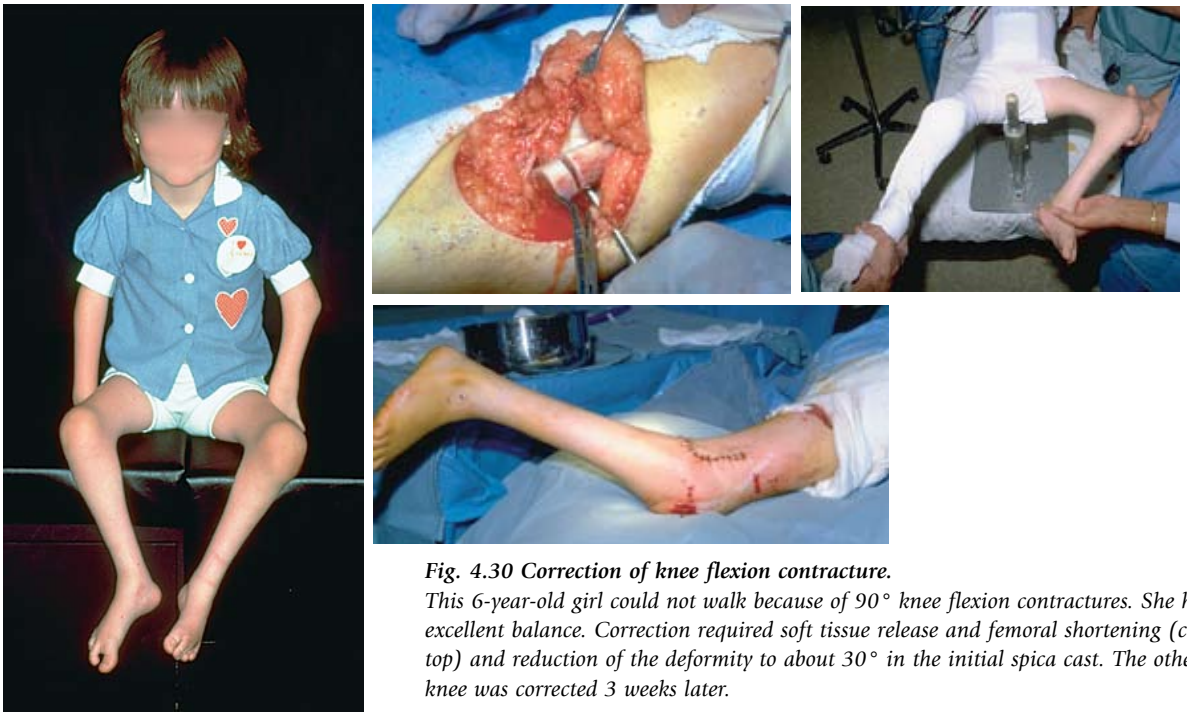


Fig. 4.30 Correction of knee flexion contracture.
This 6-year-old girl could not walk because of 90° knee flexion contractures. She had excellent balance. Correction required soft tissue release and femoral shortening (center top) and reduction of the deformity to about 30° in the initial spica cast. The other knee was corrected 3 weeks later.

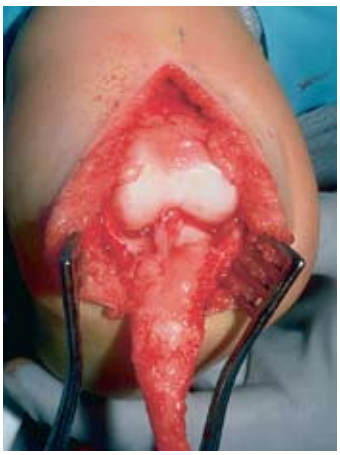


Fig. 4.31 Quadricepsplasty.
The quadriceps has been divided in an inverted V in preparation for lengthening.

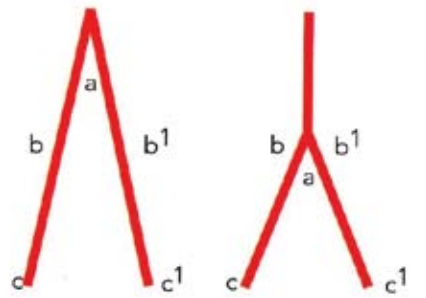


Fig. 4.32 V-Y-plasty.
Lengthening of the quadriceps is achieved by this technique. The inverted V is converted to an inverted Y, and in the process the soft tissue is lengthened.

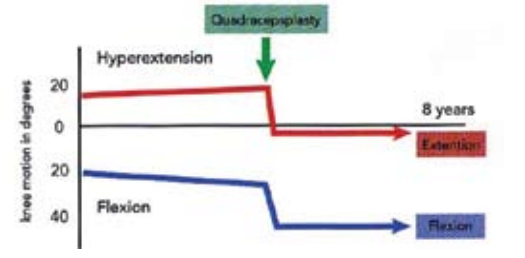


Fig. 4.33 Effects of quadricepsplasty.
This sequence shows the change in the arc of knee motion into a more functional position. The 40° arc of motion is not increased. Walking and especially sitting are improved with the increased flexion achieved.

Correction in the Older Child

Employ femoral extension osteotomy at the end of growth to correct recur rent or persisting deformity. As this procedure is frequently complicated by neurovascular compromise, shorten the femur to avoid excessive tension on the popliteal artery.

Knee Extension Contracture

About 20% of children with amyoplasia have extension or hyperextension deformities of the knee. The deformity is often bilateral, although one side may be more severe. Pathology includes shortening of the quadriceps tendon, a tight anterior capsule, and hypoplasia of the suprapatellar bursa. Valgus deformity of the knee is common.

Hyperextension deformity usually requires correction, whereas simple extension deformity may be acceptable, as the knee is stable and the child can walk. The objective is to optimize the child's ability to walk and sit.

Neonatal Period

The deformity is most pronounced at birth. First, correct the positional component of the deformity by gentle ranging of the knee into the maximum degree of flexion. It may be difficult to determine the true axis of joint motion. The knee is small and deep in subcutaneous fat, and the patella is difficult to palpate. Stretching of the collateral ligaments rather than the quadriceps contracture is a risk. Be aware of the lateral rotation of the femur, and adjust the arc of stretching accordingly. Carefully evaluate the hips, as dislocations are more common in infants with hyperextended knees.

Infancy

Correct unacceptable residual extension deformity between 3 and 6 months of age. Combine correction with open reduction of the hips. Correction is also easily combined with correction of clubfeet. The knee flexion gained by operation is helpful in maintaining the rotational correction of the clubfoot deformity.

Operative Technique

For bilateral procedures, mark the incision sites to be certain that the scars are symmetric in position and length. Make a vertical incision centered over the superior pole of the patella. Deepen the incision directly to the quadriceps fascia. Expose the quadriceps and the patella. Incise the fascia with a long inverted V incision. Attempt to avoid cutting muscle fibers. Reflect the base of the V and patella to expose the joint (Fig. 4.31). Remove obstructing joint contents. Often the lateral patellar retinaculum must be incised to flex the knee. Place the knee in the maximum degree of flexion desired and repair the quadriceps fascia in a Y fashion (Fig. 4.32). Modify the quadriceps reconstruction to position the quadriceps in the most functional position. Close the skin with subcuticular absorbable sutures. Immobilize in the midrange of the arc of flexion achieved after lengthening. If postoperative bracing is necessary, remove the cast at 3 weeks, make the mold for a brace, and then reapply a cast. Plan for the orthoses to be completed and available when the cast is removed at 5 weeks. Night splinting is not required following correction of extension deformity.

Lengthening of the quadriceps usually just repositions the arc of motion. The arc of motion is usually not increased. The objective is to place the arc in the most functional position (Fig. 4.33). The knee is usually best positioned in about 10° of flexion.

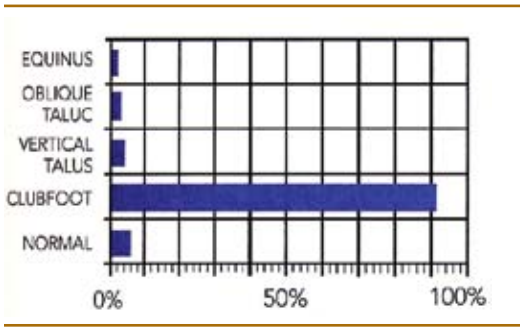


Fig. 4.34 Incidence of foot deformities in amyoplasia. *Over 90% of the subjects have clubfeet.*

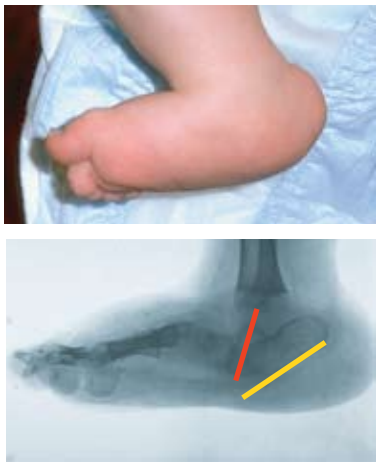


Fig. 4.35 Vertical talus.
The foot has a convex plantar surface. Radiographs show a near vertical orientation (red) of the talus and plantarflexion (yellow) of the calcaneus. The talohorizontal angle is 70°.

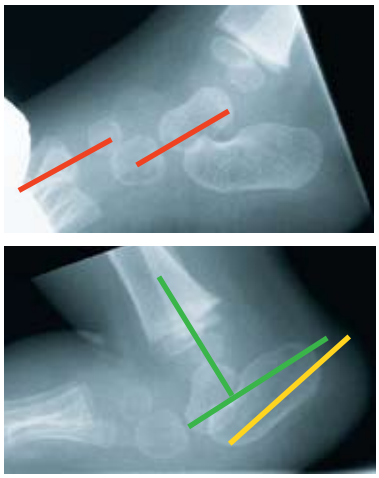


Fig. 4.36 Stiffness of the vertical talus.
(Above) Plantarflexion radiograph shows a failure of reduction of the midtarsal joint (red). The lines fail to align. (Below) Dorsiflexion radiograph shows a failure of the calcalneus (yellow) to dorsiflex. The perpendicular lines show the axis of the tibia (green).

Foot deformity occurs in the majority of infants with amyoplasia (Fig. 4.34). Correction of these deformities is essential regardless of the anticipated ambulatory level. Even in nonambulatory patients, a deformed foot makes shoeing difficult, positioning of the foot on the wheelchair foot rest uncomfortable, and the appearance unacceptable.

Vertical and Oblique Talus

The vertical and less severe oblique talus deformities are not common and are managed like those seen in infants with spina bifida or who are otherwise normal. For this reason, the evaluation and management are presented only briefly.

Diagnosis

The oblique talus is plantarflexed beyond the normal range but is flexible. In contrast, the foot with a vertical talus is stiff. Both the anterior and posterior musculature is contracted, producing a midfoot dislocation. The calcaneus is flexed and limited in dorsiflexion. The talonavicular and calcaneocuboid joints are subluxated or dislocated, and plantar flexion of the forefoot is limited.

The diagnosis is suggested by a prominence of the talar head in the sole of the foot (Fig. 4.35). Lateral radiographs show a vertical orientation of the talus with an increased talar-metatarsal angle. The rigidity is confirmed by lateral radiographs of the foot taken in maximum flexion and extension (Fig. 4.36). These demonstrate a failure of the midfoot to reduce on plantar flexion and a lack of dorsiflexion of the calcaneus with dorsiflexion of the foot.

Treatment

The oblique talus does not require treatment. It tends to improve with time and is unlikely to cause any disability.

The vertical talus requires correction. You may try to reduce the midtarsal dislocation by casting the foot in plantar flexion. Unfortunately, this procedure is usually not successful. Correct in a single stage (Ogata et al., 1979) between 3 and 6 months of age. The procedure is easily combined with correction of other deformities during the same anesthesia.

Operative Technique

The infant is positioned with the affected limb laterally rotated. The Cincinnati incision provides excellent exposure. The neurovascular bundle and lateral sural cutaneous nerve are identified, mobilized, and protected. Divide the posterior tibial tendon 2 cm from its attachment. Open the talonavicular joint to expose the head of the talus. Rarely, the deformity is so severe that naviculectomy is required to align the hindfoot and midfoot. In most feet, reduction of the talonavicular joint is possible (Fig. 4.37). This is facilitated by the introduction of a 1.6-mm smooth K-wire through the

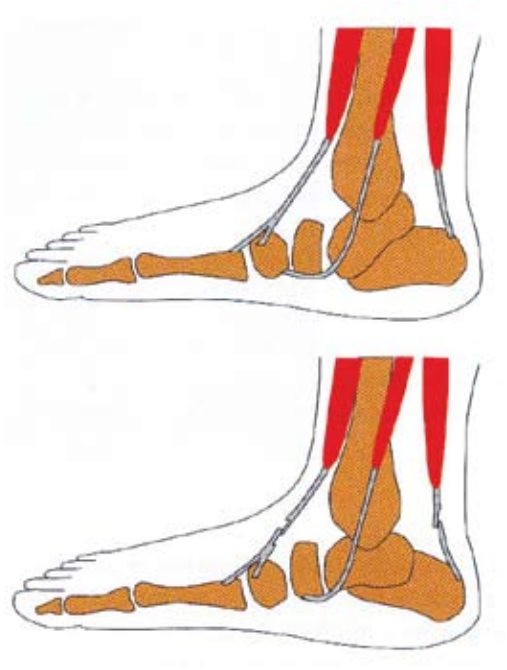


Fig. 4.37 Operative correction of the vertical talus.
The contractures of the triceps and extensors have been lengthened. The talonavicular joint has been reduced, and the posterior tibial tendon has been rerouted to support the navicular.

Fig. 4.38 Clubfoot severity spectrum.
Clubfeet fall into a spectrum of severity. Arthrogrypotic feet (right) are at the severe end of the spectrum.

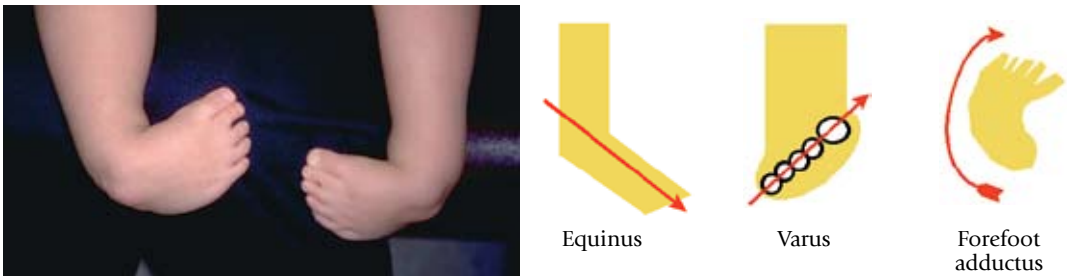


Fig. 4.39 Clubfoot components.
The equinus, varus, and forefoot adductus components of the clubfoot are illustrated.

head and into the body to provide a means of manipulating the talus. Lengthen the Achilles tendon by Z-plasty. Open the posterior joint capsules of the ankle and subtalar joints. Reduce the talonavicular joint and fix the reduction with a second 1.6-mm smooth K-wire placed through the body of the talus, navicular, and first metatarsal to exit adjacent to the great toe. Bend the wire at right angles and leave it outside the skin.

Repair the posterior tibialis in a shortened position and the tendoachilles in a lengthened position. Lengthen the anterior tibialis and toe extensors as necessary. Close with subcuticular sutures. Place in a long leg cast with the foot in a neutral position. Continue immobilization for 6 weeks. Remove the wire in clinic. Continue with night splinting. Anticipate recurrence serious enough to require reoperation in 30%-50% of feet.

Clubfoot

Clubfoot deformity occurs in more than 90% of infants with amyoplasia and is common in other forms of arthrogryposis. Clubfoot is also one of the most disabling deformities, as if it is uncorrected, it will cause severe disability. Management of clubfoot is challenging, as the feet are rigid, and the condition tends to recur. It occupies the extreme end of the clubfoot severity spectrum (Fig. 4.38).

The clubfoot deformity, also referred to as talipes equinovarus, includes several components (Fig. 4.39). Medial rotation is a prominent feature of clubfeet. This is due to medial deviation of all of the elements of the foot from the subtalar joint to the tarsal metatarsal areas. Medial rotation is not due to medial tibial torsion.

Equinus is severe and includes a contracture of all of the posterior musculature. In addition, the posterior capsules of the ankle and subtalar joints are shortened.

The hindfoot is in varus position, with the calcaneus positioned medially under the talus. The forefoot is adducted and supinated.

The foot and lower leg are smaller than normal (Fig. 4.40). The hypoplasia is proportional to the severity of the deformity. It is most pronounced in the foot. For children with unilateral clubfoot, shoes of different sizes may be required. The hypoplasia of the calf is a feature of the disease and not of treatment. The parents should be made aware of this feature of clubfoot. It will be of greatest concern to the patient during adolescence. Limb shortening is mild and is not severe enough to require correction.

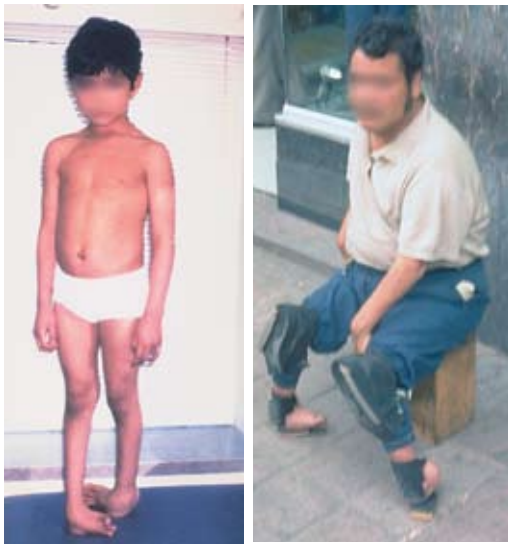


Fig. 4.40 Limb hypoplasia.
These photographs show the hypoplasia of the calf of untreated individuals. Left, a pubescent child and, right, an adult with amyoplasia showing calf hypoplasia and severe disability from the uncorrected clubfoot deformity.

Management Overview

The flowchart (Fig. 4.41) provides an overview of management. Management is complicated by the tendency for the clubfoot to recur (Drummond and Cruess, 1978; Williams, 1978). The risk of recurrence continues throughout the period of growth but is more pronounced in infancy. As an objective of management is to perform the least number of operative procedures, delay correction of recurrent deformity until the disability becomes unacceptable. A maximum of three procedures is required.

Diagnosis

The clubfoot deformity is readily recognized. Separation of amyoplasia from the other congenital contracture disorders is the major challenge. Management of all of the congenital contracture group generally will follow this same general management scheme.

Cast Correction

Cast correction of most clubfeet is started soon after birth. It is important that this process does not interfere with bonding. Make certain that the parents are comfortable holding and interacting in a normal fashion with the infant. Ask the parents to gently stretch the foot with each diaper change. This gentle stretching is continued for about 15 minutes per session. The cast is applied with the parent comforting the infant. A bottle or feeding helps. Casting is stressful for the family. It consumes their energy and resources. It should not be continued if improvement has plateaued.

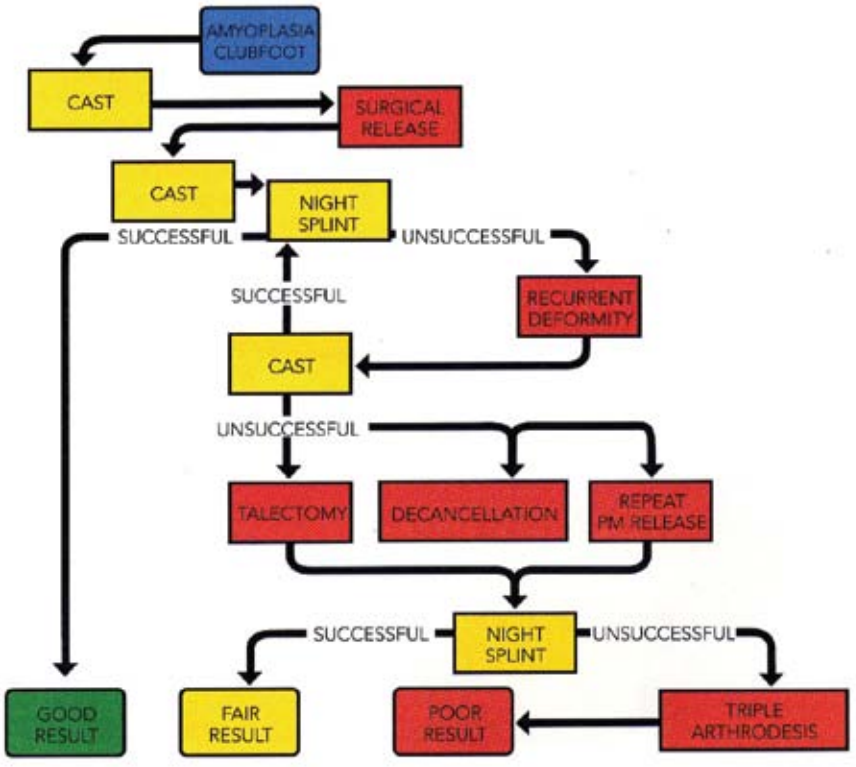


Fig. 4.41 Clubfoot management flowchart.
This is a general scheme for clubfoot management in amyoplasia.

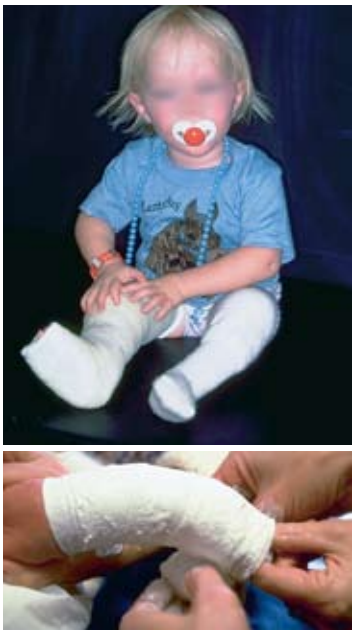


Fig. 4.42 Cast management.
Plaster casts are useful in the presurgical management and in correcting recurrent deformity. Plaster casts can be removed by soaking in water. Casts should extend above the knee to control the rotational component of the clubfoot.

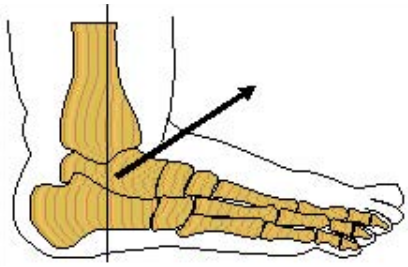


Fig. 4.43 Talectomy.
Removal of the talus is an alternative method for primary correction.

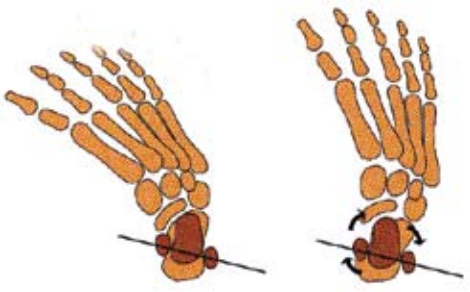


Fig. 4.44 Hindfoot correction.
The typical clubfoot deformity as seen in the transverse plane on the left. With release of the subtalar joint, rotation correction may occur between the talus-tibial unit and the calcaneus, navicular, and rest of the foot (right).

Apply the cast with the foot held in a position of maximum correction (Fig. 4.42). A long leg cast is most effective in correcting the medial rotation component of the deformity, laterally rotated relative to the thigh. The long leg component may also help correct knee flexion deformity at the same time. Change casts at 1-2 week intervals.

Continue cast applications as long as progress is being made. Progress can be assessed either clinically or by radiography. Casting very rarely achieves adequate correction. It reduces the contracture and stretches the skin in preparation for operative correction.

Choice of Primary Procedure

A major decision in management of the arthrogrypotic clubfoot is choosing between soft tissue release procedures and talectomy (astragalectomy). Although most published reports favor talectomy (Menelaus, 1971; Hsu et al., 1984; Guidera and Drennan, 1985; Solund et al., 1991), a recent poll at a European Pediatric Orthopedic Society meeting revealed that most performed a posterior medial release primarily. Talectomy was reserved as a backup or salvage operation.

Talectomy relaxes the contracture of the hindfoot and midfoot, allowing immediate plantigrade positioning of the foot (Fig. 4.43). The normal ankle joint is lost, and recurrence following talectomy is difficult to manage as the primary salvage procedure has already been performed.

The choice between primary talectomy and soft tissue release remains unsettled. I recommend that this choice be made based on the severity of the clubfoot, the preference of the family and surgeon, and the practicality of a continuous postoperative night splinting program.

Skin Expanders

The preoperative subcutaneous placement of a balloon to gradually stretch the skin has had mixed success (Buebendorf et al., 1992) and is probably rarely appropriate.

Primary Soft Tissue Release

There are many approaches to clubfoot surgery. Releases on the medial, posterior, and lateral aspects of the foot are usually required. McKay's correction of subtalar rotational deformity (Fig. 4.44) is a useful principle to employ in these stiff feet.

The optimum position for surgery depends on what procedures are being combined. If only the feet are to be corrected, position the infant prone. The surgeon and assistants can be seated.

A transverse incision is made (Fig. 4.45) just proximal to the posterior skin crease.

This surgery differs from traditional clubfoot correction only in that it is more extensive. First, isolate and protect the neurovascular bundle medially and the lateral sural cutaneous nerve (Fig. 4.45). Lengthen the heelcord, posterior tibialis, toe flexors, and adductor of the great toe. Lengthen the flexor to the great toe by a percutaneous tenotomy at the MTP joint (Fig. 4.46). Open the posterior ankle and subtalar and talonavicular joints. Release the spring and calcaneofibular ligaments. Repair the skin with interrupted nylon sutures. Keep the procedure brief and the tourniquet time less than 60 minutes.

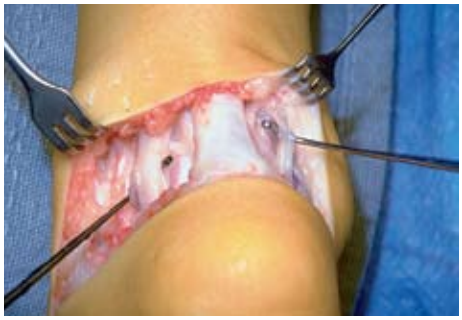


Fig. 4.45 Posteriomedial-lateral release.
This intraoperative photograph of the back of the foot shows the excellent exposure provided by the Cincinnati incision. The plantar and lateral sural cutaneous nerves are identified.

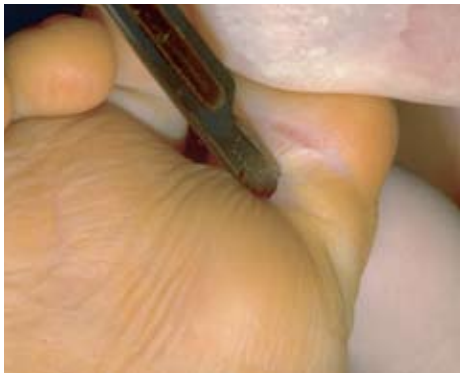


Fig. 4.46 Percutaneous lengthening of the flexor hallicus tendon.
The tendon is divided in its sheath.

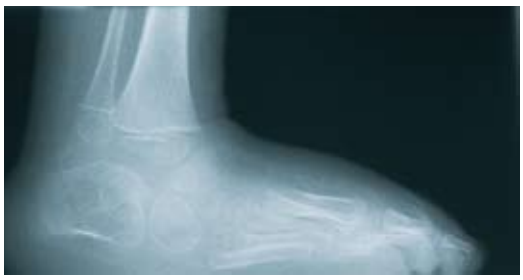


Fig. 4.47 Radiograph after talectomy.
The child had a talectomy to correct recurrent deformity. The foot is plantagrade but stiff.

Apply a well-padded cast with the foot in a position of maximum ankle extension that will not place undue tension on the skin. Usually, this is in neutral or slight plantarflexion. Extend the cast above the knee to control rotation and reduce the risk of the cast being kicked off by the infant. At 2-3 weeks following surgery, reapply the foot portion of the cast to achieve more correction. Change the cast weekly until the foot is plantigrade. Remove the sutures only after the correction has been achieved to avoid dehiscence of the wound.

Talectomy

Removal of the talus allows correction of equinus and hindfoot varus. The talus is excised through an anterolateral approach. Be certain to remove the entire bone. The foot should be displaced posteriorly and fixed with a longitudinal K-wire to hold the foot in the proper position during healing. Immobilize in a cast for 6 weeks. Night splinting is essential, as recurrence is not uncommon. Recurrence following talectomy is difficult to manage and is best prevented. Most surgeons reserve talectomy (Fig. 4.47) as a means of salvaging the foot following recurrence after soft tissue release procedures.

Night Splinting

As clubfoot in amyoplasia tends to recur, night splinting is essential. For very severe feet, postoperative splints should be worn during part of the day as well as at night for a period of several months (Fig. 4.48). In less severe deformity, only nighttime splinting is required. Night splints should be continued as long as the tendency for recurrence remains. Night splinting throughout infancy and early childhood is usually necessary. Although night splinting is a bother, it is best for the child, as it frees the child of the need for daytime bracing. When possible, avoid daytime bracing, as braces hamper play and only reinforce the message that the child has a disability.

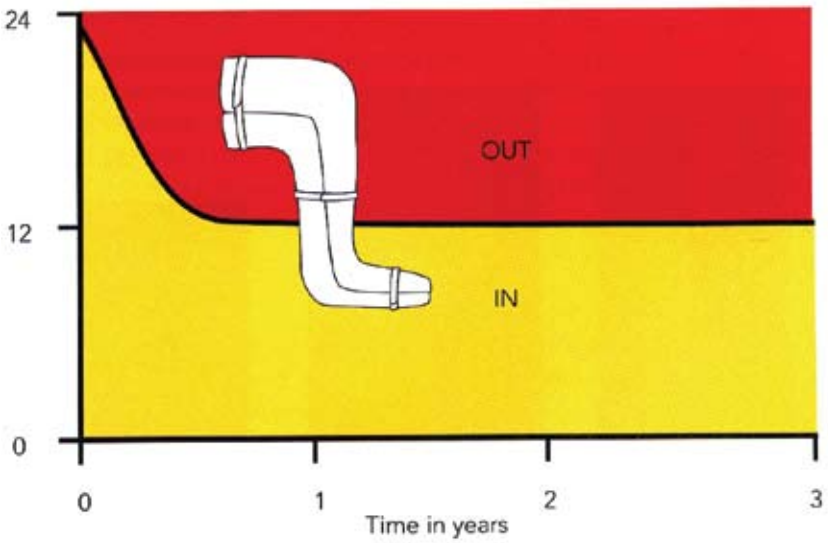


Fig. 4.48 Night splint use.
The use of postoperative splinting is tapered during the first few months after surgery from full-time to nighttime use. Splinting is continued at night for several years.

Fig. 4.49 Fabrication of night splints.
Night splints are made of fiberglass or plaster, cut into front and back halves, and lined with foam for padding.

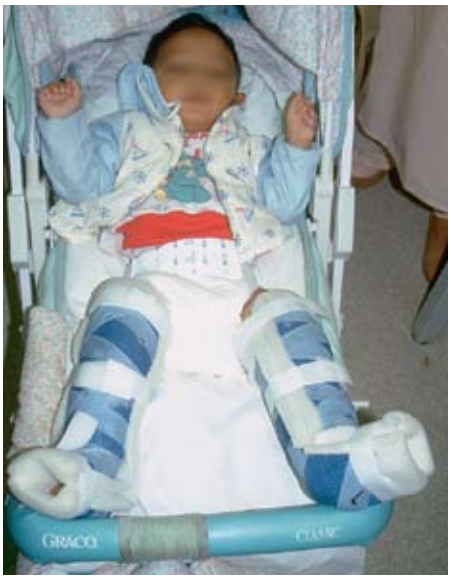
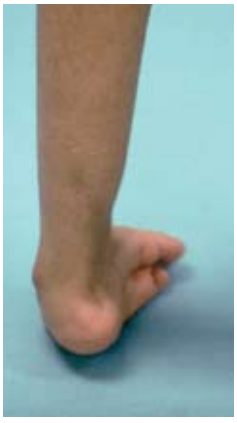


Fig. 4.50 Clubfoot splinting.
This photograph shows a child with the night splints applied.



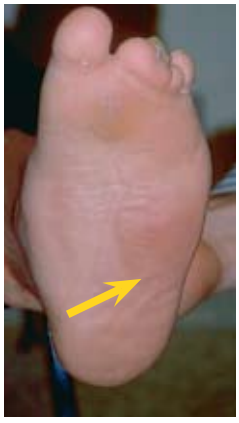


Fig. 4.51 Recurrent deformity.
Recurrent deformity usually includes varus and equinus. The base of the fifth metatarsal becomes prominent and often a source of discomfort (arrow).

Fabrication of Night Splints

Make night splints from long leg casts. The foot should be positioned in maximum dorsiflexion, valgus, and lateral rotation. We use a fiberglass cast lined with foam (Fig. 4.49). The splints should be comfortable (Fig. 4.50).

Recurrent Deformity

Recurrent deformity (Fig. 4.51) sometimes occurs even when night splints have been consistently applied by the parents. In most cases, recurrence usually follows some problem in the splinting program. Most early recurrent deformity can be corrected or substantially improved with a series of long leg casts (Fig. 4.52). Change the casts every other week. Continue casting as long as correction is being achieved. Discontinue casting when the deformity has been corrected, progress has plateaued, or the stress is just too great for the family. Following correction, reinstitute a vigorous night splinting program.

Recurrent deformity usually causes a prominence over the base of the fifth metatarsal with calluses and often discomfort. If recurrent deformity is not corrected by casting and is causing discomfort, treatment is necessary.

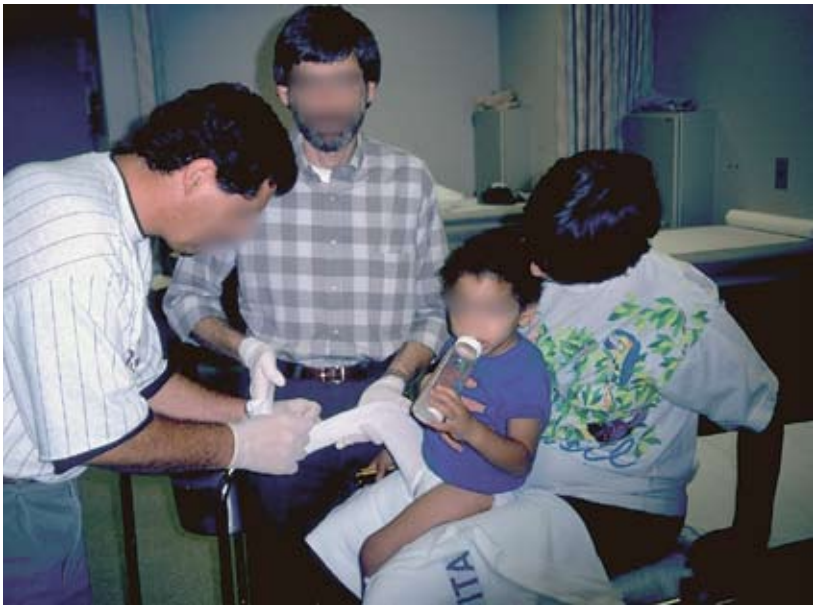


Fig. 4.52 Cast correction of recurrent deformity.
Recurrent deformity can usually be improved by casting. Hold the foot in dorsiflexion and lateral rotation while the long leg cast is applied.

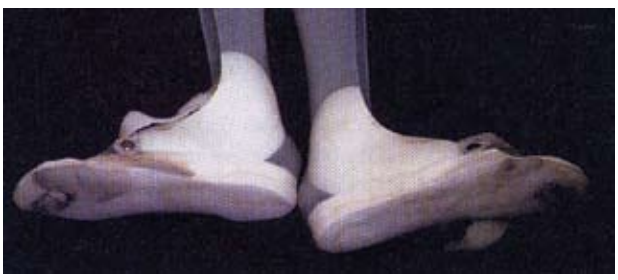


Fig. 4.53 Braces with relief over bony prominence.
The use of molded braces or inserts may unweight the areas of excessive loading and reduce discomfort.

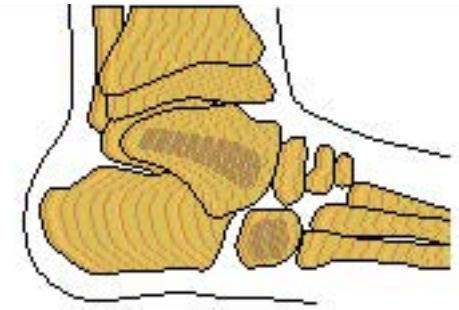


Fig. 4.54 Decancellation.
Removal of the bone of the talus and cuboid allows correction by manual manipulation of the foot.

Fig. 4.55 Triple arthrodesis.
Removal of joint cartilage and bone wedges allows correction of even severe deformities.

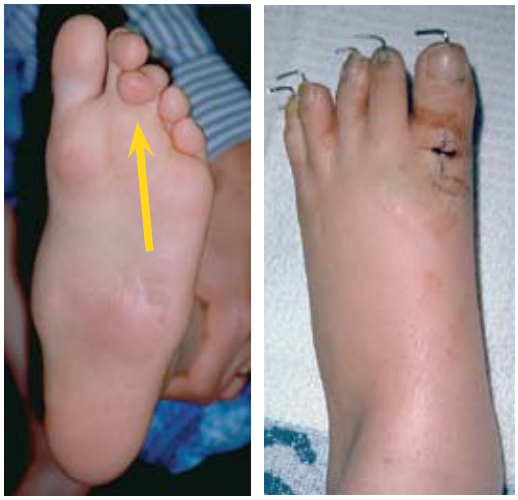


Fig. 4.56 Toe deformities.
Toe deformities are common and, if severe, require surgical correction. Temporary use of fixation pins is often necessary to hold the corrected position of the toes until healing occurs.

To avoid an excessive number of procedures, try to delay corrective surgery as long as possible. Orthotics or molded AFO are useful to relieve the areas of excessive loading with reduction in discomfort (Fig. 5.53). Make certain that the orthotist is aware of the need for molding which is rather extreme. Often only minimal relief is provided and the pain remains. Operative correction is indicated if the disability cannot be managed by the molded orthosis or if the child is at the end of growth, and significant deformity is present.

Secondary Operative Procedure

Talectomy

If the first procedure was a soft tissue release, removal of the talus is a good alternative in the young child.

Repeat Soft Tissue Release

Redo of soft tissue releases is very difficult in arthrogryposis. The soft tissue is contracted and ungiving. The neurovascular structures are encased in scar. It is usually best to avoid this procedure unless the deformity is unusually mild and the foot is flexible.

Chondroplasty

Chondroplasty is sometimes referred to as a *soft tissue triple*. An anterolateral-based wedge of bone and cartilage is removed from the foot to correct the deformity. The procedure is basically the same as the triple arthrodesis but removes cartilage as well as bone rather than only bone, as in the triple arthrodesis. This procedure is effective and may be performed in the growing child.

Decancellation

Removal of bone from the antero-lateral aspect of the foot may be performed with a curet. This is a well-established method of reducing the width of the cuboid. Combining a decancellation of the cuboid and talus is referred to as the Verebelyi-Ogston procedure (Fig. 4.54) (Gross, 1985).

Triple Arthrodesis

Removal of bone and articular cartilage from the subtalar and midtarsal joints (Fig. 4.55) is referred to as a triple arthrodesis. The procedure is one of the oldest and most effective foot operations in orthopedics. Nearly any deformity can be corrected. The procedure is best performed after the age of 10 years, when the majority of foot growth is completed. The procedure shortens the foot, eliminates any subtalar motion, and limits subsequent growth. This is the procedure of choice for correcting severe residual deformities during late childhood and adolescence.

Toe Deformity

Flexion deformity of the toes is common during late childhood and adolescence. The toe often is stiff, making wearing shoes difficult and walking uncomfortable. Severe deformity requires operative correction (Fig. 4.56) by tenotomy or bone excision, or both. Fix with smooth K-wires, and protect with a short leg cast.

K.M. Jaffe, M.D.

Fig. 5.1 Goals.
Rehabilitation focuses on functional activities.

Introduction

The characteristic features of arthrogryposis (limitation of movement of two or more joints in different body areas) can and usually do result in the dramatic reduction of a child's ability to function at an age-appropriate level. Although orthopedic management can improve the underlying deformity, by itself it is not sufficient to expand a child's functional capacity or improve his or her functional performance. Rehabilitation, provided in concert with traditional pediatric and orthopedic care, addresses a broad set of common issues that relate to the functional performance of activities of daily life. These issues focus on the whole child and the child's interaction with home, school, and community environments as well as society at large. They include not only health-related concerns about physical functioning, but also the domains of psychologic, emotional, social, educational, and vocational development. This chapter provides a framework for the next three chapters by introducing the goals, services, strategies, and principles of rehabilitation.

Goals of Rehabilitation

The principal objective of rehabilitation is to facilitate and promote maximal independent function in the activities of daily life (Thompson and Bilenker, 1985) (Fig. 5.1). The broad categories or domains of performance customarily addressed by rehabilitation include personal care, mobility, communication, and social function. Figure 5.2 provides a sampling of major activities within each domain. Many of these activities are common to all children. Others are highly specific and relate to the unique characteristics and desires of an individual child.

Through maximizing independent function, the long-term goal of rehabilitation is to enable children with physical impairments to achieve their fullest potential and so improve their quality of life. The outlook for children with arthrogryposis is excellent (Gibson and Urs, 1970; Drummond et al., 1974; Carlson et al., 1985; Hahn, 1985; Sarwark et al., 1990; Sells et al., 1996). They have the potential to mature into competent adults and assume their roles as self-sufficient, productive citizens. However, children with arthrogryposis or other congenital impairments may not achieve their full potential if their health care providers do not understand the likelihood of ensuing dis-

Self-Care
Eating, dressing, grooming, bathing, personal hygiene, toileting

Mobility
Bed mobility, bed transfer, toilet transfer, floor transfer, car transfer, chair transfer, developmental positions and transitions, sitting, indoor ambulation, stairs, wheelchair propulsion, outdoor ambulation, running, body movements, climbing, driving, public transportation

Communication and Social Function
Expression, comprehension, problem solving, safety, recreation and leisure, household chores, community activities, work activities, school activities

Fig. 5.2 Domains of functional performance.

ability (functional limitations). Many disabilities are preventable; others can be eradicated or lessened. Improved function, independence, and quality of life can be achieved through the early provision and integration of rehabilitative care into traditional health services.

For example, when the functional capacity of a child with severe arthrogryposis is diminished to the point of total dependence, acute medical and surgical care may ensure survival, but a chronic state of partial (Fig. 5.3, curve B) or total (Fig. 5.3, curve A) dependence may persist. The addition of limited rehabilitative care can aid in the achievement of a higher level of function, but this may not be sustained (Fig. 5.3, curves C and D). An ideal comprehensive rehabilitative program and plan should include sufficient training, education, and long-term monitoring to enable the child to attain self-sufficiency as well as an optimal level of functioning throughout life (Fig. 5.3, curve E) (Kottke et al., 1990).

Although the child must be the focus of our efforts, broader attention must also be paid to the physical and psychosocial environment (Sloper and Turner, 1993; Daniels et al., 1987; Hamlett et al., 1992) in which the child and family function. It is not enough to simply understand the physical abilities that lead to the successful performance of an activity. The child must also be viewed in the context of the varied environments through which his or her life passes each day. For example, a child who is able to walk successfully with crutches in a school building may not be able to handle the challenges posed on the playground or on a field trip. Only through an understanding of the interaction between the child and the environment can a comprehensive, holistic rehabilitation plan be formulated and implemented. Rehabilitation plans for children must allow for and incorporate developmental changes. Children's functional needs are being expanded continuously by the growing array of activities associated with development.

Rehabilitation Services

Effective rehabilitation is not the domain of any single provider or discipline. The interaction of many different health care professionals is necessary. Professionals must understand and respect one another's expertise as well as their own limitations and must be willing to work together to achieve commonly identified short-term objectives and long-term goals. Services that commonly comprise rehabilitation for children with arthrogryposis are discussed here.

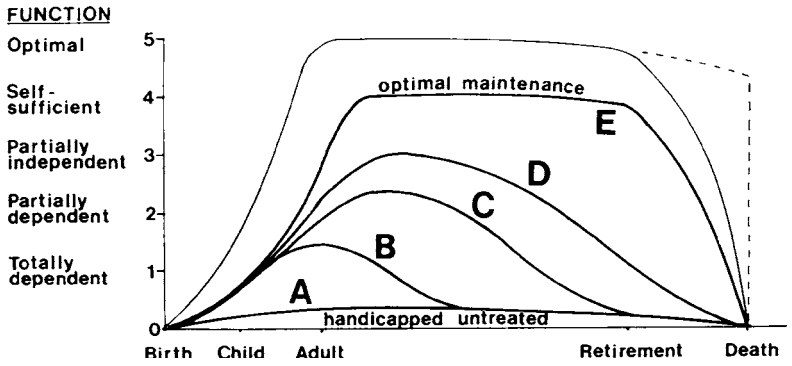


Fig. 5.3 Functional performance over the life span as it relates to the provision of rehabilitative care and services.
(With permission from Kotke et al., 1990, W.B. Saunders.)

Fig. 5.4 Rehabilitation nursing.
When possible, a home visit can add immeasurably to the nursing assessment.

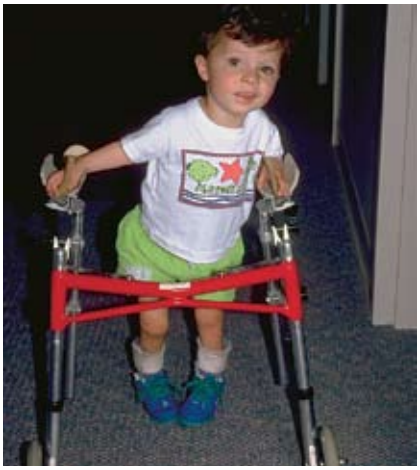


Fig. 5.5 Physical therapy.
Physical therapists help select mobility aids, such as this front-wheeled walker.

Fig. 5.6 Occupational therapy.
Occupational therapists work to help children improve their upper extremity function and fine motor skills.

Rehabilitation Nursing

Rehabilitation nursing addresses comprehensive care management and coordination, ensuring that children with disabilties and their families receive appropriate services, support, and education (Fig. 5.4). Positioned at the hub of the multidisciplinary team, its specific efforts include:

- Educating the family about the child's condition and its implications.
- Monitoring the child's general health.
- Assessing the child's and family's needs and triaging to appropriate service providers and community resources.
- Promoting the child's and family's adjustment to the underlying condition.
- Advocating for the child and family.

Physical Therapy

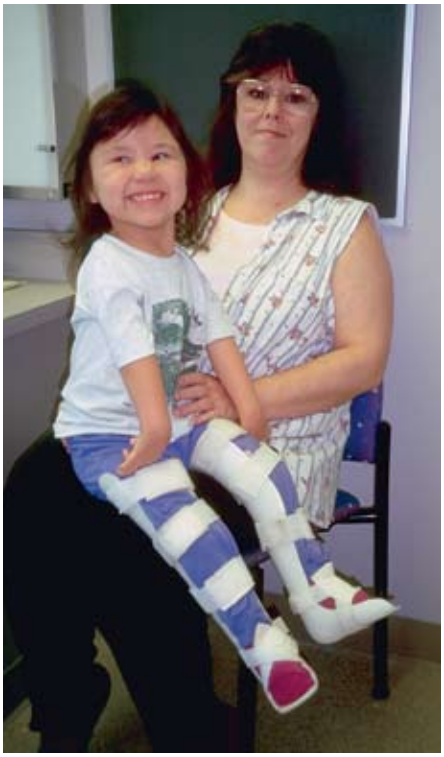
Physical therapy focuses on impairments that interfere with gross motor skills and mobility. Activities affected include crawling, rolling, transferring, walking, running, stair climbing, and bicycle riding. Treatment is directed at:

- Improving strength and endurance.
- Improving posture (seated or standing), transfers, gait, balance, and coordination.
- Monitoring and maintaining joint range of motion, particularly in the lower limbs.
- Selecting appropriate mobility aids and devices (Fig. 5.5).
- Monitoring function, fit, and proper use of lower limb splints and mobility aids.
- Family education and support.

Occupational Therapy

Occupational therapy focuses on impairments that interfere with fine motor skills and functioning in daily life. These include activities of daily living, such as eating, dressing, grooming, bathing, personal hygiene, and toileting; school and work skills, such as writing (Fig. 5.6), drawing, and the use of computers, scissors, books, and paper; driving; and the diverse tasks needed for play and leisure activity. Treatment is directed at:

- Improving strength and endurance to enhance head control and upper-body function (upper trunk, arms, and hands).
- Improving eye-hand coordination and manual dexterity.
- Monitoring and maintaining joint range of motion, particularly in the upper limbs.
- Splinting of upper limbs.
- Selecting appropriate adaptive equipment.
- Monitoring function, fit , and proper use of splints and adaptive equipment.
- Family education and support.



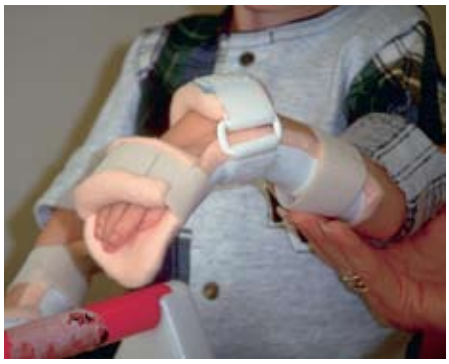


Fig. 5.7 Orthoses.
(Top) These knee-ankle-foot orthoses were fabricated to maintain joint position following orthopedic surgery. (Bottom) This wrist-hand orthosis is designed to maintain wrist and finger range of motion and is intended for nighttime use only.

Speech Therapy

Speech therapy is not usually necessary for children with arthrogryposis. However, when impairments interfere with oral-pharyngeal function, including eating and swallowing, or speech articulation, speech therapy is indicated (Paugh et al., 1988; Robinson, 1990; Quinn et al., 1994). Treatment is directed at:

- Proper trunk and head positioning during meals.
- Manipulation of food texture, consistency, and temperature to facilitate safe swallowing.
- Enhancement of chewing, swallowing, and tongue placement.
- Improving speech articulation.

Nutrition Services

Nutrition services, like speech therapy, are not usually needed for children with arthrogryposis. But for those who have eating and swallowing difficulties (Paugh et al., 1988; Robinson, 1990), dietary counseling and monitoring of caloric intake and weight are especially important. When oral intake is insufficient to maintain normal growth, feeding through a nasogastric or gastrostomy tube may be needed. When a mobility restriction significantly reduces daily energy expenditure, weight control measures can prevent obesity and further compromise of a child's mobility.

Orthotic Services

Orthoses are devices (splints or braces) applied to the external surface of the limbs or trunk to promote stability, maintain joint alignment, and improve function. They are frequently used for children with arthrogryposis. Although a variety of orthoses are manufactured for off-the-shelf use, children with arthrogryposis require individually fabricated models to accommodate their unique limb deformities. Fabrication materials include lightweight metal and plastic and silicone rubber (Bell and Graham, 1995). The time of day that the orthosis will be worn is determined by its purpose and therapeutic goal. Those that provide support and enhance function are intended for use during daytime activities, such as walking, eating, or writing. Others are designed to help maintain range of motion, and their use may actually interfere with function (Fig. 5.7). These are commonly used at bedtime or at other times when function can be sacrificed. The professionals who fabricate orthoses are called orthotists. Physical and occupational therapists sometimes fabricate orthoses.

Recreational Therapy

Recreational therapy helps children with disabilities socialize and learn to use leisure and recreation time productively (Fig. 5.8). Treatment is directed at:

- Developing the skills, knowledge, and attitudes necessary for satisfactory leisure experiences.
- Remedying functional problems that limit participation in leisure activities.

Fig. 5.8 Recreational therapy.
Games, toys, or the environment in which they are used can be adapted so that children can play successfully.

Rehabilitation Counseling

The goal of rehabilitation counseling is to help children become self-sufficient, productive citizens (Fig. 5.9). The services provided by qualified rehabilitation counselors address:

- Career development and employment preparation.
- Achieving independence.
- Integration in the workplace and community.
- Counseling regarding the transition from high school to post-school activities.

Social Work and Counseling Services

Social workers address child welfare in the broadest sense by focusing on home, school, and community life. The duties of social workers include:

- Addressing problems in a child's living situation that affect emotional and social adjustment by mobilizing school and community resources.
- Providing group or individual counseling to the child or family or both.
- Providing parents with referrals to support groups.
- Identifying resources for financial assistance.
- Recommending referral to a psychologist or psychiatrist if needed to address more serious mental health assessment and intervention needs.

Child Clinical Psychology Services

Child clinical psychologists specialize in the assessment and treatment of children experiencing emotional, behavioral, or learning difficulties. Their efforts include:

- Providing individual or family psychotherapy.
- Assisting with adjustment to disability.
- Gaining cooperation for necessary medical treatment plans.
- Assessing a child's cognitive and developmental level.
- Assisting in the design of special education programs.

Fig. 5.9 Rehabilitation counseling.
The University of Washington recruits high school students with disabilities through Project DO-IT (Disabilities, Opportunities, Internetworking, and Technology). This program helps them to explore careers in science, engineering, and mathematics and to gain prerequisite knowledge to enter these fields of study and employment.

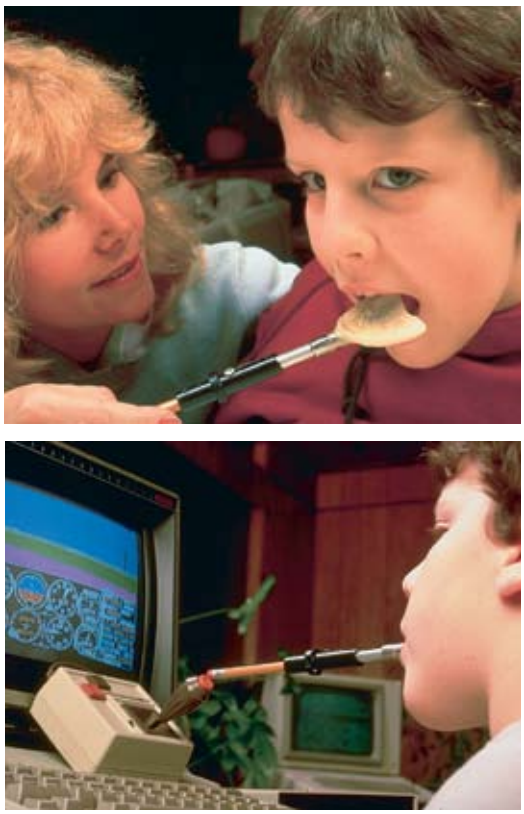


Fig. 5.10 Enhancing unaffected systems.
Oral motor skills can compensate for limited upper extremity function.

Strategies for Rehabilitation

There are six groups of treatment strategies employed by rehabilitationists to improve function and minimize disability (Stolov, 1982). Examples for each strategy are provided to illustrate this approach in the context of arthrogryposis.

Prevention or Correction of Additional Impairment or Disability

Examples include:

- Health care maintenance, including the provision of immunizations and monitoring of growth and development.
- Feeding via nasogastric or gastrostomy tube to prevent malnutrition.
- Passive joint range of motion (ROM) exercises to reduce contractures.
- Splinting to prevent recurrence of joint deformity after orthopedic surgery.
- Screening of vision and hearing to rule out associated sensory impairments that can further compromise function.
- Injury prevention strategies for both family and child.

Enhancement of Systems Unaffected by the Pathologic Process

These include:

- Strengthening normal musculature to enhance a specific and meaningful functional outcome.
- Increasing oral motor skills to substitute for reduced fine motor hand skills (Fig. 5.10).

Enhancement of the Functional Capacity of Affected Systems

This can be done by:

- Strengthening weak muscles when there is realistic hope of improved function.
- Training dysarthric speakers to improve intelligibility.
- Use of a hearing aid to compensate for associated partial hearing loss.

Use of Adaptive Equipment to Promote Function

Examples include:

- Use of crutches or orthoses to achieve ambulation.
- Wheelchair training when walking is not realistic as the only source of mobility.
- Use of equipment to improve upper limb and hand function.

Modification of the Social and Educational Environment

This can be done by:

- Moving to a single-level home without entry steps for an ambulatory child unable to climb stairs.
- Providing ramp entry and widening doorways to permit wheelchair access (Fig. 5.11).
- Providing caregiver assistance at home or in school for physical dependency.
- Redesigning classrooms to accommodate wheelchair users.

Fig. 5.11 Accessibility.
A public transportation system with specially designed buses enables people with disabilities to have equal access to their communities.

Psychologic Techniques to Enhance Patient Functioning and Adaptation

These include:

- Cognitive-behavioral interventions to improve coping, compliance with medical treatment, social skills, and assertiveness.
- Parenting techniques to support the child's development and independent functioning.
- Consultation with schools to address cognitive, behavioral, or social-emotional concerns in the academic setting.

Principles of Rehabilitation

The principles discussed not only are applicable to the field of rehabilitation or to the diagnosis of arthrogryposis, but also can be readily applied to other health care disciplines and diagnostic entities and are designed to be a framework for family-centered, sensitive, and effective care.

Provide an Accurate and Specific Diagnosis

The importance of an accurate and specific diagnosis cannot be overemphasized. Although there are four major causes for congenital limitation of joint movement, there are more than 150 specific entities that result in multiple congenital contractures (Hall, 1985a). It is the specific diagnosis that provides information on associated findings, natural history, and prognosis (Thompson and Bilenker, 1985). Ultimately, treatment and management decisions, as well as the setting of short-term and long-term goals, will be derived from the diagnosis (Drummond et al., 1974; Hahn, 1985; Shapiro and Specht, 1993).

Provide Ongoing, Comprehensive, and Coordinated Multidisciplinary Care

Developmental changes due to the physical processes of growth, maturation, use and disuse, injury, and degeneration (or senescence) occur over the entire life span and not only during childhood years. For children with arthrogryposis, the long-term impact of many of these processes is not entirely understood. Ongoing monitoring of an individual's physical condition, psychologic status, and physical and social environments will help to reduce disability and promote opportunity. These goals can all be achieved by teamwork — the efforts of many individuals working collaboratively for a common purpose (Hahn, 1985; Thompson and Bilenker 1985; Sarwark et al., 1990).

Establish Community-Based Care and Services

Families should be assisted in developing a community-based, family-oriented care and support system. Such a system is not meant to replace expert and knowledgeable management, but to discourage over-reliance on the tertiary care center. Many families and children do not live in close proximity to centers of excellence. Major operative procedures and periodic consultations can be center-based, but routine care and support should be provided closer to home. Good communication among the center, the family, and community-based providers will foster continuity of care and commonality of purpose.

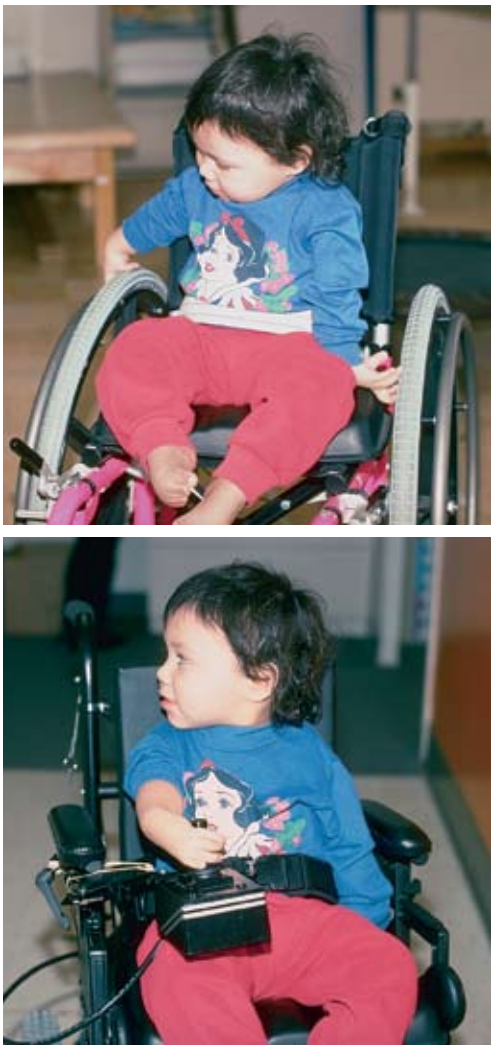


Fig. 5.12 Self-initiated mobility.
This 2-year-old girl with arthrogryposis could neither ambulate nor effectively propel a manual wheelchair, but she quickly learned to skillfully control a power wheelchair for independent mobility.

Establish a Means of Independent, Self-Initiated Locomotion; Manage the Child's Movement to Allow Maximal Function and Environmental Interaction

During the first few years of life, the development of gross motor skills enables children to interact, influence, and thereby learn from their environment (Piaget and Inhelder, 1969). Mobility is an important vehicle for learning, socialization, and the promotion of independence (Piaget and Inhelder, 1969). Children with motor impairments should be afforded developmentally appropriate opportunities to achieve independent locomotion through the timely prescription of mobility aids, including powered mobility devices. Three groups of children with arthrogryposis are candidates for powered mobility (Fig. 5.12): those who will never walk, those with inefficient ambulation (i.e., who walk but lack the speed or endurance to be considered fully functional in all contexts), and those who have the potential to walk, but whose potential may not be achieved for many years (Hays, 1987). Children as young as 20 months can quickly and skillfully learn to drive powered mobility devices (Butler et al., 1984). Their provision does not deter the eventual achievement of independent bipedal ambulation in those children who have this capability.

Be Mindful of Post-Childhood Goals

Many pediatric health care professionals focus attention on the childhood and adolescent years and lose sight of the fact that adulthood approaches rapidly. In the United States, the Individuals with Disabilities Education Act (IDEA) was signed in 1990, placing increased attention on transition services, a coordinated set of activities that promote movement from school to post-school activities and settings. Transition services are meant to facilitate post-secondary education, vocational training, integrated employment, continuing and adult education, adult services (health, social, housing, transportation), independent living, and community participation. The preteen years are an appropriate time to begin consideration of these critical issues (Fig. 5.13).

Normalize the Child's Appearance as Much as Possible

Western culture maintains an idealized conception of attractiveness and places a high value on physical appearance. Even young children display a tendency, at least initially, to avoid children with observable physical differences (Harper et al., 1986). During adolescence, peer acceptance and fitting in gain paramount importance. As rehabilitationists, we must be mindful of the impact that our recommendations have on a child's physical appearance (Harper, 1991a). We must also be open to helping children cope with issues related to their looks.

Fig. 5.13 Transition services.
Planning for life after high school must begin early and requires the concerted efforts of parents, educators, and counselors to enable young people to succeed as adults.

Focus Efforts on Effective, Meaningful, and Functional Interventions

In the prescription of rehabilitation services, particularly occupational and physical therapy, care must be taken to identify specific goals. These should be stated in terms of measurable functional outcomes, and whenever possible, realistic estimates for treatment time frames should be provided. Open-ended or indefinite treatment, without the benefit of critical reevaluation of effectiveness, must be discouraged. Adaptive technology, durable medical equipment, and environmental control systems that improve function, reduce dependency, and improve quality of life should be employed (Fig. 5.14).

Minimize Economic Cost and Disruption of the Child's and Family's Lives

The provision of rehabilitation services must be balanced with a host of competing child and family needs (Beavers et al., 1986; Patterson et al., 1990; Spinetta et al., 1988). For the child, treatment of the underlying physical condition must be weighed against other normal developmental priorities. Time spent in therapy means time not spent in other activities (i.e., education, socialization, recreation). Family members must acknowledge that limited resources need to be shared. The costs of professional recommendations include direct economic expenditures as well as indirect costs, such as time off from work and sacrificed relationships, activities, or other opportunities.

Respect the Uniqueness of Each Child and Family

Paternalism, or the attitude that "we know what is best for you," should be avoided. Families know themselves best and should be empowered through education and support to retain control over their lives and medical decision making. By conveying this respect, the treatment team will contribute to the development of their self-sufficiency.

Fig. 5.14 Adaptive technology in the workplace.
The appropriate use of adaptive techology can promote vocational success.

Fig. 5.15 Family-focused care.
A child with arthrogryposis affects all members of a family.

Respect the Family's and Child's Confidentiality

Many professionals are involved in the care and management of children with arthrogryposis. Before privileged and potentially sensitive information is disseminated to other members of the treatment team, efforts must be made to clearly understand the family's wishes on this matter.

Families Are More Than Nuclear Units

The birth of a child with arthrogryposis obviously has a tremendous impact on the parents. Siblings, grandparents, and other family members are also affected and deserve to be recognized in our approach (Fig. 5.15). The inclusion of family members in clinic visits can dispel misunderstandings about the condition and its course and management, and provide support in addressing the fear, guilt, and sadness frequently associated with the birth of a child with physical differences.

Remain Flexible with Recommendations

There are very few situations in the management of a child with arthrogryposis for which there is only one approach. The treatment team must remain flexible and help the family be flexible. Recommendations that set the stage for parent-child or spousal conflict are to be avoided. Often, compliance with overly rigid professional recommendations comes at the expense of harmonious child and family functioning (Patterson, 1991).

Communicate Competently

In communicating with families of children with arthrogryposis, it is important to remain positive, hopeful, and optimistic about the child's future. Hope and optimism, however, must be balanced with the reality that raising a child with a developmental disability poses a unique set of demands and challenges that might not otherwise be encountered. To imply otherwise is an injustice, one that trivializes the parenting responsibility. All information should be presented to families in everyday language that is easily understood. Professional jargon and the words "never" and "always" are to be avoided.

It is only fair for health care professionals to share with families not only what we know, but also the limitations of our personal knowledge. Families who desire second opinions should be encouraged to obtain them, particularly if it will give them increased peace of mind in reaching responsible decisions. Because the families of children with arthrogryposis face such complex burdens, professionals should make particular efforts to ensure that their time with these families is not rushed. It is important that there be adequate time to answer questions, share information, and educate. Families can also be encouraged to acknowledge and express feelings associated with their particular circumstances. Such open expressiveness is associated with better child and family outcomes and is more likely to occur in an atmosphere that does not feel rushed (Borrow et al., 1985).

Fig. 5.16 Multidisciplinary clinic.
A multidisciplinary clinic facilitates the delivery of coordinated, comprehensive care.

The Multidisciplinary Clinic

A multidisciplinary clinic for children with arthrogryposis can enhance patient care and promote understanding of this condition by professionals and families alike (Fig. 5.16). The organization and operation of our clinic is presented in the hope that it will encourage development of other such clinics and programs.

The clinic meets three to four times yearly. It is organized through the Department of Rehabilitation Medicine and coordinated by a rehabilitation nurse clinician. It is staffed by pediatric physiatrists/pediatricians, a pediatric orthopedic surgeon, a hand orthopedic surgeon, a pediatric geneticist, a pediatric neurologist, and occupational and physical therapists. The full services of our 200-bed hospital and medical center are available to meet each child's individual needs.

The clinic is scheduled for 1 full weekday, but some appointments, particularly with therapists, can be spread over 2 or 3 days. This creates a more relaxed and less frenetic schedule for those with multiple appointments. The staff addresses questions of diagnosis, management of deformity, and rehabilitation. A midday luncheon affords families and children an opportunity to network and to both seek and provide mutual support.

Referrals are accepted from any source, including parents, primary care providers, medical and surgical specialists, therapists, nurses, and school personnel. The intake process includes a review of the child's needs, current level of functioning, past medical history, and parents' expectations. Past medical records are usually requested. Following this, a mutually agreed upon set of appointments is established and scheduled.

On clinic day, the clinicians individually share their recommendations with the family as they see the child. However, at the conclusion of the clinic day, a team meeting provides a means to collectively review each child, discuss recommendations, and ensure appropriate follow-up and coordination of services. Copies of reports are forwarded to physicians and other care providers, as requested.

Conclusion

This chapter provides an introductory framework for the rehabilitation section of the book *Arthrogryposis: A Text Atlas*. It reviews the principal objectives of comprehensive rehabilitative management, the services and disciplines that are necessary to achieve successful outcomes, and the strategies and principles that service providers must keep in mind when serving children with arthrogryposis and their families. In the next three chapters, more detailed discussion focuses on occupational and physical therapy, promoting social and emotional well-being, and educational services.

Chapter Contents **Page**

C.S. Graubert, P.T.
D.L. Chaplin, M.S., O.T.
K.M. Jaffe, M.D.

Introduction

Physical therapy and occupational therapy play important roles in the management of children with arthrogryposis. The ultimate goals of therapy are to enable children with arthrogryposis to achieve maximal independence and function (Drummond et al., 1974; Thompson and Bilenker, 1985). Of equal importance to hands-on treatment is the role the therapist plays as an educator and facilitator. Not all parents and families intuitively know how they can best help their children to realize their fullest potential in the realm of physical functioning. Physical and occupational therapists incorporate treatment strategies with teaching sessions to show parents how to work toward functional goals at home and at school (Lloyd-Roberts and Lettin 1970).

In general, the recommended frequency of therapy changes during a child's life. It tends to be more intensive during the first year and decreases during the preschool and elementary years. Later, therapists tend to serve more as consultants than direct providers of therapy. The frequency of therapy may also increase for short periods during the preadolescent and adolescent years to work on specific, mutually agreed-upon goals. For example, an orthopedic surgical procedure may provide a child with the capacity to acquire a new functional skill. A short burst of therapy may also be indicated when and if a child is interested in improving independence in some aspect of self-care or mobility.

This chapter sets out to sequentially describe assessment and intervention strategies at four stages of child development: birth to one year, the toddler/preschool years, the early school years, and the teenage years. Throughout the chapter, physical and occupational therapy considerations are described together. This interweaving of disciplines reflects real world practice and is a model that fosters collaboration for the best interests of the child. For example, a child who needs a walker may be best served if a physical therapist consults with an occupational therapist when developing the exact prescription. Will an optimal hand position be achieved by the

handles on the walker, or would custom molded hand and wrist supports work better?

Above all, it is imperative that therapy programs remain focused on issues and goals that are important to the child and the family. The goals should be meaningful and attainable. Periodic evaluation will help determine if progress is occurring, if different techniques are indicated to attain goals, or if new goals need to be established.

Birth to One Year

The general goals of treatment for infants with arthrogryposis are to:

1. Increase range of motion at joints where this is possible.
2. Maintain newly acquired range of motion through splinting.
3. Position infants appropriately.
4. Help parents and caregivers feel comfortable and knowledgeable about handling and holding their infants

The frequency of therapy will depend on a child's individual needs. Some parents want to be the primary providers of treatment for their infants, whereas others prefer to have a physical or occupational therapist provide as much of the intervention as possible. In either case, parents should be encouraged to take an active role in establishing goals and making decisions about their infant's treatment.

Throughout the evaluation, the interaction between the parent and the infant is observed. The parent's ease in handling and holding the infant is noted, and parents are encouraged to voice any concerns or questions about their infant's assessment and care needs.

Range of Motion Assessment

For reasons not entirely understood, restriction in joint range of motion can be best overcome during the first year of life through various techniques, including stretching, positioning, splinting, and casting (Thompson and Bilenker, 1985; Sarwark et al., 1990; Palmer et al., 1985). The neck, spine, and joints of the upper and lower limbs are carefully evaluated. Range of motion is measured passively (Fig. 6.1). Through an understanding of range of motion, therapists can begin to understand a child's potential for future functional tasks. In this manner, specific interventions can be prioritized.

Shoulders:	flexion, abduction, extension, internal and external rotation
Elbows:	flexion, extension, forearm pronation and supination
Wrists:	flexion, extension, radial and ulnar deviation
Hands:	MCP flexion, PIP and DIP flexion, and thumb motion including abduction and ability to oppose the fingers
Hips:	flexion, extension, abduction, adduction, internal and external rotation
Knees:	flexion, extension
Ankles:	dorsi and plantar flexion, eversion, inversion
Neck:	flexion, extension, rotation, lateral flexion

Fig 6.1 Range of motion measurements.

Range of Motion Interventions

Passive Range of Motion

Ideally, daily gentle passive range of motion (PROM) exercises, or stretching of the joints, are started during the first weeks of life. Stretching is usually done for all joints exhibiting limitation, even those with little or no motion. Increased joint motion is needed to improve positioning for function and to allow for greater movement that can be achieved through strengthening, substitution, or orthopedic surgery.

When stretching joints, it is important to let the infant's response serve as a guide. It is counterproductive to stretch if the infant is upset or tense. When done correctly, stretching may be uncomfortable, but it should not be painful. Stretching is always done gently and held at the end of range for only a few repetitions rather than many quick repetitions. The act of stretching an infant can be emotionally exhausting to the parents, and therapists need to be sensitive to this possibility and acknowledge it when it occurs.

Fig. 6.2 Foam positioning wedge.
Infant positioned to provide stretch to hips and shoulders.

Fig. 6.3 Spine hyperextension deformity.
Soft foam positioning wedge can be an initial strategy to improve range of motion.

Ideally, gentle stretching should occur two to three times daily. It is easier for parents or other caregivers to remember to do the stretching if it is incorporated into daily routines. For instance, hip and knee flexors could be stretched two or three times each time the diaper is changed, and wrists can be extended each time clothing is put on or removed. For babies who enjoy bathing, this can be an optimal time to stretch.

It may work well to begin PROM by gently stretching the infant's hands. Passive flexion and extension of the fingers must be done carefully to prevent tissue damage. Stretching of the hands should also include the long finger flexors by extending the fingers with the wrist extended. When forearm rotation is done, the elbow should be held in 90° of flexion. Small infants enjoy the feel of their fingers in their mouth. Helping them to explore their fingers orally can facilitate passive movement of the shoulders and elbows. Older infants may be more compliant if stretching is begun by playing movement games like "So Big," in which the shoulders are flexed to bring the hands up over the head. The details of any stretching program should be taught and regularly reviewed by the infant's physical or occupational therapist.

Casting/Splinting/Positioning

In addition to manual stretching of the joints, positioning is a valuable tool to provide a stretch to joint structures. Positioning of joints in a stretched position may be maintained through the use of splints, casts, and foam wedges. Positioning provides a prolonged stretch that may be more effective for gaining range of motion in the neck, shoulders, and hips of infants. Casting often works best for feet and knees, whereas splinting is usually used on the smaller joints of the hand, wrist, and elbow (Shapiro and Specht, 1993; Hahn, 1985). Splints offer the advantage of being removable for bathing and active exercise, but casting ensures a more prolonged stretch with forces distributed over a wider area.

Other devices can be used to provide stretch on various joints at the same time (Figs. 6.2 and 6.3). Neck and trunk supports can be fabricated with firm foam or low-temperature plastics to provide a stretch while supporting the infant in a more midline position. Foam wedges can be used to position infants on their stomachs while providing a stretch on hips, knees, or shoulders with gravity assisting. Serial positioning, in which the angle of a foam wedge is steadily reduced under the hips while in a prone position, has been effective (Fig. 6.2). Infants with hip abduction contractures who are sitting in highchairs, carseats, and strollers can be positioned with foam blocks or rolls alongside their thighs to encourage more adduction.

During the initial evaluation session, newborn infants with wrist or hand contractures are commonly fitted with full hand splints (Carlson et al., 1985). It is recommended that the splints be worn a minimum of 18 to 20 hours each day. Parents are instructed to remove the splints only for bathing and hygiene, while stretching and exercising, and during brief periods of play. The extensive use of splints will maximize the inherent capacity of the newborn's tissues to respond to stretching at a time when they will not interfere with function (Palmer et al., 1985).

The occupational therapist checks the splint fit every other week. New splints are fabricated as needed to accommodate growth and improvement in motion. After 2 1/2 to 3 months, wearing time can be decreased as passive movement increases. At 4 to 5 months of age, the infant may be fitted with functional wrist splints that leave the thumb and fingers free for grasping and hand use. The functional wrist splint is worn during the day, and the full hand splint is worn at night.

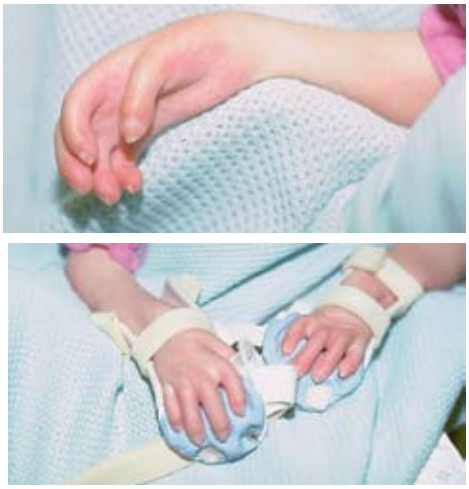


Fig. 6.4 Hand splint with elastomer.
Ulnar deviation and overlapping of fingers are minimized when elastomer putty is incorporated into a full hand splint.

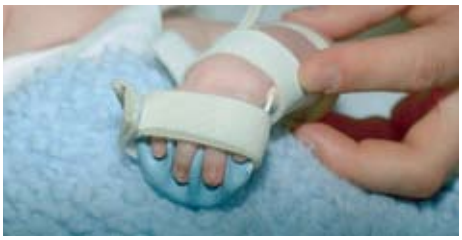


Fig. 6.5 Full hand splint.
Used to extend wrist and fingers, abduct thumb, and separate fingers.

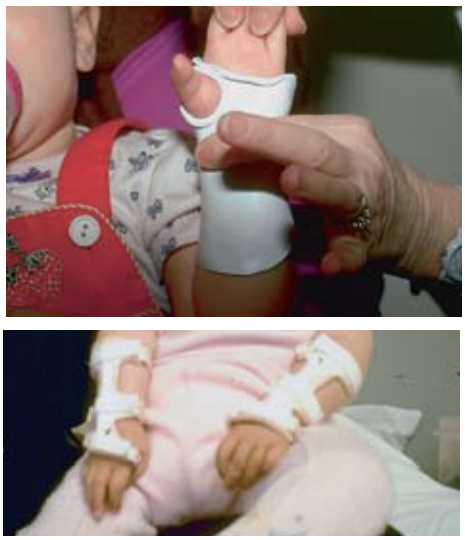


Fig. 6.6 Volar style splint *(top).*
This functional wrist splint allows for full MCP flexion.

Fig. 6.7 Dorsal style splint *(bottom).*
This functional splint allows increased tactile input to the palm.

Splinting to increase elbow flexion is challenging because of the relative strength of the triceps, which can push the forearm out of the splint. Initially, an anterior or posterior shell-style splint may be used because either shell can be fabricated to accommodate the hand splint. Other styles, discussed subsequently, are usually worn on an alternating basis with the hand splints (Lloyd-Roberts and Lettin, 1970).

Full Hand Splint

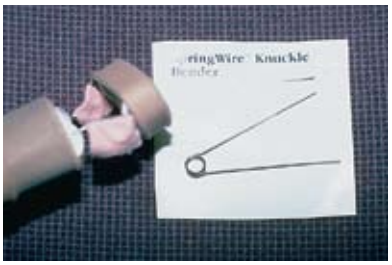
This is a forearm-based hand splint that is designed to extend the wrist and support the fingers and thumb. It can provide the best leverage for decreasing wrist and finger flexion contractures. A wide thumb post is incorporated into the design of the splint to position the thumb out of the palm and into abduction. For stretching the wrist into greater extension, a padded strap that secures with a D-ring can be riveted to the wrist area of the splint. Elastomer putty has been used successfully to minimize ulnar deviation and maintain finger separation in a full hand splint (Figs. 6.4 and 6.5). Similar materials have also been used with good outcomes (Bell and Graham, 1995).

Functional Wrist Splint

This type of splint is used to support the wrist in neutral or slight extension while allowing functional hand use. A volar style is usually chosen because it provides optimal support (Fig. 6.6). However, when less support is needed, a dorsal style splint that supports the wrist with a strap across the palm can also be used successfully (Fig. 6.7). A dorsal wrist splint weighs slightly less than a volar splint and permits greater tactile input to the palm. Both styles extend to just below the proximal palmar crease to allow for maximal MCP flexion.

Dynamic Wrist Extension Splint

Dynamic or active splinting makes use of a force similar but opposite to that which produces the deformity; i.e., it provides tension to the tendons that have developed without their normal opposing muscles (Rank et al, 1973). This splint is used to increase wrist extension. Spring wires, custom-sized hinges, and other materials are used to provide sufficient external force to extend the wrist (Figs. 6.8 and 6.9). Skill is needed in fabricating the splint and adjusting the springs to give just enough force. In general, dynamic splints tend to be less durable than static splints and are, therefore, worn for shorter time periods, usually when the child can be directly supervised.



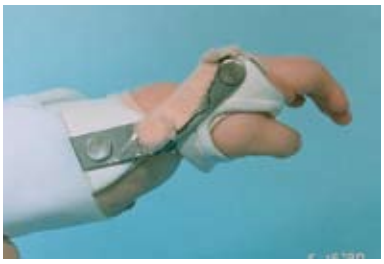


Fig. 6.8 Dynamic wrist extension splint using a spring wire *(left).*
Uses a wire to aid wrist extension.

Fig. 6.9 Custom hinge splint *(right).*
Hinge is set in desired position.

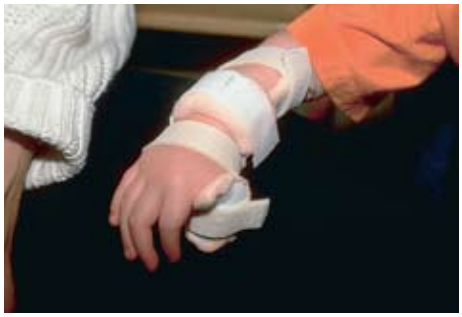


Fig. 6.10 Thumb abduction splint.

Fig. 6.11 Ulnar gutter splint.
Can be worn with an elbow splint.

Other Hand/Wrist Splints

Some children may require wrist support and thumb abduction but not finger support (Fig. 6.10). In this instance, a forearm-based thumb abduction splint is worn only at night.

The ulnar gutter splint is used when ulnar deviation needs to be minimized. It is worn on the ulnar side of the palm and forearm, usually in conjunction with an elbow splint (Fig. 6.11).

Elbow Splints

The anterior shell, fitted on the flexor surface of the upper arm, is the least complicated elbow splint to fabricate. It extends over the elbow and down the forearm to a point just above the wrist. Usually, three straps are applied to stretch the elbow. The first strap is placed just above the elbow, the second secures the upper arm, and the third is used to pull the forearm into the desired position. An anterior shell can be used to either flex or extend the elbow.

A posterior shell splint, fitted on the extensor surface of the upper arm, extends down the ulnar surface of the forearm (Fig. 6.12). This splint is usually used to position the elbow in flexion. It works well for older children but is difficult to fit on infants because of the relatively short length of the infant's arm combined with increased skin and fat folds that appear when flexing the elbow.

A crossed-strap dynamic flexion splint is used to pull the elbow into flexion (Fig. 6.13). It uses a neoprene strap to create a dynamic pull from the posterior upper arm cuff down across the anterior elbow, under the forearm cuff, and back up again, crossing the elbow to the upper arm. Because of its dynamic component and potential for improper application, this splint is used when the infant can be closely monitored and only for up to 2 hours at a time.

Fig. 6.12 Posterior shell splint.
Used to increase elbow flexion.

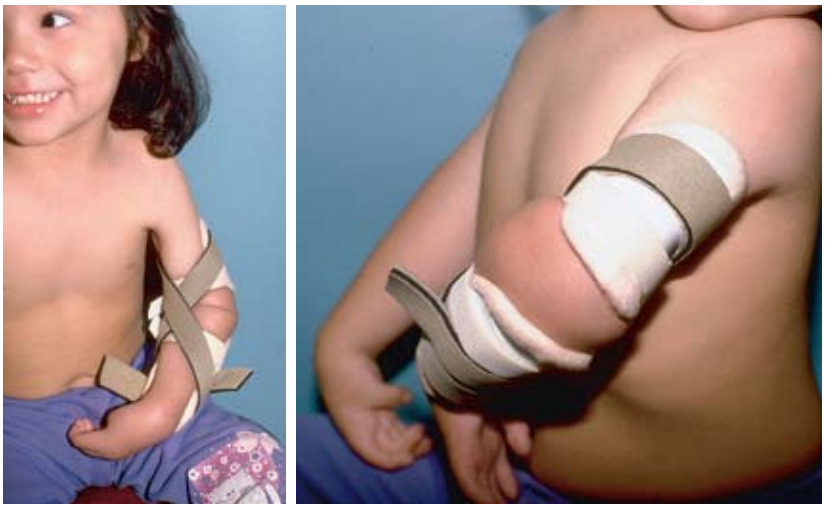


Fig. 6.13 Crossed-strap dynamic elbow flexion splint.
Front and back views.

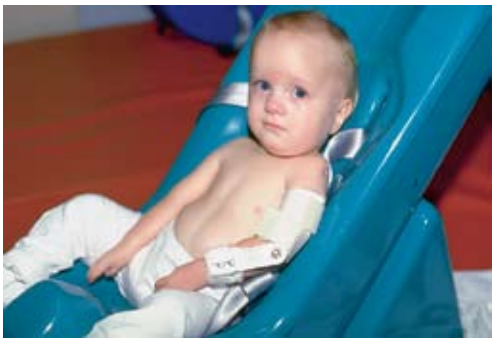


Fig. 6.14 Hinged elbow flexion splint.
With this splint, 30-45° of passive elbow flexion can be achieved.

Fig. 6.15 Rolyan locking hinge.
Allows adjustable elbow flexion.

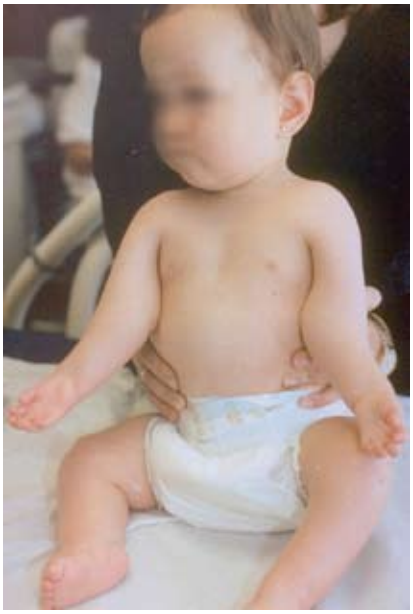


Fig. 6.16 Observing active movement in supported sitting.

A hinged elbow flexion splint can be fabricated from materials commer cially available for use in splinting adult-sized wrists. The desired degree of flexion is set with a ratchet wrench during application. Care must be taken to avoid traumatizing tissues and to correctly align the elbow joint when applying this splint (Figs. 6.14 and 6.15).

Splinting Materials

Standard thermoplastic splinting materials are used to fabricate upper extremity splints. The material chosen should be one with which the therapist is experienced. Soft foam straps with adhesive tabs are recommended because of the padding and the ease with which they can be securely attached to the splint by briefly heating the adhesive with a heat gun. They can be trimmed to an appropriate width for small infants by cutting them lengthwise. Moleskin and Hapla Fleecy Web are two materials that are used to line and reline the splints as needed. Self-adhesive contour foam padding can be cut to the exact dimensions necessary to pad D-ring style straps but must be covered with one of the lining materials to prevent skin irritation.

Strength Assessment

A first impression of an infant's strength is made through observation. Movement in a specific pattern, such as shoulder internal rotation with elbows straight and wrists flexed, shows which muscles are stronger than others. An absent or weak muscle on one side of a joint is overpowered by a stronger muscle on the opposing side. This imbalance in strength causes abnormal positioning of joints at rest. For example, when the wrist extensor muscles are weak or absent, the wrist remains in a flexed position. Strength is evaluated while watching the infant move, by placing the infant in a variety of positions to encourage movement, and by palpating muscle contractions.

Supported Sitting

Look for active movement against gravity (Fig. 6.16). What is the resting position of the hands and wrists? Does the infant exhibit a grasp reflex? Is the position of the neck and head symmetric? Do the ankles flex?

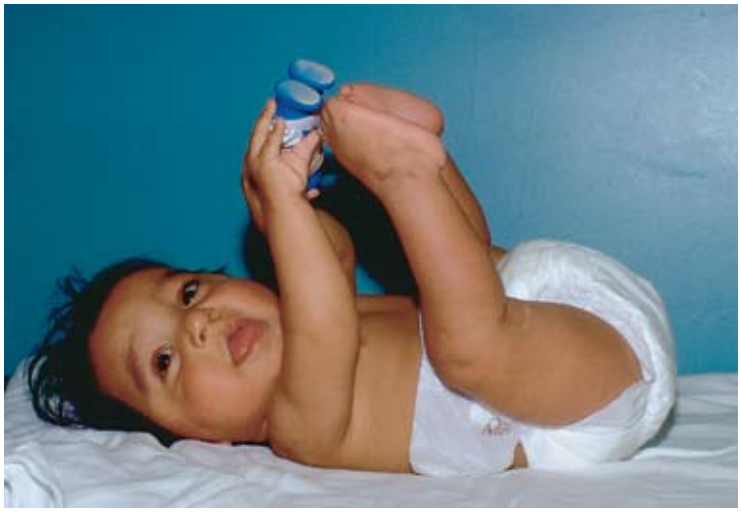


Fig. 6.17 Observing active movement in supine.

Fig. 6.18 Observing active movement in prone.

Supine

Is there active shoulder flexion or abduction? Do the elbows flex against gravity? Is there active flexion of the fingers or extension of the wrists? If the arm is held with the shoulder flexed to 90° does the elbow remain extended or does gravity cause it to flex? Is there active kicking of the legs using the hips and knees (Fig. 6.17)?

Side Lying

With gravity eliminated, is active shoulder, elbow, hip, or knee flexion observed?

Prone

Is there trunk elongation and extension? Can the infant extend his neck and lift his head? Do the arms remain at the infant's side, or does he attempt to flex the shoulders and elbows? Is weightbearing on the forearms tolerated? Is there kicking present at the knees? Do the hips extend (Fig. 6.18)?

Strength Interventions

Fig. 6.19 Encouraging play in side lying and sitting positions.

Infants cannot perform standard strengthening exercises. However, they can be encouraged to play with toys in a range of positions from side lying, where the effect of gravity is eliminated, to more challenging positions requiring movement against gravity (e.g., reaching for toys while sitting) (Fig. 6.19). Toys can be placed strategically to encourage movement of arms and legs against gravity. Baby gyms work well for this purpose by suspending toys over an infant lying on his back. Benches or boxes of various heights can also be used to position toys for the infant who is sitting. Moving from very lightweight toys to heavier toys will also help increase strength. Increasing the passive movement of joints through range of motion, splinting, casting, or positioning creates a new arc of movement that can benefit from strengthening activities.

It is important to change an infant's position frequently during the course of the day. Repositioning helps improve range of motion, encourages the development of head and trunk control, strengthens limb musculature, and facilitates functional activities. Infants must not always be placed in the most challenging positions but must be offered ample opportunities to be in relaxed positions as well. If a more challenging toy is being presented to the infant, it is wise to position him in an easier position to avoid frustration or fatigue. If a simpler, more familiar toy is being used, a more challenging posture could be tried.

Activities of Daily Living

Oral Motor/Feeding Assessment

This section addresses some of the anatomic differences and oral motor difficulties seen in children with arthrogryposis. Not all children with arthrogryposis have feeding problems. However, feeding difficulties with subsequent poor weight gain have been observed clinically and reported in the literature (Paugh et al., 1988; Robinson, 1990).

The most common oral motor structural difference in children with arthrogryposis is the presence of micrognathia (a small, posteriorly positioned jaw). The chin appears to be recessed, and there is an accompanying retroversion or posterior positioning of the tongue in the oral cavity, impairing its ability to descend appropriately. Breastfeeding may be difficult

for the infant with micrognathia because the tongue surface may not be adequately positioned beneath the nipple, resulting in insufficient compression of the milk ducts (Wolf and Glass, 1992).

Feeding problems in newborns may be due to weak or inefficient sucking or poor coordination of breathing and swallowing or both. During the latter part of the first year, difficulties with chewing may also become apparent. These problems often appear to be related to anatomic differences and mobility problems of the jaw and tongue. However, in rarer instances, difficulty in swallowing may be due to laryngopharyngeal involvement. If this is suspected, a clinical feeding evaluation may be indicated.

Some newborns have difficulty swallowing, which may be due to a delay in the swallowing reflex or inconsistent laryngeal elevation. If aspiration is suspected and the infant has a history of recurrent pneumonia, a videofluoroscopic swallowing study (Wolf and Glass, 1992) may be indicated. If the study shows frank aspiration with little or no protective cough, alternative feeding methods need to be considered.

Fig. 6.20 Postural support.
Head, arms, and trunk positioned during feeding.

Oral Motor/Feeding Interventions

Infants with feeding difficulties can benefit from a variety of therapeutic feeding strategies.

Postural support, provided by holding the infant's head and trunk in alignment and positioning the neck in slight flexion, can maximize the infant's sucking and swallowing abilities (Fig. 6.20).

Chin and cheek support can help to improve cheek strength and stability, jaw control, and lip closure for more efficient sucking. The primary feeder provides direct external support to the cheeks with the thumb and finger while providing gentle pressure to the mandible with another finger placed under the chin to facilitate jaw control (Wolf and Glass, 1992).

External pacing of breathing (Wolf and Glass, 1992) may be helpful for the infant having problems coordinating sucking, swallowing, and breathing. This is done by carefully counting the number of suck/swallows without a spontaneous breath and removing the bottle after three to four sucks to impose a pause to breathe.

The type of artificial nipple used can affect tongue and lip position and movement during sucking. A longer nipple or firm nipple may help facilitate forward movement and central grooving of the tongue for infants with tongue retraction (Wolf and Glass, 1992).

Thickening formula with rice cereal may help to create a more cohesive bolus. Chilled formula provides thermal stimulation. Either technique may help to facilitate swallowing in infants with arthrogryposis who exhibit an inconsistent or delayed swallowing reflex. Any changes to an infant's formula may warrant a referral to a nutritionist to assist with cereal-to-formula ratios.

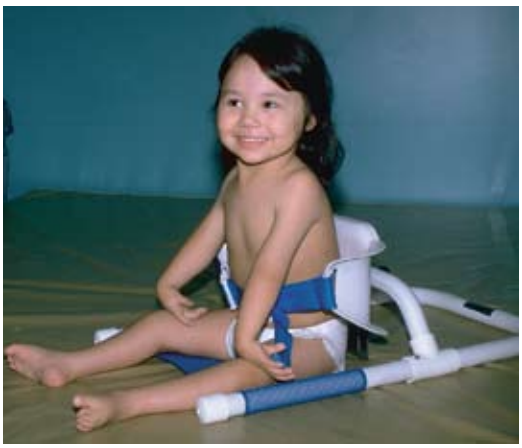


Fig. 6.21 Bathing support.
Commerically available equipment can be used to provide safety in the bathtub.

Bathing

Safe upright positioning when bathing some infants with arthrogryposis may present difficulties for the parent or caregiver. The use of an appropriately fitted bath support may be very helpful, especially for those infants needing greater support for head and trunk alignment. A variety of supports are available commercially (Fig. 6.21).

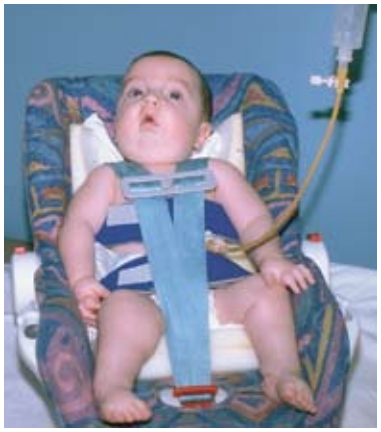


Fig. 6.22 Safe transportation.
Foam seat insert provides postural support in an infant carseat.

Safe Transportation

All infants should be transported in safety-approved carseats. Occasionally, it is necessary to add some extra supportive material to a carseat to maintain neck and trunk alignment. A foam seat insert can be fabricated with extra trunk and head supports. Added material should not, however, interfere with the proper functioning of the carseat. It should not change the approved method of securing the carseat to the car, or the system that harnesses the infant into the carseat. Added material should compress minimally to avoid changing the baby's position relative to the harness in the event of an accident (Fig. 6.22). Material with more cushioning qualities can be used as an insert for a highchair or stroller.

If an infant is immobilized in a spica cast after lower extremity surgery, a stroller or highchair can be adapted with extra material to support the cast securely and comfortably. It may not be possible to adapt the car seat for this period, but an alternative safety-approved harness could be used with the infant lying down along the back seat.

Gross Motor Skills Assessment

Gross motor skills are assessed with a normal developmental sequence in mind. This can be done either through careful clinical observation or occasionally through standardized developmental evaluations. Head and trunk control are key to later movement skills and are evaluated in a variety of positions. During the second half of the first year, sitting balance and mobility skills are evaluated. Infants with arthrogryposis do not always go through the normal sequence of mobility: rolling, creeping on stomach, crawling on hands and knees, cruising, walking. Some will learn to scoot on their backs or scoot while sitting instead. The pattern of muscle weakness and joint contracture that each infant develops will have significant impact on how he or she learns to move.

Gross Motor Skills Interventions

Head and Trunk Control

The attainment of gross motor skills normally proceeds along a predictable sequence, with rapid changes occurring in the first year. Head control in various positions against gravity is critical to the future development of other skills. If head control is not emerging in the first few months of life, this should be specifically targeted for intervention (Fig. 6.23).

Trunk control is more complex, as there are many planes of movement through which the trunk must move. There must be adequate control of the trunk muscles before complex, antigravity movements of the arms and legs are possible. At a few months of age, effort should be directed at promoting trunk balance. Parents can be instructed in ways to hold and move their infants to encourage trunk use and thereby strengthen muscles. At first, infants may require considerable assistance, with the parent's hands placed high on the trunk for support and guidance. In time, supporting hands can move further down the trunk so the infant receives less support and is progressively challenged to work the trunk muscles to maintain positions and move.

Rolling

If an infant is not rolling by approximately 5 months, parents should encour age this activity by providing physical assistance. Once the infant begins to initiate this motion, he or she can be encouraged to roll by reinforcement with attractive toys or sounds.

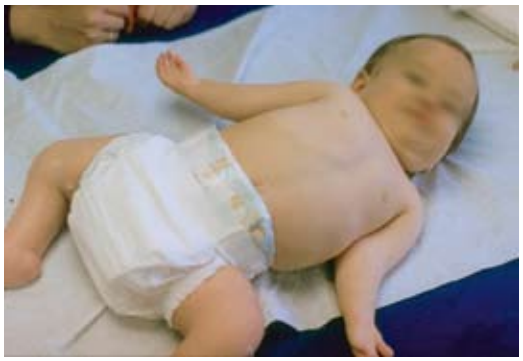


Fig. 6.24 Scooting in supine.

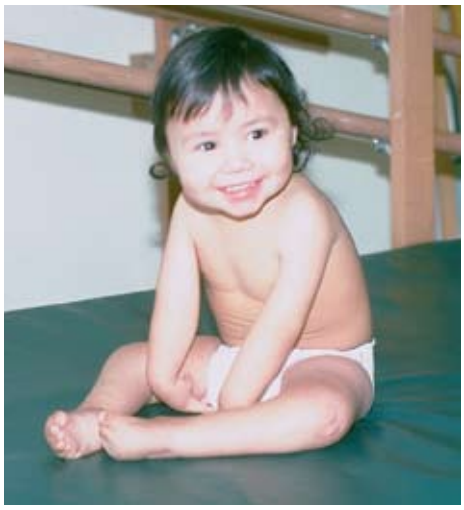


Fig. 6.25 Scooting in sitting.

Compensatory Movement

Again, depending on the level of joint and muscle involvement, the infant with arthrogryposis usually will not move spontaneously through the normal developmental sequence of motor milestones. Those infants with relatively more involvement often choose to move in alternative ways. For example, infants who do not develop good control of their flexor muscles, including abdominals, may not roll across the floor from stomach to back but may instead learn to use their stronger extensor muscles to scoot about in a back-lying position (Fig. 6.24). This activity should be encouraged as a first attempt at independent movement and exploration. Infants who lack good control of flexor and extensor muscles may never be able to crawl on hands and knees. However, these infants often learn to scoot on their bottoms once placed in a sitting position (Fig. 6.25). The relatively stronger extensor muscles of the back and neck are used to the infant's advantage during this type of movement.

If infants do choose alternative methods of movement, they can also be afforded the opportunity to experience more standard developmental positions. That is, if an infant scoots on his back rather than rolling, he should still be placed on his stomach for several periods of the day to allow him to work on head and trunk control and weightbearing through the arms. If joint range and strength allow a child to be positioned on hands and knees, this too is a good experience to promote head and trunk control and to allow weightbearing through the arms and legs.

Upper Extremity Use

Shoulder

The infant with limitations in shoulder range of motion may try to use trunk extension to help raise the arms. Restrictions in shoulder external rotation affect forearm and hand position. Extreme internal rotation combined with elbow extension can interfere with an infant's ability to see his fingers and bring his hands together in midline. If both arms are internally rotated, the hands may naturally oppose each other back to back. Only by crossing his arms can the child succeed in bringing his palms together for grasping.

Elbow

Lack of passive elbow motion usually indicates lack of muscle development. Extension contractures are most common, but flexion contractures also occur. Asymmetric upper limb involvement may provide an advantage. A flexed elbow can more easily reach the mouth, whereas an extended elbow can serve better for perineal hygiene and to help in mobility (Lloyd-Roberts and Lettin, 1970).

Infants who lack active elbow motion but have greater than 100° of passive flexion can use substitutions to flex their elbows. If they can actively flex their shoulders above 90° they may then use gravity to flex the elbow. Passive elbow flexion can also be achieved by wedging the arm between the edge of the table and the torso.

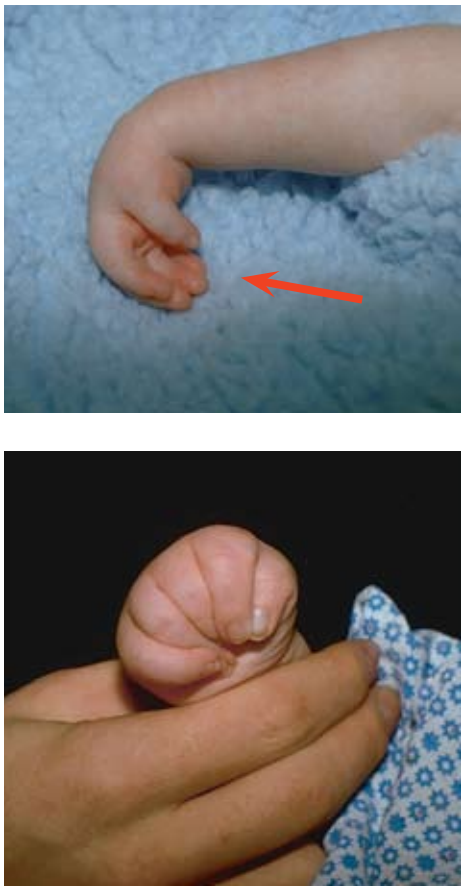


Fig. 6.26 Two subgroups of hand deformity.
(Top) In amyoplasia, typical pattern of deformity includes wrist flexion, ulnar deviation, curled fingers, and thumb in palm. (Bottom) In multiple pterygium syndrome, typical pattern of deformity includes fingers flexed and overlapped tightly and thumb adducted.

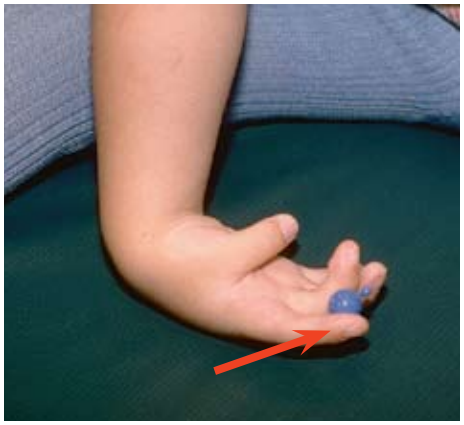


Fig. 6.27 Interdigital grasp.
A small bead is grasped using the index and middle fingers.

Wrist and Hand

Infants with amyoplasia usually have ulnarly deviated wrists with flexion contractures, stiff, slightly flexed or curled fingers, and adducted thumbs. In a subgroup of children who have trismus, or a stiff jaw, the wrists are flexed and MCP joints are hyperextended. In distal arthrogryposis and multiple pterygium syndrome, the hand is usually clenched tightly in a fist. The wrists tend to be extended, the MCP joints fully flexed and ulnarly deviated, the thumb adducted, and the fingers overlap one another (Fig. 6.26).

Some infants prefer to hold small objects between their fingers using an interdigital grasp rather than using the thumb in opposition to the fingers (Fig. 6.27). This is done for several reasons. The infant may have difficulty placing his hand where he can see the thumb and fingers (shoulder internal rotation and elbow extension contractures). The thumb may be positioned into the palm with limited abduction and decreased strength. Grasping with the fingers permits the infant to both see and hold the object. Although interdigital grasping is functional for an infant or toddler, it works poorly for grasping larger objects and may interfere with the development of the bimanual skills necessary for tool use (Fig. 6.28).

Fig. 6.28 Cylindrical grasp.
Pegs grasped in palm of hand.

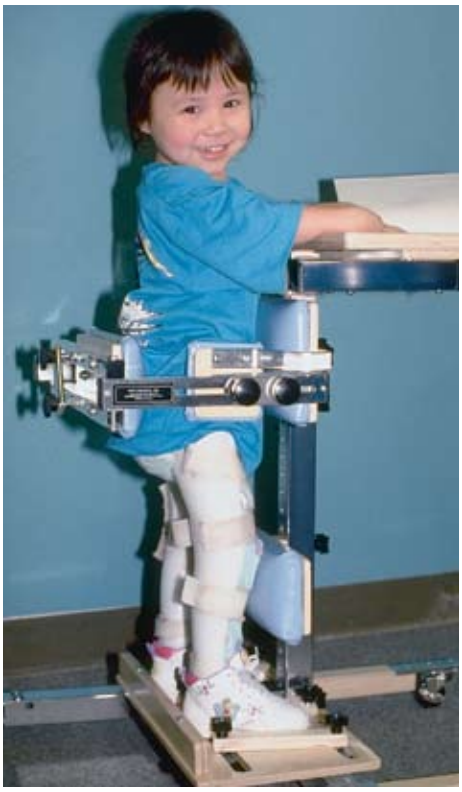


Fig. 6.29 Standing frame.
Can be used to position for fine motor and play activities, with or without long leg splints.

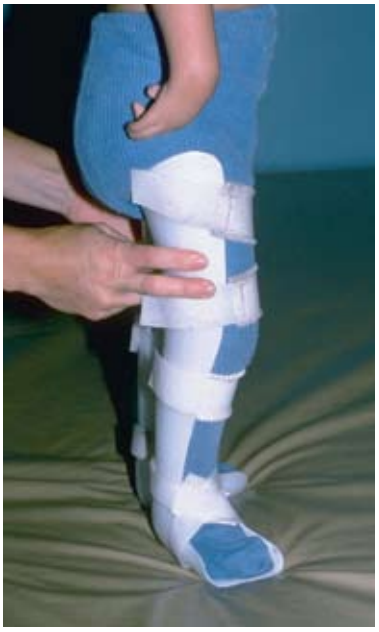


Fig. 6.30 Long leg splints.
High-temperature plastic molded in maximal knee extension and neutral ankle.

Toddler to Preschool Years

Toddler and preschool years are a time when locomotion and other motor skills develop rapidly. Interaction with peers becomes increasingly important as children begin attending day care, preschool, and other social group activities. Children learn about their world through self-initiated play and independent mobility. These important developmental motor skills may not be attained in the usual developmental sequence or time frame in children with arthrogryposis, and these motor and functional milestones become important therapeutic goals.

During these years, the frequency of therapy is usually decreased from the relatively intensive first year. The greatest gains in range of motion have usually been achieved by this time. Now the emphasis is on maintaining range of motion, increasing strength, and progressing with functional activities.

Range of Motion

Lower Extremity

Range of motion must be maintained through the growing years. Many children will continue to require gentle range of motion exercise on a daily basis. Night splinting and casting are often needed as well (Williams, 1978). Long leg night splints are worn to maintain knee extension and ankle dorsiflexion. These splints can be made of plaster, fiberglass, or plastic. Various types of standing frames are commercially available or can be fabricated to hold a child upright to stretch hip, knee, and ankle muscles. A standing frame also allows the child an alternate position for play (Fig. 6.29 and 6.30).

Upper Extremity

Full hand or wrist splints are often worn at night to maintain range of motion. Some children, especially those who have gained passive motion in wrist extension, will benefit from wearing functional wrist splints during the day (Fig. 6.31). Dynamic wrist splints work well during supervised fine motor play at home or school. Elbow splints are often still appropriate for this age group.

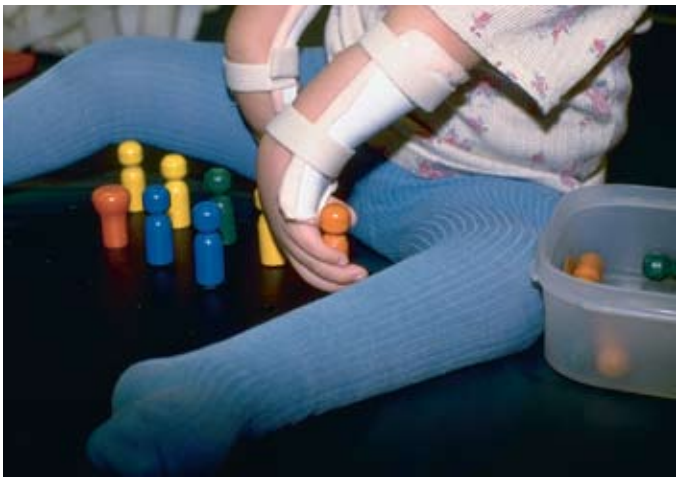


Fig. 6.31 Functional wrist splint.
Worn to minimize wrist flexion while allowing active finger and thumb motion.

Fig. 6.32 Supported tall kneeling.
(Top) Tall kneeling to evaluate and strengthen hip muscles. (Bottom) Modified position when 90° flexion is not possible.

Strength Assessment

Assessment of active motion or strength still requires observation of move ment. However, verbal requests combined with play can now be used to encourage a small child to move a body part in the desired way to assess muscle strength more accurately.

Strength Interventions

Lower Extremities

Tall kneeling is an important position for evaluating hip and pelvic muscle strength (Fig. 6.32). Control in this position helps predict future abilities in standing and walking. To both evaluate and improve this control, the child is positioned in tall kneeling, with the hips maximally extended and the forearms providing truncal support. Games can be played in this supported position to improve compliance. The eventual goal is independent, unsupported balance in tall kneeling. Sometimes this position is difficult to attain because of limited knee motion. In this case, a simple foam cut-out or wedge can be placed under the lower legs. Some children have contractures of the knees and ankles that do not allow them to attain a standing position. They may be candidates for orthopedic surgery to correct these deformities, especially if they have adequate trunk, pelvic, and hip control as demonstrated in the tall kneeling position.

Upper Extremities

Many children with arthrogryposis have hand weakness and poor grasping skills. They can benefit from activities that help to strengthen hand muscles. Various activities enjoyed by preschoolers can be incorporated into a strengthening program. Hand strengthening activities include water play with squeeze toys and sponges. Therapy putty can be used to promote finger extension, grasping, and hand strength. Some commercially available manipulative building toys, like Krinkle Blocks and Magnet Blocks, provide resistance but do not require precise alignment to connect. Cutting paper of various thicknesses with regular or adapted scissors can also help to build grip strength (Fig. 6.33).

Fig. 6.33 Hand strengthening activities.
(Left) Therapy putty. (Center) Krinkle Blocks. (Right) Adapted scissors.

Fig. 6.34 Built-up spoon handle.
The diameter of the spoon has been enlarged for easier grasp.

Fig. 6.35
Soft cuff for holding a spoon.

Fig. 6.36
Dorsal style wrist splint with palmar cuff supports the wrist in extension during eating.

Activities of Daily Living

Self-Feeding Assessment

Self-feeding is often a challenge for children with arthrogryposis, and independence in this area is often delayed. It is important to first assess the child's positioning for feeding. Has trunk stability been provided either in a highchair or at a table with an appropriately sized chair? Can the child's feet rest on the floor or other firm surface?

Developmentally, eating finger foods is the first step in the self-feeding sequence. Assessment should begin by presenting a variety of shapes and sizes of preferred finger foods in order to evaluate the child's ability to grasp with the thumb and fingers. It is important to look at whether the thumb can actively oppose the fingertips, allowing for a fine pincer grasp of a Cheerio, or whether the child tries to hold it between two fingers. Can he or she use thumb adduction to hold a cracker against the side of the hand? Grasping and holding a spoon or fork can be difficult for these children. Can the child scoop or spear food? What adapted equipment has been tried?

Next, hand-to-mouth movement is addressed. The combined motion of scooping and moving the hand to the mouth requires a sustained grasp of the utensil and movement of the elbow or shoulder or both to reach the mouth. Depending on the degree of upper extremity involvement, children with arthrogryposis may lack the muscles to combine these motions with ease. It is important to determine whether the potential to perform any part of this sequence exists. Does the child exhibit active elbow flexion? If not, can he or she use substitution to move the hand close to the mouth?

Self-Feeding Interventions

A variety of adaptive feeding aids enable children with arthrogryposis to feed themselves successfully. A few of these aids are described here. Training involves trial and error combined with patience and encouragement. With appropriate family support, many children develop their own unique style of self-feeding.

If the child has difficulty grasping a spoon or fork, a lightweight cylindrical foam or built-up handle may help (Fig. 6.34). Enlarging the diameter of the handle permits grasping with less finger flexion and less effort. Children with minimal grasping abilities can use a custom-sized universal cuff fabricated from Velfoam or neoprene to keep hold of the spoon or fork (Fig. 6.35). Children who require wrist support can use a custom-made dorsal wrist splint with palmar cuff to grasp a spoon (Fig. 6.36). It is important to look at the positioning and angle of the utensil in the cuff. A long handle or small bend or rotation of the handle may be all that is needed to keep the food on the spoon on its path to the mouth.

A diverse array of adapted plates and bowls is available to facilitate independence in self-feeding. A scoop dish provides an elevated edge against which to push a utensil. Commercially available plates designed with 2-inch-high elevated sides have allowed some children to feed themselves independently. Movement of the arm in a see-saw or lever-type motion enables the child to get hand to mouth. The child rests the forearm or wrist on the elevated plate edge and then lowers the elbow, which brings the hand up to the mouth. A 2-by-4 inch block of wood resting on the tabletop under the child's forearm can be used in much the same manner (Fig. 6.37). Both methods require problem solving and several trials to determine the correct positioning for success. Other children wedge their elbows down between the table edge and their torsos to passively flex their elbows up toward the mouth (Fig. 6.38).

Fig. 6.37
Elbow block for feeding helps position the hand closer to the mouth.

Fig. 6.38
Using table edge to flex elbow and get hand to mouth.

Fig. 6.39
Flexible straw connects nipple in mouth to bottle on table.

A table with a cutout for the plate or bowl is another design used to help a child scoop. Other unique feeding devices have been described in the literature (Wyckoff and Mitani, 1982; Hall and Hammock, 1979).

Drinking may present another challenge. Various types of straws are usual ly the simplest solution (Fig. 6.39), but no-tip cups and cups with handles are worth trying.

Dressing Assessment

Dressing presents another challenge for the child with arthrogryposis. Depending on the degree of upper limb involvement, age-appropriate independence in dressing is often not a realistic expectation. It is important to assess what tasks the child can do independently and to begin training in areas that show the potential for improvement.

Dressing Interventions

Each child will develop a unique way to dress and undress. The child often expresses a desire to participate in this process and should be allowed to help as much as possible.

Appropriately chosen clothing styles with modified closures and the use of specific dressing aids are key to the child's success in dressing. Loose-fitting pull-on style T-shirts, Velcro tabs and closures, zipper pulls, and sock aids are just a few things that may help. Other aids include a dressing frame (Fig 6.40). The dressing frame will support a shirt in an upside-down position, allowing the child to bend at the waist and slide into it. Some children with diminished grip or pinch strength can slip their hands into a loop attached to the waistband of pants or underwear to raise them. For children with good standing balance, hooks placed on a wall or other vertical surface can help with raising or lowering pants. Attaching the hook with its end pointing up at about the child's thigh height will allow him to lean against it and catch the waistband to help raise the pants. Attaching it pointing downward just below waist height will help in lowering the pants. Again, exact placement and technique must be worked out individually.

Fig. 6.40 Dressing frame.
T-shirt suspended upside down on frame makes it easier to slip into.

Fig. 6.41
Toilet grab bars.

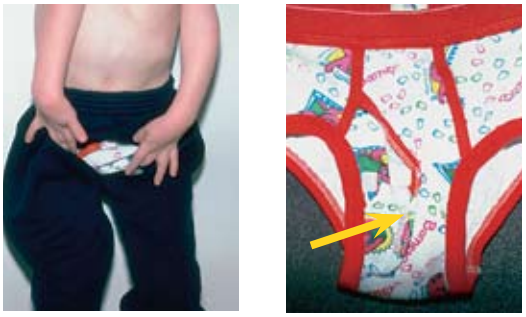


Fig. 6.42 Adapted clothing.
(Left) Front opening secured with Velcro in sweatpants. (Right) Fly of underwear enlarged along the edge of leg opening and closed with Velcro.

Fig.6.43
Scooting in prone.

Toileting Assessment

Toilet training need not be postponed in children with arthrogryposis. If the child is able to communicate the need to go "potty," it may be appropriate to begin training. However, independence in toileting directly relates to the child's mobility and lower extremity dressing skills. The caregiver may have to help the child get to the bathroom and onto the toilet, manage clothing, and wipe as needed. It is important to delineate the specific tasks for which the child requires assistance and to encourage independence whenever possible. The most common areas of difficulty seem to be in wiping after a bowel movement and raising the pants.

Toileting Interventions

The mechanics of toileting can be divided into four areas: getting to and from the bathroom, transferring on and off the toilet, managing clothing, and toilet paper access and wiping. Where to begin training for independence in toileting depends on which of these tasks the child needs help with.

For toilet transfers, a small platform, a stepstool with or without handles, or toilet grab bars can be helpful (Fig. 6.41).

Clothing management is different for boys and girls. For girls, underwear with loose elastic waistbands may be easier, especially if worn under a skirt or dress. For boys, standing to urinate minimizes the need to execute a toilet transfer and to lower and raise the pants. If the boy can learn to manage his clothing when standing, he can urinate independently. Independence in clothing management can be achieved as long as the trousers and underwear can be altered appropriately. Pants can be adapted with a large plastic zipper and a zipper-pull or Velcro tabs. Enlarging the front opening and adding a small Velcro tab will make the boy's underwear more accessible (Fig. 6.42).

Toilet paper access and wiping are often a challenge. For a toddler or preschooler, independence in wiping after a bowel movement is often not a realistic expectation. If a child of either sex wants to wipe, it may be easier to do so from front to back. Training to develop access to the perineum is done by placing a small toy under the child's bottom and asking him to reach between his legs to retrieve it. Some girls may be able to wipe after urination if an appropriate amount of toilet paper is placed within their reach.

Gross Motor Skills Assessment

All methods of mobility, such as crawling, rolling, scooting, cruising, and walking, as well as their associated transitional positions, should be reviewed (Fig. 6.43). Transitions from supine or prone to sitting up and between sitting and standing are as important as crawling and walking. If the child is not able to assume the starting position (sitting, standing) independently, he or she has not truly achieved independent mobility.

Gross Motor Interventions

Sitting

Many children at this age are able to sit well if placed in a sitting position but may not be able to attain a sitting position on their own. The transition from supine or prone to sitting up is critical in the development of independent mobility and may take many more months to achieve (Fig. 6.44). The combination of decreased abdominal strength and limited strength and range of motion in the arms makes this movement a challenge. There are a number of strategies that can help, and success depends on the child's specific limitations. Offering graduated foam wedges or pillows to lean against before pull

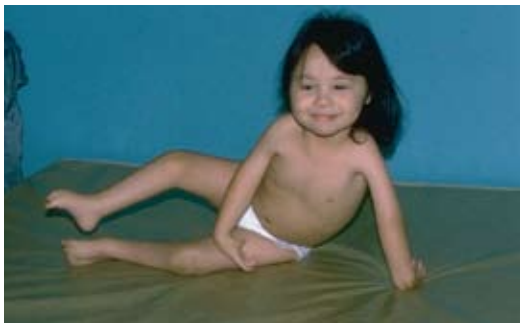


Fig. 6.44
Moving from side lying to sitting.

Fig. 6.45
Assisting child to a sitting position.

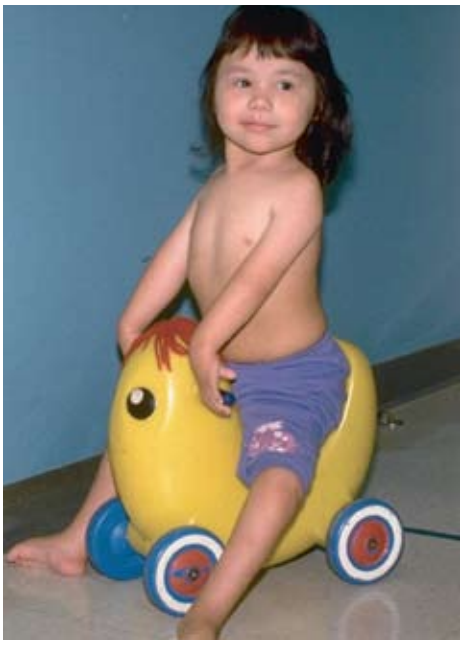


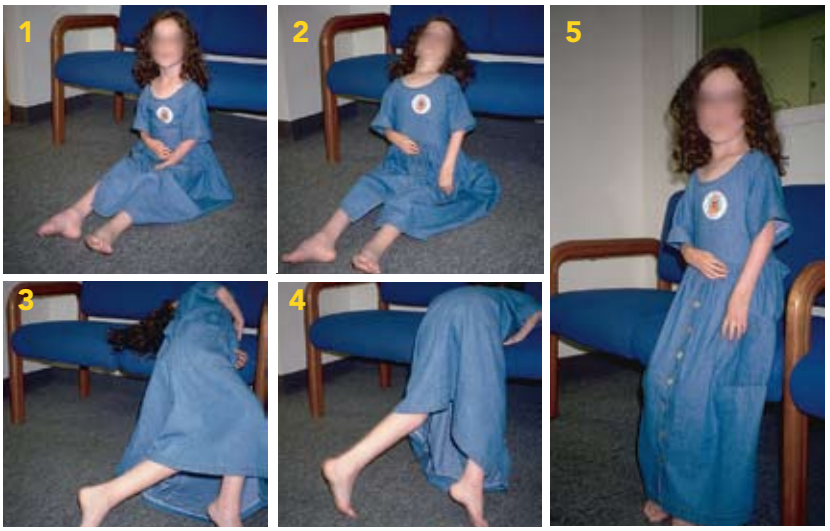
Fig. 6.46
Independent mobility using a ride-on toy.

ing to a sitting position makes this transition easier than moving all the way up from the floor (Fig. 6.45). Gradually, smaller wedges or pillows can be used until no extra prop is needed. Some children learn to move straight up to sitting from supine, whereas others do better from lying on their sides. Some children have enough hip range of motion to attain sitting by spreading their legs and pushing up from their stomachs. Others use their relatively stronger neck and trunk extensor muscles to sit up. They are able to position themselves in front of a heavy couch or a wall and sit up by pushing their heads progressively higher on the surface. It is important for parents to encourage their child to assist with this movement every time he sits up rather than allowing the child to be completely dependent and passive.

Mobility

Toddlers and preschoolers spend their time in many positions and places during the day: on the floor, crawling, sitting on furniture, standing, walking and running, playing on push toys and tricycles, moving up and down stairs, and playing outside (Fig. 6.46). These positions and movements teach children about how their bodies move and about their environment. They also permit important interactions with peers. Therefore, it is important to teach children with arthrogryposis how to make the transition from one position to another to allow this independent exploration and learning. Moving into a sitting position, from the floor onto furniture, from a wheelchair into bed, or from a chair to a standing position should all be encouraged and practiced. Some children are unable to perform all transfers independently, even with the use of adapted equipment. Through practice and problem solving, children often develop their own successful methods (Fig. 6.47) .

If children are unable to develop efficient, functional, and self-initiated mobility at approximately the same age as their peers, mobility devices should be considered seriously. Various toys and adaptive equipment can be used to improve independence. Some children are able to use ride-on toys by pushing their feet on the floor. Push toys or wagons sometimes provide enough stability for walking short distances. Children with significant leg involvement but good arm and hand control can successfully push themselves in small, lightweight wheelchairs.



6.47 Moving from floor to standing.
One method to independently stand up from the floor.

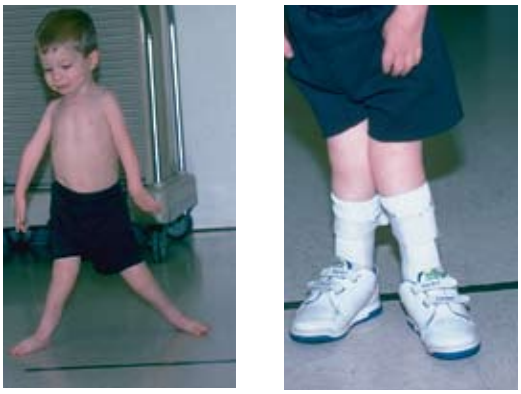


Fig. 6.48 Ankle splint (or AFO).
(Right) Provides improved foot position and stability for walking.

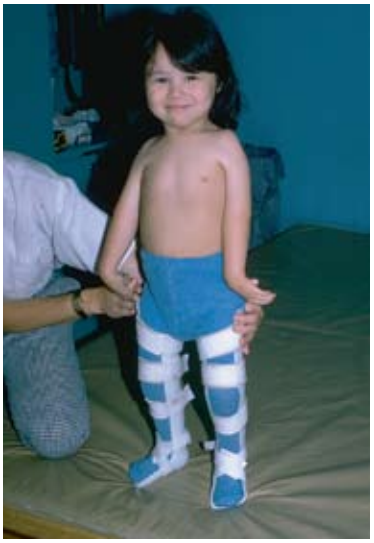


Fig. 6.49 Long leg splints (or KAFOs)
Posterior splints without knee joints provide stabilility for walking.

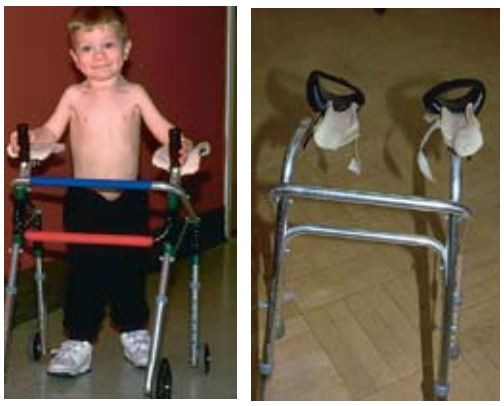


Fig. 6.50 Adapted walkers.
(Left) Walker mounted with forearm trough supports.
(Right) Walker with custom-molded full hand splints.

Assistive Devices for Ambulation

Various types of splints can be used to provide lower extremity support and stability for children who cannot walk alone. Occasionally, a foot deformity is the major limiting factor preventing standing and walking. In this case, an ankle-foot orthosis (AFO) may provide increased stability and a better weightbearing surface (Fig. 6.48). If there is some flexibility in the foot, the splint may be used to hold the foot in better alignment for walking. More often, there is insufficient ankle and knee strength for walking, necessitating the use of long leg splints or knee-ankle-foot orthoses (KAFOs). For very young children, a long plastic splint without knee joints is generally used because it is simple and lightweight (Fig. 6.49). As the child's legs grow longer, he or she can use a splint with metal knee joints that are locked in extension for walking, but can be flexed for sitting.

Children who wear KAFOs often need walkers. A walker allows weightbearing through the arms in order to maintain balance. A rolling walker with a fairly wide wheelbase is generally used to accommodate a typically wide-based walking pattern. Some children can use the standard walker grips, whereas others need arm troughs or wrist splints attached to the walker to allow weightbearing through forearms rather than hands (Fig. 6.50). Few children have sufficient strength in their arm, trunk, and pelvic muscles to use canes or forearm crutches.

Many children with arthrogryposis use different means of mobility in different environments: scooting while sitting on the floor at home, walking with a walker and splints at preschool, pushing a manual wheelchair outdoors. There may be some situations, such as taking a long walk, playing on the playground, or shopping, for which the child still does not have the endurance, balance, or strength for functional community mobility. Some children may still have no independent mobility at all. In such cases, power mobility may be a good option. Children as young as 24 months have learned to propel themselves in power wheelchairs (Butler et al., 1983). This early experience with power mobility does not appear to prevent children from making continued gains in gross motor skills. Battery-operated toy vehicles may be a first option for a child to use around the home and yard. If a power wheelchair is needed for longer distances, the child should be evaluated by a pediatric rehabilitation specialist, who can prescribe the most appropriate equipment to meet the child's needs. Whenever a young child is provided with a power device, constant adult supervision is required during its use.

Upper Extremity Function Assessment

The use of standardized assessment tools to evaluate fine motor skills may not be appropriate, depending on the degree of muscle and joint involvement. Areas to assess include grasping and bimanual activities, such as stringing beads and cutting with scissors. Does the child use both hands or tend to use only one? Is he able to oppose the thumb to the fingertips, demonstrating a fine pincer grasp to secure a small bead, or does he prefer to use an interdigital grasp? Can he grasp a marker or pencil and continue to hold it? Can he color on paper? Would the use of an adapted writing aid allow the child to maintain grasp of the marker?

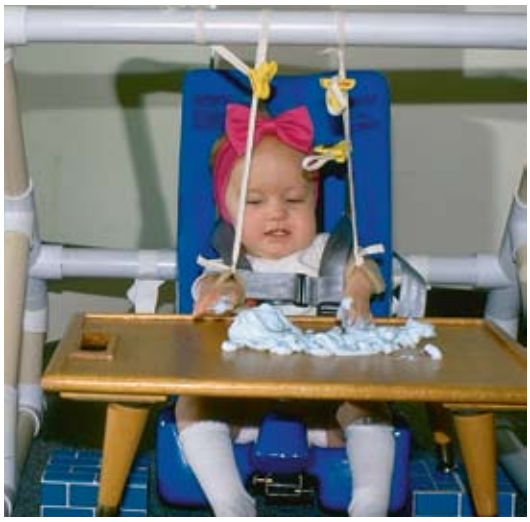


Fig. 6.51 Positioning for hand use.
Custom-made overhead support allows greater arm movement.

Upper Extremity Function Interventions

Again, positioning for hand use is extremely important for children who lack elbow or shoulder flexion. If the child has good sitting balance, placement of a puzzle or toy on the floor between the child's legs may allow greater freedom of arm movement. Play in this position allows the child to use the trunk to move the arms in a pendulum-type motion for reaching and grasping. Overhead slings or suspension support systems can help the child to see his or her hands and move them for play activities (Fig. 6.51).

Many children sit well without any adaptive equipment. Others need trunk support or foot support to provide enough stability to do precise work with their hands (Fig. 6.52). Legs that are dangling off a chair do not stabilize the trunk as well as feet that rest on a firm surface. Children who have a limited range of knee flexion may also need some support if their feet do not reach the floor.

Therapy and training to maximize fine motor development are highly individual and depend on each child's area of need. Treatment should be aimed at enhancing eye-hand coordination, facilitating grasping, and maximizing bimanual skill development. If, at an early age, the child shows a strong hand preference, he or she should be encouraged to use both hands in order to develop age-appropriate skills in the nondominant hand as well.

To encourage grasping abilities, it is important to select developmentally appropriate activities and toys for the child. Size, weight, and texture of the objects should be considered, as lighter, softer toys may be easier to grasp. If the child prefers to use an interdigital grasp for small objects, pinching and squeezing activities can be used to increase thumb opposition strength and eventually enable thumb use in grasping.

Coloring may be easier for the child with large-diameter markers or regular markers that have been built up with cylindrical foam. Adapting the markers with Velcro so they can be held in a custom-made cuff may also be needed (Fig. 6.53).

Preschool children with arthrogryposis often benefit from enrollment in a regular preschool program. Preschool can provide the opportunity to develop fine motor abilities through participation in many activities, including play with puzzles, stringing beads, building with blocks, coloring, drawing, painting, and cutting.

Fig. 6.52 Foot support at table.

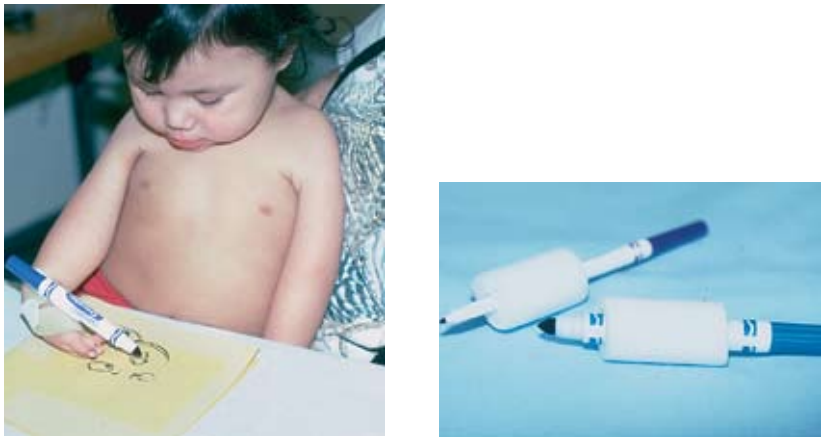


Fig. 6.53 Adapted writing devices.
(Left) Marking pen secured with a soft writing cuff. (Right) Plastazote foam will increase diameter of markers for easier grasp.

Fig. 6.54 Exploring ways to swim.

Recreation

Recreation and play comprise a large portion of toddlers' and preschoolers' days. These activities provide the child with opportunities to exercise, social ize with peers, and boost self-esteem (Sawatzky, undated). A favorite recreational activity for children with arthrogryposis is swimming, as almost all children can participate at least to some degree without the use of special equipment (Fig. 6.54). Some children have been able to ride bicycles, although these often need to be adapted. A recumbent bicycle will accommodate limitations in leg motion, or a hand-propelled bicycle may be chosen.

Early School Years

When children reach school age, it is important to include them in discussions about surgical plans, changes in therapy, and choice of adaptive equipment. Although they are not yet old enough to make these decisions independently, it is important for them to have some input and to express their opinions. Therapeutic goals will best be met if the child has some role in setting them and is given a sense of responsibility in working toward them.

Range of Motion

Passive range of motion measurements must be made to determine if children are maintaining joint motions over time. Although the importance of daily range of motion and stretching exercises begins to decrease at this age, it is still present, particularly for those children who have had surgery to change joint range. Gentle stretching, in combination with splinting to maintain the new position, is needed. Splinting is gradually decreased to night use only. Range of motion must be monitored during this time to ensure that a decrease in splinting time does not produce a loss of joint range.

Strength Assessment

A specific muscle strength evaluation should be possible at this age. Many children learn to use other muscles to substitute for their weaker muscles. These substitutions can be quite functional yet should be documented to show that a motion can be approximated, but not completed by the usual primary muscle.

Strength Interventions

The strengthening of specific muscles plays an important role in some situations. If a child demonstrates less than normal strength in a muscle, exercises may increase the strength of that muscle. Functional activities, such as scooting, standing, and swimming, when incorporated into the child's day, strengthen as well. Specific muscle strengthening may result in improvement in a new arc of movement if a child has had surgery to change joint motion. In any case, it is recommended that any muscle strengthening program be designed to improve function and continue on a regular basis for a finite period, perhaps 2-3 months. At the end of that period, muscle strength should be reevaluated to determine if muscles have responded and functional goals have been achieved.

Activities of Daily Living

Feeding

The ability to self-feed a variety of foods with or without adaptive aids is often mastered by this age. However, some children may still need to be fed specific foods, like soup or chili, that may be easily spilled. Cutting meat, opening containers, and other two-hand feeding activities are often difficult. It is important to assess how the child manages his or her lunch at school. If the child has difficulty obtaining a lunch tray or opening containers at school, he or she may be comfortable asking a peer for assistance. Other children may need an individually assigned teacher's aide or may choose to bring a lunch from home that they can manage without assistance.

Bathing

Independence in bathing becomes more of a priority as the child matures. At the same time, transferring the growing child in and out of the bathtub becomes an increasing challenge for the parent or caregiver. Assessment of bathing equipment needs may result in a recommendation for grab bars, a tub bench, or a hand-held shower. Appropriately chosen equipment will help ensure the child's safety and reduce caregiver assistance. An in-home assessment is often the most accurate way to determine what will work best. A small tub bench or larger transfer tub bench can eliminate the need for the parent to lift and lower the child in and out of the tub. Some children prefer to shower, but lack the endurance or balance to stand in the shower. For them, appropriately placed shower grab bars or a small shower bench combined with a hand-held shower will provide a safer shower environment.

Hair washing may be difficult because of limited shoulder motion and strength. Some children have enough trunk and hip flexibility to bend forward and lower their heads down to their hands. Others may find a lightweight, long-handled, angled brush helpful for reaching the top and back of the head. A wall-mounted soap dispenser may be easier to use than bar soap.

Toileting

Independence in toileting becomes increasingly important for the school-age child. School restroom access must be ensured. Can the child open the door to the restroom? Are the stalls fitted with grab bars? Can the child reach the toilet paper and wipe after a bowel movement? Is there a teacher's aide available to help the child if and when needed?

Lower extremity clothing management is critical to the child's independence in this area. Clothing adaptations can be more successful for this age group because of increased motivation to be independent at school. Providing an accessible restroom at school is the responsibility of the school district. The child's therapist and parents should be involved in order to determine exactly what the child needs (e.g., grab bars, toilet paper holder) and where it is best located. It is important that the child be able to obtain help when needed.

Fig. 6.55 Hairbrush adapted with a Velcro cuff.

Personal Hygiene

Can the child reach and control the faucet handles at the bathroom sink? Is he able to brush his teeth with or without help? Can he comb his hair? Lever style faucet handles may be easier for a child to operate than standard grip and turn handles. A Velcro cuff or built-up handles on the toothbrush and hairbrush may require less effort for the child to hold (Fig. 6.55). If reaching over the head is difficult, the brush can be mounted on a movable gooseneck secured to the counter.

Gross Motor Skills Assessment

Mobility is a focus of evaluation at this age, especially as the child begins school and increases time away from home and family. If a child is able to walk, then speed, terrain, endurance, and frequency of falling are documented. If the child uses a wheelchair, the fit and condition of the wheelchair are evaluated, as well as the child's ability to propel it over various distances and terrains. School and home accessibility are also discussed, and any areas of difficulty are noted.

Lower extremity orthopedic surgeries are sometimes performed at this age to allow or improve walking. Thorough assessments of the new joint range and strength are conducted. The need for new adaptive equipment for walking should be evaluated. The new position of the legs may also necessitate changes to chairs or wheelchairs for accommodation and to improve positioning.

Gross Motor Skills Interventions

Alternative Methods of Mobility

Children who are independent household or short-distance community ambulators and use strollers for long trips may be unable to keep up in school, where the demands on their walking increase. At this time, it is important to provide an alternative means of mobility for longer distances. This may be as simple as a rolling walker, or it may mean the addition of a manual or power wheelchair.

Children who have good sitting balance and only need a power device for occasional use do well with a three-wheel power scooter, which is easier to transport in a family car than a standard power wheelchair. Those who require more complex seating for function and need power mobility more consistently will do well with a power wheelchair at this time.

Independent access to the child's home and school is an important issue. Suggestions are often made to add ramps at entrances and rails at stairs, permit use of school elevators, or change floor coverings to allow increased independence.

Postoperative Mobility Training

For the child who is beginning to walk for the first time after orthopedic surgery, a period of intensive physical therapy is often necessary. The therapist focuses on gentle stretching and on strengthening exercises, if appropriate,

Fig. 6.56 Postoperative weightbearing in pool.

and consults on the fit and use of orthotics and other aids, as well as gait training. Pool therapy is an excellent intervention during this postoperative period, as the water can be used for support in standing, to provide resistance for muscle strengthening, and to facilitate active movement (Fig. 6.56). Involving family members in therapy sessions makes them better able to continue appropriate training with the child at home. School physical therapists can be consulted to continue the gait training program in the school setting.

Upper Extremity Function

Handwriting Assessment

Handwriting becomes more of an issue for children as the demand for written work steadily increases. Many children with arthrogryposis have difficulty with the mechanics of handwriting and are unable to keep up with the writing demands. The mechanics of handwriting involve effort, time, and legibility. The effort required to write increases when there is decreased upper extremity stability and difficulty isolating and controlling movements. Handwriting speed and endurance are diminished when a considerable amount of effort is required, and legibility or readability may be poor.

Handwriting Interventions

An adaptive writing aid can enable a child to position and sustain a grasp on the pencil and can be extremely helpful in the classroom setting. The writing aid must be comfortable and durable, and the child should be able to put it on independently if possible.

For children with severely limited hand control who tend to use their noses, lips, or tongues to type or move objects, a mouthwand held in the mouth can provide another option for keyboard typing and writing (Fig. 6.57). It has a custom-made mouthpiece that is held between the upper and lower teeth and typically has distal attachments for a pen, pencil, or paint brush.

Fig. 6.57 Mouthwand.
Used to access a computer keyboard.

Computer Access Assessment

Computer access is a topic of increasing interest to parents, therapists, and teachers. It is important to include all team members in order to ascertain the following information before evaluating the actual physical aspects of computer access. Where will the child be using the computer (in the classroom, resource room, or home)? What type of computers are available for the child's use at school, and are they compatible with what the family has or may obtain for home use? What will the child be using the computer for? Is access needed for completion of assignments in the classroom or for work at home?

Fig. 6.58
Articulating forearm supports allow easier access to the entire keyboard.

Fig. 6.59 Computer access.
(Top) A HeadMaster Plus is used to move cursor, and puff switch replaces mouse button. (Bottom) With trackball, only one finger is needed to move cursor.

Computer Access Interventions

Positioning

The child with isolated finger control may be able to use a traditional keyboard if the keyboard can be lowered to a position below the table surface, allowing him or her to activate the keys with the elbows extended. Angling the position of the keyboard or suspending it vertically will enable a mouthwand user to see the keys and screen simultaneously. Another inexpensive way to help arm positioning may be to use a foam wrist rest. If a greater amount of wrist and forearm support is needed, articulating forearm supports that clamp onto the table surface may work (Fig. 6.58). The forearm supports swivel for movement over the keyboard to help the child reach all the keys with less arm fatigue. Ergonomic systems for support of the neck, back, and feet are also available commercially.

A keyguard is a clear acrylic overlay that lets the user rest the hands directly over a standard keyboard. It allows for more accurate and less physically demanding typing.

Assistive Hardware and Software

An on-screen keyboard is a software program that replaces the traditional entry method by displaying a keyboard on the computer's monitor. Movement of the cursor to the desired key can be done with a mouse, a trackball, HeadMaster Plus, or other mouse-type device (Fig. 6.59). On-screen keyboard software is available for Macintosh, IBM, and IBM-compatible computers that use MS-DOS or Microsoft Windows. A trackball is an input device that can be used instead of a mouse, consisting of a rolling ball mounted in a solid base that can be positioned for easier reach. Trackballs have been very helpful for children with limited elbow flexion or little active hand motion, and for those who have difficulty grasping and lifting a traditional mouse. A few commercially available models include easy-to-use custom software for slowing the cursor or programming the mouse buttons so that one side is interpreted as a "double click" and the other as a "click and drag," or other options as needed. A HeadMaster Plus replaces the mouse with a lightweight headset, providing access for individuals who cannot use their hands but have good head control. It works with different brands and also with laptop computers. The HeadMaster Plus moves the cursor to where the user looks on the screen, imitating a desktop mouse. A puff switch or other external switch operates the primary mouse button.

Speech recognition may be another option for accessing the computer without using hands. A variety of voice-activated software programs is available commercially. The child's success with voice activation or any of the other options depends on the interaction of many variables, including how well the child has been trained.

6.60 Recreation.
Brothers racing together on a track.

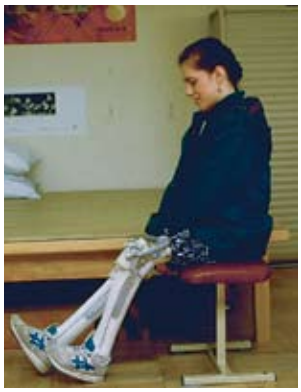


Fig. 6.61
KAFOs with knee joints for stability in walking.

Fig. 6.62
Bivalved fiberglass long leg night splints.

Recreation

Recreational activities continue to be suggested for the school-age child. In addition to swimming and perhaps bicycling, other options include snow skiing (children may benefit from consultation with an adapted skiing pro gram offering lessons in various ski techniques and adapted ski equipment), horseback riding, and wheelchair sports if the child has adequate arm and hand strength (Fig. 6.60).

Teenage Years

Teenagers with arthrogryposis are like all adolescents. They do not like to have differences in their body appearance or function pointed out. They are also struggling with issues of emancipation from their parents and prefer to make their own decisions according to their own timetables. These issues must all be considered and respected when proposing and carrying out treatments. It is important to give teenagers as much responsibility for their actions and decisions as they are able to manage. Physical and occupational therapy at this age is generally not scheduled on a regular or ongoing basis. Teenagers are usually evaluated yearly, or less often, and may receive some limited consultative services through the school district therapist to address school-related issues. An exception might be a teenager who has undergone recent orthopedic surgery to improve mobility skills and might need a short burst of more intensive therapy.

Range of Motion Assessment

As children move into the teenage years, it is important to continue to monitor range of motion and strength. If any changes have taken place in these areas and affected function, they should be addressed. A review of gross motor skills is made, with particular emphasis on independent mobility. Any adaptive equipment being used for positioning or mobility is reevaluated in terms of adequate fit and function.

Range of Motion Interventions

Self-Stretching

Joint range of motion is generally fairly static at this age and should not require much effort to maintain. A gentle self-stretching program can be learned and carried out by the more motivated teenager as a component of general conditioning and health. A more specific, targeted stretching program is used for teenagers who have recently undergone orthopedic procedures.

Splinting

In teenagers who are postoperative, splinting is often used at night and perhaps during part of the day (Figs. 6.61 and 6.62). For other teenagers with arthrogryposis, splinting is generally not used for stretching or maintaining range, but may be used if it provides an improvement in function, such as walking.

Strengthening

Specific strengthening programs are not usually advocated at this age. However, general conditioning and aerobic exercise in some form are recommended. Swimming, bicycling, and walking can be good choices. This is also an important time to address weight control through a combination of a well-balanced diet and exercise. Excess weight can mean the difference between being independent in transfers or not or being able to walk with assistive devices or not.

Activities of Daily Living

Kitchen

Evaluation of kitchen access becomes important for this age group. Reaching and grasping items from high or low cupboards may be difficult. Cutting and chopping can be impractical tasks for those with limited hand function.

The goal of intervention is to facilitate mobility and safety in the kitchen. Rearranging the cupboards can enable a teenager to better reach the items he or she needs. A small cart can be used to help move crockery, cookware, and food around the kitchen. Gadgets designed to help persons with hand weakness can be used by adolescents with arthrogryposis. Many adaptive aids are available commercially, including several styles of knives for cutting meat and chopping foods. Trial and error is needed to see what works best.

Dressing

Can the teenager independently don shoes, socks, or hose? A bra adapted with Velcro can be easier to close than one with clasps or hooks. Sewing the closure together may allow a girl to don a bra independently by pulling it on overhead. A stocking aid and long-handled shoehorn are inexpensive items to help with donning hose and shoes.

Personal Hygiene

Assessment of teenagers needs to include the management of hair and nails, shaving, and the application of makeup. An electric razor may be adapted with a Velcro cuff or mounted on a gooseneck for independence in shaving. A universal cuff or built-up handles may enable a teenage girl to grasp eyeliner or lipstick. Adaptive aids are available for flossing teeth and for holding a nail clipper and file to facilitate independence in grooming and hygiene.

Gross Motor Skills

All mobility possibilities are explored in an attempt to provide the greatest independence possible in the many settings of a teenager's life. This may mean different modes of mobility to accommodate the various settings. Assistive devices may not be needed for walking around home, but a rolling walker may help for longer distances at high school, and a power scooter may be used for community access. One teenager may be happy to use this array of equipment if it allows independence, whereas another may not like the appearance of certain equipment and may just as soon be pushed by a friend while sitting in a lightweight manual wheelchair. Provide the opportunity for the teenager to try recommended equipment and listen to his or her concerns and questions before ordering equipment to avoid abandonment of prescribed equipment (Fig. 6.63).

Computer access needs may change as children enter junior high and high school. They need to be able to use the computer efficiently in different class

Upper Extremity Function

Computer Access

rooms for a greater variety of tasks. A lightweight notebook-style computer with carrying case should be considered for this age group. Word prediction and abbreviation expansion are just two of the many software options available to improve word processing speed. Abbreviation expansion allows short sequences of letters, such as "N" and "A," to stand for longer words or phrases, such as "Name" and "Address."

Recreation/Independent Living Skills

Access to public transportation and driving are areas to be considered as independence in the community becomes important during the teenage years. Recreational activity continues to be valuable at this age. However, it is often difficult to entice teenagers into trying new activities. Opportunities are best offered at an earlier age when children are more receptive to these suggestions and eager to develop new skills. Certainly there are some teenagers who are interested in trying sports for the first time or who want to change sports. All reasonable possibilities should be explored and encouraged. Activities may include all of those previously mentioned, as well as other adapted water sports, boating, and ball sports.

Summary

This chapter has described the importance of physical therapy and occupational therapy for children with arthrogryposis. Developmentally appropriate assessments and interventions are outlined in an attempt to provide guidance and education for families, clinicians, and educators in their interactions and care of children with arthrogryposis. The unique qualities and circumstances of each child and family must be appreciated in planning the child's present therapy program and future direction.

Fig. 6.63
Independent mobility provided through use of a power wheelchair.

Chapter Contents **Page**

D.L. Hill, Ph.D.

Introduction

New parents of a child with arthrogryposis face a special set of challenges: they not only must learn to parent but also to parent a physically different child. Most find arthrogryposis an unfamiliar diagnosis that potentially limits the ability of extended family and friends to offer support and information. Grandparents, friends, and neighbors may not know how to respond. The celebration of the baby's birth is colored by a spectrum of emotional reaction to the baby's physical condition and the difficulty in seeing a positive future for the child.

From the earliest moments after the birth of a child with arthrogryposis, professional efforts can play a key role in child and family adaptation. Immediate intervention through sharing information, providing realistic hope, and dispelling misconceptions can lay the foundation for an ongoing parent-professional partnership. Such a relationship not only offers the family and child valuable support, especially during the early years, but also enhances the family's rehabilitation efforts for its physically disabled child throughout his or her development.

Every child matures within a unique social context of family, peers, school, and community, and each child has basic universal needs as well as individual and familial strengths (Fig. 7.1). For a child with arthrogryposis, the physical disability and array of interventions (surgeries, splinting, physi-

Fig. 7.1 Adopting a broad perspective.
Keep the whole child in mind throughout his or her development.

cal therapy) used to treat the condition can affect the child's emotional and social development as well as family functioning. Whether family functioning is positively or negatively affected depends largely on the manner in which intervention strategies are applied.

Clinicians and families are most effective when they appreciate the myriad factors that influence the development of a child with a disability and support the child's ever changing coping abilities (Kazak, 1989; Sameroff, 1993). Multidisciplinary, family-centered intervention, which focuses on the whole child and his or her unique strengths and challenges, leads to a more effective family-professional partnership and positive outcome than intervention focused strictly on the disability.

The information in this chapter is based on current knowledge and standards of care for children with arthrogryposis, their families, and those who care for them. In this context, "family" may refer to birth parents, grandparents or other family members, step-parents, or legal guardians. The information also may be valuable to those who work with children with arthrogryposis, including allied members of the health care team, child care providers, teachers, and others seeking information to better understand the child and help him or her realize success and personal growth in social, leisure, or career interests.

This chapter opens with a general overview of factors that influence family well-being when a child with a disability joins the family constellation. The overview is followed by three main sections that focus on the critical phases of child and family development – infancy, the preschool and school years, and the transition years from adolescence to young adulthood.

Factors Influencing Well-Being

Fig. 7.2 Perceptions of difficulty.
Every child and family may feel differently about managing daily activities.

Adaptation patterns of children with arthrogryposis and their families have not been widely studied. Most children and their families adapt successfully to arthrogryposis, a testament to the resilience of both children and families. However, numerous observations of children with other chronic physical disabilities have shown they are at greater risk for behavioral or social difficulties. Whereas behavior problems may, at best, be viewed as a normal reaction to the life challenges of a chronic physical condition, other factors, such as family functioning, often are implicated (Breslau, 1985; Wallander et al., 1988, 1989; LaGreca et al., 1992; and Lavigne and Faier Routman, 1992).

A child's or parent's self-perceptions are powerful elements in emotional health and adjustment (Fig. 7.2). What feels overwhelming to one family may seem only a minor difficulty to another. Likewise, challenges that a family finds manageable at one stage of development may seem insurmountable to the same family at another time or period in development. Each family, with the help of clinicians, may benefit from assessing its own perceptions, including the child's, of his or her level of disability as well as degree of available social support via friends, spouse, extended family, and relations with school and in the community (Lazarus and Folkman, 1984; Behr and Murphy, 1993; Ireys et al., 1994).

More often than not, a family's perception of its situation more closely reflects the family's general functioning than an objective measurement of disability severity and support systems (Wallander et al., 1990; Barakat and Linney, 1992). Hence, a family who perceives the child's disability as manageable, despite the disability severity, likely will take measures to make it manageable on a routine basis for the child and the whole family.

Conversely, a family whose functioning may not be healthy may feel overwhelmed by the child's disability even though the family may have access to the same support and resources.

Understanding what can trigger behavior problems in children with arthrogryposis can help the clinician and family focus on prevention and intervention efforts. Research has shown that well-adjusted children with disabilities report positive family and parent-child relationships, sufficient interpersonal support, and strong parent-child problem-solving and coping skills, all of which influence the child's emotional health and that of his or her family.

Conversely, children who have developmental delays or a disability that more severely impedes a child's age-appropriate functioning (or both) may experience more emotional or social difficulties. Daily stressors for parents and children resulting from physical disability, economic hardship, family conflict, or other adverse circumstances are associated with child or family coping difficulties (Varni et al., 1989a,b; Quittner et al., 1992; Thompson et al., 1992a,b).

A combination of positive and negative influences in the child's environment can affect child and family functioning (Daniels et al., 1987; Hamlett et al., 1992; Sloper and Turner, 1993; conceptual reviews in Kazak, 1989; Harper 1991a, b; Lemanek, 1994). For example, a young child who makes few friends in school may find that special time with a favorite aunt provides him the support and encouragement to try new things. Hence, the child's growing pride in being able to do new things may give him the confidence to make new friends. An older child who is uneasy about the transition to high school may find an understanding classmate with whom to connect. The interpersonal and family stress of economic hardship may be mitigated by a playful child with a sense of humor. However, if the same child faces extra challenges in school and needs more attention from parents than is readily available, emotional or behavioral problems may emerge. Periods of normal changes in a child's development, such as beginning elementary school or leaving home for college, represent a time of both increased opportunities and increased vulnerabilities (Drotar, 1981; Willis et al., 1982; Garrison and McQuiston, 1989).

Families with unhealthy functioning may benefit from professional interventions. However, any intervention must take into account a family's unique needs, strengths, resources, perceptions, and character, and be individualized for each situation.

In the Diagnostic Phase

The birth of a child with arthrogryposis presents significant parental challenges. Although antenatal diagnosis alerts some parents in advance, most learn of their child's condition at the time of delivery. Feelings of disbelief, shock, grief, anger, and guilt are common (Thompson, 1986; Levy, 1988; Davis, 1993). There is a "violation of cherished assumptions": the opportunity to parent a "normal" child was expected, forming the basis for countless future plans (Affleck and Tennen, 1993). Sadness and grief may be reflected in tearfulness and fatigue. Shock may interfere with usual eating and sleeping habits. Anger may be projected at other family members or medical staff. Lack of understanding of arthrogryposis, fears about the child's future, and for some, initial diagnostic uncertainties add to parental distress.

Fig. 7.3 Helping families cope.
Parents and extended family will have many questions.

Immediate Days After Diagnosis

To help parents cope in the first days after diagnosis, the clinician should promptly arrange appropriate referrals based on the family's needs. As parents cope with their own emotional responses, they also must care for their child or children, maintain their spousal relationship, and address immediate intervention decisions (Fig. 7.3). The goals of professional intervention are to provide calm support, structured information, suggestions for direct action, and reassurance about the wide range of emotions that are normal for parents. Social workers, psychologists, and chaplaincy services can be helpful to parents and are available at most medical centers.

Immediate education can offer hope and reduce fears. Early family education must emphasize the non-progressive nature of arthrogryposis and the child's potential for a full, active, and happy adult life. This information will bear repeating, as grief and emotional distress can compromise the parents' ability to fully absorb this news.

Parents' Needs

After the initial diagnosis, clinicians can help parents through the early years by providing a variety of services that can ease their burdens. These include providing and coordinating necessary referrals for the child's medical care, meeting parent needs for verbal and written information about arthrogryposis as well as about general developmental issues, facilitating contact with other families who have raised children with arthrogryposis, and offering general assistance with problem solving (Bailey and Simeonsson, 1988; O'Sullivan et al., 1992). Informative and reassuring reading material can be given to parents on specific topics, such as arthrogryposis treatment options, and specific issues in parenting children with disabilities. Numerous resources, many obtained at no cost, can easily be made available to share with families, particularly regarding more common topics.

Families should be encouraged to seek access to the support networks or arthrogryposis groups that are available in several countries. Group support through meetings, newsletters, and telephone contact reduces isolation, allows expression and validation of many commonly shared feelings, and facilitates education. General Parent-to-Parent support networks and other resources may offer caregiver respite services and information to help parents obtain benefits for their child in schools or other service systems (Garland, 1993; Kupper, 1993; Poyadue, 1993). Family support networks also offer a forum to help families cope with the reactions of persons in the community who may consciously or unwittingly interact poorly with children with disabilities or their families (Jones et al., 1984; Simons, 1987).

The Power of Words

Professional comments can be powerful. Parents often create their own meaning of the material presented while learning of the possible causes of arthrogryposis. Causal explanations always should be framed in ways that reduce blame. A second meeting, after the concerned parents have had time to move beyond their initial shock, provides an opportunity to explore their understanding of the condition and to clarify misconceptions (Davis, 1993). Explanations have the potential to relieve or inadvertently exacerbate parental self-blame and guilt. One parent may ruminate over how she might have erred during pregnancy, thinking, "The doctors told me I didn't make enough amniotic fluid…. I wonder what I did wrong." Clear statements that acknowledge the lack of causal connection between any prenatal

Fig. 7.4 Professional respect for the whole child. Make time to attend to the child's comfort and to the parent-child interaction.

parental action and the later occurrence of arthrogryposis can provide immeasurable relief.

Faced with the crisis that arthrogryposis presents, parents search for meaning and rational explanations. For some, spirituality provides solace and a way to reduce their sense of vulnerability. Others may seek multiple medical opinions in their search for support and meaning. Clinicians who make the time for respectful and compassionate discussions about the etiology of arthrogryposis can assist parents with these issues.

Infancy: Balancing Needs

Parental functioning and the quality of the parent-child relationship have a profound impact on child functioning (Bowlby, 1982; Jacobson and Wille, 1986; Dawson et al., 1992; Lyons-Ruth and Zeanah, 1993). In this early developmental phase, frequent separation of the infant from parents or inability of a depressed parent to offer consistent and appropriate nurturing can be difficult for both the baby and the parents (Barnard et al., 1993; Sameroff, 1993).

Professionals who take time to play with the baby during evaluation or treatment and who share positive regard for the parent-baby interaction reinforce positive, holistic views of the child and of the parents' growing competence (Fig. 7.4). Professional attention to parents' concerns and problem-solving abilities related to feeding, playing, or caregiving routines promotes the development of parental confidence and healthy parent-child interactions. For example, discussion of how range of motion exercises can be incorporated into daily care or play routines will help minimize the intrusiveness of this activity.

During medical procedures, attention to the comfort and relaxation of the baby and parent will ease the experience. To minimize stress, consider scheduling medical and therapy appointments to accommodate the baby's routine feeding and sleeping schedules as well as the family's schedules. When hospitalization is required, caregivers should be encouraged to be present and participate in care routines, especially since most hospitals today offer more open and flexible caregiving policies (Thompson, 1985; Minde, 1993).

Promoting Optimal Development

All infants, with or without physical disabilities, need to explore the physical world and feel secure in their relationship with mother or father (Bowlby, 1982). Exploration of the physical environment contributes to a baby's overall development and awareness of his or her ability to affect and master the environment (Piaget and Inhelder, 1969). The infant's or toddler's ability to satisfy curiosity and explore the world partly depends on his or her physical abilities and how well parents nurture a sense of love and security. These important developmental experiences contribute to later childhood and adult psychosocial adjustment (Erikson, 1963; Schore, 1994).

In exploring the physical world, children with arthrogryposis may be less able to engage in oral and manual exploration of objects. Difficulties with crawling or walking may impede exploration of their physical world. Adapting toys and the environment permits independent, child-directed exploration and activity. Such independence enables children to grow in self-mastery and social pride and, as important, allows caregivers moments of respite and time for home care activities and for other family members.

As the child matures, further adaptations that permit age-appropriate self-care skills in feeding, toileting, and dressing will help meet the growing need for independence. Understanding normal developmental needs to explore

Fig. 7.5 Meeting every child's developmental needs.
Consultation with families to adapt the home environment, activities, and toys is important.

and attempt independent problem solving will help parents creatively adapt situations for their child's developmental benefit (Fig. 7.5). Guided (structured or supervised) peer or sibling play groups also can help the preschool child experience positive social interactions that enhance self-confidence.

If Problems Arise

Whether a child has a disability or not, parenting young children can be stressful and normally presents certain challenges in family or marital functioning. Whereas many families move through these preschool years with positive coping abilities and adaptation, others experience difficulties. Clinicians and families should regularly monitor family well-being. Families are encouraged to provide balanced attention among the child with the disability and his or her siblings. When conflicts arise concerning the child or a sibling, families may need a referral for professional guidance to address such issues as parent-child communications, marital conflict, and child-management techniques, including setting limits.

Parental overprotection is common among families with children with a disability. If parents seem to seriously limit the child's acquisition of independent daily living skills, a clinician may choose to discuss appropriate developmental expectations in a manner that is sensitive to the parents' concerns. Attention to general child-management techniques also is important (Crary, 1979; Dinkmeyer and McKay, 1982; Ames, 1992). Setting developmentally appropriate behavioral limits for children can be difficult, especially for parents or other caregivers who feel guilty or concerned for their child with a disability. However, inconsistent behavior management can create increasing noncompliance and other child behavior problems. Family counseling that specifically addresses such parental concerns may be warranted. Genuine respect for each family's strengths and choices will help maintain a positive parent-professional partnership that supports family coping.

Fig. 7.6 New world of peers.
As children move into the school setting, new skills in social relationships and cooperative problem solving are necessary.

Preschool and School Years

School entry is a time of preparation and transition for children and their families and a time to further develop competence and social networks. A child's world view enlarges. From a base of parents, family, and home, the child emerges into a world of new peers and new rules (Fig. 7.6). The child must learn to establish social relationships apart from the immediate security and guidance of parents and behave according to teacher-established rules that often are different in emphasis from rules at home. The acquisition and demonstration of academic and social skills is paramount. Development of skills that promote social competence, and positive self-esteem are central themes at these ages (Sullivan, 1953; Erikson, 1963; Cowen et al., 1973). How does the child with arthrogryposis move through these formative years, and what interventions promote psychosocial adaptation in the school-age child?

Fig. 7.7 Appearance and peers.
The social importance of personal appearance as well as function and appearance of adaptive equipment must be considered.

Visibility, Mobility, and Peers

As early as the preschool years, peers notice and react to physical differences in playmates (Harper 1991a; Cohen et al., 1994). The ways in which teachers, parents, friends, and children model comfortable, pleasant social interactions are essential in educating naive observers and encouraging new interactions. Being with friends transmits the unspoken message of acceptance by other children. Teachers who comfortably incorporate all children into classroom activities provide powerful lessons on many levels.

Children with arthrogryposis must develop an age-appropriate understanding of their condition and ways of explaining their physical limitations to others. They need to cope with teasing and their own "Why am I different?" questions. Beginning as early as preschool, open age-appropriate discussion, guided play, modeling behavior, acting or role-playing, and active social practice are all techniques that may be used by teachers, parents, or counselors to address these issues (Pope et al., 1988; LaGreca, 1990). Children may enjoy communicating with others who share the experience of arthrogryposis through the Avenues pen pal program. Structured summer camp experiences and computer networking, telephoning, or letter-writing may allow the child with arthrogryposis a more relaxed opportunity to build friendships and gain social support without the same concerns for physical status and acceptance children sometimes experience at school.

Attention to aesthetics in physical rehabilitation efforts can facilitate a child's social development as well as functional independence. The color, size, and obtrusiveness of adaptive appliances, such as braces, walkers, and wheelchairs, may draw attention to the visible intervention instead of the child or may elicit unwelcome, negative attention to the child. To minimize the impact of splints on the child's social relations, part-time splinting schedules that allow for nighttime placement, as long as sleep can be maintained, may be preferable to daytime splinting. Adaptive devices or orthotics that are used during the school day can be the least restrictive or cumbersome in design and colored in neutral or less visible tones or, conversely, adorned with fashionable colors or patterns of the child's choice. Promoting unaffected aspects of appearance via attention to current preferences in hairstyles, clothing, and accessories benefits the child's self-confidence and ability to fit in with peers (Fig. 7.7). The child's own personality, sparkle, and interests thus can be permitted to be prominent features in social interactions.

Beyond appearance and activities, access and exposure to common cultural experiences, such as current music, toys, games, sports events, and awareness of popular entertainers, clothing fashions, and food fads, are important in building and maintaining peer relations. After-school peer interactions, such as playing at a friend's house or hanging out with friends at the corner store or mall, also are basic to developing friendships in the school years. Resourceful parents will find ways for their mobility-impaired child to have these normal experiences.

Fig. 7.8 Recreational pursuits.
A variety of games and leisure activities can be adapted to allow important social time with friends.

Children's Friendships

Both at home and at school, social development proceeds at an intense pace. Feeling part of a peer group is important, as are individual friendships (Furman and Gavin, 1989; Parker and Asher, 1993). Through friends, children gain social support and many opportunities for socialization experiences.

During the school-age years, peer groups, such as Boy Scouts or Girl Scouts, provide structured and supervised social activities. By belonging to a club or group, particularly one organized around a common interest or goal, a child can earn acceptance and build friendships while contributing to group efforts. Scouting projects that earn individual and group badges and science or environmental groups that promote group learning and cooperative projects are examples of activities in which an interested child might build a social support network. A variety of sports, recreation, and leisure activities may be adapted to suit children with arthrogryposis (Sawatsky, undated) (Fig. 7.8).

Any individual activity or interest can be an area of strength and self-satisfaction and can provide important ways to connect with peers. One child may become an expert at remembering sports statistics or playing the latest video game. Another may develop a native sense of humor, cartooning skill, musical talent, dramatic flair, a passion for computers – the possibilities are endless. Developing individual interests yields satisfaction and increases social competence and avenues to friendship. By expressing their interest in a child's pursuits, parents, teachers, and clinicians all have the opportunity to foster the further development of special areas of competence.

Improving Social Skills

An important factor in gaining peer support is the child's ability to display age-appropriate social interactional skills. Some children may have limited positive social experience and may genuinely lack social skills. The ability to initiate and maintain friendships through openness and interest in others, conversation and listening skills, and assertiveness are important for any child. A number of cognitive-behavioral techniques can be used to help the child improve cooperative problem solving and other social skills, as well as self-esteem. Training in social competence may be incorporated into an individualized educational program or be part of the curriculum for an entire classroom or grade (Gresham, 1986; Pope et al., 1988; Walco and Varni, 1991).

Improving Coping Ability

Coping skills are important not only for social interactions, but also in the child's ability to manage everyday stress. The daily realities of living with physical impairment can be more or less difficult for each child at different times. How the child perceives this level of difficulty strongly influences adaptation. A child's sense of his or her level of "daily hassles," family conflict or support, and peer support is associated with the child's self-esteem and vulnerability for depressive symptoms (Varni et al., 1989a,b). These daily "microstressors" may be minimized through the collaborative efforts of parents, teachers, and clinicians. For example, the child who is frustrated by the need to carry books between classrooms may be relieved when a parent or teacher arranges for him to transport the books in a backpack whose design he can select. Coping strategies, such as reframing or altering beliefs that cause distress, can also be helpful. The child who believes that she must perform superiorly in all subjects to compensate for her disability causes herself distress in this rigid self-expectation. She can learn strategies that allow her more flexible thoughts and self-expectations, improving her self-esteem and overall adjustment.

The child's caregivers influence the child's knowledge of coping strategies and problem-solving skills (Halberstadt, 1986; Ladd and Price, 1986; Pettit et al., 1988; Quamma and Greenberg, 1994). Professional efforts to assist parents in these areas can have a meaningful impact. Parents who cope and adapt in their daily lives model and teach their children these valuable abilities and attitudes. Conversely, family conflict and dysfunction are major stresses for children. Family therapy or marital therapy can improve family functioning, reduce family conflict, and promote a family's ability to support its children.

Parental Overprotection

Parents commonly struggle with urges to overprotect. For optimal development, children must be permitted to engage in activities with the least restrictions needed for their safety and developmental abilities. This undoubtedly poses an adaptive challenge in parenting for which clinicians can offer ongoing consultation and respectful discussion.

When a family becomes overprotective or the child is overanxious, the child may refuse to go to school or may have frequent school absences due to unsubstantiated somatic complaints. (The latter behaviors also may be seen in a child who is having peer problems or finds the school day difficult.) In these situations, a family should seek consultation with a child mental health professional, such as a pediatric psychologist, so that difficulties may be addressed before much school is missed. A school-based counselor may also be helpful.

The Importance of Siblings

Siblings are key players in each other's social and emotional development as well as family functioning. Children who have a sibling with a disability frequently experience stresses as well as positive experiences with each other and outside of the family constellation. For example, siblings may be the target of teasing on behalf of the child with arthrogryposis, or they may be asked to alter their own schedule to assist this child or the family with care routines. Siblings may perceive the child with arthrogryposis as receiving extra attention or other resources that cause the sibling to feel resentment, guilt, or jealousy. On the positive side, many siblings and families report a special closeness and greater affection, which they attribute to their experience of growing with a special child. Many parents report that all their children seem to be more compassionate and tolerant than children who do not have siblings with a disability (Simons, 1987; Ambler, 1988).

Parent support groups may provide access to sibling networks or an opportunity for parents to receive support for parenting dilemmas and compare notes on how best to parent all of their children. It can be helpful for children (and adults) to have an accepting opportunity for feelings to be acknowledged and normalized, not only loving feelings but the full range of normal emotions, including guilt, anger, and hurt or sad feelings. Sibling rivalry for parental attention exists to some extent in all families. Parents can dedicate regularly scheduled individual quality time (as little as 15 minutes, or more, most evenings) in which each child has an opportunity to choose or direct his or her special time with a parent, such as reading a story, going for a walk, or talking about the school day over dessert. This can be quite beneficial if siblings feel a lack of parental attention. A number of excellent resources are available, including national workshops that specifically promote sibling adaptation. These resources can be offered along with general consultation on parenting and sibling issues (Meyer et al., 1985; Ambler, 1988; Meyer, 1993).

Fig. 7.9 The teenage years.
Adolescence is a time of increased freedom as well as challenge.

Adolescence and Beyond: The Transition to Adulthood

As children enter high school, a new level of independence and self-sufficiency is demanded by the school day structure, and the academic workload. The adolescent must plan for an adult life that is more separate from family. Peer relations become increasingly important as the focus shifts from the child's family to the larger adult world. Additionally, as teenagers strive for greater autonomy, they must learn to assert or advocate for their own rights, needs, and desires. Mobility impediments may seem more distressing as peers prepare for a broad range of vocational possibilities and readily obtain driver's licenses, which enhance social and vocational access and independence. Dating and relationship issues become prominent, and in those realms where physical prowess can provide greater access to social opportunities, youths with physical disabilities may feel increasingly disadvantaged.

The family constellation and the child's successes and vulnerabilities from preceding years provide a base from which an adolescent will negotiate this period. With social and community support, children with arthrogryposis and their families can adapt to these transitional challenges. Professionals may assist by raising relevant issues for the teen and family, normalizing the challenges of this developmental phase, and providing confidence that an adult future with clear goals and rewards indeed lies ahead. During this time, it is especially important to respect, empower, and involve the adolescent in decision making and in his or her own medical care.

Social Concerns

During high school, appearance plays a prominent role as teens become more involved with peers and dating (Fig. 7.9). At any age, part of a child's self-concept emerges from the quality of social interactions he or she experiences. As the child matures, social experiences outside the immediate family have greater influence on his or her self-concept: first within peer groups, and as the adolescent years pass, more so in individual friendships or relationships in the educational, personal, and work worlds. Group social activities, particularly in a teen's areas of interest, provide solid opportunities to relate with other boys and girls and to build friendships that form the basis for developing intimacy. Contact with other teens and young adults with disabilities provides another forum for social experience, as well as the opportunity to learn from others' experiences. Outside of academic efforts, a paid or volunteer job at school, in a library, office, or other work setting, offers valuable social and workplace experience.

From an early age, social learning involves the risk of experiencing a social rejection or lost friendship. Caregivers may wish to shield their children from emotional discomfort but need to balance protective concerns by supporting their child in his or her involvement in socially challenging growth opportunities. Excellent written resources are available for parents and teenagers that address relationships, sexuality, and safety issues for teens with physical disabilities, as dating and other social concerns emerge (Hopper and Allen, 1980; Shaman, 1985; Kroll and Klein, 1992; Kupper, 1992).

Fig. 7.10 Planning for the future.
Early attention to postsecondary educational and vocational goals is essential.

Planning for Adulthood

Adolescents with arthrogryposis and their families will benefit from knowing the range of educational, vocational, and financial assistance options available after high school. Information can enable adolescents to explore personal interests, develop goals, maximize focused educational efforts, and make better use of available resources. Contact with other young adults with disabilities who are engaged in innovative employment and independent lifestyles gives the adolescent a more vivid and realistic picture of opportunities he or she may be excited to work toward and first-hand knowledge of special programs, such as those that enhance college accessibility for students with disabilities.

Vocational goals provide hope for a more independent adult lifestyle and add immeasurably to a person's sense of life satisfaction, competence, and ability to contribute as a productive citizen. In addition to consideration of academic pursuits, such as postsecondary or graduate programs, educational attention in the high school years can be directed at prevocational and vocational skills (Fig. 7.10). For adolescents who do not choose to go to college, but have finished high school in the United States, federal programs may provide job access. The federal Department of Vocational Rehabilitation, which has a central office in each state, can assist youth with disabilities in defining suitable employment goals and coordinating the necessary training and assistance.

When children reach the age of 18, determination of eligibility for U.S. Social Security Administration benefits, such as SSI (Supplemental Security Insurance) and SSDI (Social Security Disability Insurance), is no longer based on parental resources. Families and youths are encouraged to verify their eligibility for basic assistance from these federal sources at this time and to clarify the effect of potential employment earnings on benefits.

Advocacy Issues

Postsecondary goals to attend college or enter the work force raise questions about accessibility. Excellent publications that discuss post-high school educational, vocational, and financial assistance and other resource issues are available from NICHCY [The National Information Center for Children and Youth with Disabilities, 1(800) 999-5999] and from the HEATH Resource Center [The National Clearinghouse on Postsecondary Education for Individuals with Disabilities, 1(800) 544-3284]. Many of these materials are available at no cost and can be distributed to families.

Young adults with arthrogryposis also benefit from contact with a broader community of persons with physical disability, not only for social support but also as a way to stay abreast of civil rights issues and changing laws affecting citizens with disabilities. The Americans with Disabilities Act (ADA), which became law in 1990, is a comprehensive ban on discrimination against persons with disabilities in housing, education, employment, and other major areas. Young adults and their families are encouraged to learn how relevant laws affect them. Community-based individual or group advocacy may be necessary to create local compliance with the ADA. As an added benefit, participants meet other young adults and families through advocacy efforts, making new friends while increasing their knowledge of important civil rights (Fig. 7.11).

Adaptational Challenges

Some children may take more or less time to make the transition from adolescence to young adulthood. All young adults – whether they have a disability or not – face challenges in any kind of living situation. Common challenges include managing independently, and securing sufficient emotional separation to allow further development while living near their parents. Parents, too, may face some adaptational challenges, as they must either allow their child to experience normal risks of living independently or struggle with having a young adult child remain at home.

Self-sufficiency must be defined individually for each adolescent and young adult, and the goals must be adjusted accordingly. One teenager may look forward to living in an apartment or college dormitory room and arranging for necessary assistance on her own. Another teen who requires significant assistance with daily living skills may not find such goals appropriate, and a group living situation or extended period of residence with family may be the setting of choice.

Families and adolescents can be reassured that goals they set now may evolve with experience. For example, a youth who begins his young adult years living at home may find he is increasingly able to arrange for assistance as needed and that communication with a network of disabled peers enables necessary learning about independent living and advocacy issues unique to physically disabled persons.

Fig. 7.11 Learning about rights.
Young adults working together on advocacy issues can meet new people and share common problems and solutions.

Fig. 7.12 Support of the whole family.
A child's ability to adapt depends greatly on family well-being.

If Difficulties Arise

Adolescence is a time of major transition. Parental overprotection may leave a teenager few positive avenues for independent development. Some adolescents may be unable to engage in independence-seeking behaviors out of significant fears. Social difficulties or family imbalances increase a teen's vulnerability for acting-out and exhibiting risky behaviors, as well as internalizing sadness and worries. The close, sustained interest of adults in an arranged Big Brother or mentoring relationship with a teen offers additional support and guidance outside the immediate family. A member of the child's extended family, such as an uncle or older cousin, sometimes can be engaged in this kind of mentoring. A troubled youth – one in the midst of a conflictual family environment or one whose school performance or psychosocial functioning is declining – can be referred for formal intervention to improve adaptation.

If the usual timeline for an adolescent leaving home to live independently is delayed, parents' normal reactions may include frustration or a recurrence of grief. Parents may direct subtle or open anger at the adolescent despite their love and pride in their child and his or her development to date. Families may benefit from family therapy that addresses coping, problem solving, communication skills, and management of anger and other emotions during this transition.

Conclusion

Promoting well-being in children with arthrogryposis requires collaboration with, and support of, the whole family (Fig. 7.12). Family connections to larger networks of social support, such as extended family, other families parenting similar children, special school services and staff, and economic resources, are important in family adaptation and can be facilitated through comprehensive medical care. Interventions for arthrogryposis may require long-term involvement of the child and family. Clinicians can accord a child patient an age-appropriate level of involvement in treatment decisions and actions. Intervention can be designed in ways that address a child's developmental needs for independence, competence, making friends, and finding adult vocations. A respectful partnership among patients, their families, and professionals permits the most effective interventions (Turnbull and Turnbull, 1990).

Professionals' attitudes and knowledge will have a profound bearing on how a child or family perceives their evolving experience with a disability. The ability to cope or adapt is strongly influenced by the child or family's understanding and by the meaning they impart to events and circumstances. Indeed, families may ask questions for which answers are not readily available. However, the professional can offer support and confidence at critical moments by willingness to listen, to acknowledge the limits of current information, to make appropriate referrals, to collaborate with families to generate solutions, or simply to acknowledge that a problem exists and that the family is managing as best it can. Discussing information and support services transmits resource facts, as well as an attitude of normalcy, acceptance of community support, and readiness to discuss family needs and welfare. With experience, professionals can interact with a calmness born of first-hand knowledge of the many ways in which children and families can achieve developmental milestones, pursue education and other services that enhance a child's independence, and enjoy creative lives – challenged by physical disability, but not limited in options for happiness.

B. Ross, M.Ed.

Introduction

Since the 1950s, noticeable changes have occurred within economically developed countries in both the concept and provision of educational services to children with special needs. Initially, these children, if they received any education at all, were identified on the basis of a medical treatment model that emphasized their functional impairment(s) and need for treatment in special schools isolated from nondisabled peers. This continues to be a predominant service model in many parts of the world (UNESCO, 1988). The isolation of children with disabilities in special schools is increasingly being viewed as problematic, however, since these schools are limited in size and leave many children with disabilities without services (Jan Pijl and Meijer, 1994.)

It is now generally accepted in economically developed countries that children with impairments occupy one end of a continuous distribution of abilities, and these children's educational outcome is the result of a complex interaction among individual, home, and community variables. Their special education needs can best be met through a continuum of services that are integrated into one educational system that serves students with a wide range of abilities (Fig. 8.1). This approach acknowledges that learning difficulties

Fig. 8.1 Children with disabilities in the mainstream of education.

that these children experience in school can be traced, in many instances, to the ways in which schools are organized, the curriculum available, teacher experience and training, and the school's respect for and nurturance of individual differences (Ainscow et al., 1995). Although countries vary considerably in how they identify and provide educational services, many have policies that support increased integration of students with disabilities within regular education programs (Evans et al., 1995a).

This focus on integrating students with disabilities into mainstream practice (Fig. 8.1) was stimulated in part by the concept of "least restrictive environment," or LRE, which is mandated in the United States by the Individuals with Disabilities Education Act (IDEA) and supported by other U.S. civil rights legislation. Through this act and acts that amended the statute, the U.S. federal government provides financial assistance to states that provide early intervention services to children ages birth to 3 years and mandates that all children ages 3 to 21 years must be provided a free, appropriate, public education no matter what their disability. In the 1992-1993 school year, over 5 million children with disabilities from birth through age 21, including many of those with arthrogryposis, were served under this federal law (U.S. Department of Education, Office of Special Education, 1994). Section 504 of the Vocational Rehabilitation Act of 1973 ensures that children with disabilities receive appropriate modification within their classroom program to accommodate their special needs, regardless of whether or not their placement is in special or general education classes (American Academy of Pediatrics: Committee on Children with Disabilities, 1993). Most recently, the Americans with Disabilities Act (ADA), signed into law in July 1990, assures children and youths with disabilities that their efforts during the school years will be productive. They can strive for any professional career, knowing that employers must make reasonable accommodations within the workplace (Chaikind, 1992).

The United States has traditionally been viewed as a country that has made significant progress in moving toward an integrated model for delivering education and related services across a wide age range of children with disabilities. This chapter reviews the status of special education practice in the United States to illustrate some of the central issues that many countries, in one form or another, are addressing within their respective educational systems. It supports the view that providing an appropriate education for a child or youth with arthrogryposis may be seen within the broader context of how best to achieve quality educational outcomes for students with disabilities in general. Recent developments in the field of education in the United States, specifically the philosophy of inclusion and the educational reform movement, are discussed as they relate to educational outcomes for children with disabilities.

There are two underlying themes throughout this discussion. First, children with arthrogryposis are likely to need continuing therapies and medical follow-up throughout the school years and thus require a well-coordinated, multidisciplinary, and long-term approach to educational planning. This must be family focused and include mechanisms for communication and collaboration across the disciplines of education, rehabilitation, and orthopedics, as well as other community services, such as vocational counseling. Second, the importance of parents' and caregivers' involvement in the educational process discussion must be respected and acknowledged, and professionals, particularly those from the health care profession, have many opportunities to support them in this endeavor.

Fig. 8.2 Early intervention starts in infancy.

Early Intervention

Children with arthrogryposis are generally identified in infancy by health care professionals, with treatment focusing on increasing functional gait and independence with activities of daily living in the home (Fig. 8.2). In the United States, federal support for states to provide early intervention services to very young children with disabilities is available under the IDEA, signed into law in October 1990. This act mandates a free, appropriate public education, or FAPE, for children and youths with disabilities between the ages of 3 and 21 years. Part H of the IDEA, also referred to as the Program for Infants and Toddlers with Disabilities, offers financial assistance to states (or territories) to help them design and implement systems of statewide, comprehensive, multidisciplinary, and interagency programs that provide early intervention services to eligible infants and toddlers from birth through 2 years. In the 1992-1993 school year, all states were participating and serving roughly 140,000 infants and toddlers, or 1.2% of the resident population in this age range (U.S. Department of Education, Office of Special Education, 1994). The distinguishing feature of early intervention services is its emphasis on providing care to infants and toddlers with disabilities within the context of the needs, concerns, and priorities of the family.

Prior to this legislation, few states were providing educational and ancillary services to infants with disabilities. The IDEA offered financial assistance to states and greatly expanded the range of services available to infants and toddlers with disabilities and their families. Federal support for early childhood education programs is based on the premises that early intervention can enhance the acquisition of more complex skills useful for later functioning, maximize the potential for independent functioning and thereby produce long-term economic and social benefits to the individual and society, reduce the intensity or need for special education and related services on reaching school age, and enhance family functioning by enabling families to meet the special needs of a child with disabilities (Umbreit, 1983).

Under the law, each state is required to determine to what extent a physical or health impairment is negatively affecting the normal development of an infant or toddler based on the concept of developmental delay. States must identify the criteria used to document the existence of a delay in each of the following areas: cognitive development, physical development (including a statement on vision, hearing, and health status), language and speech development, psychosocial development, and self-help skills. Children who have a diagnosed physical or mental condition that has a high probability of causing developmental delay (e.g., Down syndrome, sensory impairments, and other chromosomal abnormalities that are likely to result in mental retardation) are also eligible for services. In some states, children at risk for developmental delay based on biologic or environmental risk factors (e.g., poverty, intrauterine drug exposure) are also eligible for services.

Children identified as at risk receive a multidisciplinary assessment that includes a description of the child's performance in each of the areas noted in the preceding paragraph and also a review of pertinent records describing the child's medical history and current health status. The assessment must also include a statement of the family's strengths and needs that relate to enhancing the child's development. The law offers flexibility to states by allowing assessments to occur with the child and family in mind and where results are most likely to produce valid and reliable information useful for program planning. Thus, a child may be assessed at home or within a hospi

Components of the Individual Family Service Plan (IFSP)

- Child's present levels of development based on professionally accepted, objective criteria.
- Family's strengths and needs relating to enhancing the development of their child with disabilities.
- Targeted outcomes with criteria, procedures, and timelines for review of progress.
- Specific intervention services required to meet the unique needs of the child and family.
- Projected dates for initiation and duration of services.
- Identification of a services coordinator who will implement and coordinate the plan with other service agencies.
- Procedures for transition of the child to a preschool program.

Fig. 8.3. The Individual Family Service Plan

tal, day care setting, or therapy center (Fewell, 1991). No one test should be the single source of information for the multidisciplinary assessment, and in general, selection of a particular test or battery should be guided by which skills should be the primary focus of intervention. Assessment procedures are generally based on surveys of normal development, with the most common practice being to use one-test, multidomain instruments, such as the Batelle Developmental Inventory, Early Learning Accomplishment Profile, and Early Intervention Development Profile, among others (Fewell, 1991; Haring and McCormick, 1990).

Following assessment, an Individual Family Service Plan, or IFSP (Fig. 8.3), is developed for eligible infants and their families. This is a written document that must be evaluated annually and reviewed at 6-month intervals. The IFSP provides opportunities for physicians who manage the care of a newly diagnosed infant with arthrogryposis to become directly involved with several aspects of the early intervention process.

Identification and Referral

Although initial care of the newborn with disabilities occurs within the hospital or in other health care institutions, the majority of ongoing treatment and therapies will take place within the home and community settings. Under Part H of the IDEA, each state must have in place a central directory of services, and many states use regional service coordinators who can provide a single point of entry into a state's early intervention system. Since physicians are the first individuals involved in making a diagnosis of arthrogryposis, they, as well as nurse practitioners or other primary health workers, play a key role in identifying resources within the community and making a referral so that an infant with arthrogryposis can benefit from early intervention services within the community (Nader, 1993).

Assessment

Physicians are also an important part of the assessment process in determining a child's eligibility for services, as well as in developing guidelines for program intervention (Purvis, 1991). For the child, pertinent assessment information includes specific health care issues, such as information about the condition and its impact on learning and development, the probable medical course, how to coordinate services with anticipated hospitalizations and surgical interventions, management of emergencies, and any specific health care needs within the early intervention setting (Nader, 1993). Physicians are often in a good position, due to the longitudinal nature of their involvement, to offer information useful for completing an evaluation of the parents or guardians. Areas to consider include how the child's specific needs might affect family functioning, parent-child interactions, family needs, critical events, and family strengths (Haring and McCormick, 1990). Throughout the assessment process, parents and caregivers will benefit from a knowledgeable source of medical advice and counsel.

Resource Support to Families

Physicians can be an initial source of information about relevant laws governing the provision of services, availability and location of services within the community, financial assistance available to parents or caregivers, and advocacy strategies (Ziegler, 1989; Summers et al., 1990; American Academy of Pediatrics, 1992; Decker, 1992). To meet this need, physicians must know how their state manages early intervention services and also become acquainted with local early intervention staff and programs. Unlike services for school-aged children, the responsibility for managing early intervention services may reside outside the state education system. In the 1992-1993 school year, for instance, 19 states specified Education as the lead agency, 22 specified the Department of Health, and the rest had other agencies, such as social or rehabilitative agencies (U.S. Department of Education, Office of Special Education, 1994). Even if the physician is not a member of the multidisciplinary team, he or she can act as a consultant to families by reviewing the appropriateness of the IFSP, particularly the goals and objectives, and if the health-related services proposed are sufficiently comprehensive (American Academy of Pediatrics, 1992).

Service Coordination

Finally, the IFSP must include the name of a service coordinator from the profession most relevant to the child's or family's needs. This individual will be responsible for implementation of the IFSP and for coordination with other agencies and personnel. Service coordination is seen as the key to successful entry of families and children into multidisciplinary services. Studies suggest three levels of physician involvement in service coordination. These range from direct treatment and care of the child with complex medical needs to acting as a consultant to a multidisciplinary team serving a child with less medical involvement. For children in between these extremes, the physician is seen as serving as a medical manager, coordinating the work of various other physicians providing subspeciality expertise, while another professional, for instance, a nurse practitioner, social worker, or educator, coordinates services across agencies, offers resources to the family, and completes paperwork (Fullagher et al., 1992).

After the IFSP has been developed, the multidisciplinary team, together with the parent or caregiver, determines which specific program and services will meet the unique needs of the infant and family and achieve the specified outcomes. They must state the frequency, intensity, duration, method of service provision, and location of services. Unlike an Individual Education Program (IEP) for school-age children with disabilities, the IFSP can include a wide range of specific primary services other than specialized instruction. These include family training, counseling, and home visits, speech pathology and audiology, occupational therapy, physical therapy, psychologic services, medical services for diagnostic or evaluation purposes, and health services necessary to enable the infant or toddler to benefit from the other early intervention services (excluding, for instance, surgical interventions). The initial evaluation and assessment to determine eligibility is free, as are, in most instances, the direct services specified in the IFSP. Most states use a variety of different state and federal funding sources to support Part H services, including Medicaid, maternal and child block grant programs, and special supplemental programs for Women, Infants and Children (WIC), among others (U.S. Department of Education, Office of Special Education 1994). Some states charge families a sliding fee based on their yearly income (Brown et al., 1993).

The majority of intervention services for infants or toddlers with disabilities are home based (U.S. Department of Education, Office of Special Education, 1994). Teachers, therapists, or other interventionists schedule weekly or biweekly visits at the child's home, providing special toys, materials, and instruction. The home is the natural environment of the child and family, and services delivered in this setting are most likely to enhance learning and adaptation of skills and promote active parent involvement. Other frequently reported sites for early intervention services include a center-based or classroom program and an outpatient service facility. Center-based programs provide direct instruction or therapy or both to the child. Children may be served individually or in groups, from 3 to 5 days per week. These groups are usually facilitated by teachers or therapists and can provide opportunities for social interaction with nondisabled young children through integrated settings. An outpatient center is a clinic or hospital where the child and family come for short periods of time (e.g., 45 minutes) to receive therapy. Infants and toddlers with significant medical needs or who are chronically ill may be served in hospital-based programs (Meyen, 1990).

Transitions are an important aspect of family life, particularly for families that include a child with disabilities. Fowler et al. (1991) note that for these families, the birthday of a child or achievement of an important developmental milestone may also mark a transition between service options. Birthdays "may serve as prompts that it is time for another professional evaluation of their child's developmental progress, as dates for determining eligibility for special education services, as deadlines for choosing new service programs or providers, and as reminders that their child is developing differently from other children in their family or neighborhood" (p. 136). An important component of the IFSP is a description of what steps will be taken to support the transition of the infant who requires continuing special education services to preschool services. These steps include discussions with and training of parents or caregivers regarding future school placements, procedures to prepare the child for changes in placement and services, and with parental consent, the transfer of information about the child to ensure continuity of services, including evaluation and assessment information and copies of the IFSP that have been developed (Education of the Handicapped Act Amendments of 1986, §303.344h). The intent of transition planning is to ensure that there are no gaps in service as the child moves from early intervention services to preschool programs, and financial responsibilities for evaluations and transfer of information are clarified (Fowler et al., 1991).

Preschool Services

Many countries have developed preschool programs for children with special education needs. In the United States, children ages 3 to 5 years who continue to demonstrate developmental delays as defined by individual states' criteria are entitled to special education services through state education agencies. In the 1992-1993 school year, all states provided services under the Preschool Grants program of the IDEA to a total of almost 450,000 children, at no cost to families (U.S. Department of Education, Office of Special Education, 1994). Many states are still in the process of developing specific policies regarding the transition of children from early intervention to special preschool services. The prevailing belief guiding public policy is to develop seamless systems directed toward the needs of children in the birth through 5 year age range rather than more narrowly in either the birth through age 2 or the 3 to 5 year age range. Consequently, states have the option of using the IFSP to guide services until the child's sixth birthday.

Fig. 8.4 Peer interactions.
Opportunities for cooperative peer interactions are an important factor in selecting a preschool program.

The concept of LRE, as applied to the school-aged child, refers to removal from the regular classroom in order to receive special education services. As applied to the preschool population, this concept is less clear because many schools do not operate programs for preschool children without disabilities. Preschool children with disabilities have a number of program options. These include placement in school-sponsored preschools and kindergartens, reverse mainstream options where non-disabled children are enrolled in specialized programs to act as peer models for social interaction, enrollment in programs for children without disabilities where specialized services such as occupational, physical, or speech therapy are available to implement the IFSP (e.g., Head Start programs for children from disadvantaged or low-income families), or participation in a family-based or center-based setting (Meyen, 1990).

Many parents of preschool-aged children with arthrogryposis want their child to receive appropriate special education, but have difficulty finding a regular child care setting that could accommodate the physical needs of their child. The ADA, which became effective in 1992, specifies that child care settings are public facilities and consequently must make reasonable accommodations to the needs of a preschooler with disabilities. These include increasing access through removal of physical barriers, additional staff training or adjustment of staff ratios, and the availability of certain types of equipment.

In supporting parents in selecting a preschool, Winton and Turnbull (1981) found that factors of greatest importance to parents included location and ease of transport, respite care, parent-professional relationships, parent involvement activities, and the availability of a peer group for discussion and support. Parents are likely to need at least 6 months to 1 year to decide on a placement for preschool services, obtain information, and tour the potential program facility. They may also desire staff follow-up from the originating program to the new program (McDonald et al., 1986). Additional information that may be useful for deciding on a particular preschool or early childhood setting is available from the National Association for the Education of Young Children (NAEYC). This organization specifies recommendations for appropriate group sizes, child-to-adult ratios, developmental activities, facility design, and qualification of personnel.

In general, interventions for the preschool child with disabilities are likely to focus on improving functioning both in the present setting, such as the home and community environment, and in the regular kindergarten or first grade classroom. Important readiness skills include independent work skills, participating in groups, following class schedules and routines, following directions, functional communication, and social/play skills (Haring and McCormick, 1990) (Fig. 8.4).

Fig. 8.5 Public school education.
Approximately 20,000 children with arthrogryposis are currently served in school settings, primarily in regular classrooms.

The School Years

At age 6 (or as early as age 3, at states' discretion), children who have been participating in early childhood education programs are evaluated to determine their eligibility for special education services. These are defined in U.S. federal statutes as "specially designed instruction, at no cost to the parent, to meet the unique needs of a child with disabilities, including classroom instruction, instruction in physical education, home instruction and instruction in hospitals and institutions" [The Individuals with Disabilities Education Act, 1990 §1401(a)(16)]. There is a two-pronged criterion that determines a child's eligibility for special education. First, the child must have one or more impairments in intellectual, physical, socioemotional, or sensory abilities. Second, the child's disability must significantly interfere with his or her ability to learn in a regular classroom environment, thus creating the need for specially designed instruction. The application of specific disability categories (e.g., orthopedic impairment) to children who require specialized instruction is currently under Congressional review in the United States. Many countries in the world have abandoned the use of specific categories to describe a child's unique learning characteristics, favoring the term "special education needs" or SEN (Evans et al., 1995b). This latter approach reduces the negative impact of labeling children and the likelihood of their subsequent placement into substandard educational programs separate from their nondisabled peers.

Estimates of the prevalence of students with arthrogryposis within the U.S. general school-age population are difficult to make, but based on a reported incidence of 1 per 3000 births applied to the total number of children ages 3 to 21 years, one might project that over 20,000 children with arthrogryposis are served in public school settings (Fig. 8.5). It is unclear how many of these children are currently included in federal counts of children receiving special education services, since the federal government does not require states to keep disability counts of children served based on medical diagnoses. However, collectively, students receiving special education services within current IDEA disability categories that would typically be applied to students with arthrogryposis (e.g., multiply disabled, orthopedically impaired, and other health impaired) account for just over 4.5% of the total population of students with disabilities (U.S. Department of Education, Office of Special Education, 1994). These figures suggest that most schools, and consequently many teachers, will have had little direct experience or training on the educational implications of arthrogryposis.

Individual Education Plan

The cornerstone for delivering specialized services to school-age children with disabilities (and at individual states' discretion, children at age 3) is the Individual Education Program (IEP) (Fig. 8. 6). Like an IFSP, the IEP provisions for eligible children consist of two parts: meetings where parents, school personnel, and other professionals can jointly make decisions about the program for a child with disabilities, and a written plan that requires multidisciplinary involvement in assessment and implementation that specifically addresses the unique needs of the child (Decker, 1992).

Components of the Individualized Education Program (IEP)

1. Statement of the child's present level of educational performance.

2. Statement of annual goals, including short-term educational objectives stated in terms that can be measured, expected levels of performance, and schedules for their accomplishment.

3. Statement of specific education and related services, and the extent to which the child will be able to participate in the regular education programs.

4. Statement of needed transition services based on a functional vocational evaluation and anticipated postschool outcome, beginning no later than age 16 or sooner if determined appropriate.

5. Projected dates for initiation and duration of all special education and related services.

6. Appropriate objective criteria and evaluation procedures and schedules for determining, at least on an annual basis, whether the short-term objectives are being achieved.

Fig. 8.6 The Individualized Education Program

There are no federal standards for the length or amount of detail that may appear in the IEP, and consequently, the appearance of the document varies from state to state. However, an IEP can serve many purposes or functions, including (1) a vehicle for communication between schools and parents or caregivers, (2) a focal point for reviewing any differences between the parents and the schools, (3) a stated commitment of resources to enable the child with disabilities to obtain an appropriate education, (4) a management tool to ensure that individualized services are being provided, (5) a compliance or monitoring document to ensure that school systems are following state and federal guidelines under the IDEA, and (6) an evaluation tool to measure a child's progress toward projected outcomes (Individualized Education Programs, 1980). The IEP is not, however, a legally binding contract between school districts and families, in that it does not require that teachers or other school personnel be held accountable if a child with disabilities does not achieve the goals and objectives specified in his or her IEP.

Although education for a very young child with arthrogryposis is likely to address developmental functioning and family support, the primary focus during the school years is on the student's academic achievement, peer interactions, and preparation for transition to adulthood. Discussions with parents of children and youths with arthrogryposis suggest the following areas of program concern.

Specialized Services

Parents need to know what types of specialized services the school may provide to enable their child to participate in the general education class. Part B of the IDEA specifies that related services are not designed to supplant therapies required for the medical or health management of the child with disabilities. Rather, they are developmental, corrective, and other support services that may be needed to enable a child to benefit from education [The Individuals with Disabilities Education Act, Public Law 101-476 1990 § 1401 (17)]. These include speech pathology and audiology, occupational and physical therapies, counseling, school health services, social work and psychologic services, rehabilitation counseling, and recreation.

Medical Services

As defined by the IDEA, these are services provided by a licensed physician. They are considered a related service but are provided solely for diagnostic or evaluative purposes that contribute to determining a child's eligibility for special education based on a medically related disability. In practice, few schools actually employ full-time or part-time medical consultants, preferring to rely on informal relationships with physicians within the community. Some children with disabilities may need a particular service that is not specifically stated in the IDEA. However, if the service is "developmental, corrective or supportive," it is considered a related service and must be stated in the student's IEP. Examples include the requirement of an instructional aide, certain equipment, and assistive technology.

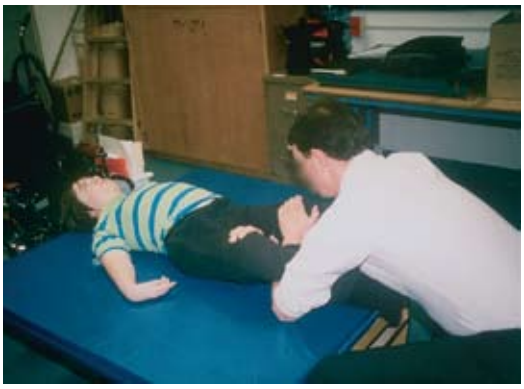


Fig. 8.7 School therapy.
In the school setting, therapy is often provided by an aide under the supervision of the physical therapist.

School Health Services

This is a another service that can support participation in the regular class room of a student with arthrogryposis, particularly those with more severe medical conditions. In the United States, schools employ an estimated 30,0000 nurses who are the major provider of school health services in many schools in this country (Cluff, 1985). School nurses act as an important link between the school and the student's primary or specialist physician. They coordinate student health education programs, act as the primary teacher for children who need to learn how to care for special bodily needs (e.g., catheterization), act as a medical resource for teachers who request information about a child's particular medical condition, and assist families in identifying important community health resources. School nurses working full time are most frequently found at the secondary level. However, many elementary schools have a school nurse available no more than one half-day per week (Meyen, 1990). Thus, schools frequently rely on health aides to carry out screening procedures (where permitted by law), recording health information and general record keeping about a student's daily participation in school (Nader, 1993).

Therapy

In the school setting, children with arthrogryposis most commonly use school-based occupational therapists (OT) and physical therapists (PT) as related services. These professionals provide treatment through a prescription from a physician that will enhance the ability of the student with disabilities to participate in educational activities.

School-based OT and PT function as members of a multidisciplinary service team that includes as regular members a special education teacher, school counselor (at secondary levels), the building principal, and a school psychologist. They have at least four distinct roles or responsibilities in serving students with disabilities, such as arthrogryposis: (1) providing direct, but often limited, therapy activities consistent with the child's overall medical treatment plan, (2) supervising the activities of trained paraprofessionals who may implement a therapy program on a daily basis (Fig. 8.7), (3) serving as a link between the student's health care providers and school personnel, and (4) acting as a consultant to classroom teachers by offering specific recommendations on how to incorporate therapy goals into the academic environment (and vice versa), as well as increase physical accommodation and accessibility to classroom instruction and materials. Together with medical rehabilitation specialists, school-based OT and PT are responsible for recognizing when a student's physical impairment, such as decreased hand strength and dexterity, is likely to interfere with academic achievement. If no further functional improvement is likely despite direct therapy, these professionals can perform assessments of the student's assistive technology needs and recommend compensatory aids that might increase independence within the classroom setting. Ideally, these individuals can be available to evaluate the student's performance with compensatory aids within his or her customary environments and provide training in the use of such aids.

Fig. 8.8 Assistive technology.
Students with arthrogryposis often require adaptations to access computers.

Assistive Technology

Many children with physical disabilities or other special needs can increase their participation in general education programs by using technology aids, such as voice synthesizers or other augmentative communication devices, electric wheelchairs, microswitches, environmental control units, robotic prostheses, and many others. The IDEA defines an assistive technology device as "any item, piece of equipment, or product system, whether acquired commercially off-the-shelf, modified, or customized, that is used to increase, maintain, or improve the functional capabilities of individuals with disabilities" [The Individuals with Disabilities Education Act, Public Law 101-476, 1990 § 1401 (a)25].

Assistive technology services means any service that directly assists a student with a disability in the selection, acquisition, or use of an assistive technology device. Services also include training or technical assistance for the student, the family (where appropriate), and professionals, employers, or other individuals who are involved in major life functions of the individual with disabilities. Furthermore, these services must be coordinated with other therapies or intervention services, such as those associated with existing education and rehabilitation plans or programs.

Who pays? The IDEA requires that if the IEP team determines that a student requires assistive technology devices in order to receive a free, appropriate public education, the IEP must designate the technology services and devices required as special education or related services and that these services must be provided at no cost to the student (Chandler, 1991). Families requesting that the school pay for a particular assistive device must be prepared to show how the device will enhance the child's ability to obtain an appropriate education within the least restrictive classroom setting (Exceptional Parent, 1993). Determining appropriate inclusive technologies is a team decision requiring close communication among classroom teachers, school-based therapists, and technology professionals within health care settings.

Probably the most frequently applied technology device for students with arthrogryposis is the personal computer (Fig. 8.8). Decreased upper extremity functioning inhibits fluent handwriting and can significantly impair the ability to keep notes, complete assignments, and organize work within the classroom. The introduction of keyboarding skills early in curriculum planning may provide these students with opportunities for increased academic achievement and employment. How early should assistive technology be introduced? Several projects in the Handicapped Children's Early Education Program (HCEEP) are using computer technology with infants and young children with disabilities. One example is Project ACTT: Activating Children Through Technology, which is using customized switches, music and voice synthesizers, and other access peripheral devices to increase the ability of very young children to control their environments. Some students with severe arthrogryposis may never be fluent keyboarders. However, recent advances in adaptive access devices, such as voice recognition, touch screens, expanded keyboards, and word prediction programs, can be effective alternatives for the school-age student.

The availability of home computers can reinforce skills learned within the school setting for children and youths with disabilities. Compatibility in software and hardware will be the most important factor in determining what families should buy. Computers within the home setting can also offer increased social and recreational opportunities by leveling the playing field between children with physical disabilities and nondisabled peers. By using a

modem, children with arthrogryposis can form pen pals with individuals in other parts of the country (or world) and access the information highway. The Disabilities, Opportunities, Internetworking, and Technology program (DO-IT) at the University of Washington is one example of how to incorporate computer technology into peer interactions and career planning. This innovative program enables high school students with disabilities in the Northwest region to explore careers in engineering, science, and mathematics through summer study courses and Internetworking throughout the year with mentors (e.g., college students, professors, scientists, and engineers), most of whom have disabilities themselves.

Appropriate Education

The bottom line in developing an appropriate education program for a student with arthrogryposis is that it must meet the unique learning characteristics of the student rather than the needs of the school district. The IDEA does not include language that specifically defines what combination of special education and related services or placement constitutes an appropriate education for a student with disabilities. In general, an IEP must be designed to confer on a disabled student meaningful educational benefit that is consistent with the student's overall abilities. For students with mild disabilities, this implies receiving passing grades and advancing from grade to grade in accordance with federal and state standards for educational quality. For students with severe disabilities, reasonable outcomes include greater independence and self-sufficiency (Osborne, 1992). As in many countries that support integration, the U.S. education system must provide students with disabilities physical access to school buildings, classrooms, and facilities. Perhaps most important, the IDEA specifies that states must also ensure that students with disabilities have the same access to the variety of educational programs and services that nondisabled students have, including such enrichment courses as art, music, home economics, health and physical education, among others (Ordover and Boundy, 1991). An appropriate curriculum for students with arthrogryposis should address each of the following areas.

Academic Achievement

In the United States, both the IDEA and Section 504 of the Vocational Rehabilitation Act require that students with disabilities, including those in public and private institutions, be educated, to the maximum extent possible, with students who do not have disabilities. Although almost all students with mild disabilities are educated in regular classrooms, adherents to the principle of inclusion stress the need for schools to increase their capacity to serve students with severe disabilities within the context of the regular classroom.

This means that the majority of students with arthrogryposis will be held to the same anticipated outcomes for education as their nondisabled peers. Passage of the Goals 2000: Educate America Act in March 1994 provided resources to states to help them develop and implement comprehensive educational reforms that will enable all students to reach high academic standards and occupational skill levels (U.S. Department of Education, 1994). This Act is fully consistent with both the IDEA and the ADA in its intention that students with disabilities are entitled to the same "expectations, treatment and leadership" available to nondisabled students (Exceptional Parent, 1993). School districts must provide appropriate support for students with such disabilities as arthrogryposis in the regular education setting. This may include the use of an instructional aide, peer tutors, classroom notetakers, use

of assistive technology like computers or augmentative communication devices, modification of the regular curriculum, resource room time, or use of a special education consultant to assist the regular education teacher in adapting instruction.

In general, students with disabilities are entitled to the same accommodations in standardized evaluations and test taking as they receive in instruction. Examples include the use of work portfolios rather than standardized tests, extended time limits, individual testing, and use of a reader, scribe, tape recorder, or other assistive devices. Many of these accommodations are available for students with disabilities taking college entrance examinations, based on a written request from the school counselor or testing officer (College Entrance Examination Board, 1994).

Physical Education

Students with arthrogryposis who participate in regular or adaptive physical education programs will have access to activities that can extend the benefits of rigorous occupational and physical therapies. For these students, physical activity can strengthen muscles, maintain joint range, and improve overall conditioning. There are secondary benefits as well, since developing a regular exercise habit can increase these students' feelings of competence and self-esteem, provide recreational opportunities for social interaction, and offer an emotional outlet in times of stress. Students with arthrogryposis should be encouraged to develop physical competence within the limits of their disability (Sawatzky, undated). Family and school personnel should work closely together to develop an individualized physical education program for the student with arthrogryposis that specifically describes what types of individual and group activities (e.g., swimming or wheelchair sports) will be provided during the school year.

Social Interaction

Children with disabilities can be at increased risk for psychosocial problems, particularly those with significant disabilities (Armstrong et al., 1992). Adolescent students are likely to be concerned that their physical limitations may restrict freedom by increasing dependence on parents, limit intimacy in coed relationships, and affect important mobility concerns, such as driving an automobile. Students with impairments like arthrogryposis can develop important social skills by participating in extracurricular activities through school or community groups. Longitudinal studies of youths with disabilities have shown that this type of involvement can increase the probability of postsecondary academic enrollment, residential independence, and full community participation following graduation from high school (SRI International, 1993). Special education students who are experiencing significant difficulty in adjusting to school life are eligible for counseling services. These are defined as a related service under the IDEA and include services provided by qualified social workers, psychologists, guidance counselors, or other qualified personnel. The extent and types of services that the individual student may need should be included in his or her IEP.

Family Involvement

Having an extraordinary child almost inevitably guarantees that parents or caregivers will have an extraordinary relationship with a school program. Generally speaking, they will be afforded many more opportunities to interact with teachers than parents of students without disabilities. At times, this

relationship will be collaborative and mutually supportive. In other cases, it will be adversarial and detrimental to establishing a positive dialogue between home and school (Leff and Walizer, 1992). The entire family, not just the student with disabilities, is likely to need support and services during the school years, and these needs will evolve over time (Alper et al., 1995; Covert, 1992). In some countries, like the United States, parents' involvement in both the evaluation and subsequent placement of their child in special education programs is carefully prescribed in legislation (Hegarty, 1995).

Students with actively involved parents are likely to demonstrate less school absenteeism and higher academic achievement during the school years. Parent support for education is also positively related to postsecondary outcomes for students with disabilities. This support can be demonstrated either directly (e.g., as volunteers in the classroom, home-based teachers, or committee members and by attending parent-teacher conferences) or indirectly by expressing interest and encouragement throughout each step of the educational process for their child. Longitudinal studies of students with disabilities have shown parent expectations to be highly correlated with academic and vocational postsecondary attendance, independent living, and community participation, particularly for students with orthopedic or health impairments (SRI International, 1993).

Involved, informed, and responsible parents are the health or education professional's most important asset. Parents of children with arthrogryposis can best support their child's participation in educational programs by keeping in mind the following points. Parents must be familiar with their rights and responsibilities as specified within their country's education laws. In the United States, the IDEA contains a number of procedural safeguards to ensure that families are involved throughout the planning and provision of educational services for their child (Haring and McCormick, 1990; Ordover and Boundy, 1991).

Informed Consent

A school system must give parents prior written notice before conducting an evaluation of their child's need for special education. Parents must be informed of the rationale for evaluation, what the evaluation consists of, and what rights they have as parents under the provisions of the IDEA. This must be in language that they can understand. Written notice must also be provided if the district refuses a parent's request for an evaluation. Parents have the right to an independent evaluation performed by a professional outside the school district and at public expense if it can be shown to be relevant to placement decisions.

IEP Participation

Parents have the right to participate in educational planning. One or both parents must be present at any IEP meeting and must be given a meaningful opportunity to attend. This includes scheduling the meeting at a time convenient for the parent(s), advance notice so that parents can arrange their schedule, and a description of the purpose, time, and location and individuals who will attend. If parents cannot attend directly, they must be given alternative methods, such as telephone conferencing. The IEP must be developed at the meeting, with parents' participation in planning. IEPs are reviewed periodically, usually on an annual basis, and school districts must follow the procedures described.

Due Process

Before an initial IEP can be implemented, parents must agree that it is an appropriate plan, including the educational placement, and sign it. If they disagree with the proposed program (or any other part of the process, including identification and evaluation, access to school records, or disabling category), they have a right to a hearing to resolve these differences. States vary in the nature and format of this meeting, but it generally involves the use of an independent authority outside the school district.

Parents are not alone. In the United States, there are numerous resources for parents at the local, state, and federal levels to support their involvement in their child's special education program. Many school districts have organized groups for parents of special education students to facilitate positive communication between home and school. Although the names of these groups may vary from state to state, they may be referred to as parent advisory councils, special education parent advisory councils, or special education advisory councils. The U.S. Department of Education also provides financial assistance to help each state operate Parent Training and Information Programs (PTI). These programs offer training and information to parents to enable them to participate more effectively with professionals in meeting the special educational needs of children with disabilities. Parent-to-parent groups are an important service offered by PTIs. These groups provide one-to-one emotional and information support to parents of children with disabilities by matching experienced, or veteran, parents with parents who are newly referred to the program.

Parents are in many cases the best resource for school personnel on how to care for their child's medical condition within the school setting. Parents of children with arthrogryposis can expect to be the primary vehicle whereby information is exchanged between their child's school and the health care facility. Given the low incidence of arthrogryposis among the school-age population, parents can act as an important resource to teachers and other school-based professionals regarding the specific aspects of their child's disability and its impact on their child's participation in the school setting. One parent of a student with arthrogryposis attributed the success of her child in a secondary level school program to the fact that each year she met with each of his teachers and reviewed his medical condition and subsequent physical limitation, and what types of accommodations and adaptive instructional techniques had been used previously to enable her son to successfully complete school assignments.

Preparation for the Future

The transition from adolescence to adult life and responsibilities is a difficult task for any student, particularly for a youth with disabilities. Schools have traditionally been responsible for preparing students for continuing education, independence, and employment (Hallahan and Kauffman, 1991). Education is the key to a person with disabilities becoming self-supporting, as shown by studies reporting that four times as many Americans with disabilities who work have at least a 4-year college education (Bowman and Marzouk, 1992). Students with disabilities who graduate from high school are more likely to be employed than those who do not, but they often earn less than minimum wage (Darrow and Stephens, 1992). Numerous studies

have shown, however, that many students with disabilities do not finish high school and make a successful transition to adult living (Hasazi et al., 1985; Wagner et al., 1991; Sitlington et al., 1993). To this author's knowledge, there are no research findings available that describe the specific postschool outcomes of youths with arthrogryposis. However, since 1985, the U.S. Department of Education has commissioned several longitudinal studies on the "occupational, educational and independent living outcomes of students with disabilities after graduating from high school or otherwise exiting from special education" [The Individuals with Disabilities Education Act, Public Law 101-476, 1990 §1418, (e)(2)(A)]. Findings suggest that no more than 16% of youths with disabilities were enrolled in academic or vocational postsecondary education 2 to 3 years out of secondary school. Students with orthopedic or other health impairments were among the highest groups to pursue higher education, with roughly one-third enrolled in postsecondary academic programs. However, only 26% of youths with orthopedic impairments were employed, and only 1 in 6 was living independently (SRI International, 1993).

In recognition of these and other findings, U.S. federal law mandates that states provide transition services to students 16 years or older (or as early as 14 years) who are receiving special education. These are defined as "a coordinated set of activities…which promote movement from school to post-school activities, including post-secondary education, vocational training, integrated employment…continuing and adult education…independent living or community participation" [The Individuals with Disabilities Education Act, Public Law 101-476, 1990, §1401, (a),(19)]. An Individual Transition Plan (ITP) may be part of the IEP but can be a separate written document that specifies the skills and supportive services that the student will need in the future. It should include short-term and long-term objectives that address what skills the student with disabilities requires to function as independently as possible within the home, community, or work setting. An ITP should clearly state what activities the school will undertake to best meet the postschool needs of the student with disabilities. This can include contacting the state vocational rehabilitation agency, vocational training programs, job placement programs, and prospective employers.

Since the majority of students with arthrogryposis have normal intelligence, their ITP should specify postsecondary education as a goal, and it should include arrangements for formal contact between the secondary school and prospective 2- to 4-year colleges, since this has been shown to significantly increase the likelihood of enrollment. The ADA provides increased opportunities for students with disabilities, since it prohibits discrimination in all aspects of postsecondary services, including recruitment and admissions, academic and athletic programs, student examinations and evaluations, housing, financial aid, counseling and career planning, and placement (West et al., 1993). Follow-up studies of college-age students with disabilities, however, show that far too often, they learn of the availability of services and accommodations by chance or late in their academic careers when poor grades are already on their permanent transcript. For this reason, an ITP for students with arthrogryposis should include information for parents or caregivers and the student that will help them select a college based on its academic program, accessibility, and the services available to meet the individual needs of the student with disabilities. If postsecondary plans specify attending college within the community, a representative from the college or university can become part of the student's transition planning team.

In Closing

Collaboration is not a treasonable act. Children with medically related disabilities, such as arthrogryposis, will require long-term, comprehensive, and coordinated delivery of services that extend well beyond a particular setting, such as the classroom, physician's office, hospital, or home setting (Fig. 8.9). Whereas in the past, many of these children were served in special schools isolated from their nondisabled peers, most countries today actively embrace the concept of integration and are focusing attention on how to best achieve this ideal. Historically, a collaborator was defined as one who "cooperated with or assisted an enemy" (Webster, 1959). Largely as a result of community-based, grassroots efforts by families beginning in the mid-1960s, the U.S. federal government has redefined this term by enacting considerable legislation and committing financial resources to improve educational opportunities for children with disabilities. The resulting framework provides opportunities for increased collaboration between professionals in the fields of medicine and education. Support and respect for families of children with disabilities like arthrogryposis should enhance the quality of this relationship.

Fig. 8.9 A bright future.
Children with arthrogryposis can look forward to a rewarding educational experience, given an effective collaborative effort among parents, school, and health care professionals.

A

Agenesis Absence of an organ usually from failure during embryonic development.
Amyoplasia The most common type of arthrogryposis. Abnormal development of muscle or skin resulting in contracted joints during fetal development.
Ankylosis Immobility and consolidation of a joint due to disease, injury, or surgical procedure.
Arachnodactyly Abnormally long, slender toes and fingers.
Articular Pertaining to the surface of a joint.
Aspiration Act of inhaling.
Asymmetric Uneven, as from one limb to another.
Autosomal dominant In genetics, a dominant trait is expressed when it is carried by only one member of a pair of chromosomes.
Autosomal recessive In genetics, a recessive trait cannot be expressed unless it is carried by both members of a pair of chromosomes.

C

Calcaneocuboid Articulation between the heel bone and cuboid bone in the foot.
Calcaneus The heel bone.
Caudal Pertaining to an anatomic position away from the head toward the tail.
Central nervous system The brain and spinal cord.
Chrondroplasty Plastic surgery of cartilage.
Cleft palate Elongated opening in the roof of the mouth resulting from failure of parts to fuse during embryonic development.
Clubfoot Also known as talus equinovarus. Inward turning heel and forefoot with increased toe down motion of the foot at the ankle.
Collagen synthesis Cell production of protein material that is the supporting structure in connective tissue.
Congenital Present at birth.
Contracture Permanently shortened muscle tissue from paralysis, spasm, or fibrosis of tissue at a joint.
Craniofacial abnormalities Malformations of the bones of the head and face.
Curettage Removal of growths or other material from the wall of a cavity or other surface with a spoon-shaped instrument (curet).
Cutaneous Pertaining to the skin.

D

DIP flexion Flexion at the distal interphalangeal joint.
Distal Away from the point of reference, origin, or attachment.
Dorsal Back.
Dorsiflex Upward bend of a body part.
Dynamic splint Allows for or provides motion by use of outside forces such as springs, rubber bands, or electricity or by transfer of movement from other body parts.

E

Empiric Based on scientific experience.
Epicanthic fold A vertical fold of skin on either side of the nose.
Epiphysis End of a bone that lies between the joint surface on one side and the epiphyseal plate on the other.
Equinovarus Also known as clubfoot.
Eversion Outward motion of the heel.
Extension Straightening of a joint.
External rotation Rotation of limb away from midline of the body.

F

Femoral Pertaining to the femur or thigh.
Femur Thigh bone.
Fibrosis Degeneration or excessive formation of normal tissue resulting in thickened and scarred connective tissue.
Fixator In orthopedics, the use of metallic devices inserted in bone to hold the position during healing.
Flexion Bending across a joint resulting in decreased joint angle.
Flexion contracture Fixed deformity in which a joint retains a permanent degree of bending.

G

Gastrocnemius Calf muscle leading to the Achilles tendon that flexes both knee and ankle.
Genu recurvatum Also known as backknee. Ability of the knee to bend backward.
Grasp reflex A reflex consisting of a grasping motion of the fingers or toes in response to stimulation.

H

Hemangioma A tumor of blood vessels.
Hindfoot Heel bone.
Hypoplasia A reduction in size of a body part due to arrested development.

I

Inguinal hernia Protrusion of a loop of an organ or tissue through an abnormal opening in the groin.
Internal rotation Rotation of a limb toward the midline of the body.
Interphalangeal joint A joint between two fingers.
Inversion Inward motion of the heel, flexion, supination, and adduction of the foot.

J

Joint capsule Connective tissue housing a joint.

K

Kyphoscoliosis Curvature of the spine in two directions, side to side and back to front.
Kyphosis Also known as hunchback. Abnormal convex curvature of the spine as viewed from the side.

L

Laryngeal Pertaining to the larynx or voice box.
Laryngopharyngeal Pertaining to the larynx and the pharynx.
Lateral flexion Bending to the side.
Lumbrosacral Pertaining to the spine.

M

MCP flexion Flexion at the metacarpophalangeal joint.
Metacarpals The five long bones of the palm.
Metaphyseal Pertaining to the widened end of the tubular bone shaft, the site of active bone formation.
Metatarsals The five long bones of the foot.
Metatarsus adductus Turning in of the forefoot.
Micrognathia Unusually small jaw.
Midtarsal dislocation Dislocation of the midfoot.
Mitochondrial Components found in the cytoplasm of cells which contain RNA and DNA by means of which they independently replicate and code for the synthesis of some proteins.

N

Naviculoectomy Surgical removal of the navicular, a small bone in the hand or foot.
Neurovascular bundle Any grouping of nerves and blood vessels that supply a specific region.

O

Oblique talus Deformity characterized by an oblique position of the ankle bone.
Oral motor Pertaining to movements of the mouth.
Osteotomy Surgical division of a bone.

P

Palmar skin crease Skin crease in the palm caused by natural folds in the skin.
Palpation Use of the fingers to determine firmness, shape, and motion of a body part.
Patella Kneecap.
Pathogen Any disease-producing microorganism.
Pes equinovarus Also known as clubfoot.
Phalanges Any of the bones of the fingers or toes.
Pinnae External ears.
PIP flexion Flexion at the proximal interphalangeal joint.
Plantar flexion Toe down motion of the foot.
Popliteal Pertaining to the back or posterior surface of the knee.
Posterior tibialis Muscle that rotates the foot inward under the ankle.
Pronation Inward rotation of the hand or foot.
Proximal Near the point of reference, which is usually the trunk or main part of the body.
Pterygium Also called webbed joint.

R

Radial deviation Abnormal angulation of the wrist or fingers toward the radius.
Radiograph X-ray.
Ranging Moving a joint through its full range of motion.

S

Scoliosis Side to side curvature of the spine.
Serial casts Any sequence of casts to progressively correct a deformity.
Spinal stenosis A developmental malformation that produces a narrow bony spinal canal with nerve root compromise.
Static splint Has no moving parts; maintains a joint in a desired position.
Subcuticular Thick layer of skin below the outermost layer of skin.
Subluxated Also known as a partial dislocation. Malaligned opposing joint surfaces with a partial loss of contact.
Subtalar joint Joint between the ankle bone (talus) and heel bone (calcaneus).
Supination Outward rotation of the forearm or foot.

T

Tactile input Stimuli entering the body through skin contact.
Talectomy Surgical removal of the talus.
Talonavicular joint Articulation between the talus and the navicular bone.
Talus Bone beneath the tibia that is a part of the ankle joint.
Tendo Achilles Achilles tendon situated at the back of the ankle and inserting into the heel bone.
Tibia Also known as the shin bone. Medial and larger bone of the lower leg.
Triceps Arm muscle that extends the elbow.
Trismus Spasm of chewing muscles, with difficulty in opening the mouth.

U

Ulnar deviation Abnormal angulation of the wrist or fingers toward the ulna.

V

Valgus Turning away from the midline of the body.
Varus Turning toward the midline of the body.
Vertical talus Deformity characterized by a vertical position of the ankle bone (talus).
Visceral anamolies Malformations of any of the organs of the three great cavities of the body, especially in the abdomen.
Volar Underneath surface, palm or sole.

Australia
The Australian Arthrogryposis Group

c/- 28 Stewart Avenue
Curl Curl, NSW 2096
Australia
02-452-1846

Canada
Canadian Arthrogryposis Support Team (CAST)

Joyce Jeppesen
365 Fiddlers Green Road
Ancaster, Ontario. L9G 1X2
Canada
800-567-2873 or 905-648-2007

England
The Arthrogryposis Group

1,The Oaks
Common Mead Land
Gillingham, Dorset SP8 4SW
England
01747-822655

France
Agnes and Frances Novaille (Contact only)

52, bis route de St. Cyr.
Marly le Roi
78160
France

Germany
IGA

Cornelia Umber
Haupstrasse 130
D-79713 Bad
Germany

Ireland
The Arthrogryposis Association

Christine Healy
19 Lower Beechwood Avenue
Ranelagh Dublin 6
Ireland
01-962915

Italy
G.I.S.A

Viale Dell'Esperanto, 71
01144
Rome, Italy

Netherlands
Support Group for AMC

Manja van den Elsen
Kamillestraat 15
5741 VN Beek en Donk
Netherlands

New Zealand
The Arthrogryposis Group of New Zealand (NZ)

Marianne Devenoges
40 Eversham Road
Mt. Mauganui
New Zealand
07-575-4416

Spain
Gillian Rowland-Carrascal (Contact only)

Calle Urbanizacion Zulema
Caceres 78, Villabilla
Madrid, Spain

United States

Alabama Based Support Group for AMC

Debbie and Robert Adams
700 Redwood Drive
Maylene, AL 35114
205-664-3196

Arthrogryposis Utah Support Group

Sue Bryson
2241 S. 500 West
Bountiful, UT 84010
801-292-8323

Avenues

Mary Anne and Jim Schmidt
P.O. Box 5192
Sonora, CA 95370

Georgia Chapter

National Arthrogryposis Foundation AMC
2347 Melinda Drive
Atlanta, GA 30345
404-325-8982

National Arthrogryposis Foundation

Jerry and Elaena Faraino
3356 S. Cove Trace
Birmingham, AL 35216
205-823-0786

National Foundation for AMC

Elaine Muzzo and Bev Barnes
P.O. Box 382
Chicago Heights, IL 60411
312-755-7072

Northern Westchester Support Group

Allida Stauber
95 Oliver Road
Bedford, NY 10506
914-234-7628

Southern Arizona Support Group for AMC Birth Defect

Georgia McLaughlin
232 James Drive N.E.
Sierra Vista, AZ 85635
602-458-2306

Exceptional Parent Magazine

120 State Street
Hackensack, NJ 07601
1-800-E-PARENT

A magazine primarily for families of children and young adults with disabilities and special health care needs that is an authoritative source of information and help for professionals as well. It publishes an informative resource guide each January that is a directory of national associations, products, and services of interest to these families and professionals.

Abiding Hearts

P.O. Box 5245
Bozeman, MT 59717
406-587-7421

Support for parents continuing a pregnancy after a prenatal diagnosis of fatal or nonfatal birth defects.

STOMP (Specialized Training of Military Parents)

c/o Washington PAVE
12208 Pacific Highway South
Tacoma, WA 98499
206-984-7520

Provides information to military families about obtaining early intervention and special education services for their children.

Aarskog, D. 1971. Pterygium syndrome. BDOAS. 7(6):232-233.

Aase, J.M., and Smith, D.W. 1968. Dysmorphogenesis of joints, brain, and palate: A new dominantly inherited syndrome. J Pediatr. 73(4):606-609.

Abbott, L.C., Finnell, R.H., Chernoff, G.F., Parish, S.M., and Gay, C.C. 1986. Crooked calf disease: A histological and histochemical examination of eight affected calves. Vet Pathol. 23(6):734-740.

Abeliovich, D., Carmi, R., Karplus, M., Bar Ziv, J., and Cohen, M.M. 1979. Monosomy 21: A possible stepwise evolution of the karyotype. Am J Med Genet. 4:279-286.

Adams, C., Becker, L.E., and Murphy, E.G. 1988. Neurogenic arthrogryposis multiplex congenita: Clinical and muscle biopsy findings. Pediatr Neurosci. 14(2):97-102.

Affleck, G., and Tennen, H. 1993. Cognitive adaptation to adversity: Insights from parents of medically fragile infants. In Cognitive Coping, Families, and Disability, ed. A.P. Turnbull, J.M. Patterson, S.K. Behr, D.L. Murphy, J.M. Marquis, and M.J. Blue-Banning. Baltimore: Paul H. Brookes.

Agapitos, M., GeorgiouTheodoropoulou, M., Koutselinis, A., and Papacharalambus, N. 1988. Arthrogryposis multiplex congenita, Pena Shokeir phenotype, with gastroschisis and agenesis of the leg. Pediatr Pathol. 8(4):409-413.

Ainscow, M., Jangira, N.K., and Ahuja, A. 1995. Education: Responding to special needs through teacher development. In Disabled Children & Developing Countries, ed. P. Zinkin and H. McConachie, pp. 131-146. Mac Keith Press: London.

Al Awadi, S.A., Naguib, K.K., Teebi, A.S., Farag, T.I., Devarajan, L.V., and El Khalifa, M.Y. 1986. Lethal multiple pterygium syndrome: Report of two sporadic cases from Kuwait. J Kwt Med Assoc. 20:135-140.

Albisetti, W., Facchini, R., Prina, A., Scotti, L., and Borzani, M. 1989. Congenital multiple arthrogryposis. Report of a case. Minerva Pediatr. 41(9):477-480.

Alexiou, D., Manolidis, C., Papaevangellou, G., Nicolopoulos, D., and Papadatos, C. 1976. Frequency of other malformations in congenital hypoplasia of depressor anguli oris muscle syndrome. Arch Dis Child. 51(76):891-893.

Al Gazali, L.I., and Lytle, W. 1994. Otospondylomegaepiphyseal dysplasia: Report of three sibs and review of the literature. Clin Dysmorpho. 3: 46-54.

Almeida, L., AnyaneYeboa, K., Grossman, M., and Rosen, T. 1988. Myelomeningocele, Arnold Chiari anomaly and hydrocephalus in facial dermal hypoplasia. Am J Med Genet. 30:917-923.

Alper, S., Scholoss, P.J., and Scholoss, C.N. 1995. Families of children with disabilities in elementary and middle school: Advocacy models and strategies. Exceptional Children. 62(3):261-270.

Altman, H.S., and Davidson, L.T. 1939. Amyoplasia congenita (arthrogryposis multiplex congenita). J Pediatr. 15:551-557.

Alward, W.L., Krachmer, J.H., and Macsai, M.S. 1990. Arthrogryposis multiplex congenita with Peter's anomaly. J Pediatr Ophthalmol Strabismus. 27(6):329.

Ambler, L. (ed) 1988. Children with disabilities: Understanding sibling issues. NICHCY News Digest. (11).

American Academy of Pediatrics: Committee on Children with Disabilities 1992. Pediatrician's role in the development and implementation of an individual education plan (IEP) and/or individual family service plan (IFSP). Pediatr. 89(2):340-342.

American Academy of Pediatrics: Committee on Children with Disabilities 1993. Provision of related services for children with chronic disabilities. Pediatr. 92(6):879-881.

American Nurses Association. 1983. Standards of School Nursing Practice. Kansas City, MO: Author.

Ames, L.B. 1992. Raising good kids: A developmental approach to discipline. New York: Dell.

Amick, L.D., Johnson, W.W., and Smith, H.L. 1967. Electromyographic and histopathologic correlations in arthrogryposis. Arch Neurol. 16:512-523.

Anderson, R.A., Koch, S., and Camerini Otero, R.D. 1984. Cardiovascular findings in congenital contractural arachnodactyly: Report of an affected kindred. Am J Med Genet. 18:265-271.

Andre, M., Vigneron, J., and Didier, F. 1981. Abnormal facies, cleft palate, and generalized dysostosis: A lethal X-linked syndrome. J Pediatr. 98(5):747-752.

Andrews, A.D., Barrett, S.F., Yoder, F.W., and Robbins, J.H. 1978. Cockayne's syndrome fibroblasts have increased sensitivity to ultraviolet light but normal rates of unscheduled DNA synthesis. J Invest Dermat. 70:237-239.

Andrisano, A., Manfrini, M., Zucchi, M., and Mignani, G. 1988. Arthromyolysis of the elbow in arthrogryposis. Ital J Orthop Traumatol. 14(2):239-242.

Anichini, C., Tomaccini, D., Scarinci, R., and Vivarelli, R. 1986. Multiple pterygium syndrome. Pediatr Med Chir (Italy). 8(6):881-884.

Antich, J., Iriondo, M., Lizarraga, I., Manzanares, R. and Cusi, V. 1993. Radiohumeral synostosis, femoral bowing, other skeletal anomalies and anal atresia, a variant example of Antley-Bixler syndrome? Genet Counsel 4:207-211.

Antley, R., and Bixler, D. 1975. X trapezoidocephaly, midfacial hypoplasia and cartilage abnormalities with multiple synostoses and skeletal fractures. BDOAS. 11(2):397-401.

Anyane Yeboa, K., Collins, M., Kupsky, W., Maidman, J., Malin, J., and Yeh, M. 1987. Hydrolethalus (Salonen Herva Noria) syndrome: Further clinicopathological delineation. Am J Med Genet. 26:899-907.

Arduini, D., Rizzo, G., Giorlandino, C., Missone, A., Nava, S., Dell'Acqua, S., Valensise, B., and Romanini, C., 1985. The fetal behavioural states: An ultrasonic study. Prenat Diagn. 5:269-279.

Argov, Z., Gardner Medwin, D., Johnson, M.A., and Mastaglia, F.D. 1984. Patterns of muscle fiber type disproportion in hypotonic infants. Arch Neurol. 41:53-57.

Armstrong, R.W., Rosenbaum, P.L., and King, S. 1992. Self-perceived social function among disabled children in regular classrooms. Dev Behavioral Pediatr. 13(1):11-16.

Asha Bai, P. V., and John, T.J. 1979. Congenital skin ulcers following varicella in late pregnancy. J Pediatr. 94(1):65-67.

Atkins, R.M., Bell, M.J., and Sharrard, W.J.W. 1985. Pectoralis major transfer for paralysis of elbow flexion in children. JBJS. 67B(4):640-644.

Aughton, D.J., and Cassidy, S.S. 1987. Hydrolethalus syndrome: Report of an apparent mild case, literature review, and differential diagnosis. Am J Med Genet. 27:935-942.

AVENUES: Newsletter of the National Support Group for Arthrogryposis Multiplex Congenita. Mary Ann and Jim Schmidt (eds.), P.O. Box 5192, Sonora, CA 95370.

Avlves, A.F., and Azevedo, E.S. 1977. Recessive form of Freeman-Sheldon syndrome or "whistling face." J Med Genet. 14:139-141.

Awwaad, S. 1958. Amyoplasia congenita. Review of literature and report of three cases. Arch Pediatr. October:421-430.

Aziz, M.A. 1979. Muscular and other abnormalities in a case of Edwards' syndrome (18 trisomy). Teratology. 20:303-312.

Aziz, M.A. 1981. Possible "atavistic" structures in human aneuploids. Am J Phys Anthropology. 54:347-353.

Bacino, C.A., Platt, L.D., Garber, A., Carlson, D., Pepkowitz, S., Lachman, R.S., Sharony, R., Rimoin, D.L., and Graham, J. M. 1993. Fetal akinesia/hypokinesia sequence: Prenatal diagnosis and intra-familial variability. Prenatal Diag 13:1011-1019.

Bailey, D.B., and Simeonsson, R.J. 1988. Assessing needs of families with handicapped infants. Special Education. 22:117-127.

Baines, D.B., Doublas, I.D., and Overton, J.H. 1986. Anaesthesia for patients with arthrogryposis multiplex congenita: What is the risk of malignant hyperthermia? Anaesth Intensive Care. 14(4):370-372.

Baird, H.W., III. 1964. Kindred showing congenital absence of dermal ridges (fingerprints) and associated anomalies. J Pediatr. 64(5):621-631.

Bajnoczky, K., and Meggyessy, V. 1985. Coincidence of paternal 13pYq translocation and maternal increased 13p NOR activity in a child with arthrogryposis and other malformations. Acta Paediatr Hung. 26(2):151-156.

Bakkeren, J., Carpay, I., Weemaes, C., and Monnens, L. 1976. Cellular immunity in cerebrohepatorenal syndrome of Zellweger. Lancet. 6 Nov:1029.

Balestrazzi, P., Corrini, L., Villani, G., Bolla, M.P., Casa, F., and Bernasconi, S. 1980. The Cohen syndrome: Clinical and endocrinological studies of two new cases. J Med Genet. 17:430-432.

Bamforth, J.S. 1992. Amniotic band sequence: Streeter's hypothesis reexamined. Am J Med Genet. 44:280-287.

Bamshad, M., Watkins, W.S., Zenger, R.K., Bohnsack, J.F., Carey, J.C., Otterud, B., Krakowiak, P.A., Robertson, M., and Jorde, L.B. 1994. A gene for distal arthrogyrposis type I maps to the pericentromeric region of chromosome 9. Am J Hum Genet. 55:11531158.

Banker, B.Q. 1985. Neuropathologic aspects of arthrogryposis multiplex congenita. Clin Orthop. (194):30-43.

Banker, B.Q. 1986. Arthrogryposis multiplex congenita: Spectrum of pathologic changes. Hum Pathol. 17(7):656-672.

Baraitser, M. 1982. A new camptodactyly syndrome. J Med Genet. 19:40-43.

Baraitser, M., Burn, J., and Fixsen, J. 1983. A recessively inherited windmillvane camptodactyly / ichthyosis syndrome. J Med Genet. 20:125-127.

Baraka, A. 1981. Antagonism of succinylcholine induced contracture of denervated muscles by D-tubocurarine. Anesth & Analgesia. 60(B):605-607.

Barakat, L.P., and Linney, J.A. 1992. Children with physical handicaps and their mothers: The interrelation of social support, maternal adjustment, and child adjustment. J Pediatr Psychol. 17:725-739.

Barna, J. 1988. Knee deformities in patients with Larsen syndrome. Magy Traumatol Orthop Helyreallito Seb. 31(1):13-19.

Barnard, K.E., Morisset, C., and Spieker, S. 1993. Preventive interventions: Enhancing parent infant relationships. In Handbook of Infant Mental Health, ed. C.H Zeanah. New York: Guilford.

Barrera, M. 1986. Distinctions between social support concepts, measures, and models. Am J Community Psychol. 14:413-445.

Bartsocas, C.S., and Papas, C.V. 1972. Popliteal pterygium syndrome: Evidence for a severe autosomal recessive form. J Med Genet. 9:222-226.

Barylak, A., and Kozlowski, K. 1972. Dyggve Disease. Aust. Paediat. J. 8:338-341.

Bass, H.N., Sparkes, R.S., Crandall, B.F., and Marcy, S.M. 1981. Congenital contractural arachnodactyly, keratoconus, and probable Marfan syndrome in the same pedigree. J Pediatr. 98(4):591-593.

Baty, B.J., Cubberley, D., Morris, C., and Carey, J. 1988. Prenatal diagnosis of distal arthrogryposis. Am J Med Genet. 29(3):501-510.

Bawle, E., and Quigg, M.H. 1992. Ectopia lentis and aortic root dilatation in congenital contractural arachnodactyly. Am J Med Genet. 42:19-21.

Bayne, L.G. 1985. Hand assessment and management of arthrogryposis multiplex congenita. Clin Orthop. 194:68-73.

Beals, R.K., and Hecht, F. 1971. Congenital contractural arachnodactyly: A heritable disorder of connective tissue. JBJS. 53A(5):987-993.

Beavers, J., Hampson, R.B, Hulgus, Y.F., and Beavers, W.R. 1986. Coping in families with a retarded child. Fam Process. 25:365-378.

Beckerman, R.C., and Buchino, J.J. 1978. Arthrogryposis multiplex congenita as part of an inherited symptom complex: Two case reports and a review of the literature. Pediatr. 61(3):417-422.

Begleiter, M.L., Callenbach, J.C., Hall, R.T., and Harris, D. 1980. Atypical amniotic band syndrome. Lancet. 19 Jul:153.

Behr, S.K., and Murphy, D.L. 1993. Research progress and promise: The role of perceptions in cognitive adaptation to disability. In Cognitive Coping, Families, and Disability, ed. A.P. Turnbull, J.M. Patterson, S.K. Behr, D.L. Murphy, J.M. Marquis, and M.J. Blue-Banning. Baltimore: Paul H. Brookes.

Beighle, C., Karp, J.W., Hall, J.G., and Hoehn, H. 1977. Small structural changes of chromosome 8: Two cases with evidence for detection. Hum Genet. 38:113-121.

Bell, D.R., and Smith, D.W. 1972. Myotonic dystrophy in the neonate. J Pediatr. 81(1):83-86.

Bell, E., and Graham, H.K. 1995. A new material for splinting neonatal limb deformities. J Pediatr Orthop. 15:613-616.

Bellon, J.M., and Filipe, G. 1987. Spinal complications encountered in Larsen's syndrome. Apropos of 3 cases. Rev Chir Orthop. 73(1):57-62.

Bender, L.H., and Withrow, C.A. 1989. Arthrogryposis multiplex congenita. Orthop Nurs. 8(5):29-34.

Bendon, R., Dignan, P., and Siddiqi, T. 1987. Prenatal diagnosis of arthrogryposis multiplex congenita. J Pediatr. 111:942-946.

Bennett, J.B., Hansen, P.E., Granberry, W.M., and Cain, T.E. 1985. Surgical management of arthrogryposis in the upper extremity. J Pediatr Orthop. 5(3):281-286.

Berk, P.D., Wolkoff, A.W., and Berlin, N.I. 1975. Inborn errors of bilirubin metabolism. Med Cl N Amer. 59(4):803-816.

Bethem, D., Winter, R.B., and Lutter, L. 1980. Disorders of the spine in diastrophic dwarfism. JBJS. 62(4):529-536.

Bettman, A.G. 1946. Congenital bands about the shoulder girdle. Plast Reconst Surg.:205-215.

Bharucha, E.P., Pandya, S.S., and Dastur, D.K. 1972. Arthrogryposis multiplex congenita part 1: Clinical and electromyographic aspects. J Neuro, Neurosurgery, & Psychia. 35:425-434.

Bianchi, D.W., and Van Marter, L.J. 1994. Approach to ventilator-dependent neonates with arthrogryposis. Pediatrics 94:682.

Bieber, F.R., Mostoufizadeh, M., Birnholz, J.C., and Driscoll, S.G. 1984. Amniotic band sequence associated with ectopia cordis in one twin. J Pediatr. 105(5):817-819.

Bixler, D., Poland, C., and Nance, W.E. 1973. Phenotypic variation in the popliteal pterygium syndrome. Clin Genet. 4:220-228.

Bjerkrein, I., Skogland, L.B., and Trygstad, O. 1976. Congenital contractural arachnodactyly. Acta Orthop Scand. 47:250-253.

Blattner, R.J. (ed.). 1969. Bell's palsy in children. J Pediatr. 74(5):835-837.

Blau, E.B. 1985. Familial granulomatous arthritis, iritis, and rash. J Pediatr. 107(5):689-693.

Bockel, J., Grassl, F., Pfeiffer, R.A., Ruprecht, K.W., and Heidbreder, E. 1984. Connatal ptosis: A symptom of the syndrome of multiple pterygium and arthrogryposis. Klin Monatsbl Augenheilkd (Germ). 185(2):123-125.

Bofinger, M.K., Dignan, P., Schmidt, R.E., and Warkany, J. 1973. Reduction malformations and chromosome anomalies. Am J Dis Child. 125:135-143.

Bonafede, R.P., and Beighton, P. 1978. The Dyggve Melchior Clausen syndrome in adult siblings. Clin Genet. 14:24-30.

Bonaventure, J., Lasselin, C., Mellier, J., Cohen Solal, L., and Maroteaux, P. 1992. Linkage studies of four fibrillar collagen genes in three pedigrees with Larsen-like syndrome. J Med Genet. 29(7):465-470.

Booke, M.H., and Engel, W.K. 1969. The histographic analysis of human muscle biopsies with regard to fiber types 4 children's biopsies. Neurology. 19:591-605.

Borlum, K.G. 1984. Amniotic band syndrome in second trimester associated with fetal malformations. Prenat Diagn. 4:311-314.

Borrow, E.S., Avruskin, T. W., and Siller, J. 1985. Mother daughter interaction and adherence to diabetes regimens. Diabetes Care. 8:146-151.

Bowen, J.R., Ortega, K., Ray, S., and Mac Ewen, G.D. 1985. Spinal deformaities in Larsen's syndrome. Clin Orthop. 197:159-163.

Bowen, P., Lee, C.S.N., Zeljaveger, H., and Lindernberg, R. 1964. A familial syndrome of the multiple congenital defects. Bull Johns Hopkins Hosp. 114:402-414.

Bowlby, J. 1982. Attachment and Loss (Volume I): Attachment, 2nd ed. New York: Basic Books.

Bowman, O.J., and Marzouk, D.K. 1992. Implementing the Americans with Disabilities Act of 1990 in higher education. Am J Occupational Ther. 46(6):521-533.

Bowser Riley, S., and Bain, A.D. 1975. Chromosome abnormalities in Dupuytren's disease. Lancet. 27 Dec:1282-1283.

Boylan, K.B., Ferriero, D.M., Greco, C.M., Sheldon, R.A., and Dew, M. 1992. Congenital hypomyelination neuropathy with arthrogryposis multiplex congenita. Ann Neurol. 31(3):337-340.

Breslau, N. 1985. Psychiatric disorder in children with physical disabilities. J Am Acad Child Adol Psychiat. 24:87-94.

Brewerton, D.A. 1988. Causes of arthritis. Lancet. 5 Nov:1063-1066.

Brooke, M.H., Carroll, J.E., and Ringel, S.P. 1979. Congenital hypotonia revisited. Muscle & Nerve. 2:84-100.

Broome, D.L., Ebbin, A.J., Jung, A.L., Feinauer, L.R., and Madsen, M. 1976. Aberrant tissue bands and craniofacial defects. BDOAS. 12(5):65-79.

Brown, C., Goodman, S., and Kupper, L. 1993. The unplanned journey: When you learn that your child has a disability. NICHCY News Digest. 3(1):5-15.

Brown, F.R. III, McAdams, A.J., Cummins, J.W., Konkol, R., Singh, I., Moser, A.B., and Moser, H.W. 1982. Cerebrohepatorenal (Zellweger) syndrome and neonatal adrenoleukodystrophy: Similarities in phenotype and accumulation of very long chain fatty acids. Johns Hopkins Med J. 151:344-361.

Brown, L.M., Robson, M.J., and Sharrard, W.J.W. 1980. The pathophysiology of arthrogryposis multiplex congenita neurologica. JBJS (Br). 62B(3):291-296.

Browne, D. 1955. Congenital deformities of mechanical origin. Arch Dis Child. 50:37-41.

Bruhn, J.G., and Phillips, B.U. 1987. A developmental basis for social support. J Beh Med. 10:213-229.

Brumback, R.A., Yoder, F.W., Andrews, A.D., Peck, G.L., and Robbins, J.H. 1978. Recognition and relationship to neurological abnormalities in Cockayne's syndrome. Arch Neurol. 35:337-345.

Buchanan, P.D., Rhodes, R.L., and Stevenson, C.E., Jr. 1983. Interstitial deletion 2q31> q33. Am J Med Genet. 15:121-126.

Buchler, U. 1993. Arthrogryposis multiplex congenita of the upper extremity. Handchir Mikrochir Plast Chir. 25(1):3-11.

Buebendorf, N.D., Concannon, M.J., Gaines, R.W., and Puckett, C.L. 1992. Skin expansion as preparation for an opening wedge osteotomy of the midfoot in arthrogryposis. Mo Med. 89(9):671-674.

Bui, T.H., Lindholm, H., Demir, N., and Thomassen, P. 1992. Prenatal diagnosis of distal arthrogryposis type I by ultrasonography. Prenat Diagn. 12(2):1047-1053.

Burhan, S., and Meyer, J. 1981. Familial trigonocephaly associated with short stature and developmental delay. Am J Dis Child. 135:711-712.

Butler, C., and McKay, T.M. 1984. Motorized wheelchair driving by disabled children. Arch Phys Med Rehabil. 65:95-97.

Butler, C., Okamoto, G.A., and McKay, T.M. 1984. Powered mobility for very young disabled children. Dev Med Child Neur. 25:472-474.

Camera, G., Serra, G., and Selicorni, A. 1990. "C" trigonocephaly syndrome: Two additional cases. Am J Med Genet. 37:463-464.

Cantu, J.M., Garcia Cruz, D., Gil Viera, J., Nazara, A., Ramirez, M.L., Sole Pujol, M.T., and Sanchez Corona, J. 1985. Guadalajara camptodactyly syndrome type II. Clin Genet. 28:54-60.

Cantu, J.M., Rivera, H., Nazara, A., Rojan, Q., Hernandez, A., and Garcia Cruz, D. 1980. Guadalajara camptodactyly syndrome: A distinct probably autosomal recessive disorder. Clin Genet. 18:153-159.

Carey, J.C., and Hall, B.D. 1978. Confirmation of the Cohen syndrome. J Pediatr. 93(2):239-244.

Carlson, W.O., Speck, G.J., Vicari, V., and Wenger, D.R. 1985. Arthrogryposis multiplex congenita: a long term follow up study. Clin Orthop. 194:115-123.

Carnevale, A., Hernandez, M., Limon Toledo, I., Frias, S., Castillo, J., and Del Castillo, V. 1982. A clinical syndrome associated with dup(5p). Am J Med Genet. 13:277-283.

Carroll, R.E. 1962. Restoration of elbow flexion by transplantation of the sternocleidomastoid muscle. JBJS (A). 44:10-39.

Carroll, R.E., and Hill, N.A. 1970. Triceps transfer to restore elbow flexion: A study of fifteen patients with paralytic lesions and arthrogryposis. JBJS (AM). 52:239-244.

Cartlige, I. 1984. Observations on the epidemiology of club foot in Polynesian and Caucasian populations. J Med Genet. 21:290-292.

Cawston, T. 1991. Arthritis and the collagen connection. New Scientist. 8 Jun:39-41.

Cayler, G.G. 1968. Cardiofacial syndrome, congenital heart disease and facial weakness, a hitherto unrecognized association. Arch Dis Child. 69-75.

Cazzato, G., and Walton, J.N. 1968. The pathology of the muscle spindle: A study of biopsy material in various muscular and neuromuscular diseases. J Neurol Sci. 7:15-70.

Cetta, G., Lenzi, L., Ruggeri, A., Tenni, R., and Boni, M. 1979. Biochemical and structural abnormalities of the connective tissue in Larsen's syndrome. Int Orthop. 3(1):47-53.

Chaikind, S. 1992. Children and the ADA: The promise of tomorrow. Exceptional Parent. 22(2):M8-M10.

Chandler, B.E. 1991. Providing assistive technology services within the schools. OT Week. 5:8.

Chappard, D., and Lauras, B. 1983. Unusual morphodysplasia as a result of early amnion rupture: Umbilicocephalic adherence. J Genet Hum. 31(4):329-335.

Charnas, L., Trapp, B., and Griffin, J. 1988. Congenital absence of peripheral myelin: Abnormal Schwann cell development causes lethal arthrogryposis multiplex congenita. Neurology. 38(6):966-974.

Chitayat, D., Hall, J.G., Couch, R.M., Phang, M.S., and Baldwin, V.J. 1990. Syndrome of mental retardation, facial anomalies, hypopituitarism, and distal arthrogryposis in sibs. Am J Med Genet. 37(1):65-70.

Chitayat, D., Hodgkinson, K. A., Blaichman, S., Chen, M.F., Watters, G.V., Khalife, S., and Hall, J.G. 1991. Syndrome of mental retardation and distal arthrogryposis in sibs. Am J Med Genet. 41(1):49-51.

Chitayat, D., Hodgkinson, K.A., Ginsburg, O., Dimmick, J., and Watters, G.V. 1992. King syndrome: A genetically heterogeneous phenotype due to congenital myopathies. Am J Med Genet. 43:954-956.

Chonmaitree, T., Menegus, M.A., Schervish Swierkosz, E.M., and Schwalenstocker, E. 1981. Enterovirus 71 infection: Report of an outbreak with two cases of paralysis and a review of the literature. J Pediatr. 67(4):489493.

Chou, M., and Nonaka, J. 1978. Werdnig Hoffmann disease: Proposal of a pathogenetic mechanism. Acta Neuropath. (Berl). 41:45-54.

Chowdhary, U.M., Ibrahim, A.W., and Dawodu, A.H. 1989. Tectocerebellar dysraphia with occipital encephalocele. Surg Neurol. 31:310-314.

Christ, F., and Anders, G. 1981. The radiological features of arthrogryposis multiplex congenita. ROFO Fortschr Geb Rontgenstr Nuklearmed. 135(5):592-596.

Christiaens, GCML, Van Baarlen, J., Huber, J., and Leschot, N.J. 1989. Fetal limb constriction: A possible complication of CVS. Prenat Diagn. 9:67-71.

Christian, J.C., Andrews, P.A., Conneally, P.M., and Muller, J. 1971. The adducted thumbs syndrome: An autosomal recessive disease with arthrogryposis, dysmyelination, cranioastenosis, and cleft palate. Clin Genet. 2:95-103.

Clancy, R.R., Kelts, K.A., and Oehlert, J.W. 1980. Clinical variability in congenital fiber type disproportion. J Neurol Sci. 46:257-266.

Clarren, S.K., and Hall, J.G. 1983. Neuropathologic findings in the spinal cords of 10 infants with arthrogryposis. J Neurol Sci. 58(1):89-102.

Cluff, L.E. 1985. Chronic disability of infants and children: A foundation's experience. J Chronic Disabilities. 38 (1):113-124.

Cohen, M.E., Duffner, P.K., and Heffner, R. 1978. Central core disease in one of identical twins. J Neurol, Neurosurg, Psychiat. 41:659-663.

Cohen, M.M., Hall, B.D., Smith, D.W., Graham, C.B., and Lampert, K.J. 1973. A new syndrome with hypotonia, obesity, mental deficiency, and facial, oral, ocular, and limb anomalies. J Pediatr. 83(2):280-284.

Cohen, M.M., Lerner, C., and Balkin, N.E. 1983. Duplication of 16p from insertion of 16p into 16q with subsequent duplication due to crossing over within the inserted segment. Am J Med Genet. 14:89-96.

Cohen, R., Nabors, L.A., and Pierce, K.A. 1994. Preschoolers' evaluations of physical disabilities: A consideration of attitudes and behavior. J Pediatr Psychol. 19:103-111.

Colacino, S.C., and Pettersen, J.C. 1978. Analysis of the gross anatomical variations found in four cases of trisomy 13. Am J Med Genet. 2:31-50.

College Entrance Examination Board. 1994. SAT Services for Students with Disabilities. Princeton, NJ: Author.

Compas, B.E., Malcarne, V.L., and Fondacaro, K.M. 1988. Coping with stressful events in older children and adolescents. J Consult Clin Psychol. 56:405-411.

Conrad, E.U., and Rang, M. 1986. The evaluation of gait disturbances in children. Paediatric Med. 1:235-240.

Cook, L.C.1936. Amyoplasia congenita associated with mongolism. Arch Dis Child. 11:261-270.

Cote, G.B., Adamopoulos, D., and Panetlakis, S. 1982. Arthrogryposis and ectodermal dysplasia. Hum Hered. 32(1):71-72.

Coverdate, O.R., Cybinski, D.H., and St. George, T.D. 1978. Congenital abnormalities in calves associated with akabane virus and aino virus. Aust Vet J. 54:151-152.

Covert, S.B. 1992. Supporting families. In Natural Supports in School, at Work, and in the Community for People with Severe Disabilities, ed. J. Nisbet, pp. 121-163. Baltimore: Paul H. Brookes.

Cowen, E.L., Pedersen, A., Babigian, H., Izzo, L.D., and Trost, M.A. 1973. Longterm followup of early detected vulnerable children. J Consult Clin Psychol. 41:438-443.

Cox, A.D., and Lambrenos, K. 1992. Childhood physical disability and attachment. Dev Med Child Neur. 34:1037-1046.

Crandell, R.A., Livingston, C.W., Jr., and Shelton, M.J. 1989. Laboratory investigation of a naturally occurring outbreak of arthrogryposis hydranencephaly in Texas sheep. J Vet Diagn Invest. 1(1):62-65.

Crane, J.P., and Heise, R.L. 1981. New syndrome in three affected siblings. Pediatrics. 68(2):235-237.

Crary, E. 1979. Without Spanking or Spoiling: A Practical Approach to Toddler and Preschool Guidance. Seattle: Parenting Press.

Cremer, R., and Kunzer, W. 1988. Arthrogryposis multiplex congenita with associated abnormalities. Case report of a fatal course in a premature triplet. Monatsschr Kinderheilkd. 136(8):464-466.

Currarino, G., and Friedman, J.M. 1986. A severe form of congenital contractural arachnodactyly in two newborn infants. Am J Med Genet. 25:763-773.

Cusi, V., Antich, J., Vela, A., and Vila, J. 1993. Neural tube defect and amniotic band sequence. Genet Couns. 4(3):203-205.

Daher, Y.H., Lonstein, J.E., Winter, R.B., and Moe, J.H. 1985. Spinal deformities in patients with arthrogryposis. A review of 16 patients. Spine. 10(7):609-613.

Dangles, C.J., and Bilos, Z.J. 1981. Surgical correction of thumb deformity in arthrogryposis multiplex congenita. Hand. 13(1):55-58.

Daniels, D., Moos, R.H., Billings, A.G., and Miller, J.J. 1987. Psychosocial risk and resistance factors among children with chronic illness, healthy siblings, and healthy controls. J Abnorm Child Psychol. 15:295-308.

Danks, D.M., Tippett, P., Adams, C., and Campbell, P. 1975. Cerebrohepatorenal syndrome of Zellweger. A report of eight cases with comments upon the incidence, the liver lesion, and a fault in pipecolic acid metabolism. J Pediatrics. 86(3):382-387.

Darrow, D., and Stephens, S. 1992. Interferences in psychosocial development of seriously health impaired and physically disabled children: Educational implications. Acta Paedopsychiatrica. 55:41-44.

Davidson, J., and Beighton, P. 1976. Whence the arthrogrypotics? JBJS. 58B(4):492-495.

Davis, H. 1993. Counselling Parents of Children with Chronic Illness or Disability. Leicester: British Psychological Society (Baltimore: Paul H Brookes, North American distributor).

Davis, J.E., and Kalousek, D.K. 1988. Fetal akinesia deformation sequence in previable fetuses. Am J Med Genet. 29:77-87.

Dawson, G., Grofer, L., Panagiotides, H., Hill, D., and Spieker, S. 1992. Frontal lobe activity and affective behavior of infants of depressed mothers. Child Development. 63:725-737.

De Almeida, J.C.C., Llerena, J.C., Jr., and Alonso, M.R. 1992. C syndrome and omphalocele: Another example. Am J Med Genet. 43:385.

De Koster, J., Legius, E., de Zegher, F., Devlieger, H., and Fryns, J.P. 1990. Opitz C syndrome and pseudohypoaldosteronism. Am J Med Genet. 37:457-459.

De Mattos, J.P., and Martins, W.N. 1982. Neuromedullary amyotrophy of Charcot -Marie-Tooth associated with congenital multiplex arthrogryposis. Report of a case and review of the literature. Arq Neuropsiquiatr. 40(3):281-288.

De Paepe, A., and De Bie, S. 1991. Genetic counseling of a couple presenting respectively terminal transverse defects and congenital arthrogryposis. Genet Couns. 2(4):195-203.

De Smet, L., Legius, E., Fabry, G., and Fryns, J.P. 1993. The Larsen syndrome. The diagnostic contribution of the analysis of the metacarpophalangeal pattern profile. Genet Couns. 4(2):157-164.

De Villemeur, T.B., Beauvais, P., and Richardet, J.M. 1992. Bowen syndrome: Congenital glaucoma, flexion contracture of fingers and facial dysmorphism withour peroxisomal abnormalities. Eur J Pediatr. 151:145-152.

Decker, B. 1992. A comparison of the individualized education plan and the individualized family service plan. Am J Occupational Ther. 46(3):247-252.

Degreif, J., and Rudigier, J. 1987. Distal type I arthrogryposis surgical possibilities of the hand. Handchir Mikrochir Plast Chir. 19(4):226-229.

DelBello, D.A., and Watts, H.G. 1996. Distal femoral extension osteotomy for knee flexion contracture in patients with arthrogryposis. J Pediatr Orthop. 16(1):122-126.

Del Torto, U., Bianchi, O., Pone, G., and Sante, G. 1983. Experimental study on the etiology of congenital multiple arthrogryposis. Ital J Orthop Traumatol. 9(1):91-99.

DeMyer, William, and Baird, I. 1969. Mortality and skeletal malformations from amniocentesis and oligohydramnios in rats: Cleft palate, clubfoot, microstomia, and adactyly. Teratology. 2(1):33-38.

DeNicola, L.K., and Hanshaw, J.B. 1979. Congenital and neonatal varicella. J Pediatr. 94(1):175-176.

Deschavanne, P. J., Diatloff Zito, C., Macieira Coelho, A., and Malaise, E.P. 1981. Unusual sensitivity of two Cockayne's syndrome cell strains to both UV and y irradiation. Mutation Research. 91:403-406.

Desesso, J.M. 1976. Lectin teratogenesis: Defects produced by concanavalin A in fetal rabbits. Teratology. 19:15-26.

Di Rocco, M., and Borrone, C. 1991. Metabolic defect in arthrogryposis multiplex congenita with renal and hepatic abnormalities. J Pediatr. 118(5):828-829.

Di Rocco, M., Callea, R., Pollice, B., Faraci, M., Campiani, F., and Borrone, C. 1995. Arthrogryposis, renal dysfunction and cholestasis syndrome: Report of five patients from three Italian families. Eur J Pediatr 154:835-839.

Di Rocco, M., Erriu, M.I., and Lignana, E. 1991. Distal arthrogryposis, mental retardation, whistling face, and Pierre Robin sequence: Another case. Am J Med Genet. 38(4):557-561.

Di Rocco, M., Reboa, E., Barabino, A., Larnaout, A., Canepa, M., Savioli, C., Cremonte, M., Borrone, C., 1990. Arthrogryposis, cholestatic pigmentary liver disease and renal dysfunction: Report of a second family. Am J Med Genet. 37(2):237-240.

Diab, M., Wu, J.J., Shapiro, F., and Eyre, D. 1994. Abnormality of type IX collagen in a patient with diastrophic dysplasia. Am J Med Genet. 49(4):402-409.

Diamond, L.S., and Alegado, R. 1981. Perinatal fractures in arthrogryposis multiplex congenita. J Pediatr Orthop. 1(2):189-192.

Dias, L.S., and Stern, L.S. 1987. Talectomy in the treatment of resistant talipes equinovarus deformity in myelomeningocele and arthrogryposis. J Pediatr Orthop. 7(1):39-41.

Dieppe, P. 1989. Familial osteoarthrosis and type II collagen gene. Lancet. 17 Jun:1396.

Dietz, F.R. 1985. Dogma disputed on the pathogenesis of clubfoot. Lancet. 15 Feb:388-390.

Dimmick, J.E., Berry, K., MacLeod, P.M., and Hardwick, D.F. 1977. Syndrome of ankylosis, facial anomalies, and pulmonary hypoplasia: A pathologic analysis of one infant. BDOAS. 13(3D):133-137.

Dinkmeyer, D., and McKay, G.D. 1982. The Parent's Handbook: Systematic Training for Effective Parenting. Circle Pines, MN: American Guidance Service.

Dobyns, W.B., Gilbert, E.F., and Opitz, J.M. 1985. Letter to the editor: Further comments on the lissencephaly syndromes. Am J Med Genet. 22:197-211.

Dodinval, P. 1979a. Oligodactyly and multiple synostoses of the extremities: Two cases in sibs. Hum Genet. 48:183-189.

Dodinval, P. 1979b. Facial asymmetries: Problems in genetic counselling. J Genet Hum Belg. 27(3):189-203.

Dodsworth, H. 1992. Chorionic villus sampling and limb abnormalities. Lancet. 4 Apr:339.

Domey, S.F. A., Byrne, W.J., and Ament, M.E. 1986. Case of congenital short small intestine: Survival with use of long term parenteral feeding. Pediatr. 77(3):386-389.

Donnenfeld, A.E., Dunn, L.K., and Rose, N.C. 1985. Discordant amniotic band sequence in monozygotic twins. Am J Med Genet. 20:685-694.

Dorchy, H., Baran, D., and Richard, J. 1976. Association of asymmetric crying facies, malformation of the ear and pulmonary agenesis. Acta Paediatr Belg. 29:255-256.

Dorn, U., Rosenkranz, U., and Bosch, P. 1980. Diastrophic dwarfism. Z Orthop. 118(3):359-366.

Doyle, J.R., James, P.M., Larsen, L.J., and Ashley, R.K. 1980. Restoration of elbow flexion in arthrogryposis multiplex congenita. J Hand Surg. 5(2):149-152.

Drachman, D.B., and Coulombre, A.J. 1962. Experimental clubfoot and arthrogryposis multiplex congenita. Lancet. (2):523.

Drachman, D.B., and Sokoloff, L. 1966. The role of movement in embryonic joint development. Developmental Biology. 14:401-420.

Drotar, D. 1981. Psychological perspectives on chronic illness. J Pediatr Psychol. 6:211-228.

Drummond, D.S., and Cruess, R.L. 1978. The management of the foot and ankle in arthrogryposis multiplex congenita. JBJS. 60B(1):96-99.

Drummond, D.S., and MacKenzie, D.A. 1978. Scoliosis in arthrogryposis multiplex congenita. Spine. 3:146-151.

Drummond, D.S., Siller, T.N., and Cruess, R.L. 1974. Management of arthrogryposis multiplex congenita. A.A.O.S.: Instructional Course Lectures. 23:79-95.

Dubowitz, V., and Platts, M. 1965. Central core disease of muscle with focal wasting. J Neurol, Neurosurg, Psychiat. 28:432-437.

Dubowitz, V., and Roy, S. 1970. Central core disease of muscle: Clinical, histochemical and electron microscopic studies of an affected mother and child. Brain. 93:133-146.

Dubowitz, V., and Sharrard, J. 1968. Congenital clubfoot with central core disease of muscle. Proc Roy Soc Med. 61:1258-1260.

Ealing, M.I. 1944. Amyoplasia congenita causing malpresentation of the foetus. J Obstet & Gynaecol of Brit Emp. 51:144-146.

Ebinger, G., Six, R., Bruyland, M., and Somers, G. 1986. Flexion contractures: A forgotten symptom in Addison's disease and hypopituitarism. Lancet. 11 Oct:858.

Education of the Handicapped Act Amendments of 1986, Public Law 99457.20 USC (Sections 1462-1485).

Edwards, J.F., Livingston, C.W., Chung, S.I., and Collisson, E.C. 1989. Ovine arthrogryposis and central nervous system malformations associated with in utero Cache Valley virus infection: Spontaneous disease. Vet Pathol. 26(1):33-39.

Edwards, M.J. 1986. Hyperthermia as a teratogen: A review of experimental studies and their clinical significance. Terat Carcino Mutat. 6:563-582.

Eegani, S., Shapiro, I., Lewinsky, R., and Sharf, M. 1989. Prenatal ultrasound diagnosis of isolated arthrogryposis of feet. Acta Obstet Gynecol Scand. 68(5):461-462.

Elias, S., Boelen, L., and Simpson, J.L. 1978. Syndromes of camptodactyly, multiple ankylosis, facial anomalies, and pulmonary hypoplasia. BDOAS. 14(6B):243-251.

Emed, A. 1956. Pterygium syndrome. J Pediatr. 48:73-76.

Emery, A.E., and Nelson, M.M. 1970. A familial syndrome of short stature, deformities of the hands and feet, and an unusual facies. J Med Genet. 7:379-382.

Engle, E.C., Marondel, I., Houtman, W.A., de Vries, B., Loewenstein, A., Lazar, M., Ward, D.C., Kucherlapati, R., and Beggs, A.H. 1995. Congenital fibrosis of the extraocular mus cles (autosomal dominant congenital external ophthalmoplegia): Genetic homogeneity, linkage refinement, and physical mapping on chromosome 12. Am J Hum Genet 57:1086-1094.

Epstein, C.J., and Hodgkin, W.E. 1968. Hereditary dysplasia of bone with kyphoscoliosis, contractures, and abnormally shaped ears. J Pediatr. 73(3):379-386.

Epstein, J.B., and Wittenberg, G.J. 1987. Maxillofacial manifestations and management of arthrogryposis: Literature review and case report. J Oral Maxillofac Surg. 45(3):274-279.

Erikson, E.H. 1963. Childhood and Society, Second Ed. New York: W.W. Norton.

Eronen, M., Somer, M., Gustafsson, B., and Holmberg, C. 1985. New syndrome: A digitorenocerebral syndrome. Am J Med Genet. 22:281-285.

Escobar, V., Bixler, D., Gleiser, S., Weaver, D.D., and Gibbs, T. 1978. Multiple pterygium syndrome. Am J Dis Children. 132:609-611.

Escobar, V., and Weaver, D. 1978a. Popliteal pterygium syndrome: A phenotypic and genetic analysis. J Med Genet. 15:35-42.

Escobar, V., and Weaver, D.D. 1978b. The faciogenitopopliteal syndrome. BDOAS. 14(6B):185-192.

Eteson, D.J., Beluffi, G., Burgio, G.R., Belloni, C., Lachman, R.S., and Rimoin, D.L. 1986. Pseudodiastrophic dysplasia: A distinct newborn skeletal dysplasia. J Pediatr. 109(4):635-641.

Eulert, J. 1984. Clinical aspects and treatment of arthrogryposis multiplex congenita. Lower extremity. Z Orthop. 122(5):661-669.

Evans, J., Evans, P., and McGovern, M.A. 1995a. Statistics. In Integrating Students with Special Needs into Mainstream Schools, pp. 33-52. Paris: Organization for Economic Cooperation and Development.

Evans, J., Labon, D., McGovern, M.A. 1995b. Principles and Practice. In Integrating Students with Special Needs into Mainstream Schools, pp. 15-21. Paris: Organization for Economic Cooperation and Development.

Evers, K.G., and Groneck, P. 1983. Asymmetric crying facies: Which is the 'right wrong' side? Pediatr. 71(1):144-145.

Exceptional Parent. 1993a. Finding funding for assistive technology. Exceptional Parent. 23(3):18-28.

Exceptional Parent. 1993b. Future goals application of the goals 2000: Education America act to individuals with disabilities. Exceptional Parent. Sept:25-28.

Fagan, J., and Schor, D. 1993. Mothers of children with spina bifida: Factors related to maternal psychosocial functioning. Am J Orthopsychiat. 63:146-152.

Fahy, M.J., and Hall, J.G. 1990. A retrospective study of pregnancy complications among 828 cases of arthrogryposis. Genet Couns. 1(1):3-11.

Falco, N.A., and Eriksson, E. 1990. Facial nerve palsy in the newborn: Incidence and outcome. Plastic Reconstr Surg. 85:1-2.

Falls, H.F., and Kertesz, E.D. 1964. A new syndrome combining pterygium colli with developmental anomalies of the eyelids and lymphatics of the lower extremities. Trans. Am Ophth Soc. 62:248-279.

Farag, T.I., Teebi, A.S., and Al Awadi, S.A. 1986. Brief clinical Report: Nonsyndromal anencephaly: Possible autosomal recessive variant. Am J Med Genet. 24:461464.

Farrell, K., and McGillivray, B.C. 1983. Arthrogryposis following maternal hypotension. Dev Med Child Neurol. 25(5):648-650.

Fedrizzi, E., Botteon, G., Inverno, M., Ciceri, E., D'Incerti, L., and Dworzak, F. 1993. Neurogenic arthrogryposis multiplex congenita: Clinical and MRI findings. Pediatr Neurol. 9(5):343-348.

Feldman, G.M., Baumer, J.G., and Sparker, R.S. 1982. Brief clinical report: The dup(17p) syndrome. Am J Med Genet. 11:299-304.

Fenichel, G.M., and Bazelon, M. 1966. Mypopathies in search of a name: Benign congenital forms. Dev Med & Child Neurol. 8:532-538.

Fenoll, B., Rigault, P., Maroteaux, P., Padovani, J.P., Guyonvarch, G., and Durand, Y. 1990. Multiple pterygium syndrome in children. 7 cases. Rev Chir Orthop Reparatrice Appar Mot (France). 76(2):102-111.

Fewell, R.R. 1991. Trends in the assessment of infants and toddlers with disabilities. Exceptional Children. 58(2):166-173.

Fidzianska, A., Goebel, H.H., and Burck Lehmann, U. 1989. Myopathic form of arthrogryposis and microcirculation lesion. J Neurol Sci. 92(2):337-348.

Fishbein, J.F., Shadravan, I., Hebert, L., and Funes, R. 1974. Idiopathic bell palsy in a 2-month-old child. Am J Dis Child. 128:112-113.

Fitch, N., and Levy, E.P. 1975. Adducted thumb syndromes. Clin Genet. 8:190-198.

Fitch, N., Rochon, L., Srolovitz, H., and Hamilton, E. 1985. Vascular abnormalities in a fetus with multiple pterygia. Am J Med Genet. 21:755-760.

Fitzsimmons, J.S., Zaldua, V., and Chrispin, R. 1984. Genetic heterogeneity of the Freeman-Sheldon syndrome: Two adults with probable autosomal recessive inheritance. J Med Genet. 21:364-368.

Fleury, P., and Hageman, G. 1985. A dominantly inherited lower motor neuron disorder presenting at birth with associated arthrogryposis. J Neurol Neurosurg Psychiatry. 48(10):1037-1048.

Folkerth, R.D., Guttentag, S.H., Kupsky, W.J., and Kinney, H.C. 1993. Arthrogryposis multiplex congenita with posterior column degeneration and peripheral neuropathy: A case report. Clin Neuropathol. 12(1):25-33.

Forrester, R.M., Lees, V.T., and Watson, G.H. 1966. Rubella syndrome: Escape of a twin. Brit Med J. 1:1403.

Forsyth, C.S., Frank, A.A., Watrous, B.J., and Bohn, A.A. 1994. Effect of coniine on the developing chick embryo. Teratology. 49(4):306-310.

Fowler, S.A., Schwartz, E., and Atwater, J. 1991. Perspectives on the transition from preschool to kindergarten for children with disabilities and their families. Exceptional Children. 58(2):136-145.

Fraccaro, M., Zuffardi, O., Buhler, E., Schinzel, A., Simoni, G., Witkowski, R., Bonifaci, E., and Cavfin, D. 1983. Deficiency, transposition, and duplication of one 15q region may be alternatively associated with Prader-Willi (or a similar) syndrome. Analysis of seven cases after varying ascertainment. Hum Genet. 64:385-394.

Franceschini, P., Vardeu, M.P., Signorile, F., Testa, A., Guala, A., Franceschini, D., and Dalforno, L. 1993. Inguinal hernia and atrial septal defect in Tel Hashomer camptodactyly syndrome: Report of a new case expanding the phenotypic spectrum of the disease. Am J Med Genet. 46:341-344.

Francesco, P., and Nicola, L. 1988. Nosological difference between the Bartsocas-Papas syndrome and lethal multiple pterygium syndrome. Am J Med Genet. 29:699-700.

Frawley, J.M. 1925. Congenital webbing. Am J Dis Child. 29:799-805.

Freeman, E.A., and Sheldon, J.H. 1938. Craniocarpal dystrophy. An undescribed congenital malformation. Arch Dis Child. 13:277.

Fried, K., and Mundel, G. 1976. Absence of distal interphalangeal creases of fingers with flexion limitation. J Med Genet. 13:127-130.

Fried, K., and Mundel, G. 1977. High incidence of spinal muscular atrophy type I (Werdnig-Hoffmann disease) in the Karaite community in Israel. Clin Genet. 12:250-251.

Friedlander, H.L., Westin, G.W., and Wood, W.L. 1968. Arthrogryposis multiplex congenita. A review of 45 cases. JBJS. 50A:89-112.

Friedman, A., Bethzhold, J., Hong, R., Gilbert, E., Visaskuf, C., and Opitz, J.M. 1980. Clinicopathologic conference: A three-month-old infant with failure to thrive, hepatomegaly, and neurological impairment. Amer J Med Gen. 7:171-186.

Friedman, B.D., and Heidenreich, R.A. 1995. Distal arthrogryposis type IIB: Further clinical delineation and 54-year follow-up of an index case. Am J Med Genet 58:125-127.

Friedman, W.F., Mason, D.T., and Braunwald, E. 1965. Arthrogryposis multiplex congenita associated with congenital aortic stenosis. J Pediatr. 67(4):682-685.

Frohlich, G.S., Starzer, K.L., and Tortora, J.M. 1977. Popliteal pterygium syndrome: Report of a family. J Pediatr. 90(1):91-93.

Frostad, H. 1940. Congenital ankylosis of the elbow joint. Acta Orthopaedica. 11:296-305.

Froster, U.G., and Baird, P.A. 1993. Amniotic band sequence and limb defects: Data from a population-based study. Am J Med Genet. 46:497-500.

Froster, U.G., Rehder, H., Hohn, W., and Oberheuser, F. 1993. Craniofacial anomalies, abnormal hair, camptodactyly, and caudal appendage (Teebi-Shaltout syndrome): Clinical and autopsy findings. Am J Med Genet 47:717-722.

Froster-Iskenius, U., Curry, C., Philp, M., and Hall, J.G. 1988. Brief clinical report: An unusual bandlike web in an infant with lethal multiple pterygium syndrome. Am J Med Genet. 30:763-769.

Froster-Iskenius, U.G., Waterson, J.R., and Hall, J.G. 1988. A recessive form of congenital contractures and torticollis associated with malignant hyperthermia. J Med Genet. 25:101-112.

Fryns, J.P., Vandenberghe, K., Moerman, P., and Van den Berghe, H. 1984. Cystic hygroma and multiple pterygium syndrome. Ann Genet. 27(4):252-253.

Fryns, J.P., Volcke, P., and Van Den Berghe, H. 1988. Multiple pterygium syndrome type Escobar in two brothers. Follow up data from childhood to adulthood. Eur J Pediatr. 147:550-552.

Fuhrmann Rieger, A., Kohler, A., and Fuhrmann, W. 1984. Duplication or insertion in 15q113 associated with mental retardation, short stature and obesity: Prader-Willi or Cohen syndrome? Clin Genet. 25:347-352.

Fullagher, P., Crotser, C., Gallagher, J., et al. 1992. Provision of services to handicapped infants and toddlers with developmental delay: The health perspective on resources. Unpublished report. Chapel Hill, NC: Carolina Policy Studies Program.

Furman, W., and Gavin, L. 1989. Peers' influence on adjustment and development: A view from the intervention literature. In Peer Relationships in Child Development, ed. T.J. Berndt and G.W. Ladd, pp. 319-340. New York: Wiley.

Gacek, R.R. 1976. Abductor vocal cord paralysis. Ann Otol. 85:90-93.

Galanski, M., and Statz, A. 1978. Radiological findings in Larsen's syndrome. ROFO Fortschr Geb Rontgenstr Nuklearmed. 128(5):534-537.

Gallegos Rivera, M., Carnevale, A., Valdes, H., and Del Castillo, V. 1991. Congenital multiple arthrogryposis. Clinical and genetic study. Bol Med Hosp Infant Mex. 48(2):88-95.

Gamble, H.J. 1969. Electron microscope observations on the human foetal and embryonic spinal cord. J Anat. 104(3):435-453.

Gandolfi, A., Horoupian, D., Rapin, I., DeTeresa, R., and Hyams, V. 1984. Deafness in Cockayne's syndrome: Morphological, morphometric, and quantitative study of the auditory pathway. Ann Neurol. 15:133-143.

Garland, C.W. 1993. Beyond chronic sorrow: A new understanding of family adaptation. In Cognitive Coping, Families, and Disability, ed. A.P. Turnbull, J.M. Patterson, S.K. Behr, D.L. Murphy, J.M. Marquis, and M.J. Blue-Banning. Baltimore: Paul H. Brookes.

Garrison, W.T., and McQuiston, S. 1989. Chronic Illness During Childhood and Adolescence: Psychological Aspects. Newbury Park: Sage.

Gatrad, A.R. 1981. Congenital dislocation of the knees in a child with Downmosaic Turner syndrome. J Med Genet. 18:148-151.

Gellis, and Feingold 1974. Cerebrohepatorenal syndrome (Zellweger and Bowen syndrome) AmJ Dis Child. 127:873-874.

Gellis, S.S., ed. 1977. Tetracycline for acne? Teratogenic. Pediatric Notes. 1(32):1.

Gericke, G.S. 1991. Fragile collagen and the lethal multiple pterygium syndrome: Does heat stress play a role? Am J Med Genet. 38:630-631.

Gericke, G.S., Hall, J.G., Nelson, M.M., and Beighton, P.H. 1984. Diagnostic considerations in arthrogryposis syndromes in South Africa. Clin Genet. 25(2):155-162.

Gericke, G.S., Van Rensburg, E.J., Mitchell, D., Laburn, H., and Isaacs, H. 1991. Rejoinder by Dr. Gericke to Dr. Hartwig and coworkers. Am J Med Genet. 38:633.

Ghetti, B., Amati, A., Turra, M.V., Pacini, A., Del Vecchio, M., and Guazzi, G.C. 1971. Werdnig-Hoffmann Wohlfart Kugelberg Welander Disease. Nosological unity and clinical variability in intrafamilial cases. Acta Geneticae Medicae et Gemeliologiae. 20:43-54.

Gibson, D.A., and Urs, N.D.K. 1970. Arthrogryposis multiplex congenita. JBJS. 52B:483.

Gilchrist, K.W., Gilbert, E.F., Shahidi, N.T., and Opitz, J.M. 1975. The evaluation of infants with the Zellweger (cerebrohepatorenal) syndrome. Clin Genet. 7:413-416.

Gill, I.B., Gupta, N.P., and Oberoi, G.S. 1987. Genitourinary anomalies in arthrogryposis multiplex congenita. Br J Urol. 60(3):276-278.

Gillin, M.E., and Pryse Davis, J. 1976. Pterygium syndrome. J Med Genet. 13(3):249-251.

Gilmour, J.R. 1946. Amyoplasia congenita. J Pathol Bacteriol. 58:675-685.

Godbersen, S., Heckel, V., and Wiedemann, H.R. 1987. Brief clinical report: Pterygium colli medianum and midline cervical cleft: Midline anomalies in the sense of a developmental field defect. Am J Med Genet. 27:719-723.

Goecke, T., Majewski, F., Kauther, K.D., and Sterzel, U. 1982. Mental retardation, hypotonia, obesity, ocular, facial, dental, and limb abnormalities (Cohen syndrome). Report of three patients. Eur J Pediatr. 138:338-340.

Goff, C.W., Cercielio, R., and Holmes, G.L. 1983. Bilateral Bell's palsy. Am J Dis Child. 137:83.

Goldberg, J.D., Chervenak, F.A., Lipman, R.A., and Berkowitz, R.L. 1986. Antenatal sonographic diagnosis of arthrogryposis multiplex congenita. Prenat Diagn. 6(1):45-49.

Golden, N.L., Bilenker, R., Johnson, W.E., and Tischfield, J.A. 1981. Abnormality of chromosome 16 and its phenotypic expression. Clin Genet. 19:41-45.

Gollop, T., Dal Colletto, G.M., and Ferraretto, I. 1982. New manifestations observed in the Tel Hashomer camptodactyly syndrome. Skeletal Dysplasias. 269-277.

Gollop, T.R., and Eigier, A. 1987. Prenatal ultrasound diagnosis of diastrophic dysplasia at 16 weeks. Am J Med Genet. 27(2):321-324.

Goodlin, R.C., and Lowe, E.W. 1974. Unexplained hydramnios associated with a thanatophoric dwarf. Am J Obstet Gynecol. 118(6):873-875.

Goodman, R.M., Katznelson, M.B.M., Hertz, M., and Katznelson, A. 1976. Camptodactyly, with muscular hypoplasia, skeletal dysplasia, and abnormal palmar creases: Tel Hashomer camptodactyly syndrome. J Med Genet. 13:136-141.

Goodman, R.M., Katznelson, M.B.M., and Manor, E. 1972. Camptodactyly: Occurrence in two new genetic syndromes and its relationship to other syndromes. J Med Genet. 9(2):203-212.

Gorczyca, D.P., McGahan, J.P., Lindfors, K.K., Ellis, W.G., and Grix, A. 1989. Arthrogryposis multiplex congenita: Prenatal ultrasonographic diagnosis. JCU J Clin Ultrasound. 17(1):40-44.

Gorlin, R.J., and Sedano, H. 1973. Cerebrohepatorenal syndrome. Modern Medicine. 11 Jun:88-89.

Gorlin, R.J., Sedano, H.O., and Cervenka, J. 1968. Popliteal pterygium syndrome. A syndrome comprising cleft lip palate, popliteal and intercrural pterygia, digital and genital anomalies. Pediatrics. 41(2):503-509.

Govaerts, L., Monnens, L., Tegelaers, W., Trijbels, F., and Van RaaySelten, A. 1982. Cerebrohepatorenal syndrome of Zellweger: Clinical symptoms and relevant laboratory findings in 16 patients. Eur J Pediatr. 139:125-128.

Graham, J.M. Jr., Hoehn, H., Lin, M.S., and Smith, D.W. 1981. Diploid tripolid mixoploidy: Clinical and cytogenetic aspects. Pediatr. 68(1):23-28.

Grant, A.D., Rose, D., and Lehman, W. 1982. Talocalcaneal coalition in arthrogryposis multiplex congenita. Bull Hosp Jt Dis Orthop Inst. 42(2):236-241.

Green, A.D., Fixsen, J.A., and Lloyd Roberts, G.C. 1984. Talectomy for arthrogryposis multiplex congenita. JBJS. 66(5):697-699.

Greenough, A., Blott, M., Nicolaides, K., and Campbell, S. 1988. Interpretation of fetal breathing movements in oligohydramnios due to membrane rupture. Lancet. 23 Jan:183.

Gregory, D., Kaplan, P., and Scriver, C.R. 1984. Genetic causes of chronic musculoskeletal disease in childhood are common. Am J Med Genet. 19:533-538.

Gresham, F.M. 1986. Best practices in social skills training. In Best Practices in School Psychology, ed. A. Thomas and S. Grimes. Kent, OH: National Association of School Psychologists.

Grgic, A., Rosenbloom, A.L., Weber, F.T., and Giordano, B. 1975. Joint contracture in childhood diabetes. New Engl J Med. 13 Feb:372.

Grill, F. 1990. The hip joint in arthrogryposis. Z Orthop Ihre Grenzgeb. 128(4):384-390.

Gross, R.H. 1985. The role of the Verebelyi-Ogston procedure in the management of the arthrogrypotic foot. Clin Orthop. (194):99-103.

Gruber, M.A., Graham, T.P., Engel, E., and Smith, C. 1978. Marfan syndrome with contractural arachnodactyly and severe mitral regurgitation in a premature infant. J Pediatr. 93(1):80-82.

Gruel, C.R., Birth, J.G., Roach, J.W., and Herring, J.A. 1986. Teratologic dislocation of the hip. J Pediatr Orth. 6:693.

Guarniero, R., Montenegro, N.B., Luzo, C.A., Corsato, M., Lage, L.A., and Peixinho, M. 1991. Evaluation of treatment of the hip in arthrogryposis multiplex congenita. Rev Hosp Clin Fac Med Sao Paulo. 46(6):271-275.

Gucker, T. 1967. Muscular defects. Pediatr Clin N Am. 14(2):439-460.

Guha Ray, D.K., and Hamblin, M.H. 1977. Arthrogryposis multiplex congenita in an abdominal pregnancy. J of Reproductive Med. 18(2):109-112.

Guidera, K.J., and Drennan, J.C. 1985. Foot and ankle deformities in arthrogryposis multiplex congenita. Clin Orthop. (194):93-98.

Guidera, K.J., Raney, E., Ogden, J.A., Highhouse, M., and Habal, M. 1991. Caudal regression: A review of seven cases, including the mermaid syndrome. J Pediatr Orthop. 11(6):743-747.

Gullino, E., Abrate, M., Zerbino, E., Bricchi, G., and Rattazzi, P.D. 1993. Early prenatal sonographic diagnosis of neuropathic arthrogryposis multiplex congenita with osseous heterotopia. Prenat Diagn. 13(5):411-416.

Gupta, A., Hall, C.M., Ransley, Y.F., and Murday, V.A. 1995. A new autosomal recessive syndrome of characteristic facies, joint contractures, skeletal abnormalities, and normal development: Second report with further clinical delineation. J Med Genet 32:809-812.

Gustafsson, J., Gustavson, K.H., Karlaganis, G., and Sjovall, J. 1983. Zellweger's cerebrohepatorenal syndrome variations in expressivity and in defects of bile acid synthesis. Clin Genet. 24:313-319.

Guthrie, R.H., and Goullan, D. 1974. Congenital band of the abdomen and the amniotic etiology of bands. Am J Surg. 127:753-754.

Haaf, T., Hofmann, R., and Schmid, M. 1991. Opitz trigonocephaly syndrome. Am J Med Genet. 40:444-446.

Hageman, G., Gooskens, R.H., and Willemse, J. 1985. A cerebral cause of arthrogryposis: Unilateral cerebral hypoplasia. Clin Neurol Neurosurg. 87(2):119-122.

Hageman, G., Ippei, E.P.F., Beemer, F.A., de Pater, J.M., Lindhout, D., and Willemse, J. 1988. The diagnostic management of newborns with congenial contractures: A nosologic study of 75 cases. J Med Genet. 30:883-904.

Hageman, G., Jennekens, F.G., Vette, J.K., and Willemse, J. 1984. The heterogeneity of distal arthrogryposis. Brain Dev. 6(3):273-283.

Hageman, G., Vette, J.K., and Willemse, J. 1983. A case of asymmetrical arthrogryposis: A clinical study and a preliminary report on the value of CT scanning. Brain Dev. 5(4):407-413.

Hageman, G., and Willemse, J. 1983. Arthrogryposis multiplex congenita: Review with comment. Neuropediatrics. 14(1):6-11.

Hahn, G. 1985. Arthrogryposis: Pediatric review and habilitative aspects. Clin Orthop. 194:104-114.

Hain, D., Leversha, M., Campbell, A.D., Barr, P.A., and Rogers, J.G. 1980. The ascertainment and implications of an unbalanced translocation in the neonate. Familial 1:15 translocation. Aust Paediatr J. 16:196-200.

Hajra, A.K., Datta, N.S., Jaunson, L.G., Moser, A.B., Moser, H.W., Larben J.W., Jr., and Powers, J. 1985. Prenatal diagnosis of Zellweger cerebrohepatorenal syndrome. N Engl J Med. 312(7):445-446.

Halal, F., and Fraser, F.C. 1979. Camptodactyly, cleft palate, and club foot (the Gordon syndrome): A report of a large pedigree. J Med Genet. 16:149-150.

Halberstadt, A. 1986. Family socialization of emotional expression and nonverbal communication styles and skills. J Personality Social Psychol. 51:827-836.

Hale, M.S., Rodman, H.D., and Lipshin, J. 1974. Congenital contractural arachnodactyly. West J Med. 120:74-76.

Hall, J.G. 1981. An approach to congenital contractures (arthrogryposis). Pediatr Ann. 10(7):15-26.

Hall, J.G. 1984a. Editoral comment: The lethal multiple pterygium syndromes. Am J Med Genet. 17:803-807.

Hall, J.G. 1984b. An approach to research on congenital contractures. Birth Defects. 20(6):8-30.

Hall, J.G. 1984c. Craniofacial development in arthrogryposis (congenital contractures). Birth Defects. 20(3):99-111.

Hall, J.G. 1985a. Genetic aspects of arthrogryposis. Clin Orthop. (194):44-53.

Hall, J.G. 1985b. In utero movement and use of limbs are necessary for normal growth: A study of individuals with arthrogryposis. Prog Clin Biol Res. 155-162.

Hall, J.G. 1986a. Analysis of Pena Shokeir phenotype. Am J Med Genet. 25:99-117.

Hall, J.G. 1986b. Diagnostic approaches and prognosis in arthrogryposis (congenital contractures). Pathologica. 78(1058):701-708.

Hall, J.G. 1988. Comments on "Amyoplasia congenita-like condition and maternal malathion exposure": Is all amyoplasia amyoplasia? Teratology. 38:493-494.

Hall, J.G. 1989. Arthrogryposis. Am Fam Physician. 39(1):113-119.

Hall, J.G. 1995. Arthrogryposis. In Principles and Practice of Medical Genetics, ed. A.E.H. Emery and D.L. Rimoin. Edinburgh: Churchill & Livingstone.

Hall, J.G. 1996. Arthrogryposis associated with unsuccessful attempts at termination of pregnancy. Am J Med Genet. 63:293-300.

Hall, J.G., and Reed, S.D. 1982. Teratogens associated with congenital contractures in humans and in animals. Teratology. 25(2):173-191.

Hall, J.G., Reed, S.D., and Driscoll, E.P. 1983a. Amyoplasia: A common sporadic condition with congenital contractures. Am J Med Genet. 15:571-590.

Hall, J.G., Reed, S.D., and Greene, G. 1982a. The distal arthrogryposis: Delineation of new entities: Review and nosologic discussion. Am J Med Genet. 11(2):185-239.

Hall, J.G., Reed, S.D., McGillivray, B.C., Herrmann, J., Partington, M.W., Schinzel, A., Sharpiro, J., Weaver, D.D., 1983b. Part II. Amyoplasia: Twinning in amyoplasia. A specific type of arthrogryposis with an apparent excess of discordantly affected identical twins. Am J Med Genet. 15(4):591-599.

Hall, J.G., Reed, S.D., Rosenbaum, K.N., Gershanik, J., Chen, H., and Wilson, K.M. 1982b. Limb pterygium syndromes: A review and report of eleven patients. Am J Med Genet. 12(4):337-409.

Hall, J. G., Reed, S.D., Scott, C.I., Rogers, J.G., Jones, K.L., and Camarano, A. 1982c. Three distinct types of X-linked arthrogryposis seen in 6 families. Clin Genet. 21(2):81-97.

Hall, K.W., and Hammock, M. 1979. Feeding and toileting devices for a child with arthrogryposis. Am J Occupational Ther. 33(10):644-647.

Hallahan, D.P., and Kauffman, J.M. 1991. Exceptional Children: Introduction to Special Education, 5th ed. Englewood Cliffs, NJ: Prentice-Hall.

Hamlett, K. W., Pellegrini, D.S., and Katz, K.S. 1992. Childhood chronic illness as a family stressor. J Pediatric Psychol. 17:33-48.

Hanson, P.A., Martinez, L.B., and Cassidy, R. 1977. Contractures, continuous muscle discharges, and titubation. Ann Neurol. 1:120-124

Hanson, R.F., Szczepanik VanLeeuwen, P., Williams, G.C., Grabowski, G., and Sharp, H.L. 1979. Defects of bile acid synthesis in Zellweger's syndrome. Science. 203:1107-1108.

Hansson, L.I., Hansson, V., and Jonsson, K. 1976. Popliteal pterygium syndrome in a 74-year-old woman. Acta Orthop Scand. 47(5):525-533.

Hansson, O., Kristensson, K., Lycke, E., Solymar, L., and Sourander, P. 1975. Case report: Generalized myopathy and cerebral malformations possibly related to an enteroviral infection. Acta Paediatr Scand. 64:881-885.

Happle, R. 1981. Cataracts as a marker of genetic heterogeneity in chondrodysplasia punctata. Clin Genet. 29:64-66.

Happle, R., Stekhoven, J.H.S., Hamel, B.C.J. 1992. Restructive dermopathy in two brothers. Arch Dermatology 128:232-235.

Hariga, J., Lowenthal, A., and Guazzi, G.C. 1963. Nosological place and correlations of arthrogryposis (sensu stricto). Acta Neurol Belg. 63:766-793.

Haring, N., and McCormick, L. 1990. Exceptional Children and Youths: An Introduction to Special Education, 5th ed. Columbus, OH: Merrill Publishing Co.

Harper, D.C. 1991a. Psychosocial aspects of physical differences in children and youth. In Pediatric Rehabilitation, ed. K. Jaffe. Philadelphia: W.B. Saunders. Physical Med Rehabili Cli N Amer. 2(4):765-779.

Harper, D.C. 1991b. Paradigms for investigating rehabilitation and adaptation to childhood disability and chronic illness. J Pediatr Psychol. 16:533-542.

Harper, D.C., Wacker, D.P., and Cobb, L.S. 1986. Children's social preferences toward peers with visible physical differences. J Pediatr Psychol. 11(3):323-342.

Harris, E.D., Jr. 1990. Rheumatoid arthritis pathophysiology and implications for therapy. New Engl J Med. 322(18):1277-1289.

Harrod, M.J.E., and Sherrod, P.S. 1981. Warfarin embryopathy in siblings. Obstet & Gynecol. 57:673-676.

Hartwig, N.G., Vermeij Keers, Chr., and Bruijn, J.A. 1991. Reply to Dr. Gericke. Am J Med Genet. 38:632.

Hartwig, N.G., Vermeij Keers, Chr., Bruijn, J.A., van Groningen, K., Ottervanger, H.P., and Holm, J.P. 1989. Case of lethal multiple pterygium syndrome with special reference to the origin of pterygia. Am J Med Genet. 33:537-541.

Hasazi, S.B., Gordon, L.R., and Roe, C.A. 1985. Factors associated with the employment status of handicapped youths exiting high school from 1979-1983. Exceptional Children. 5:469.

Haselwood, D.M., and Castles, J.J. 1977. The biology of the rheumatioid synovial cell. Western J Med. 137(3):204-213.

Hauptman, A., and Thannhauser, S.J. 1941. Muscular shortening and dystrophy: A heredofamilial disease. Arch Neurol Psych. 46:645-664.

Hays, R. 1987. Childhood motor impairments: Clinical overview and scope of the problem. In Childhood Powered Mobility: Developmental, Technical and Clinical Perspectives, ed. K.M. Jaffee, p. 1. Washington, DC: RESNA.

Hecht, F. 1981. Uncommon children and common care. J Pediatr. 98(4):594-595.

Hecht, F., and Beals, R.K. 1971. "New" syndrome of congenital contractural arachnodactyly originally described by Marfan in 1896. Pediatr. 40:574-579.

Heffez, L., Doku, H.C., and O'Donnell, J.P. 1985. Arthrogryposis multiplex complex involving the temporomandibular joint. J Oral Maxillofac Surg. 43(7):539-542.

Hegarty, S. 1995. Teacher training. In Integrating Students with Special Needs Into Mainstream Schools, pp. 59-67. Organization for Economic Cooperation and Development.

Hennekam, R.C., Barth, P.G., Van Lookeren Campagne, W., De Visser, M., and Dingemans, K.P. 1991. A family with severe X-linked arthrogryposis. Eur J Pediatr. 150(9):656-660.

Hennekam, R.C.M., Befmer, F.A., Huijbers, W.A.R., Hustinx, P.A., and Van Sprang, F.J. 1985. The cerebrocostomandibular syndrome: Third report of familial occurrence. Clin Genet. 28:118-121.

Hensinger, R.N., and Jones, E.T. 1981. Arthrogryposis. Neonatal Orthopaedics, pp. 110-115. New York: Grune & Stratton.

Herbert, W. N., Seeds, J.W., Cefalo, R.C., and Bowes, W.A. 1985. Prenatal detection of intraamniotic bands: Implications and management. Obstet Gynecol. 65(3(supplement)):36S-38S.

Herring, J.A., and Banta, J.V. 1988. Arthrogryposis. J Pediatr Orthop. 8(3):353-355.

Herrmann, J., and Opitz, J.M. 1979. Syndrome delineation 2. Inborn errors of metabolism, deformities, and variant familial developmental patterns. Postgraduate Med. 65(2):231-237.

Herron, L.D., Westin, G.W., and Sawson, E.G. 1978. Scoliosis in arthrogryposis multiplex congenita. JBJS. 60A:293-299.

Herva, R., Conradi, N.G., Kallmo, H., Leisti, J., and Sourander, P. 1988. A syndrome of multiple congenital contractures: Neuropathological analysis on five fetal cases. Am J Med Genet. 29:67-76.

Heselson, N.G., Cremin, B.J., and Beighton, P. 1978. Lethal chondrodysplasia punctata. Clin Radiol. 29:679-684.

Heymans, H.S.A., Oorthuys, J.W.E., Nelck, G., Wanders, R.J.A., and Schutgens, R.B.H. 1985. Rhizomelic chondrodysplasia punctata: Another peroxisomal disorder. N Engl J Med. 313(3):187-188.

Higginbottom, M.C., Jones, K.L., Hall, B.D., and Smith, D.W. 1979. The amniotic band disruption complex: Timing of amniotic rupture and variable spectra of consequent defects. J Pediatr. 95(4):544-549.

Hittner, H.M., Kretzer, F.L., and Mehta, R.S. 1981. Zellweger syndrome: Lenticular opacities indicating carrier status and lens abnormalities. Characteristic of homozygotes. Arch Ophthalmol. 99:1977-1982.

Ho, N., and Kboo, T. 1979. Congenital contractural arachnodactyly. Report of a neonate with advanced bone age. Am J Dis Child. 133:639-640.

Hoar, D.I., and Waghorne, C. 1978. DNA repair in Cockayne syndrome. Am J Hum Genet. 30:590-601.

Hodgson, P., Weinberg, S., and Consky, C. 1988. Arthrogryposis multiplex congenita of the temporomandibular joint. Oral Surg Oral Med Oral Pathol. 65(3):289-291.

Hoefnage, D., and Penry, Capt.J.K. 1966. Partial facial paralysis in young children. New Engl J Med. 262(22):1126-1128.

Hoffer, M.M., Swank, S., Eastman, F., Clark, D., and Teitge, R. 1983. Ambulation in severe arthrogryposis. J Pediatr Orthop. 3(3):293-296.

Hogge, W.A., Golabi, M., Filly, R.A., Douglas, R., and Golbus, M.S. 1985. The lethal multiple pterygium syndromes. Is prenatal diagnosis possible?. Am J Med Genet. 20:441-442.

Holbrook, K.A., Dale, B.A., Witt, D.R., Hayden, M.R., and Toriello, H.V. 1987. Arrested epidermal morphogenesis in three newborn infants with a fatal genetic disorder (restrictive dermopathy). J Invest Dermatol. 88(3):330-339.

Honig, P.J., Yoder, M., and Ziegler, M. 1983. Acquired pyloric obstruction in a patient with epidermolysis bullosa letalis. J Pediatr. 102(4):596-597.

Hopkins, P.M., Ellis, F.R., and Halsall, P.J. 1991. Hypermetabolism in arthrogryposis multiplex congenita. Anaesthesia. 46(5):375-375.

Hopper, C.E., and Allen, W.A. 1980. Sex Education for Physically Handicapped Youth. Springfield, IL: Charles C Thomas.

Horan, F., and Beighton, P. 1976. Parastremmatic dwarfism. JBJS. 58(3):343-346.

Horoupian, D.S., and Yoon, J.J. 1988. Neuropathic arthrogryposis multiplex congenita and intrauterine ischemia of anterior horn cells: A hypothesis. Clin Neuropathol. 7(6):285-293.

Horslen, S.P., Quarrell, O.W., and Tanner, M.S. 1994. Liver histology in the arthrogryposis multiplex congenita, renal dysfunction, and cholestasis (ARC) syndrome: Report of three new cases and review. J Med Genet. 31(1):62-64.

Houston, C.S., and Chudley, A.E. 1981. Separating monosomy 21 from the "arthrogryposis basket." J Can Assoc Radiol. 32(4):220-223.

Houston, C.S., Reed, M.H., and Desautels, J.E.L. 1981. Separating Larsen syndrome from the "arthrogryposis basket." J Can Assoc Radiol. 32(4):206-214.

Houston, C.S., and Shokeir, M.H.K. 1981. Separating Pena Shokeir I syndrome from the "arthrogryposis basket." J Can Assoc Radiol. 32(4):215-219.

Houston, C.S., Zaleski, W.A., and Rozdilsky, B. 1982. Identical male twins and brother with Cockayne syndrome. Am J Med Genet. 13:211-223.

Howard, R. 1907. A case of congenital defect of the muscular system (dystrophia muscularis congenita) and its association with congenital talipes equinovarus. Proc Roy Soc Med (Lond). 1(3):157-166.

Hsu, L.C., Jaffray, D., and Leong, J.C. 1984. Talectomy for club foot in arthrogryposis. JBJS. 66(5):694-696.

Hughes, H.E., Harwood Nash, D.C., and Becker, L.E. 1983. Craniotelencephalic dysplasia in sisters: Further delineation of a possible syndrome. Am J Med Genet. 14:557-565.

Hull, R., and Pope, F.M. 1989. Osteoarthritis and cartilage collagen genes. Lancet. 10 Jun:1337-1338.

Hunter, A. 1990. The popliteal pterygium syndrome: Report of a new family and review of the literature. Am J Med Genet. 36:196-208.

Hunter, A.G.W., Cox, D.W., and Rudd, N.L. 1976. The genetics of and associated clinical findings in humeroradial synostosis. Clin Genet. 9:470-478.

Hunter, A.G.W., Woerner, S.J., Montalvo Hicks, L.D.C., Fowlow, S.B., Haslam, R.H.A., Metcalf, P.J., and Lowry, R.B. 1979. The Bowen-Conradi syndrome. A highly lethal autosomal recessive syndrome of microcephaly, micrognathia, low birth weight and joint deformities. Am J Med Genet. 3:269-279.

Huurman, W.W., and Jacobsen, S.T. 1985. The hip in arthrogryposis multiplex congenita. Clin Orthop. (194):81-86.

Ianasescu, V., Zellweger, H., Filer, L.J., and Conway, T.W. 1970. Increased collagen synthesis in arthrogryposis multiple congenita. Arch Neurol. 23:128.

Illum, N., Reske Nielsen, E., Skovby, F., Askjaer, S.A., and Bernsen, A. 1988. Lethal autosomal recessive arthrogryposis multiplex congenita with whistling face and calcifications of the nervous system. Neuropediatrics. 19(4):186-192.

Imamura, M., Yamanake, N., Nakamura, F., and Oyanagi, K. 1981. Arthrogryposis multiplex congenita: An autopsy case of a fatal form. Hum Pathol. 12(8):699-704.

Individualized Education Programs (IEP). May, 1980. Federal Register, Washington, DC: U.S. Education Department, Assistant Secretary for Special Education and Rehabilitation Services, Office of Special of Special Education.
Ioan, D.M., Belengeanu, B., Maximilian, C., and Fryns, J.P. 1993. Distal arthrogryposis with autosomal dominant inheritance and reduced penetrance in females: The Gordon syndrome. Clin Genet (Denmark). 43(6):300-302.

Ippolito, E., and Ponseti, I.B. 1980. Congenital club foot in the human fetus. JBJS. 62A(1):8-22.

Ireys, H.T., Werthamer Larsson, L.A., Kolodner, K.B., and Gross, S.S. 1994. Mental health of young adults with chronic illness: The mediating effect of perceived impact. J Pediatr Psychol. 19:205-222.

Itagaki, Y., Yoshioka, M., Sakamoto, Y., Nishitani, H., and Haebara, H. 1982. An autopsy case of severe congenital muscular dystrophy with arthrogryposis multiplex. Rinsho Shinkeigaku. 22(10):896-900.

Ito, M., Onitsuka, Y., Matsui, K., Fuisaki, S., and Mafyama, M. 1986. Craniofacial defects associated with amniotic band syndrome: A case report. Int J Gynaecol Obstet. 24:43-45.

Iukina, G.P., and Mikhailova, L.K. 1990. Diagnosis and treatment of diastrophic dysplasia and Larsen's syndrome in the 1st years of life. Ortop Travmatol Protez. (9):56-60.

Izumi, A.K., and Arnold, H.L. 1974. Congenital annular bands (pseudoainhum) association with other congenital abnormalities. JAMA. 229(9):1208-1209.

Jacobson, J.L., and Wille, D.E. 1986. The influence of attachment pattern on developmental changes in peer interaction from the toddler to the preschool period. Child Development. 57:338-347.

Jago, R.H. 1970. Arthrogryposis following treatment of maternal tetanus with muscle relaxants. Arch Dis Child. 45:277.

Jakobs, C., Dorland, L., Sweetman, L., Duran, M., Nyhan, W., and Wadman, S. 1984. Identification of methyl-branched chain dicarboxylic acids in amniotic fluid and urine in propionic and methylmalonic acidemia. Pediatr Res. 18(11):1185-1191.

James, J.I.P. 1969. The relationship of Dupuytren's contracture and epilepsy. Hand. 1:4749.

Jan, J.E., Hardwick, D.F., Lowry, R.B., and McCormick, A.Q. 1970. Cerebrohepatorenal syndrome of Zellweger. Amer J Dis Child. 119:274-277.

Jan Pijl, S., and Meijer, C.J.W. 1994. Introduction. In New Perspectives in Special Education, ed. C.J.W. Jeijer, S.J. Pijl, and S. Hegarty, pp. xi-xiv. London & New York: Routledge.

Jennings, M., Hall, J.G., and Hoehn, H. 1980. Significance of phenotypic and chromosomal abnormalities in X-Linked mental retardation (Martin Bell or Renpenning syndrome). Am J Med Genet. 7:417-432.

Jequier, S., and Kozlowski, K. 1987. Unusual facies, arthrogryposis, advanced skeletal maturation and unique bone changes. A new congenital malformation syndrome. Pediatr Radiol. 17(5):405-408.

Johnson, C., and Yngve, D.A. 1988. Answer please. Diastrophic dwarfism. Orthopedics. 11(10):1501-1502.

Johnson, R.T. 1970. Viruses and chronic neurological diseases. BDOAS. 7(6):9.

Johnson, R.T. 1972. Effects of viral infection on the developing nervous system. N Eng J Med. 287(12):599-604.

Johnson, R.T. 1982. Current concepts in neurology contribution of virologic research to clinical neurology. N Eng J Med. 307(11):660-662.

Johnston, K., Aarons, R., Schelley, S., and Horoupian, D. 1993. Joint contractures, hyperkeratosis, and severe hypoplasia of the posterior columns: A new lethal recessive syndrome. Am J Med Genet. 47(2):246-249.

Johnston, K., Curry, C.J.R., and Holbrook, K.A. 1990. Joint contractures and abnormal skin: Two new cases. Smith Workshop. 97-98.

Jones, E.E., Farina, A., Hastorf, A.H., Markus, H., Miller, D.T., and Scott, R.A. 1984. Social Stigma: The Psychology of Marked Relationships. New York: W.H. Freeman.

Jones, K.L., Smith, D.W., Hall, B.K., Hall, J.G., Ebbin, A.J., Massoud, H., and Golbus, M.S. 1974. A pattern of craniofacial and limb defects secondary to aberrant tissue bands. J Pediatr. 84(1):90-95.

Jones, R., and Dolcourt, J.L. 1992. Muscle rigidity following halothane anesthesia in two patients with Freeman-Sheldon syndrome. Anesthesiology. 77(3):599-600.

Juabeh, I.I., Thalji, A., and Dudin, A. 1987. Meckel-Gruber syndrome in one of nonidentical twins: Short case report. Acta Genet Med Gemellol. 36:571-572.

Juberg, R.C., and Touchstone, W.J. 1974. Congenital metatarsus varus in four generations. Clin Genet. 5:127-132.

Kaback, M.M., and O'Brien, J.S. 1973. Tay-Sachs: Prototype for prevention of genetic disease. Hospital Practice. March:107-116.

Kaffe, S., Hsu, L.Y.F., Sachdev, R.K., Philips, J., and Hirschhorn, K. 1977. Partial deletion of long arm of chromosome 11: del (11) (q23). Clin Genet. 12:323-328.

Kalousek, D.K., and Bamforth, S. 1988. Amnion rupture sequence in previable fetuses. Am J Med Genet. 31:6373.

Kaltenbach, G., Malherbe, V., Sari-Letet, M.L., and Kahn, M.F. 1991. The outcome at the adult age of arthrogryposis. Apropos of a case. Review of the literature. Rev Rhum Mal Osteoartic. 58(3):215-217.

Kalyanaraman, K., and Kalyanaraman, U.P. 1982. Myopathic arthrogryposis with seizures and abnormal electroencephalogram. J Pediatr. 100(2):247-250.

Kamieniecka, Z. 1973. Late onset myopathy with rod-like particles. Acta Neurol Scand. 49:547-551.

Kamil, N.I., and Correia, A.M. 1990. A dynamic elbow flexion splint for an infant with arthrogryposis. Am J Occup Ther. 44(5):460-461.

Karpati, G., Carpenter, S., and Nelson, R.F. 1970. Type I muscle fibre atrophy and central nuclei: A rare familial neuromuscular disease. J Neurol Sci. 10:489-500.

Kasai, T., Oki, T., Osuga, T., and Nogami, H. 1982. Familial arthrogryposis with distal involvement of the limbs. Clin Orthop. (166):182-184.

Kase, B.F., Bjorkhem, I., Haga, P., and Pedersen, J.I. 1985. Defective peroxisomal cleavage of the C27-steroid side chain in the cerebrohepatorenal syndrome of Zellweger. J Clin Invest. 75:427-435.

Katz, J.F. 1980. Teratological hip dislocation. Isr J Med Sci. 16:238-244.

Kawira, E.L., and Bender, H.A. 1985. An unusual distal arthrogryposis. Am J Med Genet. 20(3):425-429.

Kazak, A. 1989. Families of chronically ill children: A systems and social-ecological model of adaptation and challenge. J Consulting Clin Psychol. 57:25-30.

Keeler, R.F. 1981. Absence of arthrogryposis in newborn Hampshire pigs from sows ingesting toxic levels of jimsonweed during gestation. Vet Hum Toxicol. 23(6):416-415.

Keller, H., Neuhauser, G., Durkin Stamm, M.V., Kaveggia, E.G., Schaaff, A., and Sitzmann, F. 1978. "Adam complex" (amniotic deformity, adhesions, mutilations): A pattern of craniofacial and limb defects. Am J Med Genet. 2:81-98.

Kelley, R.I. 1983. Review: The cerebrohepatorenal syndrome of Zellweger, morphologic and metabolic aspects. Amer J Med Gen. 16:503-517.

Keppen, L.D., Husain, M.M., and Woody, R.C. 1987. X-linked myotubular myopathy: intrafamilial variability and normal muscle biopsy in a heterozygous female. Clin Genet. 32:95-99.

Khajavi, A., Lachman, R.S., Rimoin, D.L., Schimke, R.N., Dorst, J.P., Ebbin, A.J., Handmaker, S., Perreault, G., 1976. Heterogeneity in the campomelic syndromes: Long and short bone varieties. BDOAS. 12(6):93-100.

Khalifa, M.M., and Graham, G. 1994. New dominant syndrome of pterygium colli, mental retardation and digital anomalies. Am J Med Genet 52:55-57.

Kilbridge, H.W., Thibeault, D.W., Yeast, J., Maulik, D., and Grundy, H.O. 1988. Fetal breathing is not a predictor of pulmonary hypoplasia in pregnancies complicated by oligohydramnios. Lancet. 6 Feb:305-306.

Kinoshita, M., Satoyoshi, F., and Kumagai, M. 1975. Familial type I fiber atrophy. J Neurol Sci. 25:11-17.

Kite, J.H. 1955. Arthrogryposis multiplex congenitas: Review of fifty-four cases. Southern Med J. Nov:1141-1146.

Kite, J.H. 1967. Congenital metatarsus varus. JBJS. 49A(2):388-397.

Kleijer, W.J., Thoomes, R., Galjaard, H., Wendel, U., and Fowler, B. 1984. First-trimester (chorion biopsy) diagnosis of citrullinaemia and methylmalonicaciduria. Lancet. 8 Dec:1340.

Klenerman, L. 1987. Club foot. Arch Dis Child. 62:112-113.

Knobloch, W.H., and Layer, J.M. 1971. Retinal detachment and encephalocele. J Pediatr Ophthalmol. 8(3):181-184.

Kobayashi, H. Baumbach, L, Matise, T.C., Schiavi, A., Greenberg, F., and Hoffman, E.P. 1995. A gene for a severe lethal form of X-linked arthrogryposis (X-linked infantile spinal muscular atrophy) maps to human chromosome Xp11.3-q11.2. Human Molecular Genet. 4:1213-1216.

Kobayashi, T. 1979. Congenital unilateral lower lip palsy. Acta Otolaryngol. 88:303-309.

Kohn, G. 1987. The amniotic band syndrome: A possible complication of amniocentesis (Short Communication). Prenat Diagn. 7:303-305.

Kopelman, J.N. 1993. Antepartum diagnosis of arthrogryposis associated with trisomy 18. Mil Med. 158(7):498-499.

Kottke, F.J., Lehman, J.F., Stillwell, G.K. 1990. Preface. In Krusen's Handbook of Physical Medicine and Rehabilitation, 4th ed., ed. F.J. Kottke, J.F. Lehmann, and G.K. Stillwell. Philadelphia: W.B. Saunders.

Kousseff, B.G. 1981. Cohen syndrome: Further delineation and inheritance. Am J Med Genet. 9:25-30.

Kozlowski, K., Celermajer, J.M., and Tink, A.R. 1974. Humerospinal dysostosis with congenital heart disease. Am J Dis Child. 127:407-410.

Krecak, J., and Starshak, R.J. 1987. Cervical kyphosis in diastrophic dwarfism: CT and MR findings. Pediatr Radiol. 17(4):321-322.

Kretzer, F.L., Hittner, H.M., and Mehta, R. 1981. Ocular manifestations of Conradi and Zellweger syndromes. Meso Pediat Ophthal. 5:1-11.

Krieger, I., and Espiritu, C.E. 1972. Arthrogryposis multiplex congenita and the Turner phenotype. Am J Dis Child. 123:141-144.

Kroll, K., and Klein, E. 1992. Enabling romance: A Guide to Love, Sex, and Relationships for Disabled People (and the People Who Care About Them). New York: Crown Books.

Kulkarni, M.V., and Panjabi, M. 1988. Congenital glaucoma: An association with arthrogryposis multiplex congenita. A case report. Indian J Ophthalmol. 36(4):179-181.

Kupper, L. (ed). 1992. Sexuality education for children and youth with disabilities. NICHCY News Digest. I(3).

Kupper, L. (ed). 1993. Parenting a child with special needs: A guide to readings and resources. NICHCY News Digest. III(1).

Kurnit, D., Hall, J.G., Shurtleff, D.B., and Cohen, M.M., Jr. 1979. An autosomal dominantly inherited syndrome of facial asymmetry, esotropia, amblyopia, and submucous cleft palate (Bencze syndrome). Clin Genet. 16:301-304.

Kuznetsova, N.L., and Gaev, A.V. 1990. Combined examination of patient with arthrogryposis. Ortop Travmatol Protez. (12):54-56.

Lachman, R., Sillence, D., Rimoin, D., Horton, W., Hall, J., Scott, C., Spranger, J., and Langer, L. 1981. Diastrophic dysplasia: The death of a variant. Radiology. 140(1):79-86.

Lacour, J.P., Hoffman, P., Bastiani-Griffet, F., Boutte, P., Pisani, A., and Ortonne, J.P. 1992. Lethal junctional epidermolysis bullosa with normal expression of BM 600 and antropyloric atresia: A new variant of junctional epidermolysis bullosa. Eur J Pediatr. 151:252-257.

Ladd, G.W., and Price, J.M. 1986. Promoting children's cognitive and social competence: The relation between parents' perceptions of task difficulty and children's perceived and actual competence. Child Development. 57:446-460.

LaGreca, A., Siegal, L.J., Wallander, J., and Walker, C. (eds.) 1992. Stress and Coping in Child Health. New York: Guilford.

LaGreca, A.M. 1990. Social consequences of pediatric conditions: Fertile area for future investigation and intervention. J Pediatr Psychol. 15:285-307.

Lai, M.M.R., Tettenborn, M.A., Hall, J.G., Smith, L.J., and Berry, A.C. 1991. A new form of autosomal dominant arthrogryposis. J Med Genet. 28(10):701-703.

Laing, I.A., Teete, R.L., and Stark, A.R. 1988. Diaphragmatic movement in newborn infants. J Pediatr. 112:638-643.

Lake, A.M., Lauer, B.A., Clark, J.C., Wesenberg, R.L., and McIntosh, K. 1976. Enterovirus infections in neonates. J Pediatr. 89(5):787-791.

Lakshminarayana, P., Jegatheesan, T., and Venkataraman, P. 1992. Lethal multiple pterygium syndrome. Indian Pediatr. 29(10):1305-1309.

Lalatta, F., Bagozzi, D.C., Salmoiraghi, M.G., Tagliabue, P., Tischer, C., Zollino, M., Di Rocco, C., Neri, G., and Opitz, J.M., 1990. "C" trigonocephaly syndrome: Clinical variability and possibility of surgical treatment. Am J Med Genet. 37:451-456.

Lambert, D., Nivelon Chevallier, A., and Chapuis, J.L. 1977. Absence of distal interphalangeal fold causing diffuculty in extending fingers. J Med Genet. 14(6):466-467.

Lambert, J.C., Ferrari, M., Donzeau, M., Ayraud, N., Chiaramello, W., and Mariani, R. 1981. Arthrogryposis-like signs in trisomy 18. Hum Genet. 57(2):145-147.

Lambert, L.A. 1947. Congenital humeroradial synostosis with other synostotic anomalies. J Pediatr. 573-577.

Lammer, E.J., and Hayes, A.M. 1987. Isotretinoin phenocopy. Am J Med Genet. 29:675.

Landry, S.H., Robinson, S.S., Copeland, D., and Garner, P.W. 1993. Goal directed behavior and perception of self-competence in children with spina bifida. J Pediatr Psychol. 18:389-396.

Langenskiöld, A. 1985. Congenital contractural arachnodactyly. Report of a case and of an operation for knee contracture. JBJS. 67B(1):44-46.

Langer, L.O., Petersen, D., and Spranger, J. 1970. An unusual bone dysplasia: Parastremmatic dwarfism. Am J Roentgen. 110(3):550-560.

Laureano, A.N., and Rybak, L.P. 1990. Severe otolaryngologic manifestations of arthrogryposis multiplex congenita. Ann Otol Rhinol Laryngol. 99(2):94-97.

Lavigne, J.V., and Faier Routman, J. 1992. Psychological adjustment to pediatric physical disorders: A metaanalytic review. J Pediatr Psychol. 17:133-157.

Laville, J.M., Lakermance, P., and Limouzy, F. 1994. Larsen's syndrome: Review of the literature and analysis of thirty-eight cases. J Pediatr Orthop. 14(1):63-73.

Lazaro, R.P., Fenichel, G.M., and Kilroy, A.W. 1979. Congenital muscular dystrophy: Case reports and reappraisal. Muscle & Nerve. 2:349-355.

Lazarus, R.S., and Folkman, S. 1984. Stress, Appraisal, and Coping. New York: Springer.

Lebenthal, E., Shochet, S.B., Adam, A., Seelenfreund, M., Fried, A., Najenson, T., Sandbank, U., and Matath, Y. 1970. Arthrogryposis multiplex congenita: Twenty-three cases in an Arab kindred. Pediatr. 46(6):891-899.

Lee, B., Godfrey, M., Vitale, E., Hori, H., Mattei, M.G., Sarfarazi, M., Tsipouras, P., Ramirez, F., and Hollister, D.W. 1991. Linkage of Marfan syndrome and a phenotypically related disorder to two different fibrillin genes. Nature. 352:330-334.

Leff, P.T., and Walizer, E.H. 1992. Building the Healing Partnership: Parents, Professionals, and Children with Chronic Illnesses and Disabilities. Cambridge, MA: Brookline Books.

Lehmann, A.R. 1982. Three complementation groups in Cockayne syndrome. Mutation Research. 106:347-356.

Lehmann, A.R. 1985. Prenatal diagnosis of Cockayne's syndrome. Lancet. 2 Mar:486-488.

Lehmann, A.R., Kirk Bell, S., and Mayne, L. 1979. Abnormal kinetics of DNA synthesis in ultraviolet light-irradiated cells from patients with Cockayne's syndrome. Cancer Research. 39:4237-4241.

Leichtman, L.G., Say, B., and Barber, N. 1980. Primary pulmonary hypoplasia and arthrogryposis multiplex congenita. J Pediatr. 96(5):950-951.

Lemanek, K. L. 1994. Research on pediatric chronic illness: New directions and recurrent confounds. J Pediatr Psychol. 19:143-148.

Lemmon, C.B., and Vail, A.D. 1954. Amyplasia congenita: Case report and review of literature. Ann Int Med. 41:836-841.

Lenarsky, C., Shewmon, D.A., Shaw, A., and Feig, S.A. 1985. Occurrence of neuroblastoma and asymmetric cyring facies: Case report and review of the literature. J Pediatr. 107(2):268-270.

Levin, M.L., Lupski, J.R., Carpenter, R.J., Jr., Gerson, L.P., and Greenberg, F. 1993. An additional case of pachygyria, joint contractures and facial abnormalities. Clin Dysmorph. 2:365-368.

Levy, J.M. 1988. Family response and adaptation to a handicap. In The Psychiatry of Handicapped Children and Adolescents: Managing Emotional and Behavioral Problems, ed. J.P. Gerring and L. McCarthy. Boston: College Hill Press, Little, Brown.

Lewin, P. 1928. Arthrogryposis multiplex congenita. JBJS. 7:630-636.

Lewin, S.O., and Hughes, H.E. 1987. German syndrome in sibs. Am J Med Genet. 26(2):385-390.

Liebenberg, F. 1973. A pedigree with unusual anomalies of the elbows, wrists and hands in five generations. So Afr Med J. 47:745-748.

Lipson, E.H., Viseskul, C., and Herrmann, J. 1974. The clinical spectrum of congenital contractural arachnodactyly. A case with congenital heart disease. Z Kinderheilk. 118:1-8.

Livingstone, I.R., and Sack, G.R. Jr. 1984. Arthrogryposis multiplex congenita occurring with maternal multiple sclerosis. Arch Neurol. 41(11):1216-1217.

LLoyd, A.V.C., Jewitt, D.E., and Still, J.D.L. 1966. Facial paralysis in children with hypertension. Arch Dis Child. 41:292-294.

Lloyd-Roberts, G.C., and Lettin, A.W.F. 1970. Arthrogryposis multiple congenita. JBJS (Br). 52B(3):494-508.

Lomo, O.M. 1985. Arthrogryposis and associated defects in pigs: Indication of simple recessive inheritance. Acta Vet Scand. 26(3):419-422.

Lourie, J. 1983. Arthrogryposis multiplex congenita. P N G Med J. 26(3):170171.

Lowry, R.B. 1982. Invited editorial comment: Early onset of Cockayne syndrome. Am J Med Genet. 13:209-210.

Lowry, R.B., and Guichon, V.C. 1972. Congenital contractural arachnodactyly: A syndrome simulating Marfan's syndrome. C M A Jour. 107:532-533.

Lutter, L.D. 1990. Larsen syndrome: Clinical features and treatment: A report of two cases. J Pediatr Orthop. 10(2):270-274.

Lyons-Ruth, K., and Zeanah, C.H. 1993. The family context of infant mental health: I. Affective development in the primary caregiving relationship. In Handbook of Infant Mental Health, ed. C.H. Zeanah. New York: Guilford Press.

Mace, M., Williamson, E., and Worgan, D. 1978. Autosomal dominantly inherited adductor laryngeal paralysis: A new syndrome with a suggestion of linkage to HLA. Clin Genet. 14:265-270.

MacKenzie, D.Y. 1959. Arthrogryposis multiplex congenita. Proc Royal Soc of Med. 52:1101-1105.

MacLeod, P.M., and Fraser, F.C. 1978. Congenital contractural arachnodactyly. A heritable disorder of connective tissue distinct from Marfan syndrome. Am J Dis Child. 126:810-812.

Magdalena, N.I.R., and Marinoni, L.P. 1974. Parana hard-skin syndrome: Study of seven families. Lancet. 9 Feb:215-216.

Mailhes, J.B., Lancaster, K., and Sanusi, I.D. 1977. Pena Shokeir syndrome in a newborn male infant. Am J Dis Child. 31:1419-1420.

Maiti, B., Ghosh, S., Bhattacharya, I., and Deb, P. 1988. Arthrogryposis multiplex congenita with double compartment hydrocephalus. J Indian Med Assoc. 86(8):217-218.

Mankey, M.G., Arntz, C.T., and Staheli, L.T. 1993. Open reduction through a medial approach for congenital dislocation of the hip. J Bone and Joint Surg. 75A:1334-1345.

Manouvrier Hanu, S., de la Chapelle, A.C., and Farriaux, J.P. 1988. Marden-Walker syndrome. New case and discussion about its role in arthrogryposes. Pediatrie. 43(4):313-317.

Margolis, S., and Luginbeuhl, B. 1975. Eye abnormalities associated with arthrogryposis multiplex congenita. J Pediatr Ophthalmol. 12(1):57-60.

Marks, J.F., Kay, J., Baum, J., and Curry, L 1968. Uric acid levels in full-term and low-birth-weight infants. J Pediatr. 73(4):609-611.

Martin, J.R., MacEwan, D.W., Blais, J.A., McTrakos, J., Gold, P., Langer, F., and Hill, R.O. 1970. Platyspondyly, polyarticular osteoarthritis, and absent beta-2-globulin in two brothers. Arthritis and Rheumatism. 13(1):53-67.

Martin, N.J., Hill, J.B., Cooper, D.H., O'Brien, G.D., and Masel, J.P. 1986. Lethal multiple pterygium syndrome: Three consecutive cases in one family. Am J Med Genet. 24:295-304.

Martinez, A.J., Hay, S., and McNeer, K.W. 1976. Extraocular muscles light microscopy and ultrastructural features. Acta Neuropath (Berl). 34:237-253.

Martinez Frias, M.L., Frias, J.L., Vazquez, I., and Fernandez, J. 1991. Bartsocas-Papas syndrome: Three familial cases from Spain. Am J Med Genet. 39:34-37.

Martinez Lavin, M., Buendia, A., Delgado, E., Reyes, P., Amigo, M.C., Zuhaib, A., and Salinas, L. 1983. A familial syndrome of pericarditis, arthritis, and camptodactyly. New Engl J Med. 309(4):224-225.

Martini, A.K., and Banniza von Bazan, U. 1982. Surgical treatment of the hand deformity in Freeman-Sheldon syndrome. Handchir Mikrochir Plast Chir. 14(4):210-212.

Martini, A.K., and Banniza von Bazan, U. 1983. Hand deformities in Freeman-Sheldon syndrome and their surgical treatment. Z Orthop (Germany). 121(5):623-629.

Mascarello, J.T., Jones, M.C., Hoyme, H.E., and Freebury, M.M. 1983. Duplication (17P) in a child with an isodicentric (17p) Chromosome. Am J Med Genet. 14:67-72.

Massa, G., Casaer, P., Ceulemans, B., and Van Eldere, S. 1988. Arthrogryposis multiplex congenita associated with lissencephaly: A case report. Neuropediatrics. 19(1):24-26.

Matthews, S., Farnish, S., and Young, I.D. 1987. Distal symphalangism with involvement of the thumbs and great toes. Clin Genet. 32:375-378.

Mayfield, M.K. 1981. Severe spine deformity in myelodysplasia and sacral agenesis: An aggressive surgical approach. Spine. 6(5):498-509.

Mayhew, I.G. 1984. Neuromuscular arthrogryposis multiplex congenita in a thoroughbred foal. Vet Pathol. 21(2):187-192.

Mayhew, J.F. 1993. Anethesia for children with Freeman-Sheldon syndrome. Anesthesiology. 78(2):408.

McCormack, M.K., Coppola McCormack, P.J., and Lee, M.I. 1980. Autosomal dominant inheritance of distal arthrogryposis. Am J Med Genet. 6(2):163-169.

McCredie, J. 1975. Congenital malformations and embryonic neuropathy. N Eng J Med. 293(2):98-99.

McDonald, L., Kysela, T.Z., Siebert, P, et al. 1986. Parent perspective: Transition to preschool. Teaching Exceptional Children. 22(1):4-8.

McHugh, H.E., Sowden, K.A., and Levitt, M.N. 1969. Facial paralysis and muscle agenesis in the newborn. Acta Otolaryng. 89:157-169.

McKeown, C.M.E., and Harris, R. 1988. An autosomal dominant multiple pterygium syndrome. J Med Genet. 25:96-103.

McKusick, V.A., and Barranco, F.T. 1967. Osteochondritis dissecans with associated malformations in two brothers. A review of familial aspects. JBJS. 49A(5):925-937.

McLeod, J.G., Baker, W.D.C., Lethlean, A.K., and Shorey, C.D. 1972. Centronuclear myopathy with autosomal dominant inheritance. J Neurol Sci. 15:375-387.

McMenamin, J.B., Becker, L.E., and Murphy, E.G. 1982. Congenital muscular dystrophy: A clinicopathologic report of 24 cases. J Pediatr. 100(5):692-697.

McPherson, E. 1988. Renal ultrasound examination of parents in dominantly inherited renal adysplasia. A note of caution. Am J Med Genet. 29:695-696.

McPherson, E., Carey, J., Kramer, A., Hall, J.G., Paurli, R.M., Schimke, R.N., and Tasin, M.H. 1987. Dominantly inherited renal adysplasia. Am J Med Genet. 26:863-872.

McPherson, E., Hall, J.G., and Hickman, R. 1976. Chromosome 7 short arm deletion and craniosynostosis. A 7p syndrome. Hum Genet. 35:117-123.

Mead, C.A., and Martin, M. 1963. Aplasia of the trochlea. An original mutation. JBJS. 45A(2):379-383.

Menelaus, M.B. 1971. Talectomy for equinovarus deformity in arthrogryposis and spina bifida. JBJS. 53B:468.

Mennen, U. 1993. Early corrective surgery of the wrist and elbow in arthrogryposis multiplex congenita. J Hand Surg. 18(3):304-307.

Merk, H., and Bosselmann, E. 1986. Arthrogryposis multiplex congenita with femoral fracture. ROFO Fortschr Geb Rontgenstr Nuklearmed. 145(6):734-735.

Merz, E., and Goldhofer, W. 1985. Sonographic image of a form of arthrogryposis multiplex congenita. Geburtshilfe Frauenheilkd. 45(6):406-410.

Meyen, E.L. 1990. Exceptional Children in Today's Schools, 2nd ed. Denver, CO: Love Publishing Co.

Meyer, D.J. 1993. Lessons learned: Cognitive coping strategies of overlooked family members. In Cognitive Coping, Families, and Disability, ed. A.P. Turnbull, J.M. Patterson, S.K. Behr, D.L. Murphy, J.M. Marquis, and M.J. Blue-Banning. Baltimore: Paul H. Brookes.

Meyer, D.J., Vadasy, P.F., and Fewell, R.R. 1985. Living with a Brother or Sister with Special Needs: A Book for Sibs. Seattle: University of Washington Press.

Meyers, K.R., Golomb, H.M., Hansen, J.L., and McKusick, V.A. 1974. Familial neuromuscular disease with "myotubes." Clin Genet. 5:327-337.

Meyn, M., and Ruby, L. 1967. Arthrogryposis of the upper extremity. Orthop Clin North Am. 7:501-509.

Mglinets, V.A. 1992. Disorders of finger flexion crease formation in various congenital anomalies of human development. Genetika. 28(9):150-157.

Miller, A., Solimano, A., and Norman, M.G. 1987. Arthrogryposis multiplex congenita and hypotonia in a male neonate. Pediatr Neurosci. 13(5):272-277.

Miller, A.P., and Frankfi, K.A. 1969. Kyphoscoliosis. J Pediatr. 75(2):345-346.

Miller, B.A., and Pollard, Z.F. 1994. Duane's retraction syndrome and arthrogryposis multiplex congenita. Surv Ophthalmol. 38(4):395-396.

Miller, E.A. 1965. Congenital clubfoot. Surg Clin N Am. February:231-237.

Miller, M., and Hall, J.G. 1979. Familial asymmetric crying facies. Its occurrence secondary to hypoplasia of the anguli oris depressor muscles. Am J Dis Child. 133:743-746.

Miller, M., and Smith, D.W. 1980. Severe amniotic adhesion malformations. Lancet. 14 Jun:1298-1299.

Miller, M.E., Graham, J.M., Higginbottom, M.C., and Smith, D.W. 1981. Compression related defects from early amnion rupture: Evidence for mechanical teratogenesis. J Pediatr. 98(2):292-297.

Miller, P., Herndon, W.A., and Yngve, D.A. 1985. Lumbosacral agenesis. Orthopedics. 8(10):1297-1298.

Minde, K. 1993. Prematurity and serious medical illness in infancy: Implications for development and intervention. In Handbook of Infant Mental Health, ed. C.H. Zeanah. New York: Guilford Press.

Mirazimov, B.M., and Dzhuraev, A.M. 1989. Our experience in the treatment of deformations of the knee joint in arthrogryposis. Ortop Travmatol Protez. (8):12-15.

Moerman, P., Fryns, J., Cornelis, A., Bergmans, G., Vandenberghe, K., and Lwuweryns, J.M. 1990. Pathogenesis of the lethal multiple ptergium syndrome. Am J Med Genet 35:415-421.

Moerman, P., Fryns, J.P., Van Dijck, H., and Lauweryns, J.M. 1985. Congenital muscular dystrophy associated with lethal arthrogryposis multiplex congenita. Virchows Arch A Pathol Anat Histopathol. 408(1):43-48.

Moessinger, A.C. 1983. Fetal akinesia deformation sequence: An animal model. Pediatr. 72(6):857-863.

Moessinger, A.C., Blanc, W.A., Marone, P.A., and Polsen, D.C. 1982. Umbilical cord length as an index of fetal activity: Experimental study and clinical implications. Pediatr Res. 16:109-112.

Mollejo Villanueva, M., Torres Mohedas, J., Cabello Fernandez, A., Medina Lopez, C., Simon de las Heras, R., and Mateos Beato, F. 1991. Dysgenesis of the anterior horns and nuclei of the brain stem in multiple congenital arthrogryposis. Presentation of a case. An Esp Pediatr. 35(6):429-431.

Monreal, F.J. 1980. Asymmetric crying facies: An alternative interpretation. Pediatr. 65(1):146-149.

Moore, C.A., and Weaver, D.D. 1989. Familial distal arthrogryposis with craniofacial abnormalities: A new subtype of type II? Am J Med Genet. 33(2):231-237.

Morelli, G., Mesolella, C., Costa, F., Testa, B., Ventruto, V., and Santulli, B. 1982. Familial laryngeal abductor paralysis with presumed autosomal dominant inheritance. Ann Otol Rhinol Laryngol. 91:323324.

Moser, A.E., Singh, I., Brown, F.R. III, Solish, G.I., Kelley, R.I., Benke, P.J., and Moser, H.W. 1984. The cerebrohepatorenal (Zellweger) syndrome. Increased levels and impaired degradation of very long-chain-fatty acids and their use in prenatal diagnosis. N Engl J Med. 310:1141-1146.

Moutard-Codou, M.L., Delleur, M.M., Dulac, O., Morel, E., Voyer, M., and De Gammara, E.1987. Severe neonatal myasthenia with arthrogryposis. Presse Med. 16(13):615-618.

Moyer, D.B., Marquis, P., Shertzer, M.E., and Burton, B.K. 1982. Brief clinical report: Cockayne syndrome with early onset of manifestations. Am J Med Genet. 13:225-230.

Mrozek, K., Strugalska, M., and Fidzianska, A. 1970. A sporadic case of central core disease. J Neurol Sci. 10:339-348.

Mulvihill, J.J., Mulvihill, C.G., and Priester, W.A. 1980. Cleft palate in domestic animals: Epidemiologic features. Teratology. 21:109-112.

Munk, S. 1988. Early operation of the dislocated knee in Larsen's syndrome. A report of two cases. Acta Orthop Scand. 59(5):582-584.

Murakami, M., Yamatani, M., Konda, M., Konishi, T., Okada, T., and Nonaka, I. 1987. Severe type arthrogryposis multiplex congenita with pseudohypoaldosteronism. No To Hattatsu. 19(6):497-501.

Murray, J.C., Johnson, J.A., and Bird, T.D. 1985. Dandy-Walker malformation: Etiologic heterogeneity and empiric recurrence risks. Clin Genet. 28:272-283.

Nader, P.R.MD (ed.) 1993. School Health: Policy and Practice, 5th ed. American Academy of Pediatrics, Committee on School Health.

Naffah, J. 1976. The Dyggve-Melchior-Clausen syndrome. Am J Hum Genet. 28:607-314.

Naguib, K.K., Teebi, A.S., Al Awadi, S.A., Moosa, A., and Ali, N.R. 1987. Multiple pterygium syndrome in five Arab sibs. Ann Genet. 30(2):122-125.

Nakamura, Y., Yamamoto, I., Funatsu, Y., Motomura, K., Fukuda, S., Hashimoto, T., and Mortmatsu, M. 1988. Decreased surfactant level in the lung with oligohydramnios: A morphometric and biochemical study. J Pediatr. 112(3):471-474.

Narazaki, O., Hanai, T., Maeda, Y., Uchida, T., and Mitsudome, A. 1986. Arthrogryposis multiplex congenita in nemaline myopathy. Rinsho Shinkeigaku. 26(9):960-964.

Narita, M., Inui, S., and Hashiguchi, Y. 1979. The pathogenesis of congenital encephalopathies in sheep experimentally induced by akabane virus. J Comp Path. 89:229-240.

National Association for the Education of Young Children, 1834 Connecticut Ave. NW, Washington, DC 20009.

Nawrot, P.S., Howell, W.E., and Leipold, H.W. 1980. Arthrogryposis: An inherited defect in newborn calves. Aust Vet J. 56(8):359-364.

Nelson, K.B., and Eng, G.D. 1972. Congenital hypoplasia of the depressor anguli oris muscle: Differentiation from congenital facial palsy. J Pediatr. 81(1):16-20.

Nes, N., Lomo, O.M., and Bjerkas, I. 1982. Hereditary lethal arthrogryposis ("muscle contracture") in horses. Nord Vet Med. 34(12):425-430.

Neu, R.L., and Gardner, L.I. 1973. A partial trisomy of chromosome 1 in a family with a t(1q;4q+) translocation. Clin Genet. 4:47-4479.

Nevin, N.C., Hurwitz, L.J., and Neill, D.W. 1966. Familial camptodactyly with taurinuria. J Med Genet. 3:265-267.

Nezelof, C., Dupart, M.C., Jaubert, F., and Ellachar, E. 1979. A lethal familial syndrome associating arthrogryposis multiplex congenita, renal dysfunction, and a cholestatic and pigmentary liver disease. J Pediatr. 94(2):258-260.

Nicolson, T.B., Nettleton, P.F., Spence, J.A., and Calder, K.H. 1985. High incidence of abortions and congenital deformities of unknown aetiology in a beef herd. Vet Rec. 116(11):281-284.

Nonaka, I., Kikuchi, A., Suzuki, T., and Esaki, K. 1986. Hereditary peroneal muscular atrophy in the mouse: An experimental model for congenital contractures (arthrogryposis). Exp Neurol. 91(3):571-579.

Norio, R., Raitta, C., and Lindahl, E. 1984. Further delineation of the Cohen syndrome: Report on chorioretinal dystrophy, leukopenia and consanguinity. Clin Genet. 25:1-14.

North, C., Patton, M.A., Baraitser, M., and Winter, R.M. 1985. The clinical features of the Cohen syndrome: Further case reports. J Med Genet. 22:131-134.

Norum, R.A., James, V.L., and Mabry, C.C. 1969. K Pterygium syndrome in three children in a recessive pedigree pattern. BDOAS. 5(2):233-235.

Norwood, T.H., and Hoehn, H. 1974. Trisomy of the long arm of human chromosome 1. Humangenetik, Bd. 25:79-82.

Novotny, E.J., Jr. 1988. Arthrogryposis associated with connatal Pelizaeus-Merzbacher disease: Case report. Neuropediatrics. 19(4):221-223.

Oberoi, G.S., Kaul, H.L., Gill, I.S., and Batra, R.K. 1987. Anaesthesia in arthrogryposis multiplex congenita: Case report. Can J Anaesth. 34(3):288-290.

O'Brien, P.J., Gropper, P.T., Tredwell, S.J., and Hall, J.G. 1984. Orthopaedic aspects of the trismus pseudocamptodactyly syndrome. J Pediatr Orthrop. 4:469-471.

Oelberg, D.G., and Adcock, E.W. III. 1983. Oxygen hoods: An unusual cause of neonatal flexion contractures. Am J Dis Child. 137:182.

Ogasawara, H., Shimodate, Y., Matsui, M., Yodono, M., Murakawa, T., and Matsuki, A. 1990. Sevoflurane anesthesia for a patient with arthrogryposis multiplex congenita. Masui. 39(6):792-795.

Ogata, K., Schoenecker, P.L., and Sheridan, J. 1979. Congenital vertical talus and its familial occurrence: An analysis of 36 patients. Clin Orthop. (139):128-132.

Ohdo, S., Madokoro, H., and Hayakawa, K. 1982. Interstitial deletion of the long arm of chromosome 5: 46,XX,del(5) (q13q22). J Med Genet. 19:479.

Ohdo, S., Madokoro, H., Sonoda, T., Takei, M., Yasuda, H., and Mori, N. 1987. Association of tetraamelia, ectodermal dysplasia, hypoplastic lacrimal ducts and sacs opening towards the exterior, peculiar face, and developmental retardation. J Med Genet. 24:609-612.

Ohdo, S., Yamauchi, Y., and Hayakawa, K. 1981. Distal symphalangism associated with camptodactyly. J Med Genet. 18:456-458.

Oki, T., Terashima, Y., Murachi, S., and Nogami, H. 1976. Clinical features and treatment of joint dislocations in Larsen's syndrome. Report of three cases in one family. Clin Orthop. (119):206-210.

Opitz, J.M. 1987. Editorial comment: Vaginal atresia (von Mayer-Rokitansky-Kuster or MRK anomaly) in hereditary renal adysplasia (HRA). Am J Med Genet. 26:873-876.

Opitz, J.M., Johnson, R.C., McCreadie, S.R., and Smith, D.W. 1969a. The C syndrome of multiple congenital anomalies. BDOAS. 5(2):161166.

Opitz, J.M., Lowry, B.R., Holmes, T.M., and Morgan, K. 1989. Hutterite Cerebroosteonephrodysplasia: Autosomal recessive trait in a Lehrerleut Hutterite family from Montana. Am J Med Genet. 22:521529.

Opitz, J.M., ZuRhein, G.M., Vitale, L., Shahidi, N.T., Howe, J.J., Chou, S.M., Shanklin, D.R., and Sybers, H.D. 1969b. The Zellweger syndrome (cerebrohepatorenal syndrome). BDOAS. 5(2):144-160.

Ordover, E.L., and Boundy, K.B. 1991. Educational Rights of Children with Disabilities. Cambridge, MA: Center for Law and Education.

Ornay, A., Sekelec, E., and Sasovsky, E. 1974. Amniotic bands as a cause of syndactyly in a young human fetus. Teratology. 9(2):.

Osborne, A.G. 1992. Legal standards for an appropriate education in the post-Rowley era. Exceptional Children. 58(6):488-494.

Ossipoff, V., and Hall, B.D. 1977. Etiologic factors in the amniotic band syndrome: A study of 24 patients. BDOAS. 46(3D):117-132.

Ostergaard, K. 1988. Arthrogryposis multiplex congenita. Ugeskr Laeger. 150(45):2719-2722.

O'Sullivan, P., Mahoney, G., and Robinson, C. 1992. Perceptions of pediatricians' helpfulness: A national study of mothers of young disabled children. Dev Med Child Neur. 34:1064-1071.

Otto, A.W. 1841. Monstrorum sexcentrorum descriptio anatomica in Vratislavae Museus. Anat Pathol Vrat.

Padovani, J.P., Rigault, P., Pouliquen, J.C., Guyonvarch, G., and Durand, Y. 1976. L'Astragalectomie chez l'enfant. Rev Chir Orthop Reparatrice Appar Mot. 62:475.

Paez, J.H., Tuulonen, A., Yarom, R., Arad, I., Zelikovitch, A., and Ben Ezra, D. 1982. Ocular findings in arthrogryposis multiplex congenita. J Pediatr Ophthalmol Strabismus. 19(2):75-79.

Pagnan, N.A., and Gollop, T.R. 1987. Distal arthrogryposis type II D in three generations of a Brazilian family. Am J Med Genet. 26(3):613-619.

Pagnan, N.A.B., Gollop, T.R., and Lederman, H. 1988. The Tel Hashomer camptodactyly syndrome: Report of a new case and review of the literature. Am J Med Genet. 29:411-417.

Palant, D.I., Feingold, M., and Berkman, M.D. 1971. Unusual facies, cleft palate, mental retardation, and limb abnormalities in siblings. A new syndrome. J Pediatr. 78(4):686-688.

Palmer, P.M., Mac Ewen, G.D., Bowen, J.R., and Mathews, P.A. 1985. Passive motion therapy for infants with arthrogryposis. Clin Orthop. (194):54-59.

Palmer, R.M. 1964. The genetics of talipes equinovarus. JBJS. 46A(3):542-556.

Palotie, A., Vaisanen, P., Ott, J., Ryhanen, L., Elima, K., Vikkula, M., Cheah, K., and Vourio, E. 1989. Predisposition to familial osteoarthrosis linked to type II collagen gene. Lancet. 29 Apr:924-927.

Papadatos, C., Alexiou, D., Nicolopoulos, D., Mikropoulos, H., and Hadzigeorgiou, E. 1974. Congenital hypoplasia of depressor anguli oris muscle: A genetically determined condition? Arch Dis Child. 49:927-931.

Papadia, F., Longo, N., Serlenga, L., and Porzio, G. 1987. Progressive form of multiple pterygium syndrome in association with nemalin myopathy: Report of a female followed for twelve years. Am J Med Genet. 26:73-83.

Pape, K.E., and Pickering, D. 1972. Asymmetric crying facies: An index of other congenital anomalies. J Pediatr. 81(1):21-30.

Parker, J.G., and Asher, S.R. 1993. Friendship and friendship quality in middle childhood: Links with peer group acceptance and feelings of loneliness and social dissatisfaction. Dev Psychol. 29:611-621.

Parmelee, A.H. 1931. Molding due to intrauterine posture. Facial paralysis probably due to such molding. Am J Dis Child. 1155-1159.

Pascalet Guidon, M., Bois, E., Feingold, J., Mattei, J., Combes, J., and Hamon, C. 1984. Cluster of acute infantile spinal muscular atrophy (Werdnig-Hoffmann disease) in a limited area of Reunion Island. Clin Genet. 26:39-43.

Pashayan, H., Dallaire, L., and MacLeod, P. 1973. Bilateral aniridia, multiple webs and severe mental retardation in a 47,XXY/48,XXXY mosaic. Clin Genet. 4:125-129.

Passarge, E., and McAdams, A.J. 1967. Cerebrohepatorenal syndrome. J Pediatr. 71(5):691-702.

Patterson, J.M. 1991. Family resilience to the challenge of a child's disability. Pediatr Ann. 20(9):491-499.

Patterson, J.M., McCubbin, H.I., and Warwick, W.J. 1990. The impact of family functioning on health changes in children with cystic fibrosis. Soc Sci Med. 31:159-164.

Patton, M.A., Sharma, A., and Winter, R.M. 1985. The Aase-Smith syndrome. Clin Genet. 28:521-525.

Patton, R.G., Christie, D.L., Smith, D.W., and Beckwith, J.B. 1972. Cerebrohepatorenal syndrome of Zellweger. Two patients with islet cell hyperplasia, hypoglycemia, and thymic anomalies, and comments on iron metabolism. Amer J Dis Child. 124:840-844.

Paugh, D.R., Koopmann, C.F., and Babyak, J.W. 1988. Arthrogryposis multiplex congenita: Otolaryngologic diagnosis and management. Int J Pediatr Otorhinolaryngol. 16(1):45-53.

Pawlaczyk, B., Zabel, E., and Matuszak, T. 1987. Congenital arthrogryposis in a 2-year-old girl. Wiad Lek. 40(9):623-626.

Pearce, W.G. 1972. Ocular and genetic features of Cockayne's syndrome. Canad J Ophthal. 7:435-444.

Pearn, J.H., Carter, C.O., and Wilson, J. 1973. The genetic identity of acute infantile spinal muscular atrophy. Brain. 96:463-470.

Pearn, J.H., and Wilson, J. 1973. Acute Werdnig-Hoffman disease. Acute infantile spinal muscular atrophy. Arch Dis Child. 48:425-430.

Pearson, C.M., and Fowler, W.G., Jr. 1963. Hereditary nonprogressive muscular dystrophy inducing arthrogryposis syndrome. Brain. 86:75-91.

PeBenito, R., Sher, J.H., and Cracco, J.B. 1978. Centronuclear myopathy: Clinical and pathologic features. Clin Pediatr. 17(3):259-265.

Pedreira, F.A., and Long, R.E. 1971. "Arthrogryposis multiplex congenita" in one of identical twins. Am J Dis Child. 121:64-66.

Pelias, M.Z., and Thurmon, T.F. 1979. Congenital universal muscular hypoplasia: Evidence for autosomal recessive inheritance. Am J Hum Genet. 31:549-554.

Pena, C.E., Miller, F., and Budzilovich, G.N. 1968. Arthrogryposis multiplex congenita. Neurology. 18(9):926-930.

Pena, S.D., and Shokeir, M. 1970. Syndrome of camptodactyly, multiple ankylosis, facial anomalies and pulmonary hypoplasia. Further delineation and evidence for autosomal recessive inheritance. BDOAS. 21:201-208.

Peretti, G., Segre, A., and Beluffi, G. 1979. Larsen's syndrome. Case report and discussion. Ital J Orthop Traumatol. 5(1):89-96.

Perlman, M., and Reisner, S.H. 1972. Asymmetric crying facies and congenital anomalies. Arch Dis Child. 48:627-629.

Perlman, M., Williams, J., and Hirsch, M. 1976. Neonatal pulmonary hypoplasia after prolonged leakage of amniotic fluid. Arch Dis Child. 51:349-353.

Petajan, J.H., Aase, J., and Wright, D.G. 1969. Arthrogryposis syndrome (Kuskokwin disease) in the Eskimo. JAMA. 209(10):1481-1486.

Petrella, R., Rabinowitz, J.G., Steinmann, B., and Hirschhorn, K. 1993. Long-term follow-up of two sibs with Larsen syndrome possibly due to parental germline mosaicism. Am J Med Genet. 47(2):187-197.

Petrus, M., Rhabbour, M., Clouzeau, J., Bat, P., Bildstein, G., Ibanez, M.H., and Netter, J.C. 1993. Association of Moebius syndrome and hypopituitarism due to a midline anomaly. A case report. Ann Pediatr. 40(6):376-378.

Petrusewicz, I.H., Zaremba, J., and Borkowska, J. 1985. Chronic proximal spinal muscular atrophy of childhood and adolescence: Problems of classification and genetic counselling. J Med Genet. 22:350-353.

Pettit, G.S., Dodge, K.A., and Borwyn, M.M. 1988. Early family experience, social problem-solving patterns, and children's social competence. Child Development. 59:107-120.

Pfeiffer, R. A. 1982. The otoonychoperoneal syndrome. Eur J Pediatr. 138:317-320.

Pfeiffer, R.A., and Bachmann, K.D. 1973. An atypical case of Cockayne's syndrome. Clin Genet. 4:28-32.

Phillips, W.A., Cooperman, D.R., Lindquist, T.C., Sullivan, R.C., and Millar, E.A. 1982. Orthopaedic management of lumbosacral agenesis. Long-term follow-up. JBJS. 64(9):1282-1294.

Piaget, J., and Inhelder, B. 1969. The Psychology of the Child. New York: Basic Books.

Pinelli, G., Cipriani, C., and Di Stefano, A. 1983. Biochemical study of variations of collagen and elastin in arthrogryposis multiplex congenita. Minerva Pediatr. 35(6):287-290.

Pope, A.W., McHale, S.M., and Craighead, W.E. 1988. Self-Esteem Enhancement with Children and Adolescents. New York: Pergamon Press.

Popihn, H. 1980. Contribution to the surgical treatment of congenital multiple arthrogryposis of the lower extremity. Beitr Orthop Traumatol. 27(10):580-584.

Poser, S. 1986. Arthrogryposis multiplex congenita. Arch Neurol. 43(1):8-9.

Pous, J.G. 1981. Arthrogryposis in childhood. Arthrogryposis multiplex congenita. Chir Pediatr. 22(5):289-363.

Poussa, M., Merikanto, J., Ryoppy, S., Marttinen, E., and Kaitila, I. 1991. The spine in diastrophic dyslpasia. Spine. 16(8):881-887.

Poyadue, F.S. 1993. Cognitive coping at Parents Helping Parents. In Cognitive Coping, Families, and Disability, ed. A.P. Turnbull, J.M. Patterson, S.K. Behr, D.L. Murphy, J.M. Marquis, and M.J. Blue-Banning. Baltimore: Paul H. Brookes.

Poznanski, A.K., and La Rowe, P.C. 1970. Radiographic manifestations of the arthrogryposis syndrome. Radiology. 95(2):353-358. Dis Child. 8:343-354.

Price, D.S. 1933. A case of amyoplasia congenita with pathological report. Arch Project ACTT. Activating Children Through Technology, 27 Horrabin Hall, Western Illinois University, Macomb, IL 61455.

Proops, R., Taylor, A.M.R., and Insley, J. 1981. A clinical study of a family with Cockayne's syndrome. J Med Genet. 18:288-293.

Pruzanski, W. 1965. Congenital malformations in myotonic dystrophy. Acta Neurol Scand. 41:34-38.

Punnett, H.H., and Kirkpatrick, J.A., Jr. 1968. A syndrome of ocular abnormalities, calcification of cartilage, and failure to thrive. J Pediatr. 73(4):602-605.

Purvis, P. 1991. The public laws for education of the disabled. The pediatrician's role. Dev Behavioral Pediatr. 12(5):327-339.

Pyeritz, R.E. 1981. Maternal and fetal complications of pregnancy in the Marfan syndrome. Am J Med. 71:784-790.

Quamma, J.P., and Greenberg, M.T. 1994. Children's experience of life stress: The role of family social support and social problem-solving skills as protective factors. J Clin Child Psychol. 23:295-305.

Quance, D.R. 1988. Anaesthetic management of an obstetrical patient with arthrogryposis multiplex congenita. Can J Anaesth. 35(6):612-614.

Quinn, C.M., Wigglesworth, J.S., and Heckmatt, J. 1991. Lethal arthrogryposis multiplex congenita: A pathological study of 21 cases. Histopathology. 19(2):155-162.

Quinn, S.J., Bleach, N.R., and Richards, A.E. 1994. Middle ear deformity in arthrogryposis multiplex congenita. Ann Otol Rhinol Laryngol. 103(9):729-731.

Quittner, A., DiGirolamo, A.M., Michel, M., and Eigen, H. 1992. Parental response to cystic fibrosis: A contextual analysis of the diagnostic phase. J Pediatr Psychol. 17:683-704.

Radu, H., Rosu Serbu, A.M., Jonescu, V., and Radu, A. 1977. Focal abnormalities in mitochondrial distribution in muscle. Two atypical cases of so-called "central core disease." Acta Neuropath (Berl). 39:25-31.

Ramos-Arroyd, M.A., Weaver, D.D., and Beals, R.K. 1985. Congenital contractural arachnodactyly. Report of four additional families and review of the literature. Clin Genet. 27:570-581.

Rank, B.K., Wakefield, A.R., and Hueston, J.T. 1973. Surgery of Repair as Applied to Hand Injuries, 4th ed., ed. B.K. Rank, A.R. Wakefield, and J.T. Hueston. Edinburgh and London: Churchill Livingstone.

Rao, S., Israel, J., Martin, A., and Kaye, C. 1978. Hypoplasia of depressor anguli oris muscle and imperforate anus in an infant with pericentric inversion of chromosome number 15. Am J Hum Genet. 30(91A):

Raspeslagh, M. 1985. Mental retardation with pterygia, shortness and distinct facial appearance. Clin Genetic. 28:550-555.

Ray, S., Peterson, P.D., and Scott, C.I. 1986. Pelvic dysplasia associated with arthrogrypotic changes in the lower extremities. A new syndrome. Clin Orthop. 207:99-102.

Rebbeck, T.R., Dietz, F.R., Murray, J.C., and Buetow, K.H. 1993. A single gene explanation for the probability of having idiopathic talipes equinovarus. Am J Hum Genet. 53:1051-1063.

Reed, S.D., Hall, J.G., Riccardi, V.M., Aylsworth, A., and Timmons, C. 1985. Chromosomal abnormalities associated with congenital contractures (arthrogryposis). Clin Genet. 27(4):353-372.

Reed, T., and Schreiner, R.L. 1983. Absence of dermal ridge patterns: Genetic heterogeneity. Am J Med Genet. 16:81-88.

Reeve, R., Silver, H.K., and Ferrier, P. 1960. Marfan's syndrome (arachnodactyly) with arthrogryposis (amyoplasia congenita). Am J Dis Child. 99:101-106.

Reid, C., Hall, J.G., Anerson, C., Bocian, M., Carey, J., Costa, T., Curry, C., et al. 1986. Association of amyplasia with gastroschisis, bowel atresia, and defects of the muscular layer of the trunk. Am J Med Genet. 24:701-710.

Reiss, J.A., and Sheffield, L.J. 1986. Distal arthrogryposis type II: A family with varying congenital abnormalities. Am J Med Genet. 24(2):255-267.

Relkin, R. 1965. Arthrogryposis multiplex congenita: Report of two cases, review of literature. Am J Med. 39:871-876.

Renault, F., Arthuis, M., Rethore, M.O., and Lafourcade, J. 1982. Larsen's syndrome. Clinical findings and inheritance. Arch Fr Pediatr. 39(1):35-38.

Richards, B.S. 1988. Partial sacral agenesis with congenital hip dislocation. Orthopedics. 11(6):973-977.

Richards, B.S. 1991. Atlantoaxial instability in diastrophic dysplasia. A case report. JBJS. 73(4):614-616.

Rieger, M.A., Hall, J.E., and Dalury, D.F. 1990. Spinal fusion in a patient with lumbosacral agenesis. Spine. 15(12):1382-1384.

Riley, D.J., and Santiago, T.V. 1977. Blunted respiratory drive in congenital myopathy. Am J Med. 63:459-466.

Ringel, S.P., Bender, A.N., and Engel, W.K. 1976. Extrajunctional acetylcholine receptors. Alterations in human and experimental neuromuscular diseases. Arch Neurol. 33:751-758.

Rizzo, R., Pavone, L., Micali, G., and Hall, J.G. 1993. Familial bilateral antecubital pterygia with severe renal involvement in nail-patella syndrome. Clin Genet (Denmark). 44(1):1-7.

Robertson, F.W., Kozlowski, K., and Middleton, R.W. 1975. Larsen's syndrome. Clin Pediatr. 14(1):53-60.

Robertson, W.L., Glinski, L.P., Kirkpatrick, S.J., and Pauli, R.M. 1992. Further evidence that arthrogryposis multiplex congenita in the human sometimes is caused by an intrauterine vascular accident. Teratology. 45(4):346-351.

Robinow, M. 1986. Transient neonatal arthrogryposis: A presumed sequel of antenatal hypoxia. Am J Med Genet. 25(1):167-168.

Robinson, L.K., Powers, N.G., Dunklee, P., Sherman, S., and Jones, K.L. 1982. The Antley-Bixler syndrome. J Pediatr. 101(2):201-205.

Robinson, R.O. 1990. Arthrogryposis multiplex congenita: Feeding, language and other health problems. Neuropediatrics. 21(4):177-178.

Ronningen, H., and Bjerkreim, I. 1978. Larsen's syndrome. Acta Orthop Scand. 49(2):138-142.

Roscam Abbing, P.J., Hageman, G., and Willemse, J. 1985. CT scanning of skeletal muscle in arthrogryposis multiplex congenita. Brain Dev. 7(5):484-491.

Rosselli, D., and Gulienetti, R. 1961. Ectodermal dysplasia. Brit J Plastic Surg. 14:190-204.

Roth, P.B. 1926. Congenital synostosis of humerus and radius occurring in three children of one family. AUOR 1. 4 May:51-53.

Rouzbahani, L. 1995. New manifestations in an infant with Neu Laxova syndrome. Am J Med Genetics 56:239-240.

Rubin, A. 1963. Birth injuries: Incidence, mechanisms, and end results. Obstetrics. 23(2):218-221.

Rudolph, A.J., Yow, M.D., Phillips, A., Desmond, M.M., Blattner, R.J., and Melnick, J.L. 1965. Transplacental rubella infection in newly born infants. JAMA. 191(10):139-141.

Ruff, J.C., Emmanouil, D.E., and Pendzick, M.J. 1988. Mouthstick prosthesis placement in a 19-month-old arthrogryposi multiplex congenita patient: Case report. Pediatr Dent. 10(4):320-322.

Rushton, D.I. 1983. Amniotic band syndrome. Brit Med J. 286:919-920.

Russell, R.G., and Oteruelo, F.T. 1983. Ultrastructural abnormalities of muscle and neuromuscular junction differentiation in a bovine congenital neuromuscular disease. Acta Neuropathol. 62(1):112-120.

Rutledge, M.L., Hawkins, E.P., and Langston, C. 1986. Skeletal muscle growth failure induced in premature newborn infants by prolonged pancuronium treatment. J Pediatr. 109(5):883-886.

Ryoppy, S., Poussa, M., Merikanto, J., Marttinen, E., and Kaitila, I. 1992. Foot deformities in diastrophic dysplasia. An analysis of 102 patients. JBJS. 74(3):441-444.

Sack, G.H., Jr. 1978. A dominantly inherited form of arthrogryposis multiplex congenita with unusual dermatoglyphics. Clin Genet. 14:317-323.

Sack, J., and Friedman, E. 1980. Cardiac involvement in the Cohen syndrome: A case report. Clin Genet. 17:317-319.

Sadovnick, A.D. 1985. Insignificant risk for arthrogryposis multiplex cogenita. Arch Neurol. 42(6):516.

Sahgal, B., and Sahgal, S. 1977. A new congenital myopathy: A morphological, cytochemical and histochemical study. Acta Neuropath (Berl). 37:225-230.

Saint Supery, G., Wallon, P., Bucco, P., and Barnetche, J.M. 1985. Three case reports of lumbosacral agenesis: Role of the lumboiliac bone graft. Chir Pediatr. 26(3):181-186.

Saito, K., Fukuyama, Y., Ogata, T., and Oya, A. 1981. Experimental intrauterine infection of akabane virus. Pathological studies of skeletal muscles and central nervous system of newborn hamsters with relevances to the Fukuyama type congenital muscular dystrophy. Brain Dev. 3(1):65-80.

Sakamoto, F.O., Claman, L., Klabunde, M., Perry, T., and Horton, J.E. 1985. Management of arthrogryposis multiplex congenita. A case report. J Periodontol. 56(11):694-698.

Sakamoto, T., Tawara, A., and Inomata, H. 1992. Goniodysgenesis of the eye with arthrogryposis multiplex congenita. Ophthalmologica. 204(4):210-214.

Saleh, M., Gibson, M.F., and Sharrard, W.J. 1989. Femoral shortening in correction of congenital knee flexion deformity with popliteal webbing. J Pediatr Orthop. 9(5):609-611.

Salonen, R. 1984. The Meckel syndrome: Clinicopathological findings in 67 patients. Am J Med Genet. 18:671-689.

Sameroff, A.J. 1993. Models of development and developmental risk. In Handbook of Infant Mental Health, ed. C.H. Zeanah. New York: Guilford Press.

Samuel, A.W., and Davies, D.R. 1981. The Larsen syndrome with multiple congenital dislocations and a normal facies. Int Orthop. 5(3):229-232.

Saraiva, J.M., Lemos, C., Goncalves, I., Carneiro, F., and Mota, H.C. 1990. Arthrogryposis multiplex congenita with renal and hepatic abnormalities in a female infant. J Pediatr. 117(5):761-763.

Saraiva, J.M., Lemos, C., Goncalves, I., Mota, H.C., and Carneiro, F. 1992. Arthrogryposis multiplex congenita with renal and hepatic abnormalities. Am J Med Genet. 42(1):140.

Sargent, C., Burn, J., Baraitser, M., and Pembrey, M.E. 1985. Trigonocephaly and the Opitz C syndrome. J Med Genet. 22:39-45.

Sarnat, H.B., Machin, G., Darwish, H.Z., and Rubin, S.Z. 1983. Mitochondrial myopathy of cerebrohepatorenal (Zellweger) syndrome. Can J Neurol Sci. 10:170-177.

Sarwark, J.F., Mac Ewen, G.D., and Scott, C.I. Jr. 1990. Amyoplasia (a common form of arthrogryposis). JBJS. 72A(3):465-469.

Saul, R.A., and Meyer, L.C. 1985. Autosomal dominant spinal muscular atrophy in three generations. Proc Greenwood Genet Center. 4:13-15.

Savini, R., and Gualdrini, G. 1980. Report on two cases of Freeman-Sheldon syndrome ("whistling face"). Ital J Orthop Traumatol. 6(1):105-115.

Sawatzky, B. Undated. Physical activity and arthrogryposis. British Columbia Children's Hospital Orthopaedic Department booklet for patients and families. Vancouver, British Columbia.

Say, B., Barber, N.D., and Leichtman, L.G. 1979. Ankylosis, facial anomalies, and pulmonary hypoplasia syndrome. Am J Dis Child. 133:1196-1197.

Schinzel, A., Hayashi, K., and Schmid, W. 1974. Mosaictrisomy and pericentric inversion of chromosome 9 in a malformed boy. Humangenetik. 25:171-177.

Schinzel, A., Hayashi, K., and Schmid, W. 1975. Structural aberrations of chromosome 18 II. The 18q syndrome. Report of three cases. Humangenetik. 26:123-132.

Schinzel, A., Homberger, C., and Sigrist, T. 1978. Bilateral renal agenesis in 2 male sibs born to consanguineous parents. J Med Genet. 15:314-316.

Schnabel, R. 1981. Intrauterine coxsackie B infection in arthrogryposis multiplex congenita syndrome. Verh Dtsch Ges Pathol. 65:311-315.

Schnute, W.J. 1965. Congenital absence of the lower extremity. Surg Clin N Am. February:239-246.

Schochet, S.S., Zellweger, H., Ionasescu, V., and McCormick, W.F. 1972. Centronuclear myopathy: Disease entity or a syndrome? Light and electron microscopic study of two cases and review of the literature. J Neurol Sci. 16:215-228.

Schopler, S.A., and Menelaus, M.B. 1987. Subsidiary lateral femoral condyle in arthrogryposis multiplex congenita. J Pediatr Orthop. 7(4):463-465.

Schore, A.N. 1994. Affect Regulation and the Origin of the Self: The Neurobiology of Emotional Development. Hillsdale, NJ: Lawrence Erlbaum Associates.

Schrander Stumpel, C., Fryns, J.P., Beemer, F.A., and Rive, F.A. 1991a. Association of distal arthrogryposis, mental retardation, whistling face and Pierre Robin sequence: Evidence of nosologic heterogeneity. Am J Med Genet. 38(4):557561.

Schrander Stumpel, C.R.T.M., Howeler, C.J., Reckers, A.D., De Smet, N., Hall, J.G., and Fryns, J.P. 1993. Arthrogryposis, ophthalmoplegia, and retinopathy: Confirmation of a new type of arthrogryposis. J Med Genet. 30(1):78-80.

Schrander Stumpel, C.T., Fryns, J.P., Schander, J.J., and Vles, J. 1991b. Distal arthrogryposis, specific facial dysmorphism and psychomotor retardation: A recognizable entity in surviving patients with the fetal akinesia deformation sequence. Genet Couns (Switz). 2(2):69-75.

Schuring, A.G., and Gunter, J.P. 1970. Paralysis of the facial nerve in children. In early life the nerve is more susceptible to infections, trauma and tumors. Clin Pediatr. 9(2):105-109.

Scott, C.I. 1969. Pterygium syndrome. BDOAS. 5(2):232-233.

Scully, R.E.(ed.), and Galdabini, J.J. (assoc. ed.). 1976. Case records of the Massachusetts General Hospital. New Eng J Med. 295(2):92-99.

Seay, A.R., Ziter, F.A., and Petajan, J.H. 1977. Rigid spine syndrome. A type I fiber myopathy. Arch Neurol. 34:119-122.

Segal, L.S., Mann, D.C., Feiwell, E., and Hoffer, M.M. 1989. Equinovarus deformity in arthrogryposis and myelomeningocele: Evaluation of primary talectomy. Foot Ankle. 10(1):12-16.

Seitz, R.J., Wechsler, W., Mosny, D.S., and Lenard, H.G. 1986. Hypomyelination neuropathy in a female newborn presenting as arthrogryposis multiplex congenita. Neuropediatrics. 17(3):132-136.

Sellars, S., and Beighton, P. 1983. Autosomal dominant inheritance of conductive deafness due to stapedial anomalies, external ear malformations and congenital facial palsy. Clin Pediatr. 23:376-379.

Sells, J.M., Jaffe, K.J., and Hall, J.G. 1996. Amyoplasia, the most common type of arthrogryposis: The potential for good outcome. Pediatr. 97:225-231.

Sensenbrenner, J.A., Dorst, J.P., and Hungerford, D.S. 1974. S. Parastremmatic dwarfism. BDOAS. 10(12):425-429.

Sergovich, F.R., Botz, J.S., and McFarlane, R.M. 1983. Nonrandom cytogenetic abnormalities in Dupuytren's disease. New Engl J Med. 306(3):162-163.

Serville, F., Dufau Casanabe, J., and Fontan, D. 1986. Arthrogryposis and 46,XY,t(1; 16) chromosome constitution. Clin Genet. 29(5):453-455.

Shaman, E. 1985. Choices: A Sexual Assault Prevention Workbook for Persons with Physical Disabilites. Seattle: Seattle Rape Relief Crisis Center.

Shapiro, F. 1992. Light and electron microscopic abnormalities in diastrophic dysplasia growth cartilage. Calif Tissue Int. 51(4):324-331.

Shapiro, F., and Bresnan, M.J. 1982. Orthopaedic management of childhood neuromuscular disease. Part II: Peripheral neuropathies, Friedreich's ataxia, and arthrogryposis multiplex congenita. JBJS. 64A(6):949-953.

Shapiro, F., and Glimcher, M.J. 1979. Gross and histological abnormalities of the talus in congenital club foot. JBJS. 61A(4):522-530.

Shapiro, F., and Specht, L. 1993. The diagnosis and orthopaedic treatment of childhood spinal muscular atrophy, peripheral neuropathy, Friedreich ataxia, and arthrogryposis. JBJS Am. 75A(11):1699-1714.

Sheldon, W. 1932. Amyoplasia congenita (multiple congenital articular rigidity: arthrogryposis multiplex congenita). Arch Dis Child. 7:117-136.

Shepherd, N.C., Gee, C.D., Timmins, G., Carroll, S.N., and Bonner, R.B. 1978. Congenital bovine epizootic arthrogryposis and hydranencephaly. Aust Vet Journ. 54:171-177.

Shikata, J., Yamamuro, T., Mikawa, Y., Iida, H., and Nishimura, N. 1987. Kyphoscoliosis in congenital contractural arachnodactyly. A case report. Spine. 12(10):1055-1058.

Shin, Y.S., Plochl, E., Podskarbi, T., Muss, W., Pilz, P., and Puttinger, R. 1994. Fatal arthrogryposis with respiratory insufficiency: A possible case of muscle phosphorylase b kinase deficiency. J Inherit Metab Dis. 17(1):153-155.

Shved, I.A., Lazjuk, G.I., Ostrovskaya, T.I., and Cherstvoy, E.D. 1983. The popliteal pterygium syndrome (a condition of the main anatomical structures of the lower limbs). Folia Morphologica. 31:258-265.

Siebolt, R.M., Winter, R.B., and Moe, J.H. 1974. The treatment of scoliosis in arthrogryposis multiplex congenita. Clin Orthop. 103:191.

Silengo, M.C., Bell, G.L., Biagioli, M., Guala, A., Bianco, R., Strandoni, P., De Sario, P.N., and Franceschini, P. 1986. Asymmetric crying facies with microcephaly and mental retardation. An autosomal dominant syndrome with variable expressivity. Clin Genet. 30:481-484.

Simonian, P.T., and Staheli, L.T. 1995. Periarticular fractures after manipulation for knee contractures in children. J Pediatr Orthop. 15:288-291.

Simons, R. 1987. After the Tears: Parents Talk About Raising a Child with a Disability. San Diego: Harcourt Brace Jovanovich.

Simpson, C.F. 1985. Physical and occupational therapy for arthritis (topics in primary care medicine). West J Med. 142(4):562-564.

Singhi, S., Singhi, P., and Lall, K.B. 1980. Congenital asymmetrical crying facies. Clin Pediatr. 19(10):673-678.

Sitlington, P.L., Frank, A.R., and Carson, R. 1993. Adult adjustment among high school graduates with mild disabilities. Exceptional Children. 59(3):221-233.

Slacalkova, J., and Grim, M. 1984. Arthrogryposis congenita multiplex. Acta Chir Orthop Traumatol Cech. 51(1):5-11.

Sloper, P., and Turner, S. 1993. Risk and resistance factors in the adaptation of parents of children with severe physical disability. J Child Psychol Psychiatry. 34:167-188.

Smidt, W.J., and Sol, J. 1986. Congenital flexure of the forelimbs in calves. Tijdschr Diergeneeskd. 111(18):860-863.

Smit, L.M., and Barth, P.G. 1980. Arthrogryposi multiplex congenita due to congenital myasthenia. Dev Med Child Neurol. 22(3):371-374.

Smith, D.W. 1982. Approach to Arthrogryposis (Prenatal Onset of Joint Contractures). Recognizable Patterns of Human Malformation, 3rd ed., pp. 533-535. Philadephia: W.B. Saunders.

Smith, D.W., Clarren, S.K., and Harvery, M.A.S. 1978. Hyperthermia as a possible teratogenic agent. J Pediatr. 92(6):878-883.

Smith, D.W., Opitz, J.M., and Inhorn, S.L. 1965. A syndrome of multiple developmental defects including polycystic kidneys and intrahepatic biliary dysgenesis in 2 siblings. J Pediatr. 67(4):617-624.

Smith, E.M., Bender, L.F., and Stover, C. 1963. Lower motor neuron deficit in arthrogryposis: An EMG study. Arch Neurol. 8:113-116.

Smith, L.C., Lockhart, L.M., and Rouse, B.M. 1987. An unusual combination of arachnodactyly and distal arthrogryposis syndrome in a father and son. Dysmorph Clin Gen. 1:90-96.

Socol, M.L., Sabbagha, R.E., Elias, S., Tamura, R.K., Simpson, J.L., Dooley, S.L., and Depp, R. 1985. Prenatal diagnosis of congenital muscular dystrophy producing arthrogryposis. N Engl J Med. 313(19):1230.

Sodergard, J., and Ryoppy, S. 1990. The knee in arthrogryposis multiplex congenita. J Pediatr Orthop. 10(2):177-182.

Sodergard, J.M., Jaaskelainen, J.J., and Ryoppy, S.A. 1993. Muscle ultrasonography in arthrogryposis. Comparison with clinical, neuromyographic and histologic findings in 41 cases. Acta Orthop Scand. 64(3):357-361.

Solund, K., Sonne Holm, S., and Kjolbye, J.E. 1991. Talectomy for equinovarus deformity in arthrogryposis. A 13 (220) year review of 17 feet. Acta Orthop Scand. 62(4):372-374.

Sombekke, B.H., Molenaar, W.M., van Essen, A.J., and Schoots, C.J. 1994. Lethal congenital muscular dystrophy with arthrogryposis multiplex congenita: Three new cases and review of the literature. Pediatr Pathol. 14(2):277-285.

Spearritt, D.J., Tannenberg, A.E.G., and Payton, D.J. 1993. Lethal multiple pterygium syndrome: Report of a case with neurological anomalies. Am J Med Genet. 47(1):45-49.

Speltz, M.L., Morton, K., Goodell, E.W., and Clarren, S.K. 1993. Psychological functioning of children with craniofacial anomalies and their mothers. Cleft Palate Craniofac J. 30:482-489.

Spencer, D., Millar, E., and Brown, J.C. 1977. Spinal deformity in arthrogryposis multiplex congenita. Scoliosis Research Society Annual Meeting.

Spinetta, J.J., Murphy, J.L., Vik, P.J., and Day, J. 1988. Long-term adjustment in families of children with cancer. J Psychosoc Oncol. 6:179-191.

Spranger, J.W., Schnized, A., Myers, T., Ryan, J., Giedion, A., and Opitz, J.M. 1980. Cerebroarthrodigital syndrome: A newly recognized formal genesis syndrome in three patients with apparent arthromyodysplasia and sacral agenesis, brain malformation and digital hypoplasia. Am J Med Genet. 5:13-24.

SRI International. 1993. What makes a difference? Influences on postschool outcomes of youth with disabilities. In The Third Comprehensive Report from the National Longitudinal Transition Study of Spcial Education Students. Menlo Park, CA: SRI International.

Srivastava, R.N. 1968. Arthrogryposis multiplex congenita: Case report of two siblings. Clin Pediatr. 7(11):691-694.

Srivastave, R.N. 1969. Arthrogryposis or a new syndrome? J Pediatr. 7(5):840-841.

St. Clair, H.S., and Zimbler, S. 1985. A plan of management and treatment results in the arthrogrypotic hip. Clin Orthop. (194):74-80.

Staheli, L.T., Chew, D.E., Elliott, J.S., and Mosca, V.S. 1987. Management of hip dislocations in children with arthrogryposis. J Pediatr Orthop. 7(6):681-685.

Stanescu, R., Stanescu, V., and Maroteaux, P. 1982. Abnormal pattern of segment long spacing (SLS) cartilage collagen in diastrophic dysplasia. Coll Relat Res. 2(2):111-116.

Steindler, A. 1949. Arthrogryposis. J Int College Sur. 12:21-25.

Stern, W.G. 1923. Arthrogryposis multiplex congenita. JAMA. 81(18):1507-1510.

Stoll, C., Alembik, Y., Finck, S., and Janser, B. 1992. Arthrogryposis, ectodermal dysplasia and other anomalies in two sisters. Genet Couns. 3(1):35-39.

Stoll, C., Levy, J.M., Kehr, P., and Roth, M.P. 1980. Familial pterygium syndrome. Clin Genet. 18:317-320.

Stoll, C., Treisser, A., and Tranchant, C. 1991. Prenatal diagnosis of congenital myasthenia with arthrogryposis in a myasthenic mother. Prenat Diagn. 11(1):17-22.

Stolov, W.C. 1982. Evaluation of the patient. In Handbook of Physical Medicine and Rehabilitation, ed. F.J. Kotke, G.K. Stillwell, and J.F. Lehmann. Philadelphia: W.B. Saunders.

Stratton, R.F., Sykes, N.J., and Hassler, T.W. 1990. C syndrome with apparently normal development. Am J Med Genet. 37:460-462.

Strehl, E., and Vanasse, M. 1985. EMG and needle muscle biopsy studies in arthrogryposis multiplex congenita. Neuropediatrics. 16(4):225-227.

Suarez Requena, O., and Silva Sarmiento, G.E. 1986. Larsen's syndrome. Bol Med Hosp Infant Mex. 43(5):312-315.

Sugarman, G.I., Landing, B.H., and Reed, W.B. 1977. Cockayne syndrome: Clinical study of two patients and neuropathologic findings in one. Clin Pediatr. 16(3):225-232.

Sugita, T., Ikenaga, M., Suehara, N., Kozuka, T., Furuyama, J., and Yabuuchi, H. 1982. Prenatal diagnosis of Cockayne syndrome using assay of colony-forming ability in ultraviolet light irradiated cells. Clin Genet. 22:137-142.

Sul, Y.C., Mrak, R.E., Evans, O.B., and Fenichel, G.M. 1982. Neurogenic arthrogryposis in one identical twin. Arch Neurol. 39(11):717-718.

Sulaiman, A.R., Swick, H.M., and Kinder, D.S. 1983. Congenital fibre type disproportion with unusual clinicopathologic manifestations. J Neurol, Neurosurg, Psych. 46:175-182.

Sullivan, H.S. 1953. The Interpersonal Theory of Psychiatry. New York: W.W. Norton.

Sullivan, T.J., Clarke, M.P., Heathcote, J.G., Hunter, W.S., Rootman, D.S., and Morin, J.D. 1992. Multiple congenital contractures (arthrogryposis) in association with Peters' anomaly and chorioretinal colobomata. J Pediatr Ophthalmol Strabismus. 29(6):370-373.

Sumitani, S., Kameda, K., Sone, S., and Minami, R. 1994. A case of Larsen syndrome with severe cervical cord compression. No To Hattatsu. 26(4):345-348.

Summers, J., Dell Oliver, C., Turnbull, A., et al. 1990. Examining the individualized family service plan process: What are family and practitioner preferences? Topics in Early Childhood Special Education. 10:78-99.

Swift, D. 1992. Two-tiered treatment gets limbs lengthened faster. The Medical Post. 17 Mar:26-27.

Swinyard, C.A. 1963. Multiple congenital contractures (arthrogryposis). Nature of the syndrome and hereditary considerations. Proc Second Internat. Congress of Hum Genet. 3:1397-1398.

Swinyard, C.A. 1982. Concepts of multiple congenital contractures (arthrogryposis) in man and animals. Teratology. 25(2):247-258.

Swinyard, C.A., and Bleck, E.E. 1985. The etiology of arthrogryposis (multiple congenital contractures). Clin Orthop. (194):15-29.

Szabo, L., and Perjes, K. 1974a. Differential diagnosis between arthrogryposis multiplex congenita and Larsen's syndrome. Z Orthop Ihre Grenzgeb. 112(6):1275-1281.

Szabo, L., and Perjes, K. 1974b. Congenital dislocations of the major joints, multiple bone abnormalities and typical facial structure (Larsen's syndrome). Magy Traumatol Orthop Helyreallito Seb. 17(1):37-42.

Szoke, G, Staheli, L.T., Jaffe, K. and Hall, J.G. 1996. Medial-approach open reduction of hip dislocation in amyoplasia-type arthrogryplasia. J Pediatr Orthop. 16(1): 127-130.

Tanaka, K., Kawai, K., Kurnahara, Y., Ikenaga, M., and Okada, Y. 1981. Genetic complementation groups in Cockayne syndrome. Somatic Cell Genetics. 7(4):445-455.

Tardio Torio, E., Sanchez Sanchez, E., and Perez Prado, C. 1993. Larsen syndrome and idiopathic hypercalciuria. An Esp Pediatr. 39(5):467-469.

Teebi, A.S., and Daoud, A.S. 1990. Multiple pterygium syndrome: A relatively common disorder among Arabs. J Med Genet. 27:791-792.

Tellerman Toppet, N., Gerard, J.M., and Coers, C. 1973. Central core disease. A study of clinically unaffected muscle. J Neurol Sci. 19:207-223.

Temple, K., Hall, C.A., Chitty, L., and Baraitser, M 1990. A case of atelosteogenesis. J Med Genet. 27:194-197.

Teyssier, G., Damon, G., Bertheas, M.F., Freycon, F., and Lauras, B. 1982. Congenital myasthenia and arthrogryposis. Apropos of 2 cases manifesting at birth. Pediatrie. 37(4):295-298.

The Individuals with Disabilities Education Act, Public Law 101476 1990. 20 USC.

Thomas, B., Schopler, S., Wood, W., and Oppenheim, W.L. 1985. The knee in arthrogryposis. Clin Orthop. (194):87-92.

Thomas, I.T., and Smith, D.W. 1974. Oligohydramnios, cause of the nonrenal features of Potter's syndrome, including pulmonary hypoplasia. J Pediatr. 84(6):811-814.

Thompson, C.E. 1986. Raising a Handicapped Child. New York: William Morrow & Company.

Thompson, E., and Baraitsar, M. 1986. An autosomal recessive mental retardation syndrome with hepatic fibrosis and renal cysts. Am J Med Genet. 24:151-158.

Thompson, E.M., Donnai, D., Baraitser, M., Hall, C.M., Pembrey, M.E., and Fixsen, J. 1987. Multiple pterygium syndrome: Evolution of the phenotype. J Med Genet. 24:733-749.

Thompson, G.H., and Bilenker, R.M. 1985. Comprehensive management of arthrogryposis multiplex congenita. Clin Orthop. (194):6-14.

Thompson, R.H. 1985. Psychosocial Research on Pediatric Hospitalization and Health Care. Springfield, IL: Charles C Thomas.

Thompson, R.J., Gustafson, K.E., Hamlett, K.W., and Spock, A. 1992a. Stress, coping, and family functioning in the psychological adjustment of mothers of children and adolescents with cystic fibrosis. J Pediatr Psychol. 17:573-585.

Thompson, R.J., Gustafson, K.E., Hamlett, K.W., and Spock, A. 1992b. Psychological adjustment of children with cystic fibrosis: The role of child cognitive processes and maternal adjustment. J Pediatr Psychol. 17:741-756.

Tolmie, J.L., Patrick, A., and Yates, J.R.W. 1987. A lethal multiple pterygium syndrome with apparent X-linked recessive inheritance. Am J Med Genet. 27:913-919.

Tonoki, H., Kishino, T., and Niikawa, N. 1990. A new syndrome of dwarfism, brachydactyly, nail dysplasia, and mental retardation in sibs. Am J Med Genet. 36:89-93.

Toriello, H.V., Bauserman, S.C., and Higgins, J.V. 1984. Sibs with the fetal akinesia sequence, fetal edema, and malformations: A new syndrome? Am J Med Genet. 21:271-277.

Toriello, H.V., Higgins, J.V., Malvitz, T., and Waterman, D.F. 1990. Two siblings with Tel Hashomer camptodactyly and mitral valve prolapse. Am J Med Genet. 36:398-403.

Torres Aybar, F.G., and Lizasoain, J.A. 1980. Spondylohypoplasia, arthrogryposis, and popliteal pterygium. Am J Dis Child. 134(10):1001.

Toussi, T., Halal, F., Lesage, R., Delorme, F., and Bergeron, A. 1980. Brief clinical report: Renal hypodysplasia and unilateral ovarian agenesis in the Penta-X syndrome. Am J Med Genet. 6:153-162.

Tranchant, C., Ehret, C., Labouret, P., Gasser, B., and Warter, J.M. 1991. Arthrogryposis and maternal myasthenia gravis. Risk of recurrence. Rev Neurol. 147(1):62-64.

Travis, R.C., and Shaw, D.G. 1985. Congenital contractural arachnodactyly. Br J Radiol. 58:1115-1117.

Trigueros, A.P., Vazquez, V., and De Miguel, G.F. 1978. Larsen's syndrome. Report of three cases in the one family, mother and two offspring. Acta Orthop Scand. 49(6):582-588.

Trijbels, J.M.F., Berden, J.A., Monnens, L.A.H., Willems, J.L., Janssen, A.J.M., Schutgens, R.B.H., and Essen, M.V.D.B. 1983. Biochemical studies in the liver and muscle of patients with Zellweger syndrome. Pediatr Res. 17:514-517.

Tsukahara, M., and Kajii, T. 1994. Distal arthrogryposis type IIB in a girl: Autosomal recessive inheritance? Jinrui Idengaku Zasshi. 29(4):447-451.

Tsukahara, M., Sugio, Y., Kahi, Takahasbi, M, Hiroia, M., and Kato, H. 1990. Pachygyria, joint contractures, and facial abnormalities: A new lethal syndrome. J Med Genet. 27:532-535.

Tuerk, D., and Edgerton, M.T. 1975. The surgical treatment of congenital webbing (pterygium) of the popliteal area. Plast Reconstr Surg. 56(3):339-444.

Turkel, S.B., Iseri, A.L., and Fujimoto, A.O. 1980. Malformation complex. Spondylohypoplasia, arthrogryposis, and popliteal pterygium. Am J Dis Child. 134(1):42-45.

Turnbull, A.P., and Turnbull, H.R. III. 1990. Families, Professionals and Exceptionality: A Special Partnership, 2nd ed. Columbus, OH: Merrill.

Tylki Szymanska, A. 1986. Three new cases of Tel Hashomer camptodactyly syndrome in one Arabic family. Am J Med Genet. 23:759-763.

Uchida, T., Nonaka, I., Yokochi, K., and Kodama, K. 1985. Arthrogryposis multiplex congenita: histochemical study of biopsied muscles. Pediatr Neurol. 1(3):169-173.

Umbreit, J. 1983. Physical Disabilities and Health Impairments: An Introduction. New York: Merrill (Macmillan Publishing Co.).

UNESCO. 1988. Review of the Present Situation in Special Education. Paris: Unesco.

U.S. Department of Education. 1994. The Goals 2000 Educate America Act: Launching a New Era in Education. Washington, DC: Author.

U.S. Department of Education, Office of Special Education. 1994. 16th Annual Report to Congress on the Implementation of the Individuals with Disabilities Act. Washington, DC: Author.

Urich, H., and Herrick, M. Kaarsoo. 1985. The amniotic band syndrome as a cause of anencephaly. Report of a case. Acta Neuropathol (Berl). 67:190-194.

Van Allen, M.I., Curry, C., Walden, C.E., Gallagher, L., and Patton, R.M. 1987. Limb-body wall complex: II. Limb and spine defects. Am J Med Genet. 28:549-565.

Van Allen, M.I., Siegel Bartelt, J., Dixon, J., Zuker, R.M., Clarke, H.M., and Toi, A. 1992. Constriction bands and limb reduction defects in two newborns with fetal ultrasound evidence for vascular disruption. Am J Med Genet. 44:598-604.

Van Den Berghe, H., Van Eygen, M., Fryns, J.P., Tanghe, W., and Verresen, H. 1973. Partial trisomy 1, karyotype 46,XY,12,t(1q,12p)+*. Humangenetik. 18:225-230.

Van Huffe, X., Van den Hende, C., and De Moor, A. 1986. Aerobic and anaerobic metabolism of the musculus extensor carpi radialis and the musculus flexor digitorum superficialis in calves with arthrogryposis multiplex congenita (AMC) of both forelimbs. Zentralbl Veterinarmed. 33(7):551-555.

Van Huffel, X., Weyns, A., Van Nassauw, L., Cockelbergh, D., and De Moor, A. 1988. Decreased number of alpha-motoneurons in the cervical intumescence of calves with arthrogryposis multiplex congenita of both thoracic limbs. Vet Res Commun. 12(2):237-243.

Van Regemorter, N., Wilkin, P., Englert, Y., Khazen, N., Alexander, S., Rodesch, F., and Milaire, J. 1984. Lethal multiple pterygium syndrome. Am J Med Genet. 17:827-834.

Vandell, D.L., and Wilson, K.S. 1987. Infant's interactions with mother, sibling, and peer: Contrasts and relations between interaction systems. Child Development. 58:176-186.

Vanek, J., Janda, J., Amblerova, V., and Losan, F. 1986. Freeman-Sheldon syndrome: A disorder of congenital myopathic origin? J Med Genet. 23(3):231-236.

Varni, J.W., Rubenfeld, L.A., Talbot, D., and Setoguchi, Y. 1989a. Determinants of self-esteem in children with congenital/acquired limb deficiencies. Dev Behavior Pediatr. 10:13-16.

Varni, J.W., Rubenfeld, L.A., Talbot, D., and Setoguchi, Y. 1989b. Stress, social support, and depressive symptomatology in children with congenital/acquired limb deficiencies. J Pediatr Psychol. 14:515-530.

Varni, J.W., and Wallander, J.L. 1988. Pediatric chronic disabilities: Hemophilia and spina bifida as examples. In Handbook of Pediatric Psychology, ed. D.K. Routh. New York: Guilford.

Verloes, A., Dodinval, P., Retz, M.C., Schaaps, J.P., and Koulischer, L. 1991. A hydropic fetus with translucent ribs, arthrogryposis multiplex congenita and congenital myopathy: Etiological heterogeneity of A.M.C., Toriello-Bauserman type? Genet Couns. 2(1):63-66.

Verloes, A., Emonts, P., Dubois, M., Rigo, J., and Senterre, J. 1990. Paraplegia and arthrogryposis multiplex of the lower extremities after intrauterine exposure to ergotamine. J Med Genet. 27(3):213-214.

Verloes, A., Mulliex, N., Gonzales, M., Laloux, F., HermannsLe, T., Pierard, G.E., and Koulischer, L. 1993. Restrictive dermopathy, a lethal form of arthrogryposis multiplex with skin and bone dysplasias: Three new cases and review of the literature. Am J Med Genet. 47(8):1235-1237.

Vestermark, B. 1966. Arthrogryposis multiplex congenita: A case of neurogenic origin. Acta Paediatrica Scandinavica. 55:117-120.

Viljoen, D. 1994. Congenital contractural arachnodactyly (Beals syndrome). J Med Genet. 31:640-643.

Vincent, A., Newland, C., Brueton, L., Beeson, D., Riemersma, l., Huson, S.M., and Newsom-Davis, J. 1995. Arthrogryposis multiplex congenita with maternal autoantibodies specific for a fetal antigen. Lancet 346:24.

Vitale, L., Opitz, J.M., and Shahidi, N.T. 1969. Congenital and familial iron overload. N Engl J Med. 280(12):642-645.

Vogel, H., Halpert, D., and Horourpian, D.S. 1990. Hypoplasia of posterior spinal roots and dorsal spinal tracts with arthrogryposis multiplex congenita. Acta Neuropathol. 79(6):692-696.

Volpe, J.J., and Adams, R.D. 1972. Cerebrohepatorenal syndrome of Zellweger: An inherited disorder of neuronal migration. Acta Neuropath (Berl). 20:175-198.

Voorhies, T.M., Nass, R.D., and Vigorita, V.J. 1984. Arthrogryposis multiplex congenita in an infant with posterior agenesis of the corpus callosum. Brain Dev. 6(4):397-400.

Vuopala, K., Makela-Bengs, P., Suomalainen, A., Herva, R., Leisti, J., and Peltonen. 1995. Lethal congenital contracture syndrome (LCCS), a fetal anterior horn cell disease, is not linked to the SMA 5q locus. J Med Genet 32: 36-38.

Waaler, P.E., and Aarskog, D. 1980. Syndrome of hydrocephalus, costovertebral dysplasia and Sprengel anomaly with autosomal dominant inheritance. Neuropediatrics. 11(3):291-297.

Wada, H., Ryuu, A., Kito, Y., Narita, N., and Nishio, H. 1993. A case report of arthrogryposis multiplex congenita with abnormal distribution of fiber type. No To Hattatsu. 25(2):175-178.

Wagner, M., Newman, L., D'Amico, R., and et al. 1991. Youth with disabilities: How are we doing? In The First Comprehensive Report from the National Longitudinal Transition Study of Special Education Students. Menlo Park, CA: SRI International.

Wainer, S., and Vos, E.T. 1991. Congenital contractural arachnodactyly in a black African kindred. Cent Afr J Med. 37(8):262-264.

Walbaum, R. 1984. Antley-Bixler syndrome. J Pediatr. 104(5):799.

Walbaum, R., Hazard, C., and Cordier, R. 1976. Brachydactylia with symphalangism, probably autosomal recessive. Hum Genet. 33:189-192.

Walco, G.A., and Varni, J.W. 1991. Cognitive behavioral interventions for children with chronic illnesses. In Child & Adolescent Therapy: Cognitive-Behavioral Procedures, ed. P.C. Kendall. New York: Guilford.

Wallander, J.L., Pitt, L.C., and Mellins, C.A. 1990. Child functional independence and maternal psychosocial stress as risk factors threatening adaptation in mothers of physically or sensorially handicapped children. J Consult Clin Psychol. 58:818-824.

Wallander, J.L., Varni, J.W., Babani, L., Banis, H.T., and Wilcox, K.T. 1988. Children with chronic physical disorders: Maternal reports of their psychological adjustment. J Pediatr Psychol. 13:197-212.

Wallander, J.L., Varni, J.W., Babani, L., Banis, H.T., and Wilcox, K.T. 1989. Family resources as resistance factors for psychological maladjustment in chronically ill and handicapped children. J Pediatr Psychol. 14:157-173.

Warshaw, J.B. 1992. Intrauterine growth restriction revisited. Growth & Genetics & Hormones. 8(1):58.

Waterson, J.R., DiPietro, M.A., and Barr, M. 1985. Brief clinical report: Apert syndrome with frontonasal encephalocele. Am J Med Genet. 21:777-783.

Watson, G.H. 1971. Relation between side of plagiocephaly, dislocation of hip, scoliosis, bat ears, and sternomastiod tumours. Arch Dis Child. 46:203-210.

Watters, G., and Fitch, N. 1973. Familial laryngeal abductor paralysis and psychomotor retardation. Clin Genet. 4:429-433.

Weaver, D.D., and Williams, P.S. 1977. A syndrome of microcephaly, mental retardation, unusual facies, cleft palate, and weight deficiency. BDOAS. XIII(3B):69-84.

Webster's New International Dictionary of the English Language. 1959. G.&G. Merriam Co., Publishers.

Wee, A.S., Bock, H.G., and Bobo, H. 1990. Multiple pterygium syndrome: Neuromuscular findings in a case. J Miss State Med Assoc. 31(10):327-330.

Weese Mayer, D.E., Smith, K.M., Reddy, J.K., Salatsky, I., and Poznansi, A.K. 1987. Computerized tomography and ultrasound in the diagnosis of cerebrohepatorenal syndrome of Zellweger. Pediatr Radiol. 17:170-172.

Welch, J.P., and Temtamy, S.A. 1966. Hereditary contractures of the fingers (camptodactyly). J Med Genet. 3:104-112.

Wenger, F. 1977. Venezuelan equine encephalitis. Teratology. 16:359-362.

Wenner, S.M., and Shalvoy, R.M. 1989. Two-stage correction of thumb adduction contracture in Freeman-Sheldon syndrome (craniocarpotarsal dysplasia). J Hand Surg. 14(6):937-940.

West, M., Kregel, J., Zhe, M., and et al. 1993. Beyond Section 504: Satisfaction and empowerment of students with disabilities in higher education. Exceptional Children. 59(5):456-467.

Weyerts, L.K., Jones, M.C., and James, H.E. 1992. Paraplegia and congenital contractures as a consequence of intrauterine trauma. Am J Med Genet. 43:751-752.

Whitley, C.B., Thompson, T.R., Mastri, A.R., and Gorlin, R.J. 1983. Warburg syndrome: Lethal neurodysplasia with autosomal recessive inheritance. J Pediatr. 102(4):547-551.

Whittem, J.H. 1957. Congenital abnormalities in calves: Arthrogryposis and hydranencephaly. J Path Bact. 73:375.

Whittington, R.J., Glastonbury, J.R., Plant, J.W., and Barry, M.R. 1988. Congenital hydranencephaly and arthrogryposis of Corriedale sheep. Aust Vet J. 65(4):124-127.

Wiedemann, H.R., and Dibbern, H. 1980. Larsen's syndrome. Med Welt. 31(43):1548-1549.

Willems, P.J., Colpaert, C., Vaerenbergh, M., Van Thienen, M.N., Parizel, P.M., Van Marck, E., Schuerwegh, W.H., and Martin, J.J. 1993. Multiple pterygium syndrome with body asymmetry. Am J Med Genet. 47(1):106-111.

Williams, J., Cohen, D., Scolnik, B., and Zakut, C. 1978. Syndrome of camptodactyly, facial anomalies, and pulmonary hypoplasia. J Pediatr. 93(1):151-152.

Williams, P. 1978. The management of arthrogryposis. Orthop Clin N Am. 9(1):67-88.

Williams, P.E. 1985. Management of upper limb problems in arthrogryposis. Clin Orthop. (194):60-67.

Williams, P.F. 1973. The elbow in arthrogryposis. JBJS (Br). 55:834-840.

Williams, R.S., and Holmes, L.B. 1980. The syndrome of multiple ankyloses and facial anomalies: A neuropathologic analysis. Acta Neuropathol (Berl). 50:175-179.

Willis, D.J., Elliott, C.H., and Jay, S.M. 1982. Psychological effects of physical illness and its concomitants. In Handbook for the Practice of Pediatric Psychology, ed. J.M. Tuma. New York: Wiley.

Wilson, G.N., Holmes, R.G., Custer, J., Lipkowitz, J.L., Stover, J., Datta, H., and Hajra, A. 1986. Zellweger syndrome: Diagnostic assays, syndrome delineation, and potential therapy. Am J Med Gen. 24:69-82.

Winter, R.B. 1991. Congenital absence of the lumbar spine and sacrum: One-stage reconstruction with subsequent two-stage spine lengthening. J Pediatr Orthop. 11(5):666-670.

Winter, R.M., Donnai, D. and Crawford, M.D.A. 1981. Syndrome of microcephaly, microphthalmia, cataracts and joint contractures. J Med Genet. 18:129-133.

Winton, P.J., and Turnbull, A.D. 1981. Parent involvement as viewed by parents of preschool handicapped children. Topics in Early Childhood Special Education. 1:11-19.

Wlodarska Araszkiewicz, A., Araszkiewicz, H., and Chmielewski, H. 1980. Arthrogryposis multiplex congenita. Chir Narzadow Ruchu Ortop Pol. 45(3):285-289.

Wolf, L.S., and Glass, R.P. 1992. Feeding and Swallowing Disorders in Infancy. Tucson: Therapy Skill Builders.

Wrathall, A.E. 1977. Reproductive failure in the pig: Diagnosis and control. Veterinary Record. 100:230-237.

Wright, D.G. 1970. The unusual skeletal findings of the Kuskokwin syndrome. Birth Defects. 6(4):16-24.

Wyatt, S., Beach, R.C., Stuart, C., and Hallett, R.J. 1983. Cluster of cases of arthrogryposis. Lancet. 1:713.

Wyckoff, E., and Mitani, M. 1982. The spoon plate: a self-feeding device. Am J Occupational Ther. 36(5):333-335.

Wynne Davies, R. 1972. Genetic and environmental factors in the etiology of talipes equinovarus. Clin Orthop. (84):9-13.

Wynne Davies, R., Williams, P.F., and O'Connor, J.C.B. 1981. The 1960s epidemic of arthrogryposis multiplex congenita. A survey from the United Kingdom, Australia, and the United States of America. JBJS (Br). 63B(1):76-83.

Yang, M.T., Chen, C.H., Mak, S.C., Wu, K.H., and Chi, C.S. 1993. Arthrogryposis multiplex congenita: Report of a case of amyoplasia. Acta Paediatr Sin. 34(2):132-136.

Yang, S.S. 1990. ADAM sequence and innocent amniotic band: Manifestations of early amnion rupture. Am J Med Genet. 37:562-568.

Yodono, M., Taniguchi, K., Matsuki, A., and Oyama, T. 1983. Anesthesia for a patient with congenital arthrogryposis. Masui. 32(7):871-875.

Yonenobu, K., Tada, K., and Swanson, A.B. 1984. Arthrogryposis of the hand. J Pediatr Orthop. 5(4):599-603.

Yoshida, M., and Nakamura, M. 1982. Complete absence of the cerebellum with arthrogryposis multiplex congenita diagnosed by CT scan. Surg Neurol. 17(1):62-65.

Amniotic bands in connective tissue disorders. Arch Dis Child. 60:1061-1063.

Yunis, E., Fontalvo, J., and Quintero, L. 1980. X-linked Dyggve-Melchior-Clausen syndrome. Clin Genet. 18:284-290.

Zanella, B., Frenguelli, R., and Gentilini, C. 1990. A case of congenital multiple arthrogryposis. Considerations. Minerva Pediatr. 42(5):201-205.

Zeiter, J.H., and Boniuk, M. 1989. Ophthalmologic findings associated with arthrogryposis multiplex congenita: Case report and review of the literature. J Pediatr Ophthalmol Strabismus. 26(4):204-208.

Zeitoun, M.M., Ibrahim, A.H., and Hassanein, S. 1962. Arthrogryposis multiplex congenita: Report of seven cases with a review of the literature and a comment on the current concepts of aetiology. Alexandra Med J. 8(5):511-520.

Zeitune, M., Fejgin, M.D., Abramowicz, J., Aderet, N.B., and Goodman, R.M. 1988. Prenatal diagnosis of the pterygium syndrome. Prenat Diagn. 8:145-149.

Zerres, K., and Grimm, T. 1983. Genetic counseling in families with spinal muscular atrophy type Kugelberg Welander. Hum Genet. 65:24-25.

Ziegler, M. 1989. A parent's perspective: Implementing PL 99457. In Policy Implementation and PL 99457: Planning for Young Children with Special Needs, ed. J. Gallagher, P. Trohanis, and R. Clifford. Baltimore: Brookes.

Zimbler, S., and Craig, C. 1983. The arthrogrypotic foot plan of management and results of treatment. Foot Ankle. 3(4):211-219.

The term arthrogryposis describes a range of congenital contractures that lead to childhood deformities. It encompasses a number of syndromes and sporadic deformities that are rare individually but collectively are not uncommon. Yet the existing medical literature on arthrogryposis is sparse and often confusing. The aim of this book is to provide health care professionals, individuals affected with arthrogryposis, and their families with a helpful guide to better understand the condition and its therapy. With this goal in mind, the editors have taken great care to ensure that the presentation of complex clinical information is at once scientifically accurate, patient oriented, and accessible to readers without a medical background.

The book is authored primarily by members of the medical staff of the Arthrogryposis Clinic at Children's Hospital and Medical Center in Seattle, Washington, one of the leading teams in the management of the condition, and will be an invaluable resource for both health care professionals and families of affected individuals.

All cover photographs used by permission. Family photograph by David Goetze. by Ribera (1642), reproduced by permission of the Louvre Museum, Paris.

ISBN 978-1-60189-040-5
9 781601 890405

Originally published by Cambrridge University Press (1998)
Original ISBN-10: 0-521-57106-5
Dimensions: 8.5" x 11.0"

GLOBAL HELP
Health Education using Low-cost Publications
WWW.GLOBAL-HELP.ORG

CAMBRIDGE UNIVERSITY PRESS