

COMMENT. This comprehensive report on the infant with anencephaly provides medical information of importance in the analysis of social, legal, and ethical issues concerning transplantation of organs from anencephalic infants.

CONGENITAL NEUROLOGIC MALFORMATIONS

A 17-year survey of major congenital neurologic malformations among infants born in U.S. Army Hospitals worldwide from January 1, 1971 through December 31, 1987 is presented from the Neonatology Services, Walter Reed Army Medical Center, Washington, DC and Travis Grant USAF Medical Center, Travis Air Force Base, CA. From a population of 763 364 live-born and stillborn infants, 275 had anencephaly (0.36 per 1000 total births), 526 had spina bifida (0.69 per 1000), 112 had encephaloceles (0.15 per 1000), and 370 had hydrocephalus (0.48 per 1000 total births). The incidence of CNS defects among stillborn infants was 24 times greater than among live-born infants. There was a female preponderance of infants with anencephaly, spina bifida and encephalocele and a male predominance for hydrocephalus. Black infants were less likely than white infants to have spina bifida. Other congenital anomalies were associated in 20% of infants with anencephaly, 40% with encephaloceles, 37% with hydrocephalus, and 22% with spina bifida. (Wiswell TE et al. Major congenital neurologic malformations. A 17-year survey. AJDC Jan 1990; 144:61-67).

COMMENT. The racial background of the patient population in this study closely resembled that of the United States as a whole and the results may reflect those of the U.S. In the past 20 years, declines in the frequencies of anencephaly and spina bifida have been noted in many countries, particularly in the British Isles. In the present study the incidence of neural tube defects decreased only among white female infants and no etiological factor could be implicated.

CEREBELLAR VERMIS AGENESIS

The syndromes of vermal agenesis are reviewed from the Department of Pediatrics, Hopital des Enfants Malades, Paris, France. These include the Dandy-Walker syndrome and other complicated cases associated with multiple abnormalities. The Dandy-Walker syndrome consists of three abnormalities of development: 1) Partial or complete agenesis of the vermis of the cerebellum; 2) cystic formation in the posterior fossa communicating with the fourth ventricle; and 3) hydrocephalus. Enlargement of the posterior fossa and elevation of the torcular and lateral sinuses are sometimes included among the diagnostic criteria. Associated abnormalities include agenesis of the corpus callosum (7-15% of patients), occipital encephalocele (18%), cleft lip and palate, cardiac malformations, urinary tract abnormalities, and minor facial dysmorphisms. The prognosis is guarded, 75% having borderline IQ or lower, and a mortality rate of 27% in some series. Various chromosomal abnormalities have been demonstrated in a few patients but their significance is unclear. There is a 1-2% chance of recurrence in the same family. Syndromes of

agenesis of the cerebellar vermis of genetic origin are distinguished from the Dandy-Walker malformation. These include Joubert syndrome, Walker-Warburg syndrome, Meckel-Gruber syndrome, and atypical Dandy-Walker with facial angoma. Joubert syndrome includes panting respirations, abnormal eye movements, facial asymmetry and ataxia, in addition to vermian agenesis. The MRI shows an umbrella shaped fourth ventricle. Walker-Warburg syndrome includes lissencephaly, retinal abnormalities and hydrocephalus. Meckel-Gruber syndrome is characterized by occipital encephalocele, polycystic kidneys, polydactyly and hydrocephalus. Some have congenital muscular dystrophy in addition. The inheritance pattern is autosomal recessive. An MRI with median sagittal cuts is usually required in the diagnosis of partial agenesis. No reliable metabolic marker has been determined but some cases of vermian agenesis are associated with abnormal urinary excretion of succinyl-purines and pipercolic acid. Shunting operations are required when hydrocephalus develops. Operations on the posterior fossa have a high rate of failure. Prognosis depends on the occurrence of other CNS abnormalities. (Bordarier C., Aicardi J. Dandy-Walker syndrome and agenesis of the cerebellar vermis: Diagnostic problems and genetic counselling. Dev Med Child Neurol April 1990; 32:285-294).

COMMENT. In patients with agenesis of the cerebellar vermis a correct diagnosis is important in therapy, genetic counseling, and prognosis. Cases with complications which are usually autosomal recessive in inheritance and having a poor prognosis must be distinguished from the typical Dandy-Walker syndrome which is often amenable to surgical therapy.

CONGENITAL CONTRACTURAL ARACHNODACTYLY

An infant girl with arachnodactyly and spontaneously resolving contractures who died in cardiac failure is reported from the Paediatric Unit, Northern General Hospital and Department of Ophthalmology, Royal Hallamshire Hospital, Sheffield, England. In addition to the arachnodactyly the infant had dolichostenomelia, iridodonesis, and mitral and tricuspid incompetence. There was no evidence of lens subluxation on slit lamp biomicroscopy. Chromosome studies and urinary homocystine were normal. (Huggon IC et al. Contractural arachnodactyly with mitral regurgitation and iridodonesis. Arch Dis Childhood March 1990; 65:317-319).

COMMENT. Congenital contractural arachnodactyly has been described as an autosomal dominant syndrome distinct from classical Marfan's syndrome and usually unassociated with serious ocular and cardiovascular complications. This case report questions this distinction and emphasizes the importance of cardiovascular and ophthalmic assessment of patients with contractural arachnodactyly. As an editorial comment from Springfield, Illinois, I cannot omit the frequent reference to President Abraham Lincoln and Marfan's syndrome.