lower limbs is a constant feature of ataxia telangiectasia and can be taken to be one of the diagnostic characteristics. (Kwast O, Ignotowicz R. Progressive peripheral neuron degeneration in ataxia-telangiectasia: An electrophysiological study in children. Dev Med Child Neurol September 1990; 32:800-807).

COMMENT. The clinical diagnosis of ataxia-telangiectasia is based on a progressive cerebellar ataxia, ocular telangiectasia, and immunological abnormalities. Muscle weakness progresses with age and atrophy affects especially the distal leg muscles. The child is confined to a wheelchair after the 9th to the 12th year. EMG and nerve conduction studies are important in the diagnosis.

DYSTROPHIN AND DIAGNOSIS OF MUSCULAR DYSTROPHY

The value of dystrophin analysis in the early diagnosis of two patients with childhood autosomal recessive muscular dystrophy is reported from the Department of Pediatrics, Sapporo Medical College, Sapporo, Japan. The first patient, a five year old boy referred because of an elevated serum CK had developed normally until two years of age when an abnormal gait was observed. He had slight proximal muscle weakness without enlargement of calf muscles or involvement of facial muscles. Gower's sign was negative. The deep tendon reflexes in the lower limbs were slightly decreased. A muscle biopsy from the biceps showed marked variation in fiber size and a number of necrotic and regenerating fibers with proliferating connective tissue characteristic of muscular dystrophy. Dystrophin was demonstrated in all sarcolemma after immunocytochemical staining using antidystrophin antibody. The second patient, a seven year old boy with rubella, was found to have an elevated serum CK. His motor milestones of development were normal and neither muscular weakness nor atrophy were observed. There was no enlargement of calf muscles or involvement of facial muscles. Deep tendon reflexes were slightly hypoactive. The serum CK was 4250 IU. From seven to 11 years of age the clinical course was static. Muscle biopsies from the rectus femoris and biceps brachii showed a marked variation in fiber size and degenerating and regenerating fibers with proliferated connective tissue indicative of muscular dystrophy. By immunocytochemical staining with antidystrophin antibody the dystrophin was located in the sarcolemma. Both patients were diagnosed as having childhood autosomal recessive muscular dystrophy. There was neither consanguinity nor any history of neuromuscular disorder in the families. (Tachi N et al. Dystrophin analysis in the differential diagnosis of autosomal recessive muscular dystrophy of childhood and Duchenne muscular dystrophy. Pediatr Neurol July-August 1990; 6:265-268).

COMMENT. In boys with a muscular dystrophy that develops in early childhood the early differentiation of Duchenne, Becker, and the autosomal recessive limb-girdle dystrophies is important. The onset of limb-girdle dystrophy is more commonly between 10 and 20 years of age but it may present in the first decade and sometimes as early as two years of age. The course is variable but occasionally progression is as rapid as with Duchenne muscular dystrophy.