

COMMENT. In typical cases of LHON, males predominate and the fundus is characterized by circumpapillary telangiectatic microangiopathy and pseudoedema. The diagnosis should also be considered in females with an unexplained optic neuropathy, a negative family history and a normal fundus. Genetic analysis has allowed for a broader view of what constitutes the clinical phenotype of LHON. (Newman NJ. Leber's hereditary optic neuropathy. New genetic considerations. Arch Neurol May 1993; 50: 540-548).

CONGENITAL MALFORMATIONS

CORPUS CALLOSUM AGENESIS AND OSSEOUS LESIONS

A new mental retardation syndrome with agenesis of the corpus callosum and unusual bone changes is reported from the Departments of Radiology and Neurology, Royal Alexandra Hospital for Children, Camperdown, Sydney, NSW, Australia. At 11 months of age, the boy was functioning at a 5 month level, and his head circumference was at the 98th centile. His face was triangular in shape, with a broad frontal region. There was mild hypotonia, and deep tendon reflexes were exaggerated. Skeletal abnormalities included multiple Wormian bones, thin ribs, small iliac bodies, and retarded bone age. (Kozlowski K, Ouvrier RA. Agnesis of the corpus callosum with mental retardation and osseous lesions. Am J Med Genet May 1993; 48: 6-9). (Reprints: Dr K Kozlowski, RAHC, Camperdown 2050, NSW, Australia).

COMMENT. Macrocephaly and polydactyly, in Schinzel syndrome, and microcephaly and camptodactyly, in da-Silva syndrome, are additional examples of skeletal abnormalities associated with corpus callosum agenesis or hypoplasia. (see Progress in Pediatric Neurology, Millichap JG, Ed, Chicago, PNB Publishers, 1991, pp 310-312).

CONGENITAL BRACHIAL ARTHROGRYPOSIS

Two patients with congenital cervical spinal muscular atrophy and arthrogryposis limited to the upper limbs are reported from the Hospital Enschede, The Netherlands, and University Hospital RWTH Aachen, Germany. A girl, aged 1 year, born with congenital arthrogryposis multiplex, had severe bilateral hypotonia, weakness and wasting of shoulder girdles, arms and hands. Tendon reflexes were absent in the upper limbs. EMG of the triceps was silent, and a biopsy showed severe neurogenic muscle atrophy. A 31 year old man, born with severe muscle weakness and congenital contractures of the upper limbs, had herniations of the intervertebral discs C2-C4 and C5-C7 but normal signal intensity of the cord on T2-weighted images of the MRI. Both patients had normal sensory modalities, and lower limbs were not involved. (Hageman G et al. Congenital cervical spinal muscular atrophy: a non-familial,