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. Agenesis 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,

non progressive condition of the upper limbs. J Neurol Neurosurg Psychiatry April 1993; 56: 365-368). (Respond: Dr G Hageman, Department of Neurology, Medical Spectrum Twente, Hospital Enschede, PO Box 50000, 7500 KA Enschede, The Netherlands).

COMMENT. The absence of sensory deficits and normal motor nerve conduction velocities point to involvement of the anterior horn cells. The etiology of the prenatal pathology with loss of anterior horn cells in cases of neurogenic arthrogryposis is unknown, except for one case cited of rubella exposure. The limitation to the upper limbs is unusual.

METABOLIC AND TOXIC DISORDERS

SYNDROMES OF 3-METHYLGLUTACONIC ACIDURIA

The most common clinical syndromes associated with 3-methylglutaconic (MGC) aciduria are reviewed by researchers from various centers; Courtwright and Summers Metabolic Disease Center and Baylor Research Institute, Dallas, TX; Shaare Zedek Medical Center, Jerusalem; Free University of Amsterdam; Loewenstein Hospital, Tel-Aviv Univ, Raanana, Israel; and Kennedy Krieger Institute, Baltimore, MD. Three distinct syndromes are described: *Type I 3-MG-CoA Hydratase Deficiency.*- (autosomal recessive) 3 patients had delayed speech and macrocephaly, increased urinary excretion of 3-MGC acid, 3-M Glutaric(MGR) and 3-hydroxyisovaleric acids, elevated CPK and serum carnitine, and hypoglycemia. Urinary 3-MGC excretion is decreased by restriction of L-leucine intake. *Type II Barth Syndrome.*- (X-linked) dilated cardiomyopathy, recurrent infection, neutropenia, growth retardation. Improves with age. Increased urinary excretion of 3-MGC, 3-MGR, fumaric, and 2-ethylhydracrylic acids. *Type III Costeff Optic Atrophy Syndrome.*- (autosomal recessive) A Behr-like syndrome described in 39 patients of Iraqi-Jewish origin living in Israel. Optic atrophy, choreoathetosis, spastic paraparesis, cerebellar ataxia. Nonprogressive. 3-MGC and 3-MGR aciduria. An *Unclassified 3-MGC aciduria* includes patients presenting in the first year of life with neurologic impairment, seizures, retinal pathology, and a fatal course. Cardiomyopathy, hepatic dysfunction, hypoglycemia, lactic acidosis, and dysmorphia are associated manifestations. Defects of the mitochondrial respiratory chain are suggested. (Gibson KM et al. Multiple syndromes of 3-methylglutaconic aciduria. Pediatr Neurol March/April 1993; 9: 120-123). (Respond: Dr Gibson, Baylor Research Institute, 3812 Elm Street, Dallas, TX 75226).

COMMENT. This review will assist the neurologist in the differential diagnosis of patients with 3-MGC aciduria. Some cases are multisystemic and progressive, with onset from birth to several years, some are primarily neurologic and non-progressive, while others involve the heart muscle and may improve with age.