acidosis is described in two forms following a retrospective study of 16 patients at the University of Nijmegen, The Netherlands. Four patients with the fatal form died from hypertrophic obstructive cardiomyopathy in the neonatal period. In those with the relatively benign form, cardiomyopathy developed late, causing death in 7 patients at a median age of 23 years. The prognosis in patients without subvalvular aortic stenosis is dependent on the metabolic function of skeletal muscle. It varies between moderate exercise intolerance and wheelchair existence. In both forms, bilateral cataracts, lactacidemia and mitochondrial myopathy are present from birth. If a propositus has an affected sib, he will probably suffer from the same form, and genetic counselling is important. (van Ekeren GJ et al. A retrospective study of patients with the hereditary syndrome of congenital cataract, mitochondrial myopathy of heart and skeletal muscle and lactic acidosis. Eur J Pediatr 1993; 152(3): 255-259). (Respond: Dr RCA Sengers, Dept of Paediatrics, University of Nijmegen, PO Box 9101, NL-6500 HB Nijmegen, The Netherlands).

> **COMMENT.** Cataracts present at birth are the first manifestation of the syndrome in most patients. Some present with muscular hypotonia, and others with cardiac symptoms. The metabolic cause is unknown. Skeletal muscle biopsy showed abnormal mitochondrial structure or number with matrix vesicles, and the sarcoplasm contained large quantities of lipid or glycogen. Morphometric analysis demonstrated an increased volume density of subsarcolemmal mitochondria, a possible compensatory mechanism for a deficit in energy production. The findings were similar in the fatal and benign forms. Mitochondrial function showed no abnormalities. Qualitative and quatitative defects in mitochondrial DNA in infants with fatal metabolic disorders are discussed by Moraes CT (Int Pediatr 1993; 8: 40).

ATYPICAL LEBER'S OPTIC NEUROPATHY

The diagnosis of Leber's hereditary optic neuropathy (LHON) in 6 female atypical cases seen at Harvard, Emory, and Johns Hopkins Medical Centers required molecular analysis and demonstration of the 11778 mitochondrial DNA mutation for accurate confirmation. Features atypical for LHON included a negative family history, a normal fundus, bitemporal hemianopia, optic disc cupping, and premonitory episodes of transient monocular visual loss. Molecular analysis allows accurate identification of most cases. The 11778 point mutation accounts for 50%. (Weiner NC et al. Atypical Leber's hereditary optic neuropathy with molecular confirmation. Arch Neurol May 1993; 50: 470-473). (Reprints: Nancy J Newman MD, Neuro-ophthalmology Unit, Emory Eye Center, 1327 Clifton Rd NE, Atlanta, GA 30322).

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,