

COMMENT. Hyperinsulinism in infancy causes recurrent episodes of profound hypoglycemia, often <1 mmol/L, with diverse neurologic sequelae, including psychomotor retardation, learning disability, seizures, and microcephaly. Neonatal onset is associated with more complications and a greater need for surgical treatment. Early diagnosis and onset in infancy result in a slightly better prognosis.

In contrast to the frequent risk of mental impairment in neonates and infants with hyperinsulinemic hypoglycemia, young adults with insulin dependent diabetes and recurrent episodes of hypoglycemia have either a mild or negligible risk of cognitive impairment. (Deary IJ, Frier BM. BMJ 1996;313:767-8).

Offspring of diabetic Japanese mothers are at increased risk of lowered IQ scores at 3 years of age (Yamashita Y et al. Acta Paediatr 1996;85:1192-6). The risk of hypoglycemic brain damage is related inversely to the age of the patient. (See Progress in Pediatric Neurology III, 1997;p311).

MITOCHONDRIAL ENCEPHALOMYOPATHIES: INCIDENCE & DNA

The incidence, mortality, clinical features and DNA abnormalities of mitochondrial encephalomyopathies (ME) were evaluated in a population-based study of children from western Sweden conducted at The Queen Silvia Children's Hospital, Goteborg, Sweden. Thirty two patients under 16 years of age were diagnosed from 1984-1998. The incidence of ME in preschool children was 1 out of 11000, and the point prevalence in children <16 years of age was 1 out of 21000. Leigh's syndrome occurred in 1/32000 preschoolers, and Alper's syndrome and cytochrome C oxidase deficiency in 1/51000. Infantile onset of ME was frequent, the course severe, and mortality high. Patients with infantile onset ME had a median survival of 12 years. Complex I and IV deficiencies were the most common biochemical defects. The spectrum of disorders in children was different from that reported in adult hospital-based patients. Encephalopathies were more frequent in children, and mtDNA mutations were identified less frequently. Mitochondrial DNA point mutations, DNA deletions, and nuclear mutations in the SURF1 gene were identified in 4, 2, and 2 cases, respectively. (Darin N, Oldfors A, Moslemi A-R, Holme E, Tulinius M. The incidence of mitochondrial encephalomyopathies in childhood: Clinical features and morphological, biochemical, and DNA abnormalities. Ann Neurol March 2001;49:377-383). (Respond: Dr Niklas Darin, The Queen Silvia Children's Hospital, Department of Pediatrics, Sahlgrenska University Hospital-East, Goteborg University, S-416 85 Goteborg, Sweden).

COMMENT. The authors conclude that mitochondrial encephalomyopathies are relatively common neurometabolic disorders in childhood.

Among 51 patients with mitochondrial respiratory chain disease analyzed at the University of Newcastle upon Tyne, UK (Jackson MJ, Bindoff LA et al. Brain 1995;118:339-357), presenting symptoms in order of frequency included ptosis and ophthalmoplegia (20), lactic acidosis (10), seizures (6), myopathy (6), failure to thrive (6), and ataxia (5). The most useful confirmatory diagnostic test was histochemical analysis of muscle, and elevated CSF lactate was a good indicator of mitochondrial encephalopathy. (see Progress in Pediatric Neurology III, 1997;pp542-3).