Biochemical findings have included a reduction of brain noradrenaline, dopamine and seratonin, but no consistent abnormalities have been found. A genetic basis for Rett syndrome has been suggested but not satisfactorily confirmed.

MUSCLE DISORDERS

MITOCHONDRIAL MYOPATHY AND CARDIOMYOPATHY

Two siblings with infantile lactic acidosis and mitochondrial myopathy are reported from the Department of Pediatrics, Goteborg University; Ostra Hospital; Goteborg, Sweden. The first child, a girl, appeared healthy during the first four months of life. She was admitted at five months of age with feeding difficulties, vomiting and weight loss and muscular hypotonia. Her serum lactate concentration rose to 20 mmol/L (n:0.8-1.8 mmol/L), she developed edema, became comatose and died of circulatory failure eight days after admission. At autopsy, the heart was slightly enlarged and the pleurae and pericardium showed clear yellowish fluid. The second patient, the younger brother of patient one, had congenital lactic acidosis but no other symptoms until six months of age when he developed progressive muscle weakness. Treatment with dichloroacetate lowered the serum lactic acid level but did not affect his clinical condition. Cardiomyopathy was diagnosed at 13 months of age and he died of circulatory failure at 29 months. Both patients had mitochondrial myopathy with changes in skeletal muscle and the myocardium. Biochemical investigations of skeletal muscle mitochondria showed deficiencies in cytochrome c oxidase and NADH ferricyanide reductase. (Tulinius MH et al. Mitochondrial myopathy and cardiomyopathy in siblings. Pediatr Neurol May/June 1989; 5:182-188).

COMMENT. Patients with mitochondrial myopathies or cytopathies show marked heterogeneity in clinical manifestations and system involvement. Two major variants of mitochondrial myopathy and cytochrome c oxidase deficiency in infancy have been described. Most cases are rapidly progressive and fatal and are associated with renal dysfunction; occasionally the course is milder and reversible. In the present study, the heterogeneity in the mitochondrial cytochrome c oxidase activity provided clinical symptoms in proportion to the fraction of damaged mitochondria, thus explaining the different clinical course in the siblings.

CYTOCHROME C OXIDASE DEFICIENCY AND RESPIRATORY DISTRESS

A newborn male presenting with severe respiratory insufficiency, generalized muscle weakness, and lactic acidemia is reported from the Department of Pediatrics, Nagasaki University School of Medicine, Japan. Within 27 hours after birth he was markedly hypotonic, spontaneous movements and the Moro reflex were almost absent. A respirator was necessary because of respiratory arrest and he died 75 hours after birth. At autopsy there was variation in muscle fiber size and an increased number of Type 2C fibers but no ragged-red fibers by Gomori trichrome staining. Biochemical and histochemical studies showed cytochrome c oxidase activity

was decreased in skeletal muscle but not in cardiac muscle. (Takayanagi, T et al. Infantile cytochromic c oxidase deficiency with neonatal death. Pediatr Neurol May/June 1989; 5:179-81).

COMMENT. The differential diagnosis of neonatal respiratory distress syndrome should include mitochondrial myopathy. The diagnosis should still be considered even in the absence of ragged-red fibers in skeletal muscle.

Two further papers concerning mitochondrial myopathy appeared in the June 1989 issue of the Annals of Neurology. Shimozumi H et al established cultured myogenic cell lines that were defective in cytochrome c oxidase enzyme from a patient with mitochondrial encephalomyelopathy. Two kinds of myogenic cell lines, one with and one without defective enzymatic activity were demonstrated showing that a partial enzyme defect is the result of the cellular mosaicism in the tissue. The authors comment that these cloned cell lines provide an excellent system for clarifying the cause of mitochondrial myopathy and for investigating the genetic factors.

Sakuta R and Nonaka I examined muscle taken at biopsy in six patients with complex I deficiency and one patient with the clinical characteristics of mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes (MELAS). Striking abnormalities in the blood vessels were shown by electron microscopy in five patients. The authors considered that these abnormalities in small arteries might be responsible for the occasional occurence of transient cerebral ischemia causing stroke-like episodes and progressive mental deterioration in patients with mitochondrial myopathy.

MALFORMATIONS

TETHERED SPINAL CORDS

The diagnosis of tethered spinal cord by MRI in seven children with cutaneous lumbar hemangioma is reported from the Children's Hospital of Pittsburgh, University of Pittsburgh School of Medicine. The hemangiomas ranged in size from 4 x 6 cm to 8 x 20 cm and all overlapped the midline. All demonstrated tethered cords; four showed intraspinal lipomas, and two showed tight fila terminale. At surgery, all infants were found to have tethered cords and none had an intraspinal hemangioma. All patients were neurologically normal both pre and postoperatively. (Albright AL et al. Lumbar cutaneous hemangiomas as indicators of tethered spinal cords. Pediatrics June 1989; 83:977-980).

COMMENT. The lumbar hemangiomas in these patients were large and the significance of small lesions is not known. Despite a normal neurological examination, infants or children with large lumbar cutaneous hemangiomas should be suspected of having tethered cords and magnetic resonance imaging should be obtained. If neurologic deficits