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