at the University of Pittsburgh School of Medicine, PA. Of 3 patients with neonatal-onset congenital MD, 11 with childhood-onset MD, and 16 with early adult-onset MD (limb-girdle MD), only one, a 16-year-old African-American girl with childhood-onset MD, had adhalin gene mutations. All patients had autosomal recessive inheritance patterns, and all had serum CK levels higher than 1000 IU/liter. (Ljunggren A et al. Primary adhalin deficiency as a cause of muscular dystrophy in patients with normal dystrophin. Ann Neurol September 1995;38:367-372). (Respond: Dr Eric P Hoffman, BST W1211, University of Pittsburgh School of Medicine, Pittsburgh, PA 15261).

COMMENT. Primary adhalin deficiency in patients with muscular dystrophy with normal dystrophin is a relatively rare occurrence. It is not restricted to French families in which it was first reported. The phenotype is consistent with childhood-onset muscular dystrophy.

Approximately 60% of MD patients show absence or deficiency of dystrophin. Of the remaining 40% with normal dystrophin, most have the genetically heterogeneous severe childhood form of autosomal recessive MD, or limb-girdle dystrophy, and 1 in 30 may have a primary adhalin deficiency.

METABOLIC DISORDERS

CARNITINE PT II DEFICIENCY AND CEREBRAL DYSGENESIS

A newborn female infant with neonatal lethal multiorgan carnitine palmitoyltransferase II (CPT II) defiency is reported from the Departments of Medicine and Pathology, Children's Hospital, Boston, MA; and the Department of Pharmacology and Medicine, Case Western Reserve University, VA Medical Center, Cleveland, OH. The infant was referred at 4 days of age because of hyperammonemia and seizures. Pregnancy was complicated by oligohydramnios. Ultrasound on day 1 showed enlarged kidneys with cortical cysts. Cranial ultrasound revealed a periventricular cyst. On day 3, seizures with prolonged apnea were followed by unresponsiveness. Pupils were fixed and dilated. Dysmorphic features included microcephaly, long fingers and toes, extra digital creases, and joint contractures. The liver enlarged, and the infant died at 10 days with cardiac and renal failure. Long-chain acylcarnitines were elevated in blood and multiple tissues, and lipid accumulations and deficiency of CPT II activity were found in heart, liver, muscle, and kidney tissue. (North KN et al. Lethal neonatal deficiency of carnitine palmitoyltransferase II associated with dysgenesis of the brain and kidneys. J Pediatr September 1995;127:414-420). (Reprints: Kathryn N North MD, Department of Neurology, Children's Hospital, Bridge Road, Camperdown, New South Wales 2050, Australia).

COMMENT. The authors cite two previous reports of families with neonatal CPT II deficiency associated with multiple malformations. Autopsy findings include diffuse lipid accumulation, cardiomegaly, dysplastic kidneys, and brain migration defects. They conclude that this metabolic disorder should be included in the differential diagnosis of neonates dying with dysmorphism and multiple organ malformations, along with Zellweger syndrome, other disorders of peroxisomal B-oxidation, and glutaric acidemia type II.