the ability of cultured fibroblasts to esterify exogenously supplied cholesterol. This deficiency may be assayed in confirmation of the diagnosis when presentation is atypical. (Fink JK et al. Clinical spectrum of Niemann-Pick disease type C. Neurology August 1989; 39: 1040-1049.

COMMENT. Mild intellectual impairment presenting as poor school performance was the most common initial neurologic abnormality. Additional presenting signs included ataxia, dysarthria, and impaired vertical gaze. Within three years of the initial deficit most of the patients had cognitive impairment, abnormal vertical gaze and ataxia. Saccadic paresis was manifested by a complaint of difficulty in reading or in descending stairs. Hepatosplenomegaly was first noted at varying ages from birth to 24 years with a mean age of six years. It preceded neurological abnormalities in one-half the patients and was found only in the early onset rapidly progressive group.

MITOCHONDRIAL ALTERATIONS IN RETT SYNDROME

Muscle biopsy findings in two patients with Rett syndrome are reported from the Departments of Obstetrics/Gynecology, Pediatrics, and Pathology, Medical College of Ohio, Toledo, OH. Muscle biopsy was performed at 32 months of age and at 3 years 7 months of age. Light microscopy revealed fibers of uniform size with normal histochemistry. Electron microscopy revealed mitochondrial alterations including distention, vacuolation, and membranous changes. (Ruch A. Mitochondrial alterations in Rett syndrome. Pediatr Neurol Sept/Oct 1989; 5:320-3).

COMMENT. Abnormal mitochondria have been reported previously in the muscle biopsies of two patients with Rett syndrome (Eeg-Olofsson O et al. Brain Dev 1988; 10:260). The findings presented in patients with Rett syndrome did not correspond to the typical "ragged red" fibers found in mitochondrial myopathies. There are no biochemical or pathological findings specific to Rett syndrome but further studies of mitochondrial functioning in muscle may be warranted.

INFANTILE MITOCHONDRIAL DISEASE

A detailed clinical, pathologic, biochemical, and genetic analysis of a case of lethal infantile mitochondrial disease is reported from the Departments of Biochemistry, Pediatrics, Neurology and Nephrology, Emory University School of Medicine, Atlanta, GA. During the first three months of life the child showed increasing lethargy, hypotonia, difficulty in feeding and growth retardation. On admission at three months of age there was respiratory failure, bradycardia, hypotension, and severe lactic acidosis. Over the next 21 days the condition rapidly deteriorated with a progressive hypertrophic cardiomyopathy, hepatic dysfunction, and generalized seizure activity. The patient died with bradycardia and hypotension at four months of age. There were abnormalities in the striated muscles, smooth muscle, heart and liver but not in the central nervous system. Biochemical analysis revealed a combined complex I and IV

deficiency in skeletal muscle, heart and liver but not in kidney and brain. There was no abnormality in mitochondrial DNA. The disease was thought to result from a nuclear oxidative phosphorylation gene mutation. (Zheng X et al. Evidence in a lethal infantile mitochondrial disease for a nuclear mutation affecting respiratory complexes I and IV. Neurology Sept 1989; 39:1203-1209).

COMMENT. Mitochondrial encephalomyopathies attributed to mutations in the mitochondrial DNA include MERRF and Kearns-Sayre syndrome with onset in childhood through adulthood. In the neonatal period some mitochondrial myopathies have a benign course and some are lethal and a variety of oxidative phosphorylation deficiencies have been associated with these disorders.

PROGRESSIVE SPASTIC CEREBRAL ATAXIA

A syndrome of diabetes insipidus followed by progressive spastic cerebellar ataxia is reported in four boys from the Departments of Neurology, Pediatrics and Psychiatry, UCLA School of Medicine, Los Angeles, CA. In two patients central nervous system histiocytosis was detected. CT scan showed bilateral calcification of the cerebellar dentate nuclei and multiple hypodense areas in the skull; a biopsy confirmed the diagnosis of histiocytosis. A trial of Prednisone was beneficial. (Birnbaum DC et al. Idiopathic central diabetes insipidus followed by progressive spastic cerebral ataxia. Arch Neurol September 1989; 46:1001-1003).

COMMENT. Each of these patients developed idiopathic central diabetes insipidus between the ages of two and six years and all responded to intranasal Desmopressin. Spastic cerebellar ataxia developed eight to ten years later. Histiocytosis accounts for 8-16% of cases of diabetes insipidus in children. Patients with this syndrome may benefit from treatment with corticosteroids.

SEIZURE DISORDERS

NEONATAL SEIZURES

The current concepts and revised classification of neonatal seizures are the subject of a special article from the Division of Pediatric Neurology, Washington University School of Medicine, St. Louis, MO. The clinical classification includes the following: 1) subtle, 2) clonic (focal, multifocal), 3) tonic (focal, generalized), and 4) myoclonic (focal, multifocal, generalized). The focal and multifocal clonic, focal tonic, and generalized myoclonic seizures are commonly associated with simultaneous electrographic seizures. Some varieties of subtle seizures have simultaneous EEG seizure discharges. Neonatal seizures not usually accompanied by EEG seizure activity include: certain subtle seizures, most generalized tonic seizures, and the focal and multifocal myoclonic seizures. These may represent "brain stem release phenomena" and may occur in infants with hydranencephaly and anencephaly. The absence of EEG seizure activity does not rule out an epileptic origin for a clinical