heterogeneity of the molecular lesion in McArdle's disease. (Servidei S, DiMauro S et al. McArdle's disease: biochemical and molecular genetic studies. Ann Neurol Dec 1988;24:774-781).

COMMENT. McArdle's disease (muscle phosphorylase deficiency; glycogenosis type V) is manifested by exercise intolerance with myalgia, early fatigue, and muscle stiffness relieved by rest. Strenuous exercise is accompanied by acute muscle necrosis and myoglobinuria. Patients presenting in infancy or childhood may have a mild congenital muscle weakness, tiredness or poor stamina without cramps or myoglobinuria, or severe, rapidly progressive weakness soon after birth that results in respiratory failure and death in infancy. The various types of myophosphorylase protein and messenger RNA observed in the above patient population were consistent with at least 5 different mutations that give rise to McArdle's disease.

NEMALINE MYOPATHY

A boy, 5 years of age, with nemaline myopathy complicated by respiratory failure and hypertrophic cardiomyopathy is reported from the Albany Medical College, Albany, NY. He presented at 2 mos of age with failure-to-thrive, diminished suck, and hypotonia. CK was normal and EMG showed rare fibrillations and fasciculations. Muscle biopsy demonstrated variation in fiber size and electron-dense nemaline rods. He walked late at 3 yrs, fell frequently and required a walker outdoors. At 5 1/2 yrs, during an upper respiratory tract infection, respiratory distress necessitated intubation. Neurologic examination revealed hypotonia, proximal muscle weakness, mild facial weakness, absent deep tendon reflexes. Echocardiography disclosed a thickened ventricular septum consistent with hypertrophic cardiomyopathy. Because of chronic nocturnal hypoventilation, tracheostomy and assisted ventilation were required. The authors recommend routine cardiac and pulmonary function evaluations in patients with nemaline myopathy. (Van Antwerpen CL et al. Nemaline myopathy associated with hypertrophic cardiomyopathy. Pediatr Neurol Oct 1988;4:306-8).

COMMENT. Sleep hypoventilation, a rare complication of Nemaline myopathy, has been attributed to central nervous system CO2 unresponsiveness. Cardiomyopathy has not been reported previously in a child with nemaline myopathy and the authors found only 2 other references, both in adults. Neurologic conditions associated with hypertrophic cardiomyopathy include Leigh disease, Kearn-Sayre syndrome, Friedreich ataxia, neurofibromatosis, and Pompe disease.