poor-risk medulloblastoma/primitive neuroectodermal tumors (MB/PNET) has been studied at the Children's Hospital of Philadelphia, University of Pennsylvania, PA. Chemotherapy consisted of vincristine during concomitant craniospinal radiation therapy and eight 6-week cycles of vincristine, cis-platinum, and cyclohexylnitrosourea. Twenty five of 26 children (96%) treated remain alive and free of disease at a median of 24 months from diagnosis (range 6-50 mos). Actuarial disease-free survival was statistically significantly better than for control subjects who had received radiation therapy alone during an 8 year period prior to the use of adjuvant chemotherapy. The 2-year disease-free survival was 96% for patients on the protocol of adjuvant chemotherapy as compared to 59% for historical control patients treated with radiotherapy alone. (Packer RJ et al. Efficacy of adjuvant chemotherapy for patients with poor-risk medulloblastoma: a preliminary report. Ann Neurol Oct 1988;24:503-508).

**COMMENT.** Surgical excision for the treatment of medulloblastoma has less than a 6 month survival rate, and postoperative radiation of the tumor bed alone has minimal benefit. Irradiation of the tumor bed and the entire neuroaxis provides an overall chance of 30% to 50% for 5 year survival. Patients with clinical evidence of arachnoidal seeding or with gross evidence of seeding at surgery do poorly. Survival without evidence of recurrent tumor for a period exceeding the patient's age at diagnosis plus 9 months indicates a probably cure ("Collins' law"). Corticosteroids result in remarkable amelioration of root signs and symptoms and reduction of edema for a brief period. Experience with chemotherapy in the past 25 years has produced better survival rates, and used as an adjuvant these agents are indicated for the poor-risk patients, that is, those less than 5 years of age at diagnosis or with disseminated tumors (Chang stages M1 - M3). (Groover RV. In The Practice of Pediatric Neurology, Eds. Swaiman KF, Wright FS, CV Mosby, St. Louis, 1982.)

## MUSCLE DISORDERS

### McARDLE'S DISEASE

Muscle biopsy specimens from 48 patients with biochemically proven phosphorylase deficiency (McArdle's disease) have been analyzed by gel electrophoreis (SCS-PAGE), immunoblotting, and immunotitration (ELISA) at Columbia University College of Physicians and Surgeons, New York, NY. The majority had no detectable enzyme protein, 6 had markedly decreased phosphorylase protein, and only 1 had a normal amount of protein. The presence or absence of enzyme protein was not correlated with the clinical presentation or muscle glycogen concentration. In 4 patients tested, messenger RNA was normal in 2, abnormally short in 1, and absent in 1, suggesting

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.