COMMENT. This is the first report of a familial multigenerational case of Glut-1 deficiency syndrome. De Vivo et al (1991) first described Glut-1 DS in 2 children and subsequently reported 20 patients with various mutations. Symptoms present in the first year, with delayed motor and mental development, worsened by fasting, seizures refractory to antiepileptic drugs (AED) and aggravated by phenobarbital, and hypoglycorrhachia. A spinal tap and CSF glucose determination are important in a diagnostic workup of infants and young children with AED-resistant seizures, developmental delay, and ataxia. A decreased erythrocyte 3-OMG uptake and demonstration of heterozygous GLUT-1 mutations confirm the diagnosis. Barbiturates and methylxanthines, found to aggravate the seizures, should be avoided, and the ketogenic diet is used in treatment, at least in early childhood. Patients may continue to suffer from episodic limpness, ataxia, and confusion.

RESPIRATORY FAILURE IN ACID MALTASE DEFICIENCY

Sleep-disordered breathing (SDB) and respiratory failure (RF) were studied in 27 patients with juvenile and adult acid maltase deficiency (AMD) and compared with polysomnography outcomes at the University of Essen, Germany. Ventilatory restriction was present in 17/27 patients, and inspiratory vital capacity correlated with peak inspiratory muscle pressure and gas exchange by day and night. Diaphragm weakness occurred in 13/27, and was associated with SDB and RF. SDB was characterized by REM-sleep hypopneas and nocturnal hypoventilation. Treatment of RF or hypoventilation with noninvasive ventilation corrected daytime and nocturnal gas exchange. (Mellies U, Ragette R, Schwake C et al. Sleep-disordered breathing and respiratory failure in acid maltase deficiency. Neurology October (1 of 2) 2001;57:1290-1295). (Reprints: Dr Uwe Mellies, Department of Pneumology/Sleep Medicine, Ruhrlandklinik, Tuschener Weg 40, D-45239 Essen, Germany).

COMMENT. Acid maltase deficiency (AMD), Type II glycogenosis or Pompe's disease, a rare hereditary myopathy, is an autosomal recessive glycogen storage disease that is complicated by heart, CNS and skeletal muscle dysfunction. It presents in childhood or in adults, and is slowly progressive, leading to respiratory failure and obstructive sleep apnea which may be fatal. In infants, acid maltase enzyme is deficient in lysosomes of heart, liver, and skeletal muscle, and glycogen is deposited in every tissue, including the CNS. First symptoms often appear by the second month and include difficulty in feeding, dyspnea, muscle weakness, and cardiac dysfunction, with marked cardiac enlargement. In late childhood and adult onset cases, organomegaly is absent, and muscle weakness is only slowly or nonprogressive, with involvement of lower limb proximal muscles. (Menkes JH, Textbook of Child Neurology, Lea & Febiger, 1980). The above paper stresses the role of diaphragm weakness as the major cause of respiratory failure in juvenile and adult cases of AMD, and the value of noninvasive ventilation in treatment of associated respiratory disorders.

DEGENERATIVE DISORDERS

IRON METABOLISM AND HALLERVORDEN-SPATZ SYNDROME

Hallervorden-Spatz syndrome (HSS) and iron metabolism are reviewed in a scientific workshop sponsored by the NIH and HSS Association in Bethesda, MD. First reported in 1922, the syndrome is now classified in 3 clinical types: 1) early-onset childhood types, rapidly or slowly progressive; 2) late-onset, 10-18 years of age, slowly progressive; and 3) adult type, slowly progressive. Obligate