Canada).

COMMENT. Idiopathic intracranial hypertension is not "benign" and is associated with significant morbidity and short- or long-term symptoms, often resistant to therapy. None of the proposed therapies is of proven efficacy, and this extensive case series and review emphasizes the need for prospective studies.

NEUROMUSCULAR DISORDERS

PRESYNAPTIC CONGENITAL MYASTHENIC SYNDROME

Three patients (ages 7, 9, and 14 years) with a new form of presynaptic congenital myasthenic syndrome (CMS) are reported from the University of California, Davis; University of Minnesota; and the University of Chicago. This CMS was characterized by decreased quantal release with normal amplitude miniature end-plate potentials (MEPP), normal size of nerve terminals, and normal number of synaptic vesicles. Symptoms had presented with scoliosis at age 5 years in patient 1, delay in walking and easy fatigability at 17 months in patient 2, and as an infant with hypotonia and motor developmental delay in patient 3. Clinical findings included muscle weakness and fatigability, respiratory crisis, nystagmus (1 case), bulbar deficit, scoliosis - severe in one patient, and mild ataxia. No patient had ophthalmoplegia or mental delay. A similar disorder was reported in a close relative of patient 2, but patients 1 and 3 had no family history of neurologic disease. Electrodiagnostic evidence of abnormal neuromuscular transmission was obtained in all patients. Intracellular microelectrode studies showed a dramatic reduction of the endplate potentials (EPP) quantal content, indicative of presynaptic failure. Screening of reported pathogenic mutations in the CACNA1A and a mutational analysis of AChR subunit genes were negative. Treatment with prednisone and pyridostigmine was ineffective, while a combination of pyridostigmine and 3,4-diaminopyridine reduced the frequency of respiratory crises and resulted in improved muscle strength and exercise endurance in one patient. The deficiency of quantal release of neurotransmitter underlying this form of presynaptic CMS may be explained by an abnormal calcium metabolism or impaired endocytosis and recycling of synaptic vesicles. (Maselli RA, Kong DZ, Bowe CM et al. Presynaptic congenital myasthenic syndrome due to quantal release deficiency. Neurology July (2 of 2) 2001;57:279-289). (Reprints: Dr Ricardo A Maselli, UC Davis, 1515 Newton Ct, Rm 510, Davis, CA 95616).

COMMENT. Presynaptic congenital myasthenic syndrome (CMS) results from a deficiency in release of neurotransmitter from the nerve terminal. Familial infantile myasthenia (FIM) and a CMS associated with paucity of synaptic vesicles (PSV) have been fully described, and some additional isolated cases of presumed CMS have been reported. The molecular genetic defect for CMS has not been elucidated. In the 3 cases reported here due to quantal release deficiency, involvement of CACNA1A mutations is considered most likely and deserves further evaluation.

For further review of various types of congenital myasthenic syndromes, see Engel AG et al. 1993; and Progress in Pediatric Neurology III, 1997;pp346-7.

RECOVERY FOLLOWING NEONATAL BRACHIAL PLEXUS PALSY

The value of detailed strength testing monthly, up to 6 months of age, in predicting complete recovery was determined in a prospective study of 80 infants with brachial plexus injury followed at the Brachial Plexus Palsy Center, St Louis