ORAL-MOTOR FUNCTION IN RETT SYNDROME

The communication skills, oral-motor function and respiration patterns of 20 girls with Rett syndrome were studied at the Crippled Childrens Division, Oregon Health Sciences University, Portland, OR. All patients showed a regression in speech and language function by the onset of Stage II Rett syndrome. Oral-motor tone of the cheeks, lips and tongue changed from hypotonicity in Stages I and III to hypertonicity in Stage IV in a direct relationship with postural tone. Three girls in Stage IV showed fasciculations of the tongue. Most girls in Stages III and IV showed tongue deviation to the left at rest. (Budden S et al. Communication and oral-motor function in Rett syndrome. Dev Med Child Neurol Jan 1990; 32:51-55).

COMMENT. This study documents the differences in oral-motor function in the various stages of Rett syndrome. The same author has described abnormal chewing associated with tongue thrusting and involuntary undulating tongue movements in 11 of 13 girls with Rett syndrome. (Budden S. Am J Med Genet 1986; 24:99). Feeding problems included difficulty in chewing and swallowing, choking and regurgitation.

An EEG study in 52 girls with Rett syndrome (Robb SA, Harden A., Boyd SG. Neurol Ped Nov 1989; 20:192-195) showed that seizure discharges occurred in 43 patients and were not related to the onset of clinical seizures. They consisted of sharp waves or spikes, focal or multifocal, mainly limited to the central and midtemporal regions of one or both hemispheres. Light sleep enhanced the presence of discharges. Periodical hyperventilation seen frequently during EEG recordings in this study was not associated with any consistent EEG change. Sleep spindles were conspicuously absent in all records. The authors find the EEG features helpful in confirmation of the diagnosis of Rett syndrome in the appropriate clinical setting.

GENETICS OF DEGENERATIVE DISEASES

Chromosome studies of four neurodegenerative diseases are described in current literature: Huntington disease cases in Finland show linkage disequilibrium of chromosome 4rflp haplotypes (Ikonen E et al. Am J Hum Genet Jan 1990; 46:5-11). Hereditary Motor and Sensory Neuropathy Type I shows linkage to the pericentromeric region of chromosome 17. (Middleton-Price HR, Harding AE et al. Am J Hum Genet Jan 1990; 46:92-94). Friedreich's ataxia gene has been assigned to chromosome 9q13-q21. (Hanauer A et al. Am J Hum Genet Jan 1990; 46:133-137). The dystonia gene in Ashkenazi Jewish population has been located on chromosome 9q32-34. (Kramer PL, Fahn S et al. Ann Neurol Feb 1990; 27:114-120). The same gene may be responsible for idiopathic torsion dystonia in non-Jewish kindred. Most familial forms of idiopathic torsion dystonia follow autosomal dominant transmission with reduced penetrance. The frequency in Ashkenazi Jewish population is 5-10 times greater than that in other groups.