EEG PATTERNS IN ANGELMAN SYNDROME

The sensitivity of the electroencephalogram (EEG) in the diagnosis of Angelman syndrome (AS) was studied in 26 patients (3/1: F/M ratio) at the University of Sao Paulo Medical School, Brazil. EEG patterns suggestive of AS were obtained in 47 EEGs of 25 (96%) patients and they were helpful in diagnosis. The delta pattern was recorded in 41 EEGs (ages from 0.4 to 21 years; mean 5.7 years) of 22 (88%) patients. Four variants of the delta pattern were recognized: 1) Hypsarrhythmia-like variant; 2) Slow spike and wave variant; 3) Triphasic-like variant; and 4) Slow variant. Theta pattern (TP) in 8 patients was generalized or posterior. Posterior runs of sharp waves were seen in 19 patients. TP was age related, in patients younger than 8 years, and only in those with maternal chromosome 15q11-q13 deletions. The delta pattern was most specific for AS. (Valente KD, Andrade JQ, Grossmann RM et al. Angelmann syndrome: difficulties in EEG pattern recognition and possible misinterpretations. Epilepsia August 2003;44:1051-1063). (Reprints: Dr KD Valente, Laboratory of Clinical Neurophrsiology, University of Sao Paulo Medical School-R Jesuino Arruda, 901 apt 51, 04532-082, Sao Paulo, SP Brazil).

COMMENT. Diagnosis of Angelmann syndrome (AS) is made by genetics, including deletion of the maternal chromosome 15q11-q13, clinically, and by EEG. The clinical diagnostic criteria of AS include impairment of neurologic development, poor or no language acquisition, a characteristic behavioral profile, sometimes termed *happy puppet syndrome* (unprovoked laughter, happy demeanor, hand flapping, hyperactivity, and attention deficit disorder), and a wide-based ataxic gait with jerky movements. Epilepsy, characteristic EEG abnormalities, and microcephaly occur in 80% of cases. Other less constant features in 20-80% of cases include peculiar facies, brachycephaly, hypopigmentation, albinism, wide-spaced teeth, and sleep disorders. (AS Foundation Consensus for Diagnostic Criteria, 1995). The EEG in early diagnosis of AS was reported from Great Ormond Street Hospital, UK (Boyd SG, Harden A, Patton MA. Eur J Pediatr 1988;147:508-513). Three EEG patterns suggestive of AS (delta, theta, and posterior discharges) were described in 98% of cases, occurring in early life, from 4 to 9 months, and before the classic phenotype is recognized at ages 3 to 4 years.

Reserpine responsive myoclonus and hyperpyrexia in an adult with AS is described from Geisinger Medical Center, Danville, PA (Stecker MM, Myers SM. Clin Neurol Neurosurgery July 2003;105:183-187). A 32-year-old woman was admitted with refractory seizures and episodic hyperpyrexia. She had dysmorphic and clinical features of AS, and chromosomal analysis revealed a deletion at the 15q11-q13 region. She was hypotonic at birth and seizures began in childhood. Despite high dose antiepileptic medications seizures and hyperpyrexia persisted, with near continuous shaking. Baseline EEG showed diffuse slowing and occipital spikes. Myoclonus was accompanied by only muscle activity on EEG and it responded to reserpine and clonazepam.

A report of myoclonus in Angelmann syndrome described 11 unrelated patients, confirmed by genetic analysis, and the myoclonus was controlled by piracetam in 5 (Guerrini R et al. Ann Neurol 1996;40:39-48). Jerky, tremulous, or dystonic movements proved to be a cortical myoclonus, defined by video-EEG and polygraphic monitoring. (see Progress in Pediatric Neurology III, PNB Publishers, 1997;p390).