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ATAXIC SYNDROMES

ANGELMAN'S SYNDROME: PATHOLOGICAL STUDY

The first pathological descriptions of Angelman's happy puppet syndrome based on an autopsy study of a 21 year old woman are reported from the Division of Neuropathology, The Hospital for Sick Children, University of Toronto, Canada. The diagnosis was first made at age 12 on the basis of severe mental retardation, absent speech, seizures, ataxia, hyperactive reflexes, extensor plantars, inappropriate laughter, microcephaly, brachycephaly, prognathism, and widely spaced teeth. She had been treated with multiple anticonvulsants and she died of recurrent pneumonia, pulmonary abscess, and hemoptysis. Autopsy findings showed a small brain with marked cerebellar atrophy, loss of Purkinje and granule cells, and Bergmann's gliosis. Dendrite morphology was abnormal with decreases in arborization and numbers of dendritic spines. Neurochemical studies showed reduced gamma-aminobutyric acid in the cerebellar cortex and elevated glutamate content in frontal and occipital cortices. (Jay V et al. Puppet-like syndrome of Angelman: A pathologic and neurochemical study. Neurology March 1991; 41:416-422).

> COMMENT. The cause of the cerebellar pathology was unknown. Since seizures began at ten months of age and the patient had received multiple anticonvulsants, the possibility of a secondary cerebellar degeneration could not be excluded. The dendritic pathology was similar to that found in Down syndrome and could be developmental or a consequence of dendritic atrophy.

ACETAZOLAMIDE IN VESTIBULOCEREBELLAR SYNDROME

Five patients with long-standing episodic vertigo and ocular motor signs are reported from the Department of Neurology, UCLA School of Medicine, Los Angeles, CA. Because of the clinical similarity to the familial periodic ataxia syndromes, these patients and affected

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family members were treated with acetazolamide. The episodic vertigo was either abolished or markedly decreased in frequency and severity. The age of onset of the vertigo varied from age two to 32. The episodes lasted from several hours to several days and were complicated by nausea and vomiting and later, a mildly progressive ataxia. Associated symptoms included horizontal or vertical diplopia, slurred speech, and positional downbeat and rebound nystagmus between attacks. The findings suggested a lesion of the vestibulocerebellum. In seven patients examined the MRI was normal. (Baloh RW, Winder A. Acetazolamide-responsive vestibulocerebellar syndrome: Clinical and oculographic features. Neurology March 1991; 41:429-433).

COMMENT. The familial periodic ataxia syndromes have been divided into two types, 1) a vestibulocerebellar syndrome and 2) diffuse cerebellar ataxia, as described by Parker, in which vertigo is infrequent and ataxia is more pronounced. Both syndromes appear to respond to acetazolamide.

SEIZURE DISORDERS

THE ELECTROENCEPHALOGRAM IN FEBRILE SEIZURES

The clinical value of EEG investigations in children with febrile seizures is reviewed from the Section of Child and Adolescent Psychiatry, Parke Hospital for Children, Old Road, Hedington, Oxford, England. Reports of EEG abnormalities in children with a history of febrile seizures include 1) ictal, generalized spiking, or lateralized spike wave discharge; 2) postictal, slow activity, spike wave or spikes; and 3) serial EEG's showing bisynchronous theta activity, bisynchronous spike wave at rest and during over breathing, bisynchronous spike wave on photic stimulation, focal spikes or sharp waves, and hypnagogic paroxysmal spike wave. The author concludes that an early postictal standard EEG will not be helpful in: a) the distinction between clinically simple and atypical seizures, b) the identification of a cerebral infective etiology, and c) the prediction of later occurrence of either further febrile or later afebrile seizures. A limited place for EEG studies is proposed in connection with febrile seizures associated with suspect cerebral pathology. If the child showed developmental delay, if the first seizure occurred below the age of 12 months, or if the seizures are partial or focal in pattern, a possible structural brain pathology is more likely. In addition, a prolonged febrile seizure, especially if followed by residual neurologic signs or developmental regression, may be evidence of structural cerebral damage and may require further investigation. EEG can be a useful ancillary investigation suggesting a persistent brain pathology, even in the presence of a normal CT scan, if it demonstrates a slow wave abnormality with or without spike or sharp waves that is persistent. This implies the use of serial EEG recordings over a period of weeks rather than a single recording. The EEG may also be helpful in research investigations concerning genetic factors and the possible connection between febrile seizures and benign rolandic epilepsy of childhood.