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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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