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CONGENITAL MALFORMATIONS

MOEBIUS SYNDROME

Brain stem calcification on CT scan, suggesting prenatal brain stem ischemia, is reported in an infant with Moebius syndrome examined in the Department of Pediatrics and Neonatal Medicine, State University of Gent, Gent, Belgium. Attempts at oral feeding in the neonatal period resulted in cyanotic attacks and bradycardia. There were repetitive clonic flexion movements in the arms and tonic contractions of intercostal and diaphragmatic muscles leading to respiratory arrest. Anticonvulsant therapy including pyridoxine were without benefit. The face was mask-like, the tongue was small, the gag reflex weak, the voice low pitched, and there was inability to abduct either eye beyond the midline. When asleep, both eyes were incompletely closed. There was generalized hypotonia and areflexia. At six months of age head control with severe titubation was achieved but conjugate eye movements were absent. At 12 months of age she still could not sit independently and she was monitored at home because of an erratic bradycardia. EEG's were normal. Chromosome test was normal. The right facial nerve could not be stimulated and blink reflexes were absent bilaterally. Brain stem evoked response showed bilateral anomalies. CT scan of the posterior fossa showed pontomedullary calcification in the floor of the fourth ventricle and basal cisterns were large. The case provides in vivo support to a theory of disruption in the vascular territory of the subclavian artery in explanation of the Moebius syndrome. (Govaert P et al. Moebius sequence and prenatal brainstem ischemia. Pediatrics September 1989; 84:570-573).

COMMENT. Congenital nonprogressive bilateral facial palsy and external ophthalmoplegia are the clinical features for the diagnosis of Moebius syndrome. (Moebius PJ. Munch Med Wochenschr 1888; 35:91). Theories of etiology have included 1) primary brain stem nuclear hypoplasia, 2) secondary brain stem nuclear degeneration, and 3) brain stem atrophy secondary to muscular

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defect. The postmortem finding of focal brain stem mineral deposits in some patients with Moebius syndrome support prenatal ischemic necrosis as an alternative explanation. (Thakkar N et al. Arch Neurol 1977; 34:124).

An infant with Moebius syndrome and MRI evidence of calcification in the medulla died recently at our institution and at autopsy was found to have atrophic lesions in the brain stem indicative of an ischemic pathology. (Clark HB. Personal communication). A case of a 13 year old boy with autosomal dominant congenital facial diplegia is reported from the Division of Neurology, Ramos Mejia Hospital, University of Buenos Aires, Argentina. Thirteen members of his family were affected over four generations. Electrophysiologic studies showed blink reflex abnormalities suggesting functional damage to the brain stem. (Garcia Erro MI et al. Familial congenital facial diplegia: Electrophysiologic and genetic studies. Pediatr Neurol July/August 1989; 5:262-4).

AGENESIS OF THE CORPUS CALLOSUM

The postmortem findings in a two month old infant with the typical clinical features of Aicardi syndrome (i.e., infantile spasms, chorioretinal lacunae, and agenesis of the corpus callosum) are reported from the Division of Neurology, Saitama Children's Medical Center, Saitama, Japan. The rostrum of the corpus callosum was absent and the roof of the dilated third ventricle was covered with a thin leptomeningeal membrane. Cortical heterotropias were found adjacent to the anterior horn of the right lateral ventricle and consisted of small immature neurons. The article includes a review of five autopsied patients with this syndrome previously reported in the literature. (Hamano S et al. Aicardi syndrome: Postmortem findings. Pediatr Neurol July/August 1989; 5:259-61).

COMMENT. In these cases, a high incidence of EEG laterality and an asymmetry of pathological lesions are of interest. Three of six patients had focal agenesis of the corpus callosum and three had papilloma of the choroid plexus.

ARNOLD-CHIARI MALFORMATION

Intermittent symptoms of obstructive hydrocephalus in a young woman with Chiari-I malformation are reported from the Neuro-Ophthalmology Service, Wills Eye Hospital, Philadelphia, PA. A previously healthy 26 year old woman experienced episodes of intermittent pressure headaches, dizziness, tingling in the right arm, and right anterior chest pressure for two months. These episodes lasted 5-15 minutes and occurred up to 15 times a day. They were followed by blurred vision with the appearance a "green patch" inferiorly before the left eye. Bilateral optic disc elevation was identified with indirect ophthalmoscopy. Lumbar puncture showed an opening pressure of 190 mm H₂O. Intravenous fluorescein angiography demonstrated venous stasis, diffuse retinal hemorrhages, and disc edema interpreted as consistent with papillophlebitis. An MRI using multiple thin sagittal sections directed at the posterior fossa revealed the Chiari I malformation, and an intraventricular catheter