DEVELOPMENTAL ANOMALIES

NEUROANATOMY OF FRAGILE X SYNDROME

Posterior fossa abnormalities measured by magnetic resonance images of the brain were reported in a group of 14 males with fragile X syndrome from the Division of Child Psychiatry, and the Departments of Psychiatry and Radiology, The Johns Hopkins University School of Medicine, and The Kennedy Institute, Baltimore, MD. The size of the posterior cerebellar vermis was significantly decreased and the fourth ventricle significantly increased in males with fragile X syndrome compared to a group of 17 males with other causes of developmental disability and 18 males with normal IQs. The lack of a significant correlation between age and size of vermis argues against atrophy as the cause but rather a developmental hypoplasia of the posterior vermis. (Reiss AL et al. Neuroanatomy of fragile X syndrome: the posterior fossa. Ann Neurol Jan 1991; 29:26-32).

> COMMENT. Hypoplasia of the cerebellar vermal lobules VI and VII has also been described in patients with autism (Courchesne E et al. N Engl J Med 1988; 318:1349). Autistic-like behavior occurs frequently in males with fragile X syndrome and the posterior vermis hypoplasia may have a causative relation. The cerebellar vermis plays an important role in the modulation and execution of motor behavior, and hyperactivity and repetitive stereotypic movements are common behaviorisms in fragile X patients. Language dysfunction also occurs with fragile X syndrome and the posterior vermis of the cerebellum is involved in auditory processing of language. So-called "cerebellar mutism" may develop after removal of posterior fossa tumors particularly medulloblastoma, and this complication has been correlated with the amount of the posterior vermis resected. (Platenberg C et al. AJNR 1989; 10:891). A seven year old girl with a midline posterior fossa tumor developed mutism postoperatively after removal of an astrocytoma that involved the vermis of the cerebellum. Recovery was slow but speech was regained after a two month period of therapy (Millichap JG, McLone D, unpublished case report).

TUBEROUS SCLEROSIS AND NEONATAL ASTROCYTOMA

The relative value of neural imaging studies in the diagnosis of a neonatal astrocytoma associated with tuberous sclerosis is reported from the Departments of Neurosciences, Pediatrics, and Pathology, University of California, San Diego, CA. The infant was hypotonic at birth and had poor respiratory effort. Apgar scores were 3/8. Motor examination revealed decreased tone in proximal upper extremities and neck. Deep tendon reflexes were generally exaggerated and plantar responses were extensor bilaterally. Wood's lamp examination was equivocal. There was no family history of tuberous sclerosis or epilepsy and the parents showed no neurocutaneous stigmata. An echoencephalogram revealed a large mass in the left lateral ventricle arising from the left caudate nucleus and obstructing the foramen of Monroe.