VASCULAR ABNORMALTIES IN NEUROFIBROMATOSIS TYPE 1

The spectrum of cerebrovascular abnormalities (CVA), including moyamoya, was evaluated in a retrospective chart review of 353 patients with neurofibromatosis type 1 (NF1) seen at the Children's National Medical Center, Washington, DC, from 1995 to 2003. Of 316 with brain MRI, eight (2.5%) had a CVA. MR angiography at age 1 to 13 years (mean 7.3 years) identified the following: 1) narrowed left posterior cerebral artery, 2) ectasia of carotid and cerebral arteries, 3) moyamoya with infarcts, 4) aneurysm, 5) carotid arterial stenosis with moyamoya and thalamic infarction, 6) internal carotid stenosis, 7) occlusion of middle cerebral artery, and 8) hypoplastic internal carotid artery. Vasculopathy was diagnosed in 7 patients, one presenting with acute hemiparesis and seizures at age 2 years. Three children required surgery. At 5.8 years follow-up (range 10 months to 9 years), only one (with moyamoya and strokes) had residual neurologic deficits. (Rosser TL, Vezina G, Packer RJ. Cerebrovascular abnormalities in a population of children with neurofibromatosis type 1. **Neurology** February (1 of 2) 2005;64:553-555). (Reprints: Dr Tena Rosser, Department of Neurology, Children's National Medical Center, 111 Michigan Ave NW, Washington, DC 20010).

COMMENT. Early recognition of a cerebral vascular abnormality by magnetic resonance angiography may help to prevent complications. The authors cite 43 cases of CVA in the NF1 literature, 25 (58%) in children. The supraclinoid ICA is involved most frequently. Ischemia with hemiparesis, often complicated by convulsions, is the most common presenting symptom. In 7 of the authors' 8 cases, the CVA was an incidental finding.

DEGENERATIVE DISORDERS

GLIAL PROTEIN MUTATIONS IN ALEXANDER DISEASE

The role of glial fibrillary acidic protein (*GFAP)* mutations in Alexander disease was analyzed in 44 patients, including 18 with later onset, at the University of Alabama, Birmingham, AL, and at other centers in the US, UK and Europe. Missense muations had been identified previously (Brenner M et al, 2001) in the *GFAP* gene in 11 of 12 infantile Alexander disease patients. Based on age at onset, patients were classified as infantile (<2 years old, 26 patients), juvenile (2-12 yrs, 15 pts), or adult (>13 yrs, 3 pts). Age at onset ranged from birth to 1.5 yrs for infantile, and from 2 to 11.5 yrs for juvenile. Clinical presentation of infantile cases included seizures (92%) and failure to meet milestones of development, with macrocephaly (62%), spasticity (52%), bulbar signs (62%), ataxia (58%), and cognitive defects (82%). Diagnosis was confirmed by pathology (Rosenthal fibers) or MRI. Dominant missense *GFAP* mutations were found in all forms of Alexander disease, with male predominance in the juvenile variety. (Li R, Johnson AB, Salomons G et al. Glial fibrillary acidic protein mutations in infantile, juvenile, and adult forms of Alexander disease. **Ann Neurol** March 2005;57:310-326). (Respond: Dr Michael Brenner, Department of Neurobiology and CIRC, University of Alabama, Birmingham, AL).