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NEUROCUTANEOUS SYNDROMES

EYE MOVEMENT DISORDERS IN ATAXIA TELANGIECTASIA

Eye movements were examined clinically in 56 patients (age range, 2-25 years; mean, 10.7 years) with ataxia telangiectasia (A-T) at Johns Hopkins University, Baltimore, MD. Electrooculographic recordings of eye movements were obtained in 33 patients. Deficits occurred in eye movement systems that stabilize images on the retina (pursuit, gaze holding, convergence, vestibular and optokinetic slow phases, and cancellation of vestibular slow phases), and in systems that maintain fixation and shift gaze, characterized by abnormal reflexive and voluntary saccades, head movements associated with gaze shifts, ocular motor apraxia, impaired fixation, and a reduction in vestibular and optokinetic quick phases. Clinical oculomotor abnormalities increased with age and were more prevalent in patients with severe neurologic abnormalities, whereas electrooculographic signs were not age or neurologic sign related. Deficits in image stabilization are associated with dysfunction in the cerebellar flocculus and ventral paraflocculus, and fixation deficits are probably correlated with dysfunction in the cerebellar vermis or the basal ganglia which affects the superior colliculus. (Lewis RF, Lederman HM, Crawford TO. Ocular motor abnormalities in ataxia telangiectasia. Ann Neurol September 1999;46:287-295). (Respond: Dr Lewis, Department of Neurology, Harvard Medical School, 243 Charles Street, Boston, MA 02114).

COMMENT. Atrophy of the cerebellum, especially the vermis, is the most prominent structural abnormality in pathological and imaging studies of ataxia telangiectasia. Eye movement abnormalities involving stabilization of retinal images are localized in the cerebellum, whereas impairments of fixation and shifts in gaze are more likely correlated with dysfunction in the cerebellar vermis or basal ganglia.

CEREBRAL BLOOD FLOW IN STURGE-WEBER SYNDROME

Regional cerebral blood flow during seizure activity, measured by transcranial Doppler sonography, and SPECT were studied in three infants with

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The editor is Pediatric Neurologist at Children's Memorial Hospital and Northwestern University Medical School, Chicago, Illinois.
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Sturge-Weber syndrome (SWS), during work-up for possible epilepsy surgery at Great Ormond Street Hospital and The Institute of Child Health, London, UK. The infants presented with recurrent seizures, altered consciousness, and neurologic deficits, and two showed clinical and radiological progression of the encephalopathy. SPECT showed hemispheric hypoperfusion, and middle cerebral artery velocity (MCAV) was reduced by 30-60% in the affected hemisphere compared with the contralateral side. MCAV during seizures was increased from 6 to 30% in the involved hemisphere compared to 24-179% for the contralateral side. This hemodynamic response to seizures of the unaffected hemisphere appeared to decrease over time, leading to increased cognitive and neurologic deficits with further seizure episodes. Abnormalities of the venous circulation in SWS, with reduced capacity for increasing venous return, cause venous hypertension and chronic progressive ischemia. (Aylett SE, Neville BGR, Cross JH, Boyd S, Chong WK, Kirkham FJ. Sturge-Weber syndrome: cerebral haemodynamics during seizure activity. Dev Med Child Neurol July 1999;41:480-485). (Respond: Dr FJ Kirkham, Neurosciences Unit, Institute of Child Health, The Wolfson Centre, Mecklenburgh Square, London WC1N 2AP, UK).

COMMENT. Sturge-Weber syndrome is characterized by epilepsy, hemiparesis, and pial and facial angiomata. The onset of seizures is often associated with neurologic deterioration and learning disability. The hemisphere underlying the venous angioma shows progressive gliosis, atrophy, and calcification, compatible with ischemia. The present study of the cerebral hemodynamics, in both affected and contralateral hemispheres, during seizure activity elucidates the cause of the progressive ischemia and neurologic deterioration in SWS. The progressive nature of the SWS encephalopathy after seizure onset, and especially in cases complicated by status epilepticus, should prompt consideration of early surgical excision. (see Progress in Pediatric Neurology I, PNB Publ, 1991;pp38-39, for report of progressive mental impairment in SWS).

Cutaneous mosaicism of lethal mutations and the lethal gene concept is proposed for Sturge Weber and other neurocutaneous syndromes (Hamm H. Am J Med Genet Aug 1999;85:342-345). SWS is a non-hereditary disorder. The concept of autosomal lethal genes surviving only in a mosaic state is proposed to explain the genetic basis of syndromes such as SWS, characterized by sporadic occurrence, distribution of lesions in an asymmetrical pattern, lack of diffuse involvement of entire organs, and equal sex ratio.

The Sturge-Weber Foundation currently announces a new book on SWS, edited by Bodensteiner JB and Roach ES.

MAGNETIC RESONANCE SPECTROSCOPY IN NEUROFIBROMATOSIS

The functional significance of T2 hyperintense lesions (UBOs) and their role in learning disabilities associated with neurofibromatosis type 1 (NF-1) were studied by quantitative magnetic resonance spectroscopy (MRS) in 7 male patients, aged 6-19 years, and 7 controls, at Johns Hopkins University, Baltimore, MD. Choline metabolite concentrations within UBOs, globus pallidus, and thalami were elevated in younger NF-1 subjects (<10 years), and were normal in older patients. N-acetylaspartate (NAA) levels were preserved in younger subjects and reduced in older patients. NAA was also decreased in the periventricular white matter. The MRS metabolic abnormality in UBOs of NF-1 is representative of a more generalized abnormality in affected brain regions. Choline elevations, reflecting increased myelin turnover in edematous areas, is followed by axonal