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CONGENITAL DEVELOPMENTAL DISORDERS

CEREBELLAR MALFORMATIONS AND COGNITIVE DISORDERS

The behavioral developmental profile of 27 children and adults (17 males and 10 females) with congenital cerebellar malformations was determined in a clinical, neuroradiological and neuropsychological study at the Scientific Institute 'E Medea', University of Milano, Italy. Of a total of 155 patients with cerebellar lesions, 128 (82.6%) with pathologies other than malformation were excluded (eg. acquired lesions, progressive pathology, metabolic disorders, cerebral involvement, Joubert syndrome, Arnold-Chiari syndrome, seizure disorders). The malformation was classified neuroradiologically as Type I. Complete cerebellar agenesis in 1 patient; Type II, complete or partial vermal agenesis in 5; Type III, Diffuse vermis and hemisphere cerebellar hypoplasia in 17; and Type IV, agenesis, hypoplasia or dysplasia of only the cerebellar hemispheres in 4 patients. Type I and II patients showed profound retardation of cognition, affect and language, and pervasive developmental disorder. Type III patients had variable cognitive and behavioral profiles, from normal to mild or moderate degrees of retardation. Type IV patients had largely normal cognition and affect, and mild language delay. Two patients had ADHD. Malformations affecting the cerebellar vermis were associated with affective and social disorders, autistic symptoms and a less favorable outcome. Patients with cerebellar hemisphere malformations had selective neuropsychological deficits involving mainly executive functions and visuospatial and linguistic abilities. Follow-up over time showed a gradual improvement in motor abilities and neuropsychological development, especially in patients with only hemisphere involvement. (Tavano A, Grasso R, Gagliardi C et al. Disorders of cognitive and affective development in cerebellar malformations. *Brain* October 2007;130:2646-2660). (Respond: Renato Borgatti MD, Department of Neurorehabilitation I, Scientific Institute 'E Medea' Bosisio Parini (LC), Italy).

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COMMENT. Acquired cerebellar lesions have been associated with a 'cerebellar cognitive affective syndrome, [CCAS],' (Schmahmann JD, Sherman JC. **Brain** 1998;121:561-579). CCAS is characterized by disorders of executive functioning, visuospatial ability, expressive language, working memory, and affective behavior. Patients have impairment of planning, abstract reasoning, verbal fluency, working memory, impaired spatial cognition associated with distractibility, perseveration, and inattention, anomia, and personality change with blunting of affect or disinhibited or inappropriate behavior. Vermal lesions are associated with marked affective and communication disorders, or with post-surgical mutism and speech or language disorders. CCAS is described in both adults and in children with acquired cerebellar lesions. The above study has shown that CCAS also occurs with cerebellar malformations. It is noteworthy that 9 (33%) patients were diagnosed with PDD and 2 with ADHD. A structural brain pathology as the basis for ADHD is also supported by an NIH MRI study showing a smaller cerebellar vermis in boys with ADHD, particularly involving posterior inferior lobules VIII to X. (Berquin PC et al. **Neurology** 1998;50:1087-1093). The cerebellum controls not only motor functions but also cognitive, language, behavioral and affective functions.

NEUROFIBROMATOSIS 1, THALAMIC HYPERINTENSITIES, AND COGNITIVE IMPAIRMENT

The relationship between T2 weighted MRI images (T2H) and cognitive functioning in a cohort of 76 children with neurofibromatosis type 1 (NF1) was determined in a study at Children's Hospital at Westmead, University of Sydney, Australia. Patients ranged in age from 8.0 to 16.75 (mean 11.63). NF1 was sporadic in 61% and familial in 39%. One-half were macrocephalic. Ninety percent had T2H, and 71% had discrete T2H lesions. Those with T2H lesions were slightly younger than those without (11.4 vs 13.3 yrs; $p=0.029$), and a greater number of T2H were found in younger children. Discrete lesions were usually located in the basal ganglia and cerebral hemispheres, whereas diffuse lesions involved the thalamus, cerebellum and brainstem. T2H in general were not associated with cognitive dysfunction, specific learning disabilities or ADHD, but patients with discrete thalamic lesions showed lower levels of cognitive functioning than those without lesions or with lesions elsewhere (Full scale IQ 72.8 vs 91.4). Thalamic lesions were also associated with lower performance on Verbal IQ, Performance IQ, Processing Speed Index, spelling, sustained attention, fine motor coordination, and motor speed. More diffuse lesions in the thalamus were also associated with reductions in IQ but less marked than those with discrete lesions. (Hyman SL, Gill DS, Shores EA, Steinberg A, North KN. T2 hyperintensities in children with neurofibromatosis type 1 and their relationship to cognitive functioning. **J Neurol Neurosurg Psychiatry** October 2007;78:1088-1091) (Respond: Dr Kathryn North, Clinical School, the Children's Hospital at Westmead, Locked Bag 4001, Westmead NSW 2145, Australia).

COMMENT. Thalamic lesions, particularly when discrete, are associated with cognitive impairment in children with NF1. Previous studies have shown that high-signal MRI lesions in NF1 evolve over time. They either increase or decrease in size or number, dependent on their location. Lesions in the thalamus, brainstem and cerebellar peduncles expand whereas those in cerebral hemispheres and cerebellum regress. Surgery or