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NEOPLASTIC DISORDERS

CEREBELLAR COGNITIVE AFFECTIVE SYNDROME

Neuropsychological effects of cerebellar tumor resection were evaluated in 19 children at the Department of Neurology, Massachusetts General Hospital, Boston, MA. Eleven had medulloblastoma, seven astrocytoma and one an ependymoma. Ages ranged from 3 years to 14 years (mean age, 8 years) at the time of tumor resection. Eight had received chemotherapy prior to neuropsychological testing, but none had received cranial irradiation or methotrexate. The time between surgery and neuropsychological testing ranged from 1 to 22 months (mean, 5 months). Executive function, including planning and sequencing, was impaired in 5 patients, expressive language in 7 (37%), visual-spatial function in 7 (37%), visual spatial memory in 5, and modulation of affect in 6. Fourteen (74%) had mild to marked impairment of fine motor coordination, sometimes independent of cognitive deficits. Dysregulation of affect was associated with lesions of the vermis. Age was a factor, the youngest being least likely to show cognitive deficits: of 9 who were under 7 years of age, only 3 (33%) showed deficits, whereas 8 (80%) of 10 older than 7 years had deficits. (Levisohn L, Cronin-Golomb A, Schmahmann JD. Neuropsychological consequences of cerebellar tumour resection. *Cerebellar cognitive affective syndrome in a paediatric population.* Brain May 2000;123:1041-1050). (Respond: Jeremy D Schmahmann MD, Department of Neurology, Massachusetts General Hospital, VBK 915, Fruit Street, Boston, MA 02114).

COMMENT. Acquired cerebellar lesions associated with cerebellar tumor resection in children may result in impairments of neurobehavioral function. The degree and type of dysfunction are correlated with age at the time of surgery and the site of the tumor resection. The cerebellum is important in cognitive and behavioral mechanisms.

Cerebellar site specific cognitive and behavioral deficits were demonstrated in a study of 26 children treated surgically for posterior fossa tumors at the Carlo Besta National Neurological Institute, Milan, Italy (Riva D,

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Giorgi C. The cerebellum contributes to higher functions during development. Evidence from a series of children surgically treated for posterior fossa tumours. Brain May 2000;123:1051-1061). Impairments of auditory sequential memory and language processing occurred with right cerebellar hemisphere tumors, whereas deficits in spatial and visual sequential memory were associated with left cerebellar tumors. Vermis lesions resulted in post-surgical mutism and behavioral disorders, including autism.

Cerebral white matter lesions and cognitive dysfunction. An MRI study in elderly subjects at the Erasmus University Medical School, Rotterdam, showed that the more severe periventricular white matter lesions affected speed of cognitive processes more than global cognitive and memory tasks. (de Groot JC, de Leeuw F-E, Oudkerk M et al. Ann Neurol Feb 2000;47:145-151).

The genetic basis of cognition was reviewed from the Institute of Molecular Medicine, John Radcliffe Hospital, Oxford, UK. (Flint J. Brain Nov 1999;122:2015-2031). Genetic approaches are limited to exploring neuronal function. Genetic mutations with a cognitive and behavioral phenotype are characterized by specific effects, but their delineation is difficult to determine in the mentally retarded. How a specific gene determines cognitive function is poorly understood.

DEVELOPMENTAL DISORDERS

REVISED DIAGNOSTIC CRITERIA OF TUBEROUS SCLEROSIS

A consensus conference sponsored by the National Institutes of Health and the National Tuberosus Sclerosis (TS) Association in July 1998 provided a revised list of diagnostic criteria of TS. The major and minor features are as follows:

Major features: facial angiofibromas, unguinal fibroma, >3 hypomelanotic macules, shagreen patch, retinal hamartomas, cortical tuber, subependymal astrocytoma, cardiac rhabdomyoma, lymphangiomyomatosis, renal angiomyolipoma.

Minor features: dental enamel pits, rectal polyps, bone cysts, white matter migration tracts, gingival fibromas, nonrenal hamartoma, retinal achromic patch, confetti skin lesions, multiple renal cysts.

Definite TS Complex: 2 major or 1 major with 2 minor features.

Probable TSC: 1 major and 1 minor feature.

Possible TSC: 1 major feature or 2 or more minor features.

The diagnosis of TSC is frequently made on dermatological evidence, and examination with UV light is used in screening asymptomatic relatives. Genetic criteria are not included and genetic testing is unavailable. Two genes account for TSC, *TSC1* found on chromosome 9, and *TSC2* on chromosome 16, both transmitted as autosomal dominants. Affected children of asymptomatic parents are explained by germline mosaicism. Sporadic cases make up two thirds of TSC patients, and 75% are caused by *TSC2* mutations. (Hyman MH, Whittemore VH. National Institutes of Health Consensus Conference: Tuberosus Sclerosis Complex. Arch Neurol May 2000;57:662-665). (Respond: Mark H Hyman MD, University of California, Los Angeles, 11980 San Vicente Blvd, Suite 102, Los Angeles, CA 90049).

COMMENT. Dermatological features are important among the diagnostic criteria for tuberous sclerosis complex. Four major and 1 minor required criteria are dermatologic. Renal and retinal hamartomas are also major diagnostic features. Genetic criteria are not included in the list of diagnostic features.