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MENTAL RETARDATION AND DEMENTIAS

WILLIAMS AND DOWN SYNDROMES

The neurological features of the Williams (WS) and Down (DS) syndromes were compared as part of a large multidisciplinary research center study and reported from the Departments of Neurosciences and Pediatrics, University of California School of Medicine, San Diego, CA. Eight patients with Williams syndrome and six with Down syndrome were matched for age (mean ages 16.7 and 15.8 years respectively) and WISC-R or WAIS scores revealed no significant differences between the two groups (WS: 53.8 ± 7.3 ; DS: 52.5 ± 8.8). DS patients demonstrated nonspecific features of global developmental delay but functioned fairly well for their developmental ages while those with WS demonstrated impaired oromotor skills, cerebellar dysfunction, difficulty with drawing, and higher verbal abilities than expected. WS patients also were small for gestational age and were more likely to have had early feeding problems and failure to thrive. One-half of the WS patients had epilepsy. The authors consider that neurologic distinctions between these two groups may reflect an underlying metabolic defect in Williams syndrome. (Trauner, DA et al. Neurologic feature of Williams and Down syndromes. Pediatr Neurol May/June 1989; 5:166-8).

COMMENT. Williams syndrome is a disorder of unknown etiology characterized by distinctive elflike facial features, mental retardation, cardiac defects and infantile hypercalcemia. A dissociation between language and cognitive skills described in patients with this disorder suggests a specific neuropsychologic profile. Seizures as a frequent manifestation of WS have not been reported previously.

ATLANTOAXIAL INSTABILITY IN DOWN SYNDROME

Results of an investigation of 130 children with Down syndrome screened for atlantoaxial instability are reported from Our Lady's

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Hospital for Sick Children, Crumlin, Dublin, Ireland. Radiological screening at ages ranging from 1-16 years showed that seven (5.4%) had evidence of atlantoaxial instability. The incidence among children examined between one and five years of age was 5% whereas those x-rayed between six and ten years of age showed a higher incidence of 12.8%, which was closer to that reported previously. The clinical history and complete neurological examinations were of no value in detecting the presence of atlantoaxial instability. The authors recommend that children with Down syndrome have a radiological screen between the ages of five and ten years and again at 15 years. (Cullen S et al. Atlantoaxial instability in Down's syndrome: clinical and radiological screening. Irish Med J June 1989; 82:64-65).

COMMENT. The association between atlantoaxial instability and Down syndrome has been known for many years and the prevalence is reported between 10% and 30% with a preponderance of affected females. The detection of the instability is difficult since the majority of children affected have no symptoms or signs before major complications occur as the result of compression of the spinal cord. At the present time there is no evidence to suggest that repeated radiological screening is necessary in patients with Down syndrome. Children with Down syndrome who wish to participate in sports training and competitive activities should have cervical spine x-rays and those with a definite abnormality should be excluded from competitive sports. Recreational and play activities of a less strenuous nature are usually permitted. An x-ray taken before five years of age is less likely to detect the atlantoaxial instability than one performed in later childhood or adolescent. The Special Olympics Committee recommends that all children with Down syndrome should be examined for atlantoaxial spinal instability before they participate in sports training and competitive physical activities which may result in hyperextension, flexion, or direct pressure on the neck or upper spine. Those found to have atlantoaxial instability should be excluded from participation in certain sports activities in the Special Olympics.

Atlantoaxial instability and odontoid hypoplasia are found in Morquio's syndrome (mucopolysaccharidosis Type IV), and other mucopolysaccharidoses, notably Hurler's (Type I), Hunter's (Type II) and Maroteaux-Lamy syndrome (Type VI). (See Children's Memorial Medical Center Journal Club Newsletter, Ed Stockman JA. August, 1989).

ALZHEIMER DISEASE IN DOWN SYNDROME

A clinical prospective study of dementia of the Alzheimer type in 96 individuals with Down syndrome over age 35 years is reported from the Eunice Kennedy Shriver Center, Waltham, MA and the Massachusetts General Hospital and Harvard Medical School, Boston, MA. Approximately 50% had a clinical dementia and the average age at dementia onset was 54.2 years. The prevalence of dementia in institutionalized Down syndrome population in this study was 8% between 35 and 49 years, 55% between 50 and 59 years,