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,

and 75% of those over 60 years old. Seizures developed in 84% of demented individuals with Down syndrome and 20% had Parkinsonian features. Hypothyroidism had been treated in 59% of the demented patients. CT scans showed brain tissue loss most pronounced in the temporal lobes. Neuropathological examination of 12 autopsied demented cases of Down syndrome showed gyral and central atrophy especially of the temporal lobes, and large numbers of plaques and tangles distributed in the same locations (i.e. hippocampus, amygdala, neocortex) as in the non-Down syndrome cases of Alzheimer dementia. (Lai F, Williams RS. A prospective study of Alzheimer disease in Down syndrome. Arch Neurol Aug 1989; 46:849-853).

COMMENT. The early age at onset of dementia in the Down syndrome population corresponds to the average age of onset (before age 52) in several large pedigrees of familial Alzheimer's disease. The gene for this form of autosomal dominant early onset Alzheimer disease has been mapped to the long arm of chromosome 21. An increased frequency of Down syndrome has been reported among relatives of early onset Alzheimer disease probands. The neuropathology and neurochemistry of Alzheimer's disease in aging individuals with Down syndrome and in the general population seem to be identical although the clinical expression of Alzheimer disease in Down syndrome shows some distinctive features, e.g. a high incidence of seizures.

HAW RIVER SYNDROME

A newly defined familial disorder of progressive dementia, ataxia, chorea, and seizures is described from the Department of Neurology School of Medicine, the University of North Carolina at Chapel Hill and the Department of Pathology, Duke University Medical Center, Durham, N.C. The first recorded member of the family was born in 1840 and lived at Haw River, N.C. In 22 patients examined the initial symptoms were ataxia of gait, intention tremor and choreiform movements that developed usually between 15 and 30 years of age. Recurrent generalized tonic-clonic seizures and progressive dementia developed later, and 11 of the 22 died after 15-25 years of illness. Neuropathological findings in two deceased family members were: neuronal loss of the dentate nucleus, microcalcification of the globus pallidus, neuroaxonal dystrophy of the nucleus gracilis, and demyelination of the centrum semiovale. (Farmer TW et al. Ataxia, chorea, seizures and dementia. Pathologic features of a newly defined familial disorder. Arch Neurol July 1989; 46:774-779).

COMMENT. The authors list in the differential diagnosis: Olivopontocerebellar atrophy, dentatorubropallidoluysian atropy, Ramsay Hunt syndrome, familial idiopathic calcification of the basal ganglia, neuroaxonal dystrophy, Hallervorden-Spatz disease, Huntington's disease, Wilson's disease, and Gerstmann-Straussler syndrome (cerebellar ataxia, dementia, amyloid plaques). Farmer's syndrome appears to have distinctive features.

Dementia as defined by the American Psychiatric Association (1980) is a deterioration in cognitive abilities that exceeds the decline