70 patients (90%), showed infratentorial pathology in 27%, including focal maldevelopments of the cerebellum: Dandy-Walker (2), Joubert syndrome (1), encephalocele (1), and hypoplasia (1). A genetic anomaly was found in 18 patients, including Angelman syndrome in 3. Forty-seven patients (60%) had mental retardation, severe in 25 and mild in 22; sixty nine (88%) had delayed speech development; 58% had visual dysfunction; and only 17 (22%) had normal cognitive function. (Esscher E et al. Non-progressive ataxia: origins, brain pathology and impairments in 78 Swedish children. Dev Med Child Neurol April 1996;38:285-296). (Respond: Dr Eva Esscher, Department of Paediatrics, Malarsjukhuset, Eskilstuna, S-633 88, Sweden).

COMMENT. Simple non-progressive ataxias comprise less than 10% of cases of cerebral palsy. Of 50% with known pathologies, the majority were prenatal in origin, many with a genetic background. No abnormality was revealed by imaging studies in 61%.

MACHADO-JOSEPH DISEASE

The frequency, and clinical, molecular, and neuropathological features of spinocerebellar ataxia 3 (SCA3) and Machado-Joseph disease (MJD) in 125 autosomal dominant cerebellar ataxia (ADCA) families were analyzed at the Service de Neuropathologie, Hopital de la Salpetriere, Paris, and Service de Neurologie, Hopital de Haut Leveque, Pessac, France; and Service de Neurologie, Hopital des Specialites, Rabat, Morocco. Thirty four families (126 patients) carried the expanded CAG repeat in the MJD1 gene. The length of the CAG repeat influenced the age at onset and the frequency of clinical signs associated with cerebellar ataxia (abnormal DTRs, decreased vibration sense). The frequency of supranuclear ophthalmoplegia, swallowing difficulties, and amyotrophy was significantly correlated with the disease duration. The age at onset varied from 14 to 70 years, mean 36 yrs, for the SCA3/MJD. One patient with SCA2 had an onset of ataxia at 8 years, whereas the youngest with SCA1 was 21. Neuropathological lesions distinguished the varieties of SCA, eg. basal ganglia lesions were more severe in SCA3/MJD than in SCA1. (Durr A et al. Spinocerebellar ataxia 3 and Machado-Joseph disease: Clinical, molecular, and neuropathological features. Ann Neurol April 1996;39:490-499). (Respond: Dr Durr, INSERM U289, Hopital de la Salpetriere, 4 boulevard de l'Hopital, 75651 Paris Cedex 13, France).

COMMENT. Despite the infrequent occurrence of Machado-Joseph disease in children, three recent reports from different parts of the world, France, Japan, and Australia, prompted commentary. MJD is an autosomal dominant spinocerebellar degeneration, occurring mainly in people of Portuguese descent. An unstable trinucleotide CAG repeat in MJD maps to the same region of chromosome 14 as the SCA3 locus. Two of 3 patients from Japan noted gait unsteadiness at age 18 years, followed 2 years later by involuntary movements, dysarthria, dysphagia, and hand incoordination. (Sakai T et al. A family with Machado-Joseph disease, previously diagnosed as dentatorubral-pallidoluysian atrophy. Neurology April 1996;46:1154-1156). Four families of Australian aboriginal people with MJD exhibited anticipation and an earlier age of onset. (Burt T et al. Machado-Joseph disease in east Arnhem Land, Australia: Chromosome 14q32.1 expanded repeat confirmed in four families. Neurology April 1996;46:1118-1122). MJD should be included in the differential diagnosis of a progressive ataxia with onset in later childhood, adolescence, or adulthood.