

MOVEMENT DISORDERS

GENETICS OF STIFF CHILD SYNDROME

A Chinese boy with a *DYT1* gene mutation presented with muscle stiffness, painful muscle spasms, myoclonus, and dystonia, compatible with stiff child syndrome, and is reported from Queen Mary Hospital, the University of Hong Kong. Stiffness of the left ankle began at age 4 years and spread to the right lower limb over a few months. Severely painful muscle spasms of the lower limbs developed 1 year later, and gradually extended to the trunk. Spasms were abolished by rectal diazepam 5 mg, and they decreased with sleep. EMG showed normal motor unit potentials on exertion, and continuous motor unit activity during muscle spasms. Nerve conduction studies, and MRI of brain and spinal cord were normal. Improvement followed plasmapheresis, with 75% reduction in painful muscle spasms within 4 weeks. One year later, he developed progressive dystonia and painful muscle spasms, becoming wheelchair bound at age 7 years. After a third plasmapheresis, and treatment with baclofen, Artane, clobazam, and gabapentin, muscle spasms were partially controlled, and he is maintained on baclofen. Analysis of the *DYT1* gene mutation was prompted by the appearance of dystonia, and his asymptomatic mother has the same mutation. (Wong VCN MB, FRCP, Lam C-W MB, Fung CW MB, MRCP. Stiff child syndrome with mutation of *DYT1* gene. **Neurology** November (1 of 2) 2005;65:1465-1466). (Reprints: Prof Virginia CN Wong, Division of Neurodevelopmental Pediatrics, University of Hong Kong, China).

COMMENT. The "stiff man" syndrome was first described by Moersch FP and Woltman HW, at the Mayo Clinic (**Mayo Clin Proc** 1956;31:421-427). I recall a 7-year-old, African-American, male child with the syndrome at Children's Memorial Hospital, Chicago, in 1964; he responded partially to oral diazepam; the pathology was thought to be located in the spinal cord interneurons. The syndrome is rare in children, and has been described in newborns (Lingam S et al. Hereditary stiff-baby syndrome. **Am J Dis Child** 1981;135:909). The present authors recommend screening for *DYT1* mutation in a child with muscle stiffness or spasms. Stiff child syndrome is distinguished from Isaac's syndrome, characterized by muscle stiffness and cramps, with myokymia and fasciculations, and evidence of terminal motor fiber abnormality (Isaacs H. **J Neurol Neurosurg Psychiatry** 1967;30:126).

CAUDATE VOLUMES IN CHILDHOOD PREDICT SEVERITY OF TOURETTE SYNDROME IN EARLY ADULTHOOD

Basal ganglia volumes of 43 children with Tourette syndrome (TS) were measured on MRI before age 14 years, and clinical assessment conducted after age 16, an average of 7.5 years later, at Yale University, New Haven, CT, and Columbia University, New York. Caudate nucleus volumes correlated significantly and inversely with severity of tic and OCD symptoms in early adulthood, but not at the time of the MRI scan. Caudate morphological pathology is central to the persistence of TS in adults. (Bloch MH, Leckman JF, Zhu H, Peterson BS. **Neurology** Oct (2 of 2) 2005;65:1253-1258). (Reprints: Dr Bradley S Peterson, Columbia College of Physicians and Surgeons, 1051 Riverside Dr, Unit 74, New York, NY 10589).