Kerr L, Bromberg MB et al. Congenital muscular dystrophy with rigid spine syndrome: a clinical, pathological, radiological, and genetic study. Ann Neurol February 2000;47:152-161). (Respond: Dr Flanigan, Eccles Institute of Human Genetics, Room 2100, 15 N 2030 East Street, Salt Lake City, UT 84112).

COMMENT. Congenital muscular dystrophy/rigid spine syndrome is a distinct subtype of congenital muscular dystrophy with genetic heterogeneity. The syndrome linked to the RSMD1 locus is characterized by congenital hypotonia, stable or slowly progressive weakness, neck weakness, early spinal rigidity, early scoliosis, and respiratory insufficiency. MRI of thigh muscles before biopsy can be helpful in defining the location of selective involvement.

Dubowitz V discusses the development of the expanding syndrome of congenital muscular dystrophy (Ann Neurology Feb 2000;47:143-144), beginning with the classical paper of Frederick E Batten (Brain 1903;27:147-148), followed by the equally classical pathological studies of Banker BQ et al (Brain 1957;80:319-334), and the reports of syndromes by Fukuyama Y and others. Later, the merosin-deficient muscle biopsies and clinical characteristics of classical congenital muscular dystrophy were distinguished from the merosin-positive ones. One merosin-deficient syndrome of CMD combined with rigid spine syndrome in middle east consanguineous families is mapped to chromosome 1p35-36.

ATTENTION DEFICIT AND LEARNING DISORDERS

DOPAMINE TRANSPORTER DENSITY IN ADHD

Dopamine transporter (DAT) density was measured by single photon emission computed tomography (SPECT) in six adult, unmedicated patients with attention deficit hyperactivity disorder (ADHD) and compared to 30 healthy controls, in a study at the Massachusetts General Hospital, Boston, MA. Striatal accumulation of iodine-123-labelled altropane was rapid, a maximum being reached in 10-15 minutes. Independent of age, DAT density was consistently elevated by 70% in the brain of patients with ADHD compared to healthy controls. Tracer accumulations were highest in the striatum and minimal in other areas of the brain. The use of SPECT could individualize treatment with psychostimulants, evaluate new drugs for ADHD, determine the pathophysiology of ADHD, and elucidate the mechanism of action of methylphenidate and other stimulants. (Dougherty DD, Bonab AA, Spencer TJ, Rauch SL, Madras BK, Fischman AJ. Dopamine transporter density in patients with attention deficit hyperactivity disorder. Lancet December 18/25, 1999;354:2132-2133). (Respond: Dr Alan J Fischman, Massachusetts General Hospital, Boston, MA 02114 (e-mail:fischman@petw6.mgh.harvard.edu)).

COMMENT. Dopamine D4 receptor and dopamine transporter (DAT1) genes have been associated with ADHD, and DAT is the main target for psychostimulant medications. It was postulated that ADHD may be caused by an excess expression of DAT, since previous studies have shown a correlation between DAT and hyperactivity-impulsivity scores. The above SPECT study showing elevated DAT accumulation in the striatum of adults with ADHD appears to confirm this theory. See Ped Neur Briefs (Jan 2000;14:4) for a report associating DAT1 with poor methylphenidate response in ADHD. Homozygosity of the 10-repeat allele characterized nonresponse to methylphenidate.

It should be emphasized to patients that SPECT studies are a research tool and are not generally available to monitor treatment of ADHD.