MUSCLE DISORDERS

LIMB-GIRDLE MUSCULAR DYSTROPHY

Sixty-one members of a large Spanish kindred with autosomal dominant limb-girdle muscular dystrophy (LGMD), spanning 5 generations, were examined at the Hospital Vail d'Hebron, Barcelona and other centers. Characteristic proximal muscle weakness involving pelvic and shoulder girdle muscles was found in 32 patients (15 men, 17 women) between the ages of 7 and 66 years (mean 34 years). Age at onset ranged from less than 1 to 58 years (mean 16 years). A *juvenile* form (onset before 15 years) and an *adult-onset* form (30-40 year onset) were identified in 72 and 28%, respectively. Weakness extended to distal muscles late in the course in adult cases, but generalized weakness occurred at initial presentation in severely affected juvenile cases. Progression was slow in adult cases and relatively faster in juvenile-onset cases. Severity worsened in successive generations (anticipation) among 26 parent-child pairs. None had ptosis, ophthalmoplegia, dysphagia, speech disorders, calf hypertrophy, myalgia, or mental deterioration. Muscle biopsy on 5 patients showed nonspecific findings compatible with MD; rimmed vacuoles were present in 3. Linkage analysis findings were distinct from previously reported forms of LGMD (1A - E), and chromosomes 5q31, 1q11, 3p25, 6q23, and 7q linkages were identified. (Gamez J, Navarro C, Andreu AL et al. Autosomal dominant limb-girdle muscular dystrophy. A large kindred with evidence for anticipation. Neurology February (2 of 2) 2001;56:450-454). (Reprints: Dr Josep Gamez, Department of Neurology, Hospital Gral, Vail d'Hebron, Passeig Vail d'Hebron, 119-125, 08035 Barcelona, Spain).

COMMENT. A genetically distinct form of autosomal dominant limb-girdle muscular dystrophy (LGMD) is added to the previously described 5 types of AD-LGMD (1 A-E). Characteristic features of LGMD include: slowly progressive proximal symmetric weakness with predominantly pelvic onset, normal to mildly elevated CK, myopathic EMG and muscle biopsy, and negative immuno-histochemical stains for dystrophin and sarcoglycans. Extraocular, facial, and bulbar muscles are unaffected. Two clinical types are identified (juvenile and adult-onset), according to age at onset, severity of muscle involvement, and rate of progression. In almost two-thirds, onset is in childhood or adolescence. Earlier onset in children than in parents suggests genetic anticipation, but disease severity is unrelated to the parent's gender.

ATTENTION DEFICIT DISORDERS

COMORBID ADHD AND TIC DISORDER

Motor system excitability was measured in 16 children with ADHD, 16 with chronic tic disorder or Tourette's disorder (TD), 16 with comorbid ADHD and TD, and 16 healthy control children, in a study at the University of Gottingen, Germany. The technique of focal transcranial magnetic stimulation (TMS) was used in a single and paired-stimulus paradigm, to assess inhibitory mechanisms within the motor system of these 4 groups. Children with ADHD alone had a reduced intracortical inhibition compared to those without ADHD, confirming deficits in inhibitory cortical motor mechanisms in ADHD. Children with TD had a shorter cortical silent period on TMS than those without TD, correlating with a deficient motor inhibition in the sensorimotor circuit. Children with combined ADHD and TD had both a significantly reduced intracortical inhibition and a shortened cortical silent period, suggesting an additive deficit in motor system