

MUSCLE DISORDERS

CONGENITAL MYASTHENIC SYNDROME AND AChR MUTATION

A 20-year-old woman from a consanguineous Moroccan marriage, with progressive muscle weakness noted from 2 years of age and evaluated at the University of Bonn, Germany, was found to have congenital myasthenic syndrome (CMS) due to homozygosity of the 1293insG e-acetylcholine receptor subunit mutation. Compared to the original case report of a CMS with end-plate acetylcholine receptor deficiency, heteroallelic for two e-AChR subunit mutations, and affected with mild muscle weakness, in this homozygous case, the weakness was profound and was associated with muscle wasting. (Sieb JP, Kraner S, Schrank B et al. Severe congenital myasthenic syndrome due to homozygosity of the 1293insG e-acetylcholine receptor subunit mutation. *Ann Neurol* September 2000;48:379-383). (Respond: Dr OK Steinlein, Institute for Human Genetics, University of Bonn, Wilhelmst 31, Bonn D-53105, Germany).

COMMENT. Differences in the acetylcholine receptor mutation haplotype can markedly influence the severity of congenital myasthenic syndrome. This profoundly affected patient is wheel-chair bound, and has almost complete external ophthalmoplegia and progressive kyphoscoliosis. Therapy with 60 to 120 mg of pyridostigmine daily was of limited benefit.

ACETAZOLAMIDE IN HYPOKALEMIC PERIODIC PARALYSIS

The mechanism of action of acetazolamide in the K-deficient diet rat, an animal model of human hypokalemic periodic paralysis (hypoPP), was investigated at the University of Bari, Italy. In vivo administration of acetazolamide prevented paralysis and depolarization of the fibers induced by insulin. Intense sarcolemma Ca-activated K channel activity was recorded in the acetazolamide-treated animals. The serum K levels were also restored to normal by acetazolamide. (Tricarico D, Barbieri M, Camerino DC. Acetazolamide opens the muscular KCa channel: a novel mechanism of action that may explain the therapeutic effect of the drug in hypokalemic periodic paralysis. *Ann Neurol* September 2000;48:304-312). (Respond: Professor Conte Camerino, Unit of Pharmacology, Department of Pharmacobiology, Faculty of Pharmacy, University of Bari, 70126, via Orabona Number 4, Bari, Italy).

COMMENT. The observed therapeutic effect of acetazolamide in hypokalemic periodic paralysis may be mediated by the activation of the muscular KCa channel.

ATTENTION DEFICIT AND BEHAVIOR DISORDERS

MOTOR DYSFUNCTION IN CHILDREN WITH ADHD

The performance of children with attention deficit hyperactivity disorder (ADHD) in the Movement Assessment Battery for Children test was evaluated at the Motorik Lab, Department of Woman and Child Health, Karolinska Institute, Sweden. In tasks involving motor-memory representations, a special grip object recorded forces generated by the fingertips during a precision grip-lift task. Twenty-five boys, aged 11 years, with ADHD were grouped according to the presence (ADHD+) or absence (ADHD-) of movement dysfunction, and compared to