Abstract

Myotonia congenita is a muscular disease characterized by myotonia, hypertrophy, and stiffness. It is inherited as either autosomal dominant or recessive known as Thomsen and Becker diseases, respectively. Here we confirm the clinical diagnosis of a family diagnosed with a myotonic condition many years ago and report a new mutation in the CLCN1 gene. The clinical diagnosis was established using ocular, cardiac, neurological and electrophysiological tests and the molecular diagnosis was done by PCR, SSCP and sequencing of the CLCN1 gene. The proband and the other affected individuals exhibited proximal and distal muscle weakness but no hypertrophy or muscular pain was found. The myotatic reflexes were lessened and sensibility was normal. Electrical and clinical myotonia was found only in the sufferers. Slit lamp and electrocardiogram tests were normal. Two affected probands presented diminution of the sensitive conduction velocities and prolonged sensory distal latencies. The clinical spectrum for this family is in agreement with a clinical diagnosis of Becker myotonia. This was confirmed by molecular diagnosis where a new disease-causing mutation (Q412P) was found in the family and absent in 200 unaffected chromosomes. No latent myotonia was found in this family; therefore the ability to cause this subclinical sign might be intrinsic to each mutation. Implications of the structure-function-genotype relationship for this and other mutations are discussed. Adequate clinical diagnosis of a neuromuscular disorder would allow focusing the molecular studies toward the confirmation of the initial diagnosis, leading to a proper clinical management, genetic counseling and improving in the quality of life of the patients and relatives. Rev. Biol. Trop. 56 (1): 1-11. Epub 2008 March 31.

Keywords

Myotonia congenita, myotonic dystrophy, Becker myotonia, chloride channelopathy, SSCP.