Myotonia Congenita

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Myotonia Congenita Information Page.

Myotonia congenita is an inherited neuromuscular disorder characterized by the inability of muscles to quickly relax after a voluntary contraction. The condition is present from early childhood, but symptoms can be mild. Most children will be 2 or 3 years old when parents first notice their muscle stiffness, particularly in the legs, often provoked by sudden activity after rest. The disease doesn't cause muscle wasting; in fact, it may cause muscle enlargement. Muscle strength is increased. There are two forms of the disorder: Becker-type, which is the most common form; and Thomsen's disease, which is a rare and milder form. The disorder is caused by mutations in a gene responsible for shutting off electrical excitation in the muscles.