Isaac's Syndrome

Isaac's syndrome (also known as neuromyotonia, Isaac's-Merten's syndrome, continuous muscle fiber activity syndrome, and quantal squander syndrome) is a rare neuromuscular disorder caused by continuous signaling of the end regions of peripheral nerve fibers that activate muscle fibers (motor neurons). Symptoms, which include progressive muscle stiffness, continuous vibrating or twitching muscles, cramping, increased sweating, and delayed muscle relaxation, occur even during sleep or when patients are under general anesthesia. Many patients develop weakened reflexes and muscle pain, but numbness is relatively uncommon. Although symptoms can be limited to cranial muscles, in most patients stiffness is most prominent in limb and trunk muscles. Speech and breathing may be affected if pharyngeal or laryngeal muscles are involved. Age of onset is between ages 15 and 60, with most patients experiencing symptoms before age 40. There are hereditary and acquired forms of the disorder. The acquired form may develop in association with peripheral neuropathies or as an autoimmune condition. Some patients with the autoimmune-mediated disorder produce antibodies that bind to ion channels on the motor nerve fibers. This condition, when associated with confusion, hallucinations, and insomnia, is known as Morfan's syndrome.

TREATMENT

Anticonvulsants, including phenytoin and carbamazepine, usually provide significant relief from the stiffness, muscle spasms, and pain associated with Isaac's syndrome. Plasma exchange may provide short-term relief for patients with some forms of the acquired disorder.

WHAT WILL HAPPEN

There is no cure for Isaac's syndrome. The long-term prognosis for individuals with the disorder is uncertain.

RESEARCH BEING DONE

The NINDS supports an extensive research program of basic studies to increase understanding of diseases that affect the brain, spinal cord, muscles, and nerves. This research examines the genetics, symptoms, progression, and psychological and behavioral impact of diseases, with the goal of improving ways to diagnose, treat, and, ultimately, cure these disorders.

Research is continually being done to learn more about this problem.

Provided by: The National Institute of Neurological Disorders and Stroke

Document Released: 12/08/2003

ExitCare® Patient Information ©2011 ExitCare, LLC.