Spastic Paraplegia Type 7

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Keywords: genetic conditions

1. Introduction

Spastic paraplegia type 7 (also called SPG7) is part of a group of genetic disorders known as hereditary spastic paraplegias. These disorders are characterized by progressive muscle stiffness (spasticity) in the legs and difficulty walking. Hereditary spastic paraplegias are divided into two types: pure and complex. The pure types generally involve only spasticity of the lower limbs and walking difficulties. The complex types involve more widespread problems with the nervous system; the structure or functioning of the brain; and the nerves connecting the brain and spinal cord to muscles and sensory cells that detect sensations such as touch, pain, heat, and sound (the peripheral nervous system). In complex forms, there can also be features outside of the nervous system. Spastic paraplegia type 7 can occur in either the pure or complex form.

Like all hereditary spastic paraplegias, spastic paraplegia type 7 involves spasticity of the leg muscles and some muscle weakness. People with this form of spastic paraplegia can also have ataxia; a pattern of movement abnormalities known as parkinsonism; exaggerated reflexes (hyperreflexia) in the arms; speech difficulties (dysarthria); difficulty swallowing (dysphagia); involuntary movements of the eyes (nystagmus); mild hearing loss; abnormal curvature of the spine (scoliosis); high-arched feet (pes cavus); numbness, tingling, or pain in the arms and legs (sensory neuropathy); disturbance in the nerves used for muscle movement (motor neuropathy); and muscle wasting (amyotrophy). The onset of symptoms varies greatly among those with spastic paraplegia type 7; however, abnormalities in muscle tone and other features usually become noticeable in adulthood.

2. Frequency

The prevalence of all hereditary spastic paraplegias combined is estimated to be 2 to 6 in 100,000 people worldwide. Spastic paraplegia type 7 likely accounts for only a small percentage of all spastic paraplegia cases.

3. Causes

Mutations in the *SPG7* gene cause spastic paraplegia type 7. The *SPG7* gene provides instructions for producing a protein called paraplegin. Located within the inner membrane of the energy-producing centers of cells (mitochondria), paraplegin is one of the proteins that form a complex called the m-AAA protease. The m-AAA protease acts as an enzyme and is responsible for assembling ribosomes (cellular structures that process the cell's genetic instructions to create proteins) and removing nonfunctional proteins in the mitochondria. When there is a mutation in paraplegin, the m-AAA protease cannot function correctly. Nonfunctional m-AAA proteases cause a build up of unusable proteins in the mitochondria of nerve cells, which can result in swelling of the cell, reduced cell signaling, and impaired cell movement, leading to the major signs and symptoms of spastic paraplegia type 7.

3.1 The gene associated with Spastic paraplegia type 7

- SPG7

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- hereditary spastic paraplegia, paraplegin type
- spastic paraplegia 7

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