Juvenile Primary Lateral Sclerosis

Subjects: Genetics & Heredity
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Juvenile primary lateral sclerosis is a rare disorder characterized by progressive weakness and tightness (spasticity) of muscles in the arms, legs, and face. The features of this disorder are caused by damage to motor neurons, which are specialized nerve cells in the brain and spinal cord that control muscle movement.

Keywords: genetic conditions

1. Introduction

Symptoms of juvenile primary lateral sclerosis begin in early childhood and progress slowly over many years. Early symptoms include clumsiness, muscle weakness and spasticity in the legs, and difficulty with balance. As symptoms progress, the spasticity spreads to the arms and hands and individuals develop slurred speech, drooling, difficulty swallowing, and an inability to walk.

2. Frequency

Juvenile primary lateral sclerosis is a rare disorder, with few reported cases.

3. Causes

Mutations in the *ALS2* gene cause most cases of juvenile primary lateral sclerosis. This gene provides instructions for making a protein called alsin. Alsin is abundant in motor neurons, but its function is not fully understood. Mutations in the *ALS2* gene alter the instructions for producing alsin. As a result, alsin is unstable and is quickly broken down, or it cannot function properly. It is unclear how the loss of functional alsin protein damages motor neurons and causes juvenile primary lateral sclerosis.

3.1. The gene associated with Juvenile primary lateral sclerosis

- ALS2

4. Inheritance

When caused by mutations in the *ALS2* gene, juvenile primary lateral sclerosis 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

- JPLS
- juvenile PLS
- PLSJ
- primary lateral sclerosis, juvenile

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