Juvenile primary lateral sclerosis

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.

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.

Frequency

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

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.

Inheritance Pattern

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.

Other Names for This Condition

- JPLS
- juvenile PLS
- PLSJ
- primary lateral sclerosis, juvenile
Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting

- Genetic Testing Registry: Juvenile primary lateral sclerosis

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22juvenile+primary+lateral+sclerosis%22+OR+%22primary+lateral+sclerosis%22

Other Diagnosis and Management Resources

- GeneReview: ALS2-Related Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK1243

Additional Information & Resources

Health Information from MedlinePlus

- Health Topic: Degenerative Nerve Diseases
  https://medlineplus.gov/degenerativenervediseases.html

- Health Topic: Paralysis
  https://medlineplus.gov/paralysis.html

Genetic and Rare Diseases Information Center

- Juvenile primary lateral sclerosis
  https://rarediseases.info.nih.gov/diseases/4485/juvenile-primary-lateral-sclerosis

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
  https://www.ninds.nih.gov/Disorders/All-Disorders/Motor-neuron-diseases-Information-Page

- National Institute of Neurological Disorders and Stroke: Spasticity Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Spasticity-Information-Page
Educational Resources

- MalaCards: primary lateral sclerosis, juvenile
  https://www.malacards.org/card/primary_lateral_sclerosis_juvenile
- Merck Manual Consumer Version: Amyotrophic Lateral Sclerosis and Other Motor Neuron Diseases

Patient Support and Advocacy Resources

- Motor Neurone Disease Association (UK)
  https://www.mndassociation.org/
- Muscular Dystrophy Association
  https://www.mda.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/primary-lateral-sclerosis/

Clinical Information from GeneReviews

- ALS2-Related Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK1243

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28juvenile+primary+lateral+sclerosis%5BALL%5D%29+OR+%28jpls%5BTIAB%5D%29%29+NOT+%28IAHSP%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PRIMARY LATERAL SCLEROSIS, JUVENILE
  http://omim.org/entry/606353

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17566607
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19122027
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301421

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16670179

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11586297

Reprinted from Genetics Home Reference:

Reviewed: July 2013
Published: October 29, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services