Alström syndrome

Description

Alström syndrome is a rare condition that affects many body systems. Many of the signs and symptoms of this condition begin in infancy or early childhood, although some appear later in life.

Alström syndrome is characterized by a progressive loss of vision and hearing, a form of heart disease that enlarges and weakens the heart muscle (dilated cardiomyopathy), obesity, type 2 diabetes (the most common form of diabetes), and short stature. This disorder can also cause serious or life-threatening medical problems involving the liver, kidneys, bladder, and lungs. Some individuals with Alström syndrome have a skin condition called acanthosis nigricans, which causes the skin in body folds and creases to become thick, dark, and velvety. The signs and symptoms of Alström syndrome vary in severity, and not all affected individuals have all of the characteristic features of the disorder.

Frequency

More than 900 people with Alström syndrome have been reported worldwide.

Causes

Mutations in the *ALMS1* gene cause Alström syndrome. The *ALMS1* gene provides instructions for making a protein whose function is unknown. Mutations in this gene probably lead to the production of an abnormally short, nonfunctional version of the ALMS1 protein. This protein is normally present at low levels in most tissues, so a loss of the protein's normal function may help explain why the signs and symptoms of Alström syndrome affect many parts of the body.

Learn more about the gene associated with Alström syndrome

- ALMS1

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.

Other Names for This Condition

- ALMS
- Alstrom syndrome
- Alstrom-Hallgren syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Alstrom syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268425/)

Genetic and Rare Diseases Information Center

- Alström syndrome (https://rarediseases.info.nih.gov/diseases/5787/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Alström syndrome%22)

Catalog of Genes and Diseases from OMIM

- ALSTROM SYNDROME; ALMS (https://omim.org/entry/203800)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28alstrom+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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