Marinesco-Sjögren syndrome

Description

Marinesco-Sjögren syndrome is a condition that has a variety of signs and symptoms affecting many tissues. People with Marinesco-Sjögren syndrome have clouding of the lens of the eyes (cataracts) that usually develops soon after birth or in early childhood. Affected individuals also have muscle weakness (myopathy) and difficulty coordinating movements (ataxia), which may impair their ability to walk. People with Marinesco-Sjögren syndrome may experience further decline in muscle function later in life.

Most people with Marinesco-Sjögren syndrome have mild to moderate intellectual disability. They also have skeletal abnormalities including short stature and a spine that curves to the side (scoliosis). Other features of Marinesco-Sjögren syndrome include eyes that do not look in the same direction (strabismus), involuntary eye movements (nystagmus), and impaired speech (dysarthria).

Affected individuals may have hypergonadotropic hypogonadism, which affects the production of hormones that direct sexual development. As a result, puberty is either delayed or absent.

Frequency

Marinesco-Sjögren syndrome appears to be a rare condition. More than 100 cases have been reported worldwide.

Causes

Mutations in the *SIL1* gene cause Marinesco-Sjögren syndrome. The *SIL1* gene provides instructions for producing a protein located in a cell structure called the endoplasmic reticulum. Among its many functions, the endoplasmic reticulum folds and modifies newly formed proteins so they have the correct 3-dimensional shape. The SIL1 protein plays a role in the process of protein folding.

SIL1 gene mutations result in the production of a protein that has little or no activity. A lack of SIL1 protein is thought to impair protein folding, which could disrupt protein transport and cause proteins to accumulate in the endoplasmic reticulum. This accumulation likely damages and destroys cells in many different tissues, leading to ataxia, myopathy, and the other features of Marinesco-Sjögren syndrome.

Approximately one-third of people with Marinesco-Sjögren syndrome do not have identified mutations in the *SIL1* gene. In these cases, the cause of the condition is unknown.

Learn more about the gene associated with Marinesco-Sjögren syndrome

- SIL1

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

- Garland-Moorhouse syndrome
- Hereditary oligophrenic cerebello-lental degeneration
- Marinesco-Garland syndrome
- MSS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Marinesco-Sjögren syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0024814/)

Genetic and Rare Diseases Information Center

- Marinesco-Sjögren syndrome (https://rarediseases.info.nih.gov/diseases/8341/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- MARINESCO-SJOGREN SYNDROME; MSS (https://omim.org/entry/248800)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28marinesco-sjögren+syndrome%5BTIAB%5D%29+OR+%28marinesco-sjogren+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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Last updated February 1, 2015