

Legius syndrome

Description

Legius syndrome is a condition characterized by changes in skin coloring (pigmentation). Almost all affected individuals have multiple café-au-lait spots, which are flat patches on the skin that are darker than the surrounding area. Another pigmentation change, freckles in the armpits and groin, may occur in some affected individuals.

Other signs and symptoms of Legius syndrome may include an abnormally large head (macrocephaly) and unusual facial characteristics. Although most people with Legius syndrome have normal intelligence, some affected individuals have been diagnosed with learning disabilities, attention-deficit disorder (ADD), or attention-deficit/hyperactivity disorder (ADHD).

Many of the signs and symptoms of Legius syndrome also occur in a similar disorder called neurofibromatosis type 1. It can be difficult to tell the two disorders apart in early childhood. However, the features of the two disorders differ later in life.

Frequency

The prevalence of Legius syndrome is unknown. Many individuals with this disorder are likely misdiagnosed because the signs and symptoms of Legius syndrome are similar to those of neurofibromatosis type 1.

Causes

Mutations in the *SPRED1* gene cause Legius syndrome. The *SPRED1* gene provides instructions for making the Spred-1 protein. This protein controls (regulates) an important cell signaling pathway that is involved in the growth and division of cells (proliferation), the process by which cells mature to carry out specific functions (differentiation), cell movement, and the self-destruction of cells (apoptosis). Mutations in the *SPRED1* gene lead to a nonfunctional protein that can no longer regulate the pathway, resulting in overactive signaling. It is unclear how mutations in the *SPRED1* gene cause the signs and symptoms of Legius syndrome.

[Learn more about the gene associated with Legius syndrome](#)

- SPRED1

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Neurofibromatosis type 1-like syndrome
- NFLS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Legius syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1969623/>)

Genetic and Rare Diseases Information Center

- Legius syndrome (<https://rarediseases.info.nih.gov/diseases/10714/index>)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Legius syndrome%22](https://clinicaltrials.gov/search?cond=%22Legius%20syndrome%22))

Catalog of Genes and Diseases from OMIM

- LEGIUS SYNDROME; LGSS (<https://omim.org/entry/611431>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28legius+syndrome%5BTIAB%5D%29+OR+%28nfls%5BTIAB%5D%29+OR+%28neurofibromatosis+type+1-like+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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