RELN gene

reelin

Normal Function

The *RELN* gene provides instructions for making a protein called reelin. This protein is produced in the brain both before and after birth. Reelin is released by certain brain cells. After being released, it attaches (binds) to specific receptor proteins. In the developing brain, this binding turns on (activates) a signaling pathway that triggers nerve cells (neurons) to migrate to their proper locations.

After birth, reelin likely plays a role in many brain processes, including the extension of axons and dendrites, which are specialized outgrowths from nerve cells that are essential for the transmission of nerve impulses. Reelin may also regulate synaptic plasticity, which is the ability of connections between neurons (synapses) to change and adapt over time in response to experience. Additionally, reelin controls the release of chemicals that relay signals in the nervous system (neurotransmitters).

Health Conditions Related to Genetic Changes

Autosomal dominant epilepsy with auditory features

Variants (also called mutations) in the *RELN* gene can cause autosomal dominant epilepsy with auditory features (ADEAF). People with this rare form of epilepsy typically hear sounds, like buzzing or humming, during seizures.

RELN gene variants associated with ADEAF reduce the amount of reelin in the body. Research suggests that a shortage of reelin impairs the formation or function of synapses, where cell-to-cell communication occurs. Abnormal communication between neurons can lead to seizure activity in the brain.

Lissencephaly with cerebellar hypoplasia

Several variants in the *RELN* gene have been found to cause lissencephaly with cerebellar hypoplasia (LCH). This condition affects brain development, resulting in the brain having a smooth appearance (lissencephaly) instead of its normal folds and grooves. In addition, the brain region involved in coordinating movements is unusually small and underdeveloped (cerebellar hypoplasia).

The *RELN* gene variants that cause LCH lead to a complete lack of reelin. As a result,

the signaling pathway that triggers neuronal migration is not activated. Without reelin, neurons are disorganized, the normal folds and grooves of the brain do not form, and brain structures do not develop properly. This leads to intellectual disabilities, delays in overall development, movement problems, and other signs and symptoms of LCH.

Autism spectrum disorder

Certain genetic changes that reduce but do not eliminate the production of reelin may increase a person's risk of autism spectrum disorder, a condition that affects communication and social interaction. However, some studies have not supported these findings. Many genetic and environmental factors are believed to contribute to this complex condition.

Myoclonus-dystonia

MedlinePlus Genetics provides information about Myoclonus-dystonia

Other disorders

Studies have shown that certain variations (polymorphisms) in the *RELN* gene are associated with an increased risk of psychiatric disorders such as schizophrenia and bipolar disorder. Women with these polymorphisms are at particular risk of developing bipolar disorder. However, many genetic and environmental factors likely contribute to the development of these complex conditions.

Other Names for This Gene

- LIS2
- PRO1598
- RELN_HUMAN
- RL

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RELN (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5649[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RELN%5BTIAB%5D%29+OR+%28reelin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- REELIN; RELN (https://omim.org/entry/600514)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5649)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RELN[gene])

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Genomic Location

The *RELN* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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