Hypomyelination and congenital cataract

Description

Hypomyelination and congenital cataract is an inherited condition that affects the nervous system and the eyes. This disease is one of a group of genetic disorders called leukoencephalopathies. Leukoencephalopathies involve abnormalities of the brain's white matter. White matter consists of nerve fibers covered by a fatty substance called myelin. Myelin insulates nerve fibers and promotes the rapid transmission of nerve impulses. Hypomyelination and congenital cataract is caused by a reduced ability to form myelin (hypomyelination). Additionally, people with this disorder are typically born with a clouding of the lens (cataract) in both eyes.

People with this condition usually have normal development throughout the first year of life. Development slows around the age of 1. Most affected children learn to walk between the ages of 1 and 2, although they usually need some type of support. Over time they experience muscle weakness and wasting (atrophy) in their legs, and many affected people eventually require wheelchair assistance. Weakness in the muscles of the trunk and a progressive abnormal curvature of the spine (scoliosis) further impair walking in some individuals. Most people with hypomyelination and congenital cataract have reduced sensation in their arms and legs (peripheral neuropathy). In addition, affected individuals typically have speech difficulties (dysarthria) and mild to moderate intellectual disability.

Frequency

The prevalence of hypomyelination and congenital cataract is unknown.

Causes

Variants (also called mutations) in the *HYCC1* gene cause hypomyelination and congenital cataract. The *HYCC1* gene provides instructions for making a protein called hyccin, the function of which is not completely understood. Based on the features of hypomyelination and congenital cataract, researchers presume that hyccin is involved in the formation of myelin throughout the nervous system. Hyccin is also active in the lens of the eye, the heart, and the kidneys. It is unclear how variants in the *HYCC1* gene cause cataracts.

Most *HYCC1* gene variants that cause hypomyelination and congenital cataract prevent

the production of hyccin. People who cannot produce any hyccin have problems forming myelin, leading to the signs and symptoms of this condition.

People who have variants that allow some protein production tend to have milder symptoms than those who produce no protein. These individuals typically retain the ability to walk longer, although they still need support, and they usually do not have peripheral neuropathy.

Learn more about the gene associated with Hypomyelination and congenital cataract

- HYCC1

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- HCC

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hypomyelination and Congenital Cataract (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864663/)

Genetic and Rare Diseases Information Center

- Hypomyelination-congenital cataract syndrome (https://rarediseases.info.nih.gov/diseases/11980/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Hypomyelination and congenital cataract%22)

Catalog of Genes and Diseases from OMIM

- LEUKODYSTROPHY, HYPOMYELINATING, 5; HLD5 (https://omim.org/entry/610532)

Scientific Articles on PubMed

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

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