SRD5A3-congenital disorder of glycosylation

Description

SRD5A3-congenital disorder of glycosylation (*SRD5A3*-CDG, formerly known as congenital disorder of glycosylation type Iq) is an inherited condition that causes neurological and vision problems and other signs and symptoms. The pattern and severity of this condition's features vary widely among affected individuals.

Individuals with *SRD5A3*-CDG typically develop signs and symptoms of the condition during infancy or early childhood. Most individuals with *SRD5A3*-CDG have intellectual disability, vision problems, unusual facial features,low muscle tone (hypotonia), and problems with coordination and balance (ataxia).

Vision problems in *SRD5A3*-CDG often include involuntary side-side movements of the eyes (nystagmus), a gap or hole in one of the structures of the eye (coloboma), underdevelopment of the nerves that carry signals between the eyes and the brain(optic nerve hypoplasia), or vision loss early in life (early-onset severe retinal dystrophy). Over time, affected individuals may develop clouding of the lenses of the eyes (cataracts) or increased pressure in the eyes (glaucoma).

Other features of *SRD5A3*-CDG can include skin rash, unusually small red blood cells (microcytic anemia),and liver problems.

Frequency

SRD5A3-CDG appears to be a rare disorder; more than 40 affected individuals have been described in the scientific literature.

Causes

SRD5A3-CDG is caused by variants (also known as mutations) in the *SRD5A3* gene. This gene provides instructions for making the enzyme steroid 5 alpha-reductase 3, which facilitates the conversion of a compound called polyprenol to a compound called dolichol. This conversion is critical for a process called glycosylation, by which groups of sugar molecules (oligosaccharides) are attached to proteins. Glycosylation changes proteins in ways that are important for their functions.

Variants in the *SRD5A3* gene typically lead to the production of abnormally small steroid 5 alpha-reductase 3 enzyme that either has no activity or is quickly broken down.

Without any normal steroid 5 alpha-reductase 3 enzyme, dolichol production is impaired, and glycosylation cannot proceed normally.The signs and symptoms of *SRD5A3*-CDG are likely due to impaired glycosylation of proteins that are needed for the normal function of various organs and tissues.

Learn more about the gene associated with SRD5A3-congenital disorder of glycosylation

- SRD5A3

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have variants. 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

- CDG Iq
- CDG-Iq
- Congenital disorder of glycosylation type 1q
- SRD5A3-CDG

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: SRD5A3-congenital disorder of glycosylation (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4317224/)

Genetic and Rare Diseases Information Center

- SRD5A3-CDG (https://rarediseases.info.nih.gov/diseases/12397/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Iq; CDG1Q (https://omim.org/entry/612379)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=SRD5A3+AND+CDG)

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