Congenital stromal corneal dystrophy

Description

Congenital stromal corneal dystrophy is an inherited eye disorder. This condition primarily affects the cornea, which is the clear outer covering of the eye. In people with this condition, the cornea appears cloudy and may have an irregular surface. These corneal changes lead to visual impairment, including blurring, glare, and a loss of sharp vision (reduced visual acuity). Visual impairment is often associated with additional eye abnormalities, including "lazy eye" (amblyopia), eyes that do not look in the same direction (strabismus), involuntary eye movements (nystagmus), and increased sensitivity to light (photophobia).

Frequency

Congenital stromal corneal dystrophy is probably very rare; only a few affected families have been reported in the medical literature.

Causes

Congenital stromal corneal dystrophy is caused by mutations in the *DCN* gene. This gene provides instructions for making a protein called decorin, which is involved in the organization of collagens. Collagens are proteins that strengthen and support connective tissues such as skin, bone, tendons, and ligaments. In the cornea, well-organized bundles of collagen make the cornea transparent. Decorin ensures that collagen fibrils in the cornea are uniformly sized and regularly spaced.

Mutations in the *DCN* gene lead to the production of a defective version of decorin. This abnormal protein interferes with the organization of collagen fibrils in the cornea. As poorly arranged collagen fibrils accumulate, the cornea becomes cloudy. These corneal changes lead to reduced visual acuity and related eye abnormalities.

Learn more about the gene associated with Congenital stromal corneal dystrophy

- DCN

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

- Congenital hereditary stromal dystrophy of the cornea
- Congenital stromal dystrophy of the cornea
- Corneal dystrophy, congenital stromal
- CSCD
- DACS
- Decorin-associated congenital stromal corneal dystrophy
- Dystrophia corneae parenchymatosa congenita

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Congenital Stromal Corneal Dystrophy (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864738/)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CORNEAL DYSTROPHY, CONGENITAL STROMAL; CSCD (https://omim.org/entry/610048)

Scientific Articles on PubMed

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

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