DHDDS-Related Disorders (including Retinitis Pigmentosa 59)

What Your Results Mean

Test results indicate that you are a carrier of *DHDDS*-related disorders. Carriers typically show no symptoms. You and your partner or donor would both have to be carriers of *DHDDS*-related disorders for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for *DHDDS*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

DHDDS-Related Disorders Explained

What are *DHDDS*-Related Disorders?

Retinitis Pigmentosa

Retinitis pigmentosa (RP) 59 is an inherited condition that causes degeneration of the retina (light-sensitive tissue at the back of the eye) and leads to progressive vision loss. RP is caused by variants in over 60 genes, including the *DHDDS* gene. These genes play important roles in the structure and function of photoreceptor cells in the retina (called rods and cones) that are responsible for transmitting visual signals from the eye to the brain. In RP, the rod cells, which are important for night vision, break down first. As a result, RP typically starts with loss of night vision. Daytime vision is affected later when both rods and cones are lost.

Congenital Disorder of Glycosylation Type Ibb

Congenital disorder of glycosylation type Ibb is a fatal multisystem disorder. This condition may be characterized by intrauterine growth retardation, poor growth after birth, muscle weakness and rigidity, micropenis, undescended testes, liver problems, kidney failure, and seizures. Because of the limited number of individuals and the overall lack of knowledge about this subtype of congenital disorder of glycosylation, the clinical picture may vary.

Prognosis

RP typically starts with loss of night vision in childhood, followed by development of blind spots in peripheral/side vision, a merging of blind spots to produce tunnel vision, and finally a gradual loss of central vision. Many affected individuals are legally blind by adulthood.

Prognosis for congenital disorder of glycosylation type Ibb is poor as affected individuals have been reported to die in infancy.

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Treatment

Treatment is largely supportive. Some therapeutic success has been reported in RP with Diamox therapy and vitamin A palmitate supplements. Use of special lenses can help reduce glare and improve contrast. Low vision aids are also recommended.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/
National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/congenital-disorders-of-glycosylation/
National Society of Genetic Counselors
https://www.nsgc.org/

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