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Retinitis pigmentosa 59

What is retinitis pigmentosa 59?

Retinitis pigmentosa is a group of eye disorders characterized by retinal degeneration and progressive vision loss.[1] Individuals with retinitis pigmentosa 59 have defects in the dehydrodolichyl diphosphate synthase enzyme, which is needed for production of the lipid dolichol.[1,2,3] Dolichol has a role in the process of attaching sugar molecules to glycoproteins, including rhodopsin, which is necessary for normal vision.[1,3]

What are the symptoms of retinitis pigmentosa 59 and what treatment is available?

Retinitis pigmentosa 59 is a disease with variable severity and age of onset. Symptoms often start in adolescence to young adulthood[4] and may include:[1,3,4]

- Night and peripheral vision loss
- Constriction of visual fields (tunnel vision)
- Degeneration of cone receptors
- Progression to blindness

There is no cure for retinitis pigmentosa 59. Treatment is supportive.

How is retinitis pigmentosa 59 inherited?

Retinitis pigmentosa is caused by mutations in more than fifty genes.[4] Retinitis pigmentosa 59 is an autosomal recessive disease caused by mutations in the *DHDDS* gene.[1,2] An individual who inherits one *DHDDS* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *DHDDS* mutations, one from each parent, is expected to be affected with retinitis pigmentosa 59.
If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for retinitis pigmentosa 59?

Mutations causing retinitis pigmentosa 59 can be found more frequently in the Ashkenazi Jewish population where the carrier frequency is 1 in 322.[1]

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

RP Fighting Blindness: http://www.rpfightingblindness.org.uk/index.php?pageid=59&tln=aboutrp

Foundation Fighting Blindness: http://www.blindness.org/retinitis-pigmentosa

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/retinitis-pigmentosa

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References

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2. DHDDS. *Genetics Home Reference.* Available at http://ghr.nlm.nih.gov/gene/DHDDS. Accessed February 16, 2016.
3. Venturini G., Koskiniemi-Kuending H., Harper S., Berson EL., Rivolta C. Two specific mutations are prevalent causes of recessive retinititis pigmentosa in North American patients of Jewish ancestry. *Genetics in Medicine.* 2015 April; 17 (4). 285-290.
4. Zúchner S. *et al.* Whole-Exome Sequencing Links a Variant in DHDDS to Retinitis Pigmentosa. *The American Journal of Human Genetics* 2011 February; 88; 201-206.