Retinitis Pigmentosa

Retinitis pigmentosa (RP) is the umbrella term for a group of hereditary conditions that cause progressive degeneration of the retina and degrade light-sensitive cells (photoreceptors) in the eye. Individuals with this condition may have reduced peripheral vision and/or a hard time seeing in the dark. RP typically presents early on with night blindness and eventually progresses to mild or complete blindness. The onset of symptoms varies depending on the specific gene mutation.

WHAT CAUSES RP?

RP is a genetic disorder caused by pathogenic (disease-causing) variant(s) that affect the light-sensitive cells (photoreceptors) in the eye. To date, there are over 40 genes associated with RP. The condition is congenital (present at birth), even if no symptoms are present during infancy.

WHO IS THIS TEST FOR?

This panel may be appropriate for patients of any age who have a personal and/or family history of retinitis pigmentosa or have exhibited any form of night blindness or peripheral/central vision loss. RP is often diagnosed during a retinal scan, at which point, providers may recommend genetic testing to confirm the diagnosis.

4978 Santa Anita Ave, Suite 205, Temple City, CA 91780 | www.FulgentGenetics.com | info@fulgentgenetics.com | **P** +1 626.350.0537 | **F** +1 626.454.1667
 D# FLY-Retinitis Pigmentosa-2021-V1

BENEFITS OF GENETIC TESTING

Genetic testing for RP can:

- Establish or confirm the appropriate diagnosis
- Identify risks for additional health-related symptoms
- Connect patients to relevant resources & support
- Result in more personalized treatment and symptom management
- Prepare patients for the onset of vision loss
- Inform family members about their own risk factors
- Provide options for family planning

RELATED TESTS

Visit Fulgent website for most up-to-date list

Retinitis pigmentosa NGS Panel

Usher Syndrome NGS Panel

Retinopathy and Optic Atrophy NGS Panel

Usher Syndrome NGS Panel

TEST SPECIFICATIONS

Acceptable Sample Requirements

- Blood, two 4-mL EDTA tubes (lavender top)
- Extracted DNA, 3 µg in EB buffer
- Buccal swab or saliva

Turnaround Time 3-5 weeks

Coverage ≥96% at 20x

Reporting

VUS, likely pathogenic, and pathogenic variants

Customization

Customizable Gene list, VUS opt-out

GET CONNECTED

Foundation Fighting Blindness - fightingblindness.org
American Academy of Ophthalmology - aao.org
Research to Prevent Blindness - rpbusa.org
Global Genes - globalgenes.org

REFERENCES

- ncbi.nlm.nih.gov/pubmed/17296890
- ncbi.nlm.nih.gov/pubmed/22131869t
- pubmed.ncbi.nlm.nih.gov/3068820/?from_term=retinitis+pigmentosa&from_pos=2

4978 Santa Anita Ave, Suite 205, Temple City, CA 91780 | www.FulgentGenetics.com | info@fulgentgenetics.com | **P** +1 626.350.0537 | **F** +1 626.454.1667
 D# FLY-Retinitis Pigmentosa-2021-V1

A Patient's Guide to Genetic Testing

What does a genetic test check for?

Genetic testing checks the order of one's DNA sequence (coded by the letters A, T, G, C) in specific genes linked to genetic conditions. Letters that were added, missing, or changed, are known as variants and can sometimes be harmful to one's health, increasing the risk for a genetic condition.

What are the potential results?

There are three possible results from genetic testing:

Positive

A pathogenic/likely pathogenic variant is detected in one's DNA. This type of variant is known to increase one's risk of a genetic condition. Identifying the specific gene involved can help confirm a diagnosis, inform screening and management, and reveal risk factors for an individual and/or their family.

Negative

No variation known to be associated with a genetic condition was detected in one's DNA . While a result may not show an increased risk for the condition(s) tested for, one can still be at risk for disease, especially if there is a family history.

Variant of Uncertain Significance (VUS)

A variant was detected in one's DNA, however, not enough information is known about this variant to determine whether or not it is known cause the condition(s) tested for. More research is needed to better understand this variant.

What about family members?

Children, siblings, and parents of individuals who have a variant(s) identified in genetic testing could carry the same variant(s) and benefit from testing. Regardless of whether or not a variant was identified, individuals can still be at an increased risk for a genetic condition, especially with a family history.

Do genetic test results affect health insurance or employment?

No, the Genetic Information Nondiscrimination Act (GINA) was signed into law in 2008.
It protects individuals from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. GINA does not protect against life and disability insurance discrimination. For more information on GINA, go to www.ginahelp.org.

Where can I learn more?

Medline Plus/Genetics Home Reference - medlineplus.gov/genetics/understanding
National Society of Genetic Counselors - nsgc.org
Fulgent Genetics - fulgentgenetics.com/products/carrierscreening/learning.html

4978 Santa Anita Ave, Suite 205, Temple City, CA 91780 | www.FulgentGenetics.com | info@fulgentgenetics.com | **P** +1 626.350.0537 | **F** +1 626.454.1667
 D# FLY-Retinitis Pigmentosa-2021-V1