

RPGR-related X-linked Retinitis Pigmentosa: RPGR Gene Sequencing

Test Code: SRPGR

Turnaround time: 6 weeks

CPT Codes: 81479 x1

Condition Description

Retinitis pigmentosa (RP) is a group of inherited disorders characterized by abnormalities of the photoreceptors on the retinal pigment epithelium. RP disorders lead to progressive visual loss. The first symptom is usually night blindness followed by visual field constriction which eventually leads to central vision loss. Isolated RP is most often inherited as an autosomal recessive disorder (50-60% of cases), but can be autosomal dominant (30-40%), or X-linked (5-15%) as well. More than 45 different genes accounting for approximately 60% of affected individuals have been implicated in RP.

Mutations in *RPGR* (Xp21.1), also called *RP3*, account for 70-90% of X-linked RP cases. Carrier females may show mild retinal degeneration. Mutations in *RPGR* have also been identified in individuals with cone-rod dystrophy, RP with sinorespiratory infection, with or without deafness, and atrophic macular degeneration.

This testing is for sequence analysis of the *RPGR* gene only.

Visit www.ThinkGenetic.com for patient-friendly information on [retinitis pigmentosa](#).

References:

- Hartong, D. et al. Retinitis pigmentosa Lancet. 368:1795-7809.
- GeneReviews
- [OMIM #312610](#): *RPGR* gene
- [OMIM #300029](#): *RP3* gene

Genes

[RPGR](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of *RPGR*-Related X-Linked RP.
- Carrier testing in adults with a family history of *RPGR*-Related X-Linked RP.

Methodology

Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection

Clinical Sensitivity: Mutations in the *RPGR* gene account for 70-90% of XLRP cases. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Analytical Sensitivity: ~99%

Specimen Requirements

Submit only 1 of the following specimen types

Type: Saliva

Specimen Requirements:

Oragene™ Saliva Collection Kit

Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:

Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

Type: Whole Blood (EDTA)**Specimen Requirements:**

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Type: DNA, Isolated**Specimen Requirements:**

Microtainer

8µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Related Tests

- Deletion/duplication analysis of the *RPGR* gene by CGH array is available for those individuals in whom sequence analysis is negative.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.
- Sequencing analysis for the *RP2* gene is also available.