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.


### References:

- 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

* Preferred specimen type: Whole Blood

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Saliva**

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection 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.