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.

For patients with suspected X-Linked RP, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

This testing is for 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 in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of RPGR-Related X-Linked RP in whom sequence analysis was negative.

Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Type: Whole Blood

Specimen Requirements:

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

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:

Whole blood is the preferred sample. Please contact the laboratory 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.

**Related Tests**

- Sequence analysis of the *RPGR* gene is available and is required before deletion/duplication analysis.
- 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.