RP2-related X-linked Retinitis Pigmentosa: RP2 Gene Deletion/Duplication

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 RP2 (Xp11.3) (OMIM #300757) account for 10-20% of X-linked RP cases (OMIM #312600). Carrier females may show mild retinal degeneration.

This testing is for the RP2 gene only.

For patients with suspected RP2-related X-linked retinitis pigmentosa, 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.


References:
- GeneReviews
- OMIM #312600: Retinitis pigmentosa 2
- OMIM #300757: RP2 gene

Genes

RP2

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of RP2-related X-linked retinitis pigmentosa in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of RP2-related X-linked retinitis pigmentosa 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

* 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

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.

Special Instructions

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

Related Tests

- Sequence analysis of the RP2 gene is available and is required before deletion/duplication analysis.
- Sequencing and deletion/duplication analysis are available for the RPGR gene.
- 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.