

Retinitis Pigmentosa: Deletion/Duplication Panel

Test Code: MD233

Turnaround time: 2 weeks

CPT Codes: 81479 x1, 81228 x1

Condition Description

Retinitis pigmentosa (RP) is a heterogeneous group of inherited diseases that commonly results in a progressive retinal degeneration. Over 90 forms of RP have been identified. RP can be syndromic or nonsyndromic and can be inherited in an autosomal dominant, autosomal recessive, or X-linked manner. RP is characterized by progressive visual field loss, night blindness, and abnormal or nonrecordable electroretinogram (ERG). Fundus changes include pigment deposition in the retina along the blood vessels with optic nerve pallor and arteriolar narrowing in early stages advancing to characteristic "bone spicule" pigmentary pattern in later stages. Please note that *RPGR* orf15 analysis is not included in this test.

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

References:

- Daiger *et al.* (1998) *Invest Ophthalmol Vis Sci* 39:S295.
- OMIM
- GeneReviews

Genes

[ABCA4](#), [AIPL1](#), [BBS1](#), [BEST1](#), [C1QTNF5](#), [C8orf37](#), [CA4](#), [CERKL](#), [CHM](#), [CLN3](#), [CLRN1](#), [CNGA1](#), [CNGB1](#), [CRB1](#), [CRX](#), [CYP4V2](#), [DHDDS](#), [EYS](#), [FAM161A](#), [FLVCR1](#), [FSCN2](#), [GUCA1B](#), [GUCY2D](#), [IDH3B](#), [IMPDH1](#), [IMPG2](#), [KLHL7](#), [LRAT](#), [MAK](#), [MERTK](#), [NR2E3](#), [NRL](#), [OFD1](#), [PCARE](#), [PDE6A](#), [PDE6B](#), [PDE6G](#), [PRCD](#), [PROM1](#), [PRPF3](#), [PRPF31](#), [PRPF6](#), [PRPF8](#), [PRPH2](#), [RBP3](#), [RBP4](#), [RDH12](#), [RGR](#), [RHO](#), [RLBP1](#), [ROM1](#), [RP1](#), [RP2](#), [RP9](#), [RPE65](#), [RPGR](#), [RPGRI1](#), [SAG](#), [SEMA4A](#), [SNRNP200](#), [SPATA7](#), [TOPORS](#), [TTC8](#), [TULP1](#), [USH2A](#), [ZNF513](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of retinitis pigmentosa.
- Carrier testing in adults with a family history of retinitis pigmentosa.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Detection

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

Specimen Requirements

Submit only 1 of the following specimen types

Type: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

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.

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 72 hours of collection. Do not freeze.

Special Instructions

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical correlation with test results.

Related Tests

- Eye Disorders: Comprehensive Sequencing and Deletion/Duplication Panels.
- Retinitis Pigmentosa: Sequencing Panel.