Retinitis Pigmentosa: Deletion/Duplication Panel

Test Code: MD233
Turnaround time: 2 weeks
CPT Codes: S1479 x1, S1228 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.


References:
- OMIM
- GeneReviews

Genes

ABCA4, AIP1, BBS1, BEST1, C10TNF5, C8orf37, CA4, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUC1A1, GUCY2D, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERTK, NR2E3, NRI, ODF1, PCARE, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF51, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RH12, RGR, RHQ, RLBP1, ROM1, RP1, RP2, RP3, RP665, RPGR, RPGRIP1, SAG, SEMA4A, SNRNP200, SPATA7, TOPO8, 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 24 hours of collection. Do not refrigerate or freeze.

Special Instructions

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.
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.