Retina/Photoreceptor Dystrophy: Sequencing Panel

Test Code: MM239

Condition Description

The Retina/Photoreceptor Dystrophy Panel is an analysis of almost all clinically relevant genes identified as causing non-syndromic disorders affecting the retina. Disorders in this category include, but are not limited to, isolated/inherited retinitis pigmentosa, Leber congenital amaurosis, achromatopsia, congenital stationary night blindness, vitreoretinopathy, optic atrophy, and the various photoreceptor/macular dystrophies. Additionally, a select group of syndromic genes that have also been identified in causing isolated retinal disease are included in this analysis (such as PAX6, CLN3, and USH2A).

Genes

<table>
<thead>
<tr>
<th>Genes</th>
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<tbody>
<tr>
<td>ABCA4, ADAM9, AIP1L, BBS1, BEST1, C1QTNF5, C2orf71, C8orf37, CA4, CACNB4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP290, CERKL, CHM, CLN3, CLR1N1, CNGA1, CNGA3, CNGB1, CNGB3, CNMN4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CYP4V2, DHDPS, EFEMP1, ELOVL4, EYS, FAM161A, FLVCR1, FSCN2, FZD4, GNA11, GNAI2, GPR179, GRM6, GUCA1A, GUCA1B, GUCY2D, IDH3B, IMPDH1, IKBP1, IQCB1, KCNJ13, KCNV2, KLHL7, LCAS, LRAT, LRH1, LRP5, MAK, MEK1, MN2, NDP, NR2E3, NRY, OAT1, OGD1, OPA1, OPA3, OTX2, PAX6, PDE6A, PDE6B, PDE6C, PDE6H, PITPNM3, PLA2G5, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS8BP, RHOC, RIMS1, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPRG, RPGRIP1, SAG, SEMA4A, SLC24A1, SNRNPS200, SPATA7, TIMM8A, TIMP3, TMEM126A, TOPORS, TRPM1, TSPAN12, TTC8, TULP1, UNC119, USH2A, VCAN, ZNF513</td>
</tr>
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</table>

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of retina/photoreceptor dystrophy.

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: Unknown. Pathogenic variants in the promoter region, some pathogenic variants in the introns and other regulatory element pathogenic variants 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

**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: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

- In microtainer: 60 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.
Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**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.
- Retina/Photoreceptor Dystrophy: Deletion/Duplication Panel.