Retina/Photoreceptor Dystrophy: Sequencing Panel

Test Code: MM239
Turnaround time: 6 weeks
CPT Codes: 81406 x1, 81407 x1, 81408 x1, 81403 x1, 81404 x1

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 \textit{PAX6}, \textit{CLN3}, and \textit{USH2A}). Please note, this analysis does not include mitochondrial sequencing; therefore, if Leber hereditary optic neuropathy (LHON) is suspected, targeted analysis for the common pathogenic variants is recommended (see test code: QC).

Genes

\textbf{ABCA4, ADAM9, APL1, BBS1, BEST1, C1QTNF5, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNM4, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CYP4V2, DHODH, EFEMP1, ELCVL4, EYS, FAM181A, FLVCR1, FSCN2, FZD4, GAT1, GAT2, GPR179, GRM6, GUCA1A, GUCA1B, GUCY2D, IDH3B, IMPDH1, IMPG2, ICCC1, KCNJ13, KCNV2, KLHL7, LCA5, LRAT, LRIT3, LRPS5, MAK, MERTK, MFNG, NDP, NR2E3, NRL, NYX, OAT, ODF1, CPA1, CPA3, OTX2, PAX6, PCARE, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PITPNM3, PL2G5, PRCD, PROM1, PRPF3, PRPF31, PRPF8, PRPF8, PRPH2, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP2, RHO, RIMS1, RBP1, ROM1, RP1, RP2, RPR9, RPE65, RPGR, RPGRIP1, SAG, SEMA4A, SLC24A1, SNRPN200, SPATA7, TIMM8A, TIMP3, TMEM126A, TOPORS, TRPM1, TSPAN12, TTC8, TULP1, UNC119, USH2A, VCAN, ZNF512}

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 (EDTA)

Specimen Requirements:
EDTA (Purple Top)
Infants and Young Children (2 years of age to 10 years old): 3-5 ml
Older Children and 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.

Type: Saliva

Specimen Requirements:
Oragene™ Saliva Collection Kit
Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL 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.

Type: DNA, Isolated

Specimen Requirements:
Microtainer
8µg
Isolation using the PerkinElmer™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.

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.