Retina/Photoreceptor Dystrophy: Deletion/Duplication Panel

**Test Code:** MD239  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81403 x1, 81406 x1

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**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).

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**

ABCA4, ADAM9, AIPL1, BBS1, BEST1, C1QTNF5, C2orf71, C8orf37, CA4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNM4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CYP4V2, DHDUD5, EEFEMP1, ELAVL4, EYS, FAM161A, FLCN, FZD4, GATA1, GATA2, GPR179, GRM6, GUCA1A, GUCA1B, GUCA1D, IDH3B, IMPDH1, IMPG2, IQC1B1, KCNJ13, KCNV2, KLHL7, LCAS, LRAT, LRIT3, LRPS, MAK, MERTK, MFN2, NDP, NR2E3, NRY, CAT, OFD1, OPA1, OPA2, OTX2, PAX6, PDE6A, PDE6B, PDE6C, PDE6H, PITPNM2, PLAX2G, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PAX2, RBP4, RD3, RHO12, RHO5, RGS9, RGS9B, RH3, RIM51, RLP1, ROM1, RP1, RP2, RP3, RPES8, RPGR, RPGRIP1, SAG, SEMA4A, SLC24A1, SNRNP200, SPATA7, TIMM8A, TIMP3, TMEM126A, TOPORS, TRPM1, TSPAN12, TTC8, TULP1, UNC119, USH2A, VCAN, ZNF513

**Indications**

This test is indicated for:

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

**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.

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 will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**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: 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: 10 ug

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.
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: Sequencing Panel.