## Eye Disorders: Deletion/Duplication Panel

**Test Code:** MD030  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81406 x1, 81403 x1, 81405 x1

### Condition Description

The Eye Disorder Deletion/Duplication Panel is an analysis of almost all clinically relevant genes identified as causing syndromic and non-syndromic inherited retinal and choroidal dystrophies, as well as ocular developmental disorders. There is a wide range of genetic and phenotypic heterogeneity in retinal and choroidal disorders making accurate clinical diagnosis difficult especially during early phases of the disease onset.

Retinal disorders can be congenital and present at birth (as in Leber congenital amaurosis), present in early childhood (as in early onset retinitis pigmentosa), or present in mid life (as in pattern dystrophy). The clinical features of retinal disorders include vision loss, vision distortion, loss of peripheral vision, and night blindness. Fundus exam findings can range from almost normal appearance of the retina (as in Leber congenital amaurosis) to pale optic nerve, narrowed arterioles, bone spicules, photoreceptor loss, retinal pigment epithelial changes, and chorioretinal atrophy. The fundus appearance in the end stage of many retinal disorders, such as pattern dystrophy and cone-rod dystrophy, may be similar to that of macular dystrophy or chorioretinal atrophy. Electroretinogram (ERG) findings can range from non-recordable ERG to loss of rod or cone responses and be non-specific. Rarely, characteristic findings in ERG as in congenital stationary night blindness may help in arriving at a more accurate diagnosis. Detailed history and clinical examination, optical coherence tomography (OCT), pattern of visual field loss and ERG may help narrow the selection of disease causing genes or groups of genes.

Some genes on this panel are available as single gene tests or as part of a more clinically specific eye disorders sub-panel (e.g. retinitis pigmentosa). As the distinction between disorders is difficult, the Eye Disorder Deletion/Duplication Panel may be ordered as a comprehensive test. Please note that some genes may cause more than one phenotype.

General categorical overview of the eye disorders included on the panel:

- **Achromatopsia**
- **Albinism**
- **Bardet Biedl Syndrome**
- **Bradyopsia**
- **Choroideremia**
- **Cone and Cone-rod Dystrophy:** Please note, the RAB28 gene is not included on the NGS panel at this time due to the presence of at least 2 pseudogenes. For clinicians that would like RAB28 analysis if all other genes test negative, we request consultation with the EGL directly.
- **Congenital Stationary Night Blindness:** Please note, the GRK1 gene is not included on the NGS panel at this time as this gene is only partially annotated in hg19. GRK1 will be re-evaluated with the release of hg20.
- **Flecked Retina Disorders**
- **Isolated Aniridia**
- **Joubert Syndrome**
- **Leber Congenital Amaurosis:** Please note, the NMNAT1 gene is not included in the NGS panel at this time due to the presence of at least 4 pseudogenes. For clinicians that would like NMNAT1 analysis if all other genes test negative, we request that you contact the EGL directly.
- **Leber hereditary optic neuropathy (LHON)**
- **Microphthalmia, Anterior Segment Dysgenesis, and Related Anomalies**
- **Neuronal Ceroid-Lipofuscinoses**
- **Optic Atrophy**
- **Photoreceptor Dystrophy**
- **Primary Open Angle Glaucoma**
- **Retafs disease**
- **Retinitis pigmentosa and ataxia (NARP)**
- **Retinitis pigmentosa, AD, AR and X-linked**
- **Retinoschisis**
- **Senior Loken Syndrome**
- **Stargardt's Disease and Macular Dystrophy**
- **Sticker Syndrome**
- **Usher Syndrome**
- **Vitreoretinopathy**

Disclaimer: Ordering this panel may result in the identification of a genetic change that predisposes an individual to systemic disorders in addition to eye/retinal disorders. Genetic counseling by qualified genetic counselor or medical geneticist is strongly recommended before ordering any genetic test. Ordering physicians can call EGL Genetics at 470-378-2200 to speak with a laboratory genetic counselor.

### References

- OMIM.
- GeneReviews.
- Emory and Rimoin's Principles and Practice of Medical Genetics, 5th Edition.

### Genes


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TTC8
TCTN1
RPE65
PRPF31
PCARE
IDH3B
FRAS1
COL9A1
CERKL
CHM
CIB2
CLN3
CLN5
CLN6
CLN8
CLRN1
CNGA1
CNGA3
CNGB1
CNGB3
CNMM4
COL11A1
COL11A2
COL2A1
COL4A1
COL9A1
COL9A2
CPLANE1
CRB1
CRX
CTSD
CYP1B1
CYP4V2
DHDDS
EFEMP1
ELOV4
EYS
FAM161A
FLVCR1
FOXCI
FOX3
FRAS1
FREM2
FSCN2
FZD4
GNA1
GNA2
GPR143
GPR179
GRIP1
GRM6
GRN
GUCA1A
GUCA1B
GUCY2D
HARS
HCSS
IDH3B
IPDH1
IPMP2
INV5
LCG1
KCNJ13
KCNV2
KCTD7
KIF7
KLLH1
LCAS
LRAT
LRRT3
LRP5
LZTFL1
MAK
MERTK
MFN2
MFRP
MFS9D
MKKS
MK1
MTTP
MYO7A
MYOC
NDP
NPHP1
NPHP3
NPHP4
NRSE3
NRX
OAT
OCA2
OHD1
OPA1
OPA3
OTX2
PAX6
PCLD15
PDE6A
PDE6B
PDE6C
PDE6G
PEX7
PHXN
PITPM3
PITX2
PLAG25
PP1T
PRCD
PROM1
PRPF3
PRPF6
PRPF8
PRPH2
RAX2
RBP3
RBP4
ROD1
RH12
RDH4
RDH5
RGS3
RGS8BP
RH2
RIMS1
RLBP1
ROM1
RP1
RP2
RP3
RP65S
RPG1
RPGP1
RS1
SAG
SDCCAG8
SEMA4A
SLC2A1
SLC45A2
SMOC1
SNRNP200
SOX2
SPATA7
STRA6
TCTN1
TCTN2
TCTN3
TIMM8A
TIMP3
TMEM126A
TMEM216
TMEM237
TMEM67
TOPORS
TPP1
TRPM1
TSPAN12
TTC21B
TTC8
TUL1P1
TYR
TYRP1
UNC119
USH1C
USH1G
USH2A
VAX1
VCAN
VSD2
WDPCP
WFS1
WHRN
WT1
ZNFI23
ZNFP13

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of a syndromic and/or non-syndromic retinal and optic nerve disorders.
- Carrier testing in adults with a family history of a syndromic and/or non-syndromic retinal and optic nerve disorders.

**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 Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. 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 (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.

**Type: DNA, Isolated**

**Specimen Requirements:**

Microtainer

3µg

Isolation using the PerkinElmer™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.

**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**

- Achromatopsia, Cone, and Cone-rod Dystrophy: Sequencing and Deletion/Duplication Panels.
- Bardet-Biedl Syndrome: Sequencing and Deletion/Duplication Panels.
- Congenital Stationary Night Blindness: Sequencing and Deletion/Duplication Panels.
- Flecked-retina Disorders: Sequencing and Deletion/Duplication Panels.
- Albinism: Sequencing and Deletion/Duplication Panels.
- Joubert Syndrome: Sequencing Panel.
- Macular Dystrophy, Degeneration, Stargardt Disease: Sequencing and Deletion/Duplication Panels.
- Anophthalmia/Microphthalmia/Anterior Segment Dysgenesis/Anomaly: Sequencing Panel.
- Neuropilin-1-Lipofuscinosis: Sequencing Panel.
- Retinitis Pigmentosa: Sequencing and Deletion/Duplication Panels.
- Optic Atrophy: Sequencing and Deletion/Duplication Panels.
- Retina/Photoreceptor Dystrophy: Sequencing and Deletion/Duplication Panels.
- Senior-Loken Syndrome: Sequencing and Deletion/Duplication Panels.
- Stickler Syndrome: Sequencing Panel.
- Usher Syndrome: Sequencing Panel.
- Vitreoretinopathy: Sequencing and Deletion/Duplication Panels.
- Eye Disorders: Comprehensive Sequencing Panel.