Macular Dystrophy/Degeneration/Stargardt Disease: Deletion/Duplication Panel

**Condition Description**

Macular dystrophy is a general term referring to the degeneration (or atrophy) of the macula which can be accompanied by pigmentary changes, flecks, and lipofuscin-like deposits. Macular dystrophies are commonly inherited in an autosomal dominant manner. Examples of conditions in this category include Best vitelliform macular dystrophy, Stargardt disease, Sorsby’s dystrophy, and Doyne’s dystrophy.

**References:**
- OMIM
- GeneReviews

**Genes**

ABCA4, BEST1, CDH3, CNGB3, EFEMP1, ELOVL4, FSCN2, GUCA1B, PROM1, PRPH2, RBP4, RDH12, RPGR, RPGRIP1, TIMP3

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of macular dystrophy/degeneration or Stargardt disease.
- Carrier testing in adults with a family history of macular dystrophy/degeneration or Stargardt disease.

**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: 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 72 hours of collection. Do not freeze.

**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 Disorder: Comprehensive Sequencing and Deletion/Duplication Panels.
• Macular Dystrophy/Degeneration/Stargardt Disease: Sequencing Panel.