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

**Test Code:** MD138  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81479 x1

---

### 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, RGPR1P1, 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. 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 causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**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

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
- Isolation using the QiagenTM 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 Disorder: Comprehensive Sequencing and Deletion/Duplication Panels.
- Macular Dystrophy/Degeneration/Stargardt Disease: Sequencing Panel.