

Macular Dystrophy/Degeneration/Stargardt Disease: Sequencing Panel

Test Code: MM138

Turnaround time: 6 weeks

CPT Codes: 81408 x1, 81404 x1, 81406 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 Doyme's dystrophy.

References:

- Daiger et al. (1998) Invest Ophthalmol Vis Sci 39:S295.
- OMIM
- GeneReviews

Genes

[ABCA4](#), [BEST1](#), [CDH3](#), [CNGB3](#), [EFEMP1](#), [ELOVL4](#), [FSCN2](#), [GUCA1B](#), [PROM1](#), [PRPH2](#), [RBP4](#), [RDH12](#), [RPGR](#), [RPGRI1](#), [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

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 & 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: DNA, Isolated

Specimen Requirements:

Microtainer

8µg

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: 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.

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: Deletion/Duplication Panel.