Flecked-retina Disorders: Sequencing Panel

**Test Code:** MM134  
**Turnaround time:** 6 weeks  
**CPT Codes:** 81404 x1, 81408 x1, 81479 x1

### Condition Description

Flecked-retina refers to a varying distribution, and size, of yellowish lesions within the retina (optic nerve or vascular anomalies are not generally observed). Flecked-retina disorders may be stationary or progressive and range from benign to visually debilitating. The term "flecked-retina" is generally not favored by retina specialists due to the distinctive pattern and clinical course of each individual disorder. Examples of conditions in this category include benign flecked-retina, fundus flavimaculatus (Stargardt's), and fundus albipunctatus.

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

### Genes

ABCA4, PLA2G5, PRPH2, RDH5, RHO, RLBP1

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of flecked-retina disorders.  
- Carrier testing in adults with a family history of flecked-retina disorders.

### 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 Stanger-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

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: 60 ug
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.
- Flecked-retina Disorders: Deletion/Duplication Panel.