

Flecked-retina Disorders: Deletion/Duplication Panel

Test Code: MD134

Turnaround time: 2 weeks

CPT Codes: 81228 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 alpinctatus.

References:

- Daiger *et al.* (1998) *Invest Ophthalmol Vis Sci*, 39:S295.
- Walia, Fishman, and Kapur. (2009) *Ophthalmic Genet*, 30:69-75.
- 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

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: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. 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 24 hours of collection. Do not refrigerate or freeze.

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.

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: Sequencing Panel.