Vitreoretinopathy: Deletion/Duplication Panel

Test Code: MD238  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1, 81403 x1

Condition Description

Vitreoretinopathy is a general term used to describe retinal disease that also affects the vitreous body. Several types of vitreoretinopathies exist giving rise to a spectrum of phenotypic presentations such as retinal detachment (or traction), optically empty vitreous, fibrillary condensation, cataract, and neovascularization. The condition includes, but is not limited to, familial exudative vitreoretinopathy, Norrie disease, Wagner syndrome, snowflake vitreoretinal degeneration, Stickler syndrome and retinal vasculopathy with cerebral leukodystrophy. The vitreoretinopathies may be inherited in an autosomal dominant, autosomal recessive or X-linked manner (complex inheritance has also been suggested).

References:
- OMIM
- GeneReviews

Genes

COL11A1, COL2A1, COL9A1, FZD4, KCNJ13, LRP5, NDP, TSPAN12, VCAN

Indications

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

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: 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

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 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.
- Vitreoretinopathy: Sequencing Panel.