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

- 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.

### 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 uncleotted cord or cardiac blood

**Specimen Collection and Shipping:**
- Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not 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, electoretinogram (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.