The sensitivity and specificity of this method for CNV detection is highly dependent on the size of the event, sequence context, size of exon(s) involved and depth of coverage. The assay is highly sensitive for CNVs of 500 base pairs or larger and those containing at least 5 exons. Single or multi-exon deletions or duplications are expected to be below the detection limit of this analysis.

Analytical sensitivity for sequence variant detection is ~99%.

**Specimen Requirements**

**Submit only 1 of the following specimen types**

**Type: Saliva**

**Specimen Requirements:**
Orangene™ Saliva Collection Kit
Orangene™ 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.
Type: DNA, Isolated

Specimen Requirements:
Microtainer
8µg
Isolation using the Perkin Elmer™ 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: 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 72 hours of collection. Do not freeze.

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.
- Retina/Photoreceptor Dystrophy: Deletion/Duplication Panel.