Familial Mutation Testing: Targeted Sequencing

**Test Code:** KM  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81403 x1

### Condition Description

### Indications

Custom DNA sequencing testing is available for any previously characterized DNA variation in a family. This service can also confirm DNA variations identified in a research laboratory. Such confirmation allows release of research findings to the patient as well as prenatal diagnosis and carrier testing for other at-risk family members. To perform this assay we require details of the genetic variation in the family and a DNA sample from a previously tested individual as a positive control. Please call (404) 778-8500 to discuss this testing prior to sample collection.

### Methodology

The region of DNA surrounding the mutation is amplified by PCR and sequenced in both the forward and reverse direction.

### Detection

Over 99% of previously identified mutations will be detected by this assay.

### 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 72 hours of collection. Do not freeze.

**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: Saliva**

**Specimen Requirements:**

- Oragene™ 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.

### Special Instructions

Please call (404) 778-8500 to discuss testing and to obtain prices prior to sample collection.