# 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™ 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

Oragene™ 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.