# 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**

<table>
<thead>
<tr>
<th>Type: DNA, Isolated</th>
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| **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. |

<table>
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<tr>
<th>Type: Whole Blood (EDTA)</th>
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| **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 24 hours of collection. Do not refrigerate or freeze. |

<table>
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<th>Type: Saliva</th>
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| **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. |

## Special Instructions

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