**Familial Mutation Testing: Targeted Deletions/Duplications**

**Test Code:** DKMDD  
**Turnaround time:** 2 weeks - 3 weeks  
**CPT Codes:** 81228 x1

### Condition Description

### Indications

Known mutation testing for a specific deletion or duplication is available for those genes for which we offer full gene del/dup testing. (Please see our test menu for a list of genes for which del/dup testing is offered.) 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

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications.

### 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 24 hours of collection. Do not refrigerate or freeze.

#### Type: DNA, Isolated

**Specimen Requirements:**  
Microtainer 3µg  
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.