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

* Preferred specimen type: Whole Blood

#### Type: Whole Blood

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

#### Type: Saliva

Specimen Requirements:

Oragene™Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.