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) or ACD (yellow 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.