Familial Mutation Testing: Targeted Deletions/Duplications

<table>
<thead>
<tr>
<th>Test Code: DKMDD</th>
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<td>Turnaround time: 2 weeks - 3 weeks</td>
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<tr>
<td>CPT Codes: 81228 x1</td>
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**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.