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: 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.

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 uncotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

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