

Maturity Onset Diabetes of the Young (MODY): Deletion/Duplication Panel

Test Code: MD220

Turnaround time: 2 weeks

CPT Codes: 81228 x1

Condition Description

Maturity-onset diabetes of the young (MODY) is defined by the clinical features of early-onset Type 2 (non-insulin-dependent) diabetes and an autosomal dominant inheritance. Pathogenic variants in four genes have been shown to cause MODY, with each having a defined clinical presentation. In the European Caucasian population, molecular analysis of these four genes is useful in confirming the clinical diagnosis of MODY and the prediction of the future clinical course.

References:

- Hattersley. (1998), *Diabet Med*, 15:15-24.
- Stride and Hattersley. (2002), *Ann Med*, 34:207-216.

Genes

[GCK](#), [HNF1A](#), [HNF4A](#), [PDX1](#)

Indications

This test is indicated for individuals with:

- Confirmation of a clinical diagnosis of maturity-onset diabetes of the young (MODY).
- Carrier testing in adults with a family history of hereditary maturity-onset diabetes of the young (MODY).

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

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 72 hours of collection. Do not freeze.

Type: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ 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.

Related Tests

- Maturity Onset Diabetes of the Young (MODY): Sequencing Panel