Inherited Metabolic Disorders: Deletion/Duplication Panel

**Test Code:** MD310
**Turnaround time:** 2 weeks
**CPT Codes:** 81228 x1, 81401 x1, 81404 x1, 81405 x1

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### Condition Description

Inherited metabolic disorders refer to diseases caused by defects in genes that are involved in the body’s metabolism. These usually involve the production, conversion, or use of energy. Traditionally, inherited metabolic conditions were broadly classified as disorders of carbohydrate metabolism, amino acid metabolism, organic acid metabolism, or lysosomal storage diseases. This test analyses genes involved in complex metabolic processes in the body including but not limited to the above four categories.

Reference:
- OMIM.

### Genes

- ACAD9, ACADL, ACADM, ACADS, ACADVL, ACSF3, AGA, AGL, ARSA, ARSB, ASL, ASS1, ATPAF2, AUH, BCKDHA, BCKDHB, CD320, CLN3, CLN5, CLN6, CLN8, CPS1, CPT1A, CPT2, DBT, DLD, ENO3, ETFA, ETFB, ETFDH, G6PC, GAA, GALC, GALNS, GBE1, GLA, GLB1, GM2A, GNPTAB, GYS1, GYS2, HADHA, HADHB, HGSNAT, HLC5, HMGC1, HMGC2, HYAL1, IDS, IDUA, IVD, LIPA, LMBRD1, LPIN1, MAN2B1, MANBA, MCC51, MCC52, MCEE, MCOLN1, MFSD8, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGA, NAGLU, NAGS, NEU1, NPC1, NPC2, OPA1, OTC, PC, PCGA, PCGB, PFKM, POLG, PPT1, PYGL, PYGM, SERAC1, SGSH, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC27A4, SLC7A7, SMPD1, SUCGL1, SUMF1, TAZ, TMEM70, TPP1

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of inherited metabolic disorders.

### 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. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

#### 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

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

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

#### Type: Isolated DNA

Specimen Requirements:
- In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.
Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Inherited Metabolic Disorders: Sequencing Panel