Inherited Metabolic Disorders: Deletion/Duplication Panel

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

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, CLN8, CLN9, CLN10, CPT1A, CPT2, DBT, DLD, ENO1, ENO2, ETFA, ETFB, ETFDH, G6PC, GALT, GALC, GALT, GBE1, GLA, GLB1, GM2A, GNPTAB, GYPS1, GYPS2, HADHA, HADHB, HGSNAT, HLC5, HMGCCL, HMGCCL2, HYAL1, IDS, IDUA, IVD, LIPA, LMBRD1, LPIN1, MAN2B1, MANBA, MCCC1, MCCC2, MCEF, MCOLN1, MFSD8, MLYCD, MMAA, MMAB, MMACHC, MMACHF, MMATRI, MTRA, NAGA, NAGLU, NAGS, NEU1, NPC1, NPC2, OPA3, OTC, PG, PC, PI, PKH, POLG, PPT1, PYG, PYGM, SERAC1, SGSH, SLC17A5, SLC25A15, SLC25A20, SLC25A25, SLC25A4, SLC25A7, SMPD1, SUCLG1, 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.

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: 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.

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.

Related Tests

- Inherited Metabolic Disorders: Sequencing Panel