

Glycogen Storage Disorders- Muscle: Deletion/Duplication Panel

Test Code: MD152

Turnaround time: 2 weeks

CPT Codes: 81228 x1

Condition Description

Glycogen storage disorders (GSDs) are a group of inherited genetic defects of glycogen metabolism. Most of them have autosomal recessive inheritance, however there are a few exceptions. There are more than 20 subtypes classified by the specific enzyme deficiency, affected tissue, and disease phenotype. Clinical and biochemical features continue to be used reliably to assign patients to this general disease category. Identification of the precise genetic defect is important, as molecular analysis is likely to expand the clinical spectrum of GSDs, may provide data relevant to prognosis and future therapeutic intervention and is important for carrier testing and early prenatal diagnosis.

The overall incidence of GSDs as a group is estimated to be 1 in 20,000-43,000 births. GSDs primarily affect the liver, the muscle, or both. Although the phenotype range is broad, the majority of clinical manifestations are hepatomegaly, failure to thrive, hypoglycemia, hyperlactatemia, hyperuricemia, and hyperlipidemia. This GSD panel covers 15 genes in which pathogenic variants cause the muscle isoforms of GSD and disorders that have overlapping phenotype with GSDs.

References:

- Burwinkel et al. (2005), *Am J Hum Genet*, 76:1034-1049.
- Morris et al. (1995), *J Inherit Metab Dis*, 18:28-32.
- Santer, Steinmamm, and Schaub. (2002), *Curr Mol Med*, 2:213-227.

Genes

[AGL](#), [ENO3](#), [GAA](#), [GBE1](#), [GYS1](#), [LAMP2](#), [PFKM](#), [PGAM2](#), [PGM1](#), [PHKB](#), [PRKAG2](#), [PYGM](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of glycogen storage disorders (GSDs).
- Carrier testing in adults with a family history of glycogen storage disorders (GSDs).

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: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. 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

- Glycogen Storage Disorders- Muscle: Sequencing Panel
- Pompe (dry blood spot - test code DZ; leukocytes - DW).