MAYO CLINIC LABORATORIES

**NEW TEST**

Notification Date: July 20, 2022
Effective Date: August 23, 2022

# Galactosemia, *GALT* Gene, Variant Panel, Varies

**Test ID:** GALMP

## Useful for:

- Second-tier test for confirming a diagnosis of galactosemia as indicated by enzymatic testing or newborn screening
- Carrier testing family members of an affected individual of known genotype (has variants included in the panel)
- Resolution of Duarte variant and Los Angeles (LA) variant genotypes

## Methods:

Targeted Genotyping Array

## Reference Values:

An interpretive report will be provided

## Ordering Guidance:

- The recommended as a first-tier test is galactose-1-phosphate uridyltransferase enzyme analysis; order GALT / Galactose-1-Phosphate Uridyltransferase, Blood.
- This genetic variant panel is recommended for individuals with a GALT enzyme value less than 24.5 nmol/h/mg of hemoglobin.

## Specimen Requirements:

| | |
|---|---|
| **Patient Preparation:** | A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant. |
| **Specimen Type:** | Whole blood |
| **Preferred:** | Lavender top (EDTA) or yellow top (ACD) |
| **Acceptable:** | Any anticoagulant |
| **Specimen Volume:** | 3 mL |
| **Collection Instructions:** | 1. Invert several times to mix blood.<br>2. Send whole blood specimen in original tube. **Do not** aliquot. |
| **Minimum Volume:** | 1 mL |

**Note:** Specimen preferred to arrive within 96 hours of collection.

## Specimen Stability Information:

| Specimen Type | Temperature | Time | Special Container |
|---|---|---|---|
| Varies | Ambient (preferred) | | |
| | Frozen | | |
| | Refrigerated | | |

## Cautions:

- This assay will not detect all of the known disease-associated variants that cause galactosemia. Therefore, the absence of a detectable variant does not rule out the possibility that an individual is a carrier of or affected with this disease.
- Many disorders may present with symptoms similar to those associated with galactosemia. Therefore, biochemical testing is recommended to establish the diagnosis of galactosemia prior to DNA analysis.
- A negative result does not eliminate the risk of carrier status for any of the included conditions, due to the possibility that the patient carries a variant that is not interrogated with this assay or the rare chance of a false-negative result for a tested variant. For tested variants, the negative predictive value of this screen is greater than 98%. The patient's residual risk to be a carrier after a negative screen is dependent on ethnic background and family history.
- A positive control was not available for all variants targeted on this panel. For more information regarding availability of a positive control for each variant see the Table (Targeted Variants) in Clinical Information.. The negative predictive value of these targets is unknown.
- Rare variants (ie, polymorphisms) exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations.(1) This assay was designed to specifically target known pathogenic or likely pathogenic variants. In rare cases, DNA variants of undetermined significance may be identified. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.
- Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.
- An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects deidentified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

## CPT Code:

81401

**Day(s) Performed:** Thursday & Sunday **Report Available:** 14 to 42 days

## Questions

Contact Michelle Raths, Laboratory Technologist Resource Coordinator at 800-533-1710.