Lab Dept: Anatomic Pathology

Test Name: GALT FULL GENE ANALYSIS

General Information

Lab Order Codes: GALTZ

Synonyms: GALT; Galactose-I-phosphate uridyltransferase Sequencing; Galactosemia GALT Full Gene Panel

CPT Codes: 81406 – GALT (galactose-I-phosphate uridyltransferase) full gene sequence

Test Includes: Next-generation sequencing to detect single nucleotide and copy number variants in one gene associated with galactosemia, GALT. Identification of a pathogenic variant may assist with diagnosis, prognosis, clinical management, familial screening, and genetic counseling for galactosemia. Additional first tier testing may be considered/recommended.

Logistics

Test Indications: Identifying mutations in individuals who test negative for the common mutations and who have a biochemical diagnosis of galactosemia or galactose-1 phosphate uridyltransferase activity levels indicative of carrier status.

Lab Testing Sections: Pathology - Sendouts

Referred to: Mayo Clinic Laboratories (Mayo Test: GALZ)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 14 – 21 days, performance days vary

Special Instructions: N/A

Specimen

Specimen Type: Whole blood

Container: Preferred: Lavender top (EDTA) tube
Alternate: ACD yellow top tube

Draw Volume:	3 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do not centrifuge. Specimen should remain in original collection tube. Store and ship at ambient. Forward promptly. Whole blood specimen stable ambient (preferred) for 4 days, refrigerated for 14 days.
Patient Preparation:	A previous bone marrow transplant from an allogenic donor will interfere with testing. Consult with Mayo for instructions for testing patients who have received a bone marrow transplant.
Sample Rejection:	Mislabeled or unlabeled specimens. All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Interpretive

Reference Range:	An interpretive report will be provided.
Critical Values:	N/A
Limitations:	Clinical Correlations: 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. If testing was performed because of a clinically significant family history, it is often useful to first test an affected family member. Detection of at least one reportable variant in an affected family member would allow for more informative testing of at-risk individuals.To discuss the availability of additional testing options or for assistance in the interpretation of these results, contact the Mayo Clinic Laboratory genetic counselors at 800-533-1710. Technical Limitations:Next-generation sequencing may not detect all types of genomic variants. In rare cases, false-negative or false-positive results may occur. The depth of coverage may be variable for some target regions; assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered. There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. Confirmation of select

reportable variants will be performed by alternate methodologies based on internal laboratory criteria.

This test is validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47bp. Insertions/deletions (indels) of 40 or more bp, including mobile element insertions, may be less reliably detected than smaller indels.

Deletion/Duplication Analysis: This analysis targets single and multi-exon deletions/duplications; however, in some instances single exon resolution cannot be achieved due to isolated reduction in sequence coverage or inherent genomic complexity. Balanced structural rearrangements (such as translocations and inversions) may not be detected.

This test is not designed to detect low levels of mosaicism or to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

For detailed information regarding gene specific performance and technical limitations, see Method Description or contact a laboratory genetic counselor.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent heterologous blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reclassification of Variants:
At this time, it is not standard practice for the laboratory to systematically review previously classified variants on a regular basis. The laboratory encourages health care providers to contact the laboratory at any time to learn how the classification of a particular variant may have changed over time.

Variant Evaluation:Evaluation and caterorization of variants is performed using published American College of Medical Genetics and Genomics and the Association for Molecular Pathology recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. Variants classified as benign or likely benign are not reported.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and periodic updates to these tools may cause predictions to change over time. Results from in silico evaluation tools should be interpreted with caution and professional clinical judgement.

Methodology:	Sequence Capture and Targeted Next-Generation Sequencing followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing.
References:	Mayo Clinic Laboratories April 2024
Updates:	4/18/2018: Updated container options. 2/23/2021: Updated method, Limitations 4/11/2024: Removed reference to frozen blood being acceptable. Removed any anticoagulant as acceptable. Added specimen stability.