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:	Chemistry - Sendouts
Referred to:	Mayo Medical 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, performed weekly
Special Instructions:	N/A

Specimen

Specimen Type:	Whole blood
Container:	Preferred: Lavender top (EDTA) tube Alternate: All anticoagulants are acceptable

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- preferred (refrigerated or frozen are ok) temperatures. Forward promptly.
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

Interpretive

Reference Range:	An interpretive report will be provided.
Critical Values:	N/A
Limitations:	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 clinically significant family history, it is often useful to first test an affected family member. Detection of a reportable variant in an affected family member would allow for more informative testing of at risk individuals. To discuss the availability of further testing options, for assistance in general test selection, or for assistance in the interpretation of results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710. 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, but 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 specific clinical disorders are suspected, evaluation by alternate methods can be considered. There may be regions of genes that cannot be effectively amplified for 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 assay will not reliably detect insertions/deletions (indels) of 40 or more base pairs (bp), including Alu insertions, long interspersed nuclear elements (LINES), and short interspersed nuclear elements (SINES). The bioinformatics software pipeline is verified to detect 95% of deletions up to 75 bp and insertions up to 47 bp.

Additionally, low level mosaic variants may not be detected.

This test is not designed 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.

Reclassification of Variant Policy: 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 status of a particular variant may have changed over time.

Variant Evaluation: Evaluation and categorization of variants is performed using published American College of Medical Genetics and Genomics (ACMG) and Association for Molecular Pathology (AMP) 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 judgment, Intronic and synonymous sequence variants not predicted to impact splicing or otherwise contribute to disease are not reported.

Methodology: Custom Sequence Capture and Targeted Next-Generation Sequencing followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing

References: Mayo Medical Laboratories Web Page (February 2021)

Updates: 4/18/2018: Updated container options.
2/23/2021: Updated method, Limitations