

Galactosemia, *GALT* Gene, Variant Panel, Varies

Test ID: GALMP

Explanation: On the effective date, extracted DNA will be acceptable for testing. Formatting of acceptable specimen types will also be standardized. Reflex testing will be updated to include Maternal Cell Contamination testing information.

Current Algorithm

For more information see [Galactosemia Testing Algorithm](#)

New Algorithm

For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge.

For more information see [Galactosemia Testing Algorithm](#)

Current Reflex Tests

None

New Reflex Tests

Test ID	Reporting Name	Available Separately	Always Performed
MATCC	Maternal Cell Contamination, B	Yes	No

Current Method Name

Targeted Genotyping Array

New Method Name

Targeted Genotyping Array followed by Multiplex Ligation-Dependent Probe Amplification (MLPA), Polymerase Chain Reaction (PCR), Relative Quantitative PCR (qPCR), or Sanger Sequencing, as needed

Current Specimen Stability		
Specimen	Temperature	Time
Varies	Ambient (Preferred)	
	Frozen	
	Refrigerated	

New Specimen Stability		
Specimen	Temperature	Time
Varies	Varies	

Current Specimen Required
<p>Multiple whole blood tests for galactosemia can be performed on one specimen. Prioritize order of testing when submitting specimens. See Galactosemia-Related Test List for a list of tests that can be ordered together.</p> <p>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.</p> <p>Specimen Type: Whole blood Container/Tube: Preferred: Lavender top (EDTA) or yellow top (ACD) Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions: 1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. Do not aliquot.</p>

New Specimen Required
<p>Patient Preparation: A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.</p> <p>Submit only 1 of the following specimens:</p> <p>Specimen Type: Whole blood Container/Tube: Lavender top (EDTA) or yellow top (ACD) Specimen Volume: 3 mL Collection Instructions: 1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. Do not aliquot. 3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days Additional Information: 1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed. 2. To ensure minimum volume and concentration of DNA are met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate. 3. For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.</p> <p>Specimen Type: Extracted DNA Container/Tube:</p>

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Preferred: Screw Cap Micro Tube, 2 mL with skirted conical base
Acceptable: Matrix tube, 1 mL
Collection Instructions:
1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
2. Include concentration and volume on tube.
Specimen Stability Information: Frozen (preferred) 1 year/Ambient/Refrigerated
Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Current Days Performed
Thursday, Sunday

New Days Performed
Varies

Current Specimen Retention Time
Whole Blood: 2 weeks (if available); Extracted DNA: 3 months

New Specimen Retention Time
Whole Blood: 28 days (if available); Extracted DNA: 3 months

Questions
Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.