

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform galactosemia DNA testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR GALACTOSEMIA TESTING

Patient's Name _____ **Date of Birth** ____/____/____ **Gender** F M

Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____

Genetic Counselor _____ **Counselor Phone** (____) _____

Patient's Ethnicity (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

Does the patient have SYMPTOMS? No Yes (check all that apply)

- | | | | |
|--|--|--|--|
| <input type="checkbox"/> Feeding problems | <input type="checkbox"/> Diarrhea | <input type="checkbox"/> Sepsis | <input type="checkbox"/> Tremors / ataxia |
| <input type="checkbox"/> Failure to thrive | <input type="checkbox"/> Jaundice | <input type="checkbox"/> Cataracts | <input type="checkbox"/> Developmental delay |
| <input type="checkbox"/> Vomiting | <input type="checkbox"/> Liver failure | <input type="checkbox"/> Speech problems | <input type="checkbox"/> Mental retardation |
| <input type="checkbox"/> Premature ovarian failure | | | |
| <input type="checkbox"/> Other _____ | | | |

RBC Transfusion? No Yes Unknown If yes, provide date of transfusion: _____

NEWBORN SCREEN Abnormal Normal Unknown N/A

GALT enzyme testing (Galactose-1-Phosphate Uridyltransferase)? Abnormal Normal Unknown NA

Gal-1-P levels? Abnormal Normal Unknown NA
 Is the patient on a lactose-free diet? No Yes Unknown

Does the patient have a FAMILY HISTORY OF GALACTOSEMIA? No Yes Unknown

If yes, please attach a PEDIGREE or specify the **RELATIONSHIP** of family member(s) to the patient. _____
 Is the relative a healthy carrier affected with galactosemia?
 What are the GALT mutations in the family member? _____

Circle the test below you intend to order.

- 0080125: Galactose-1-Phosphate Uridyltransferase:** GALT *enzyme* testing for diagnosing galactosemia.
- 0051175: Galactosemia, (GALT) Enzyme Activity & 9 Mutations:** *Enzyme and DNA* testing; use for diagnostic or carrier testing.
- 0051176: Galactosemia, (GALT) 9 Mutations:** GALT *DNA* testing only; evaluates the presence of 7 common mutations and 2 variants (Duarte and LA). Clinical sensitivity of 80% in Caucasians.
- 0051346: Galactosemia (GALT) Sequencing:** GALT gene sequencing; clinical sensitivity 99% for all ethnicities. Recommended for affected patients who do not have 2 mutations identified by 9 mutation panel.
- 2001961: Familial Mutation, Targeted Sequencing:** A copy of the relative's DNA laboratory result is **REQUIRED** for familial mutation testing.
- 0081296: Galactose-1-Phosphate in Red Blood Cells:** Gal-1-P analyte levels for *monitoring* patients with a known diagnosis.

For questions, contact Amanda Openshaw at (800) 242-2787 ext. 3921

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