ARUP LABORATORIES

**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 Name _____ Date of Birth_____ /_____/_____ Gender [ ] F [ ] M**

**Physician _____ Physician Phone (_____) _____Practice Specialty_____**

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

**Patient's Ethnicity** (check all that apply)
[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] Other _____

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

[ ] Feeding problems [ ] Diarrhea [ ] Sepsis [ ] Tremors / ataxia
[ ] Failure to thrive [ ] Jaundice [ ] Cataracts [ ] Developmental delay [ ] Premature ovarian failure
[ ] Vomiting [ ] Liver failure [ ] Speech problems [ ] Mental retardation [ ] 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.

**2006697 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, please contact an ARUP genetic counselor at (800) 242-2787, ext. 2141**

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