ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
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:** _____ **Sex:** ☐ Female ☐ Male

Physician: _____ **Physician Phone:** _____

Practice Specialty: _____ **Physician Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

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

☐ Cataracts ☐ Feeding problems ☐ Premature ovarian failure ☐ Vomiting
☐ Developmental delay ☐ Jaundice ☐ Sepsis ☐ Other symptom(s):
☐ Diarrhea ☐ Liver failure ☐ Speech problems _____
☐ Failure to thrive ☐ Mental retardation ☐ Tremors / ataxia _____

Has the patient had an RBC transfusion? ☐ No ☐ Yes (date of transfusion: _____) ☐ Unknown

Laboratory Findings

Newborn Screen	☐ Normal	☐ Abnormal	☐ Not Performed	☐ Unknown
GALT enzyme testing (Galactose-1-Phosphate Uridyltransferase)	☐ Normal	☐ Abnormal	☐ Not Performed	☐ Unknown
Gal-1-P levels	☐ Normal	☐ Abnormal	☐ Not Performed	☐ Unknown

Is the patient on a lactose-free diet? ☐ No ☐ Yes ☐ Unknown

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown

If yes, describe the test and results: _____

_____

Is there any relevant family history of galactosemia? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. _____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing),

Indicate the *GALT* mutations in the family member: _____

The relative is: ☐ a healthy carrier ☐ affected with galactosemia

Check the test you intend to order.

☐ **3001790** Galactose-1-Phosphate Uridyltransferase (GALT Enzyme), RBC: 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 seven common mutations and two 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 two mutations identified by 9 mutation panel.

☐ **0081296** Galactose-1-Phosphate in Red Blood Cells: Gal-1-P analyte levels for monitoring patients with a known diagnosis.

☐ **2001961** Familial Mutation, Targeted Sequencing: Tests for a known familial sequence variant; a copy of relative's lab result is REQUIRED.

Master Label

For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141