ARUP LABORATORIES

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 complete and submit with the test request form or electronic packing list.

GALACTOSEMIA TESTING PATIENT HISTORY FORM

Patient Name: _____ **Date of Birth:** _____

Sex Assigned at Birth: ☐ Female ☐ Male ☐ Intersex **Gender Identity (optional):** ☐ Female ☐ Male ☐ _____

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor's Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

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

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

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

Laboratory Findings

Newborn Screen ..... ☐ Normal ☐ Abnormal ☐ Not Performed ☐ Unknown

Gal-1-P levels ..... ☐ Normal ☐ Abnormal ☐ Not Performed ☐ Unknown

GALT enzyme testing (Galactose-1-Phosphate Uridyltransferase):
..... ☐ 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 variant testing).

Indicate the *GALT* variant(s) in the family member: _____

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

Master Label

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