

CP 3.2 Galactosemia - Follow-up of Positives

NOTE: Early detection and treatment are critical, as babies with this disorder can become septic and die in the first week of life.

POLICY: All Galactosemia confirmatory testing is to be conducted at the Associated Regional and University Pathologists, Inc. Laboratories (ARUP). ARUP will measure Galactose-1-Phosphate Uridyl Transferase (GALT) and will then provide GALT genotyping on all blood specimens with low or absent GALT activity (<14.7U/g Hb). Additionally, the state NBS Program will offer parent testing (at no charge) via ARUP for parents of newborns who have one identified mutation or variant present or Duarte Galactosemia (DG) in order to provide an assessment of risk of galactosemia in future pregnancies.

NOTE: ARUP conducts GALT testing on specimens delivered before noon on Mondays, Wednesdays, and Fridays, and will call out results on the same day (unless repeat testing is required, which may occur on the following day). DNA testing is done on Tuesdays and Thursdays, with a 2-3 day turnaround. Results are posted on ARUP Connect <https://www.aruplab.com/ii/login.jsp>

CUTOFFS: A Newborn is considered screen positive when the Galactose-1-phosphate uridyl transferase is ≤ 50 enzyme units.

GUIDELINES:

- Newborns with transferase values of ≤ 50 enzyme units require confirmatory testing. **The baby should immediately be placed on lactose-free diet as a precaution.**
- The NBS test for Galactosemia (GALT) is invalidated by a red blood cell transfusion; therefore it is essential that the newborn screening specimen be collected prior to a transfusion. If the baby is only screened after a transfusion and exhibits signs/symptoms of galactosemia or if there is a family history of galactosemia, the baby should have confirmatory galactosemia testing (i.e., a specimen should be sent to ARUP). The test can also be performed without those conditions upon request of the baby's PCP (this should be noted on the TRF).
- Genotype testing (of DNA) for galactosemia mutations is not affected by transfusions.

ASSOCIATED FORMS/DOCUMENTS:

3.2.1 Instructions for Collection of Samples for Galactosemia Testing

3.2.2 ARUP Fact Sheet (Technical Bulletin)

3.2.3 Test Requisition (Sample)

3.2 Galactosemia - Follow-up of Positives

PROTOCOL FOR INITIAL POSITIVE RESULTS:

Resp. Person	Action
NAPS Lab	<ul style="list-style-type: none"> • As soon as possible but no later than the end of the same day, calls appropriate ASC when initial result is positive for galactosemia. • Enters result and C of C into SIS.
ASC NBS Coord. or Program Specialist	<ul style="list-style-type: none"> • As soon as possible but no later than 24 hours after notification, calls newborn's physician/hospital unit (if infant is still hospitalized) with results and arranges for confirmatory specimen to be obtained and sent to ARUP via overnight delivery (See 3.9.2 A). Advises that baby be immediately placed on lactose-free diet and that breastfeeding be suspended pending confirmatory results. Suggests that mother be advised to pump breasts in interim to maintain milk supply. • Faxes ARUP Galactosemia fact sheet/technical bulletin to PMD (See 3.2.2). Offers assistance as necessary. <p><u>Note: Metabolic Specialists (either the ASC's consultant, or a specialist at the metabolic center where the baby will most likely be referred) are available to provide additional information and consultation as needed. The consultation should be noted in case notes.</u></p> <ul style="list-style-type: none"> • Enters appropriate tracking event into SIS. • Sends follow-up letter to physician confirming result, the need for a confirmatory specimen, and dietary recommendations. Includes <i>Why Retest for Galactosemia</i> brochure. • Sends Parent letter, along with the brochure, "<i>Why Retest for Galactosemia</i>" • Contacts the family directly as requested by the physician to arrange for collection of the confirmatory specimen. • Notifies facility where initial specimen was obtained (or if necessary, another facility more convenient to the family) about returning newborn and which test is needed. Faxes them the partially completed ARUP manual requisition (3.9.2 C) and instructions (3.9.2 A) regarding collection, handling, and mailing. • Faxes to ARUP a copy of the partially completed manual requisition (3.2.3). • Periodically checks "ARUP Connect" website (http://www.aruplab.com/Client-Services/arup-connect-login.jsp) for results. • Verifies follow-up progress by phone contact with the physician's office daily as necessary until the confirmatory specimen is obtained.

3.2 Galactosemia - Follow-up of Positives

	<ul style="list-style-type: none"> • If the physician and the ASC coordinator are unable to contact the family within three days of the initial notification, sends letter to family by overnight mail. (Also sends a copy by “certified receipt requested” mail to maximize chance of contact.). • If contact is not made within 3-4 days of sending letter, initiates a referral to public health nurse and arranges for a home visit. • Documents all attempts at notification, interactions with physicians and parents using tracking events or case notes in SIS. • Reports unusual occurrences such as missed cases, anomalous/inconsistent results, lost to follow-up cases, delays in contacting family, delays in analysis or reporting of confirmatory results, etc., to the NBS Nurse Consultant/ASC Contract Liaison. • Refers case to Child Protective Services, as appropriate and with the approval of the ASC Contract Liaison, as needed. • Follows case closely until confirmatory testing is completed. After confirmatory specimen is processed, follows protocol for Follow-Up of Confirmatory Results (see below for protocols for negative and positive confirmatory results).
--	--

PROTOCOL FOR CONFIRMATORY RESULTS:

Resp. Person	Action
ARUP Lab	<ul style="list-style-type: none"> • E-mails or calls NBS coordinator when specimen has arrived at lab • As soon as possible but no later than 4 business hours after completion of enzyme testing, phones ASC to report GALT results, which are available on ARUP Connect on ARUP’s website. If DNA is pending, and fax is desired, ASC can request fax from ARUP. If no DNA is pending, results will automatically be faxed to ASC. • As soon as possible but no later than 4 business hours after completion of DNA testing, phones genotype to ASC. Results are available on ARUP Connect and will automatically be faxed to the ASC.
ASC NBS Coord. or Program Specialist	<p>POSITIVE CONFIRMATORY RESULTS (Classical Galactosemia and Duarte/Galactosemia)</p> <ul style="list-style-type: none"> • As soon as possible but No Later Than 24 Hours After Notification, calls the newborn’s physician with POSITIVE GALT confirmatory results of ≤ 8 U/g Hb and explains GDSP recommendations for follow-up: <ol style="list-style-type: none"> 1. Baby should immediately be placed or kept on lactose-free diet. 2. Baby should immediately be referred to CCS-approved Metabolic Center. 3. Baby should be observed for s/s of galactosemia (lethargy, jaundice, hypotonia, hepatomegaly, sepsis) and followed up accordingly.

3.2 Galactosemia - Follow-up of Positives

If GALT is >8 U/g Hb but less than 14.7, calls baby's MD with results, and informs him/her that baby most likely does not have Galactosemia but may be a carrier, and that DNA results will be sent to him/her.

Note: Medical consultants are available through each ASC to provide additional information and consultation as needed.

- Sends follow-up letter to physician, confirming notification and recommendations. Includes lab report from ARUP.
- Enters confirmatory results in SIS (Confirmatory Results screen).

For babies with GALT ≤ 8 U/g Hb or with DNA results indicative of Classical or Duarte Galactosemia, assists provider in referring baby to a NBSP and CCS-approved metabolic center for diagnosis and treatment.

- Offers parent studies to PCP/specialist if baby is found to be a carrier (i.e., one mutation) or a DG.
- Documents all attempts to notify physicians and parents and interactions with them by utilizing Tracking Events and Case Notes in SIS
- If the physician and/or the ASC coordinator are unable to contact the family within three days of the initial notification, sends letter by overnight mail (for classical Galactosemia). Also sends a copy by "certified receipt requested" mail to maximize chance of contact.
- If contact is not made within 3 days of sending letter, initiates a referral to the appropriate Public Health Department and arranges for a home visit from a public health nurse.

Because of potentially lethal consequences of baby not receiving treatment, discusses course of action with NBS Nurse Consultant/Contract Liaison.

- Follows case by maintaining contact with the physician's office daily as necessary and offering assistance until resolved, either by confirmed diagnosis and initiation of treatment, infant death, noncompliance, or lost to follow-up.
- Reports any unusual occurrences such as missed cases, lost to follow-up cases, delays in contacting family, delays in analysis, or reporting of confirmatory results, etc., to the NBS Nurse Consultant/ASC Contract Liaison.
- Refers case to Child Protective Services as appropriate and with ASC Contract Liaison approval.
- Enters confirmatory results in SIS (Confirmatory Results screen).
- Resolves case on Case Resolution Screen in SIS.

3.2 Galactosemia - Follow-up of Positives

NEGATIVE CONFIRMATORY RESULTS

- Phones physician with NEGATIVE confirmatory result
- Enters appropriate Tracking Events and case notes in SIS
- Enters confirmatory results in SIS (Confirmatory Results screen)
- Sends letter to physician to confirm phone notification of negative confirmatory test; Includes lab report from ARUP.
- Resolves case on Case Resolution Screen in SIS.