

GALACTOSEMIA

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Galactosemia is an elevation of galactose and/or a galactose variant in the blood. Galactose is one of the two monosaccharides which make up lactose, the carbohydrate of human milk, cow's milk, and most non-soy formulas. Classic galactosemia is autosomal recessive.

Clinical Features

The classic form of galactosemia is due to almost total deficiency of galactose-1-phosphate uridyl transferase (Gal-1-PUT), the enzyme which catalyzes the conversion of galactose-1-phosphate to glucose-1-phosphate. The enzyme deficiency results in elevation of galactose-1-phosphate in blood and other tissue. The clinical features include neonatal hypoglycemia, liver dysfunction, prolonged neonatal jaundice, failure to thrive, lethargy, and susceptibility to overwhelming infection particularly due to *E. coli* and other gram negative bacteria. Later manifestations include cataracts, chronic liver disease, renal dysfunction, failure to thrive, and mental retardation.

There are several genetic variants associated with lesser reductions in the activity of the transferase enzyme (e.g., Duarte Variant). Most of the variants are asymptomatic and are of little or no clinical importance. However, they are often associated with elevated blood galactose which will be detected on newborn screening and will need to be carefully distinguished from the clinically important variants. Infants suspected of having any of the forms of galactosemia need prompt specific diagnosis.

Laboratory Screening Tests

The Kansas Newborn Screening uses the Beutler test to identify children with galactosemia and its variants.

The Beutler enzyme test is an immuno fluorescence test for the presence of the normal enzyme (Gal-1-PUT) in red blood cells.

The Beutler test depends upon fluorescence produced by the normal enzyme cascade in red blood cells. The test is abnormal when the enzyme activity is reduced and little or no fluorescence is observed. Patients with classic galactosemia have no enzyme activity; patients with a variant form will have reduced, but not absent, activity and fluorescence. Gal-1-PUT is sensitive to heat, so a false positive may result if the sample has been heat damaged. Because the test assays enzyme activity in red blood cells, false negatives can result from a blood transfusion. **NOTE: The newborn screening specimen should always be obtained before an infant receives a blood transfusion.**

See Table 4 below for normal values and laboratory criteria for requesting repeat samples.

Normal Values and Laboratory Criteria for Requesting Repeat Samples

ANALYTE	NORMAL RESULTS	RESULTS REQUIRING CONSULTATION WITH SPECIALIST	RESULTS REQUIRING REPEAT NEWBORN SCREEN
Gal-1-PUT (Beutler test)	Fluorescence present	No fluorescence	Partial fluorescence

TABLE 4

All phoned results are followed by mailed or faxed confirmation. All tests are screening tests. Abnormal results need full evaluation before a diagnosis is confirmed.

Confirmatory Testing

Newborn screening tests are not diagnostic and suspected galactosemia must be specifically confirmed and referred to a specialist for galactosemia.

Treatment

Most complications of galactosemia syndromes are effectively treated by a dietary exclusion of any products that contain galactose and/or lactose additives. This diet must be followed for life and requires close supervision. The long term developmental outlook for children identified and treated early is good compared to the development expected in untreated children. However, even with early diagnosis and strict dietary restrictions, children with galactosemia are at risk for speech and language disorders, relatively mild developmental delay and in females, ovarian failure. Affected children should be followed regularly by appropriate specialists.

Screening Practice Considerations

The Beutler test is abnormal in all infants with severe (classical) galactosemia even if the specimen is obtained before lactose is ingested, unless the infant has had a recent transfusion. **If a child is to be transfused, the initial newborn screening should be obtained before transfusion.**

The enzyme is prone to damage if the sample is delayed in transit or exposed to high temperatures, so false positive Beutler tests are not uncommon, particularly in the summer. Prompt confirmatory testing is required even if there is evidence to suggest that the results may be due to a false positive screen (i.e. enzyme may have been destroyed due to heat).

The Beutler test will be unaffected by the formula given to an infant. If galactosemia is a clinical consideration, especially in an infant with non-glucose urine reducing substances and clinical symptoms of galactosemia, begin a galactose-free formula immediately. The Gal-1-PUT test will be unaffected by this potentially life-saving measure. If galactosemia is suspected, immediate consultation with a metabolic physician is strongly advised. If mom is breast-feeding, have her

continue to pump until results of confirmatory are received. In the meantime give a galactose-free formula such as Isomil, Nutramigen or Pro So bee.

Medical Consultants

Medical consultants are available to provide consultation for the follow-up, evaluation, and long term management of children with Galactosemia.

Referral to a Medical consultant is strongly recommended for assistance in definitive diagnosis, treatment and follow-up of the disorder. More information can be obtained from any of consultants listed:

Dr. Majed Dasouki
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Brenda Issa
KU School of Medicine - Wichita
Wichita, KS
Office: 316-962-7386

It is strongly recommended that prior to repeating the newborn screen practitioners should confer with one of the consultants. The consultant may recommend repeating the State Newborn Screen, or they may suggest labs to draw and analyze in a practitioner's local lab. The consultant may wish to see the newborn in the office and do the lab work and assessment there. Whatever the case, we want to prevent unnecessary lab draws or inappropriate testing on these infants. In summary, please contact the consultant in your vicinity prior to drawing the repeat blood work.

**Overview of Follow-Up Procedure
Abnormal Galactosemia Newborn Screening**

- 1) Newborn Screening follow-up team reviews the laboratory reports that are faxed overnight from the lab **or** records the information provided per telephone call from the laboratory on a white phone information sheet.
- 2) If the *GALT* result is "Present": the results are considered **normal**.
 - a) Follow-up team does not receive results.
 - b) Lab will fax or mail results to doctor listed on NBS card.
- 3) If the *GALT* result is "Diminished", the results are considered **borderline**.
 - a) Lab will contact follow-up team via phone with baby's information and test results. Follow-up team will document information on white phone slip.
 - b) Follow-up team will print out baby's information from DHEL database on a yellow sheet of paper, attach the white phone information sheet and write "*GALT*" with result on yellow sheet.
 - c) Follow-up team will enter data into Access database under *GALT*.
 - d) Follow-up team will call healthcare provider listed on report and:
 - i) Verify that they are seeing the baby.
 - ii) Inform them of results.
 - iii) Ask them to get a repeat NBS card submitted to the KS lab.
 - iv) Confirm doctor's fax number.
 - v) Inform them that a letter will be faxed to their office with the results and instructions.
 - e) Follow-up team will print *GALT* borderline letter and fax (or mail, if no fax) to healthcare provider.
 - f) Follow-up team will print *GALT* parent letter and *GALT* parent information sheet and mail to baby's parents to inform them that their child has an abnormal result.
 - g) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 1 month from date of letter.
 - h) Follow-up team will enter data into Excel spreadsheet "Presumptive Totals" located on the "H" drive.
 - i) Lab will fax or mail results to doctor listed on NBS card.
 - j) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 2 weeks from date of letter. NOTE: Name changes are documented on the copy of the lab report. Surname changes are also documented in WebIZ as an alias.
 - k) When complete, paperwork is filed by infant's date of birth.

- 4) If the *GALT* result is "Absent", the results are considered **presumptive**.
 - a) Lab will contact follow-up team via phone with baby's information and test results. Follow-up team will document information on white phone slip.
 - b) Follow-up team will print out baby's information from DHEL database on a green sheet of paper, attach the white phone information sheet and write "*GALT*" with result on green sheet.
 - c) Follow-up team will enter data into Access database under *GALT*.
 - d) Follow-up team will call healthcare provider listed on report and:
 - i) Verify that they are seeing the baby.
 - ii) Inform them of results.
 - iii) Ask them to notify parents and arrange appointment with specialist.
 - iv) Confirm doctor's fax number.
 - v) Inform them that a letter will be faxed to their office with the results and instructions.
 - e) Follow-up team will print *GALT* presumptive letter and *GALT* physician report form and fax (or mail, if no fax) to healthcare provider.
 - f) Follow-up team will print *GALT* parent letter and *GALT* parent information sheet and mail to baby's parents to inform them that their child has an abnormal result. NOTE: If infant is in the NICU, no parent letter is sent.
 - g) Follow-up team will enter data into Excel spreadsheet "Presumptive Totals" located on the "H" drive.
 - h) Lab will fax or mail results to doctor listed on NBS card.
 - i) Follow-up team will enter lab information into WebIZ and set a follow-up reminder for 2 weeks from date of letter. NOTE: Name changes are documented on the copy of the lab report. Surname changes are also documented in WebIZ as an alias.
 - j) When complete, paperwork is filed by infant's date of birth.



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ABSENT/REDUCED GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE (GALT)

CONDITION: CLASSICAL GALACTOSEMIA

DIFFERENTIAL DIAGNOSIS: Galactosemia (galactose-1-phosphate uridyltransferase deficiency); GALT heterozygotes; GALT variants; artifactual reductions due to enzyme inactivation by high temperature and/or humidity.

METABOLIC DESCRIPTION: In galactosemia, GALT deficiency results in accumulation of galactose-1-phosphate (Gal-1-P) and galactose, causing multi-organ disease.

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

- Contact the family to inform them of the newborn screening result and ascertain clinical status.
- Arrange immediate clinical evaluation; for reduced GALT with symptoms or absent GALT result, stop breast or cow milk based infant formula and initiate non-lactose feeding (powder-based soy formula)
- If reduced GALT result and infant is asymptomatic, repeat the newborn screening test.
- Consult with a specialist; refer if considered appropriate.
- Evaluate the infant (jaundice, poor feeding, vomiting, lethargy, bulging fontanel, and bleeding) and arrange diagnostic testing as directed by the specialist.
- Emergency treatment as recommended by the specialist. If baby is sick, stop breast milk and/or cow milk based formula and initiate non-lactose feedings.
- Educate family about importance of diet change.
- Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Quantification of erythrocyte galactose-1-phosphate (gal-1-P) and GALT. Classical galactosemia shows <1% GALT activity and markedly increased gal-1-P. Transfusions in infant can invalidate the results of erythrocyte enzyme assays. Enzyme variants may be distinguished by GALT electrophoresis or mutation analysis.

CLINICAL EXPECTATIONS: Classical galactosemia presents in the first few days of life and may be fatal without treatment. Signs include poor feeding, vomiting, jaundice and sometimes, lethargy and/or bleeding. Neonatal *E. coli* sepsis can occur and is often FATAL. Treatment is the withdrawal of human milk and cow milk based formula; if symptomatic, the necessary emergency management.

REPORTING: Report diagnostic result to family and Kansas NBS program.

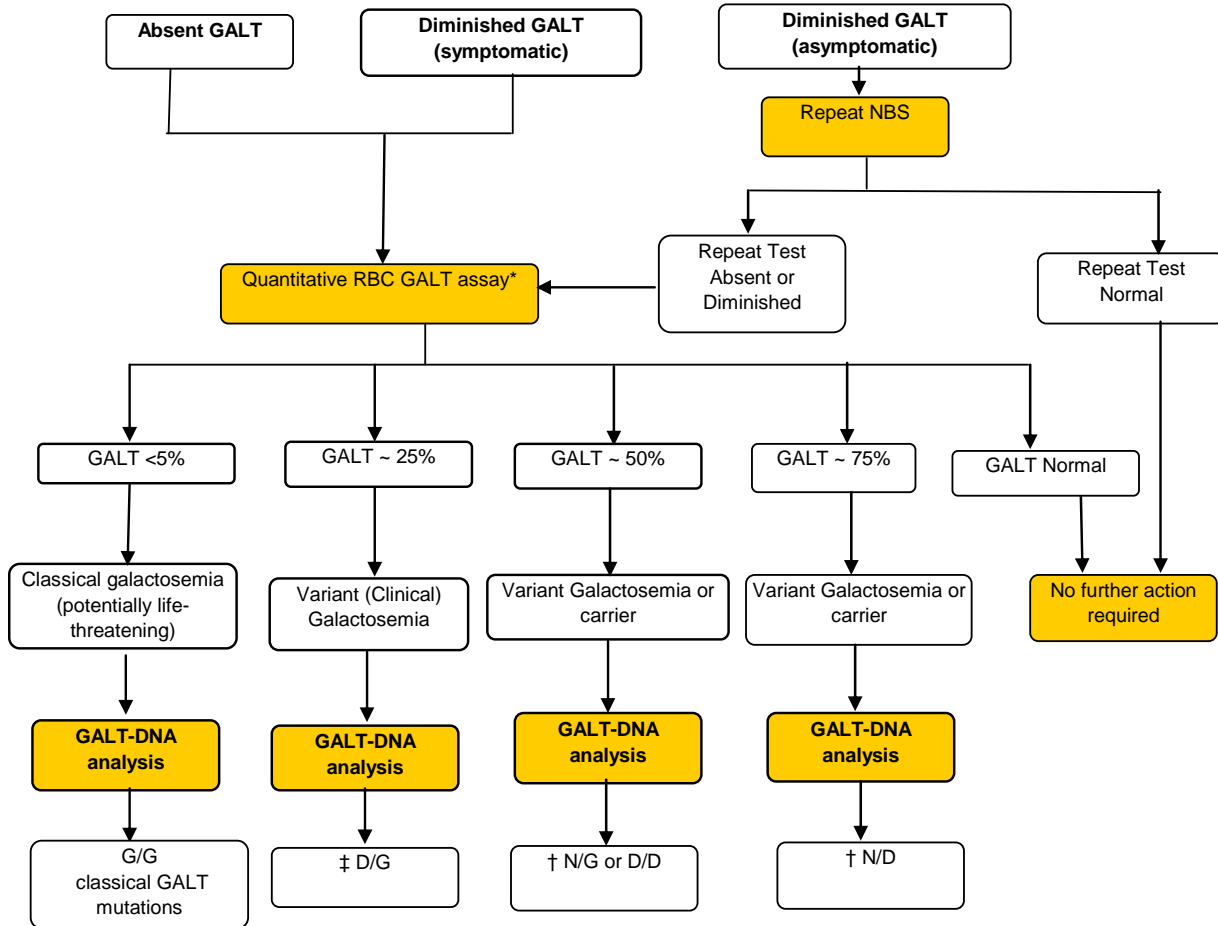
SPECIALISTS:

Dr. Majed Dasouki
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Brenda Issa
KU School of Medicine - Wichita
Wichita, KS
Office: 316-962-7386



ABSENT OR REDUCED GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE (GALT)



Abbreviations/Key

GALT = Galactose-1-phosphate uridyl transferase

NBS = Newborn Screening

RBC = Red blood cell

‡ = Clinically significant variant type

† = Benign variant type

* = Transfusions can invalidate results of RBC enzyme assays

D/D = Duarte homozygote, normal phenotype (~50% GALT activity)

D/G = Duarte/classical galactosemia compound heterozygote (~25% GALT activity)

G/G = classical galactosemia homozygote (<1% GALT activity)

N/D = normal/Duarte carrier (75% GALT activity)

N/G = normal/classical galactosemia carrier (~50% GALT activity)

Action steps are shown in gold (shaded) boxes; results are in plain boxes.

DISCLAIMER: These algorithms and guidelines were adapted from the American College of Medical Genetics algorithm sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines

EXAMPLE OF BORDERLINE GALACTOSEMIA (GALT) PHYSICIAN LETTER



Mark Parkinson, Governor
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH
AND ENVIRONMENT

www.kdheks.gov

**Abnormal Galactosemia (GALT) Lab Report
Kansas Newborn Screening Program**

Date

Doctor's Name
Address Line 1
Address Line 2

RE: Baby's Name
DOB: xx/xx/xxxx

MOTHER'S NAME: Mother's Name
MOTHER'S PHONE: xxx-xxx-xxxx

Specimen date: xx/xx/xxxx

GALT Enzyme Activity: Result
Expected Activity Value: Normal

This GALT enzyme activity is a **borderline** risk for galactosemia. Diminished GALT enzyme activity is considered to be indeterminate because galactosemia cannot be ruled out. A slightly decreased GALT enzyme level can occur if the specimen is exposed to heat for a prolonged period or due to analytical variability.

The final newborn screening lab report will be sent when all testing is completed.

RECOMMENDATION:

If the baby is asymptomatic, repeat the screening test within 1 to 3 days of receiving this notice, and send the specimen to the State lab for analysis. If baby was transfused, GALT mutation analysis may be needed. Repeat screening 120 days after blood transfusion. If the baby is symptomatic, or if the repeat screen is positive or indeterminate, additional testing and consultation with a specialist will be required.

In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.

For consultation, please contact:

Dr. Majed Dasouki
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Brenda Issa
KU School of Medicine - Wichita
Wichita, KS
Office: 316-962-7386

Additional information is available on the Kansas Newborn Screening Website at:
http://www.kdheks.gov/newborn_screening/info_professionals.htm.

You may contact the Newborn Screening Program at (785) 291-3363 or 1-800-332-6262 if you have any questions or concerns.

EXAMPLE OF PRESUMPTIVE GALACTOSEMIA PHYSICIAN LETTER



Mark Parkinson, Governor
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH
AND ENVIRONMENT

www.kdheks.gov

**Abnormal Galactosemia (GALT) Lab Report
Kansas Newborn Screening Program**

Date

Doctor's Name
Address Line 1
Address Line 2

RE: Baby's Name
DOB: xx/xx/xxxx

MOTHER'S NAME: Mother's Name
MOTHER'S PHONE: xxx-xxx-xxxx

Specimen date: xx/xx/xxxx

GALT Enzyme Activity: Result
Expected Activity Value: Normal

The absence of GALT enzyme activity is highly suggestive of variant or classical galactosemia. Untreated classical galactosemia quickly leads to symptoms of vomiting, weight loss, jaundice and hepatomegaly. Eventually, untreated patients develop permanent brain damage, cataracts and death.

The final newborn screening lab report will be sent when all testing is completed.

RECOMMENDATION:

Immediate consultation with one of the consultants listed below is essential for diagnostic testing and genetic counseling. Please call to arrange an immediate appointment.

In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.

For consultation, please contact:

Dr. Majed Dasouki
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Brenda Issa
KU School of Medicine - Wichita
Wichita, KS
Office: 316-962-7386

****PLEASE COMPLETE AND RETURN THE ENCLOSED PHYSICIAN REPORTING FORM WHEN FOLLOW UP IS COMPLETE****

Additional information is available on the Kansas Newborn Screening Website at:
http://www.kdheks.gov/newborn_screening/info_professionals.htm.

You may contact the Newborn Screening Program at (785) 291-3363 or 1-800-332-6262 if you have any questions or concerns.

EXAMPLE OF GALT PHYSICIAN'S REPORT FORM



Mark Parkinson, Governor
Roderick L. Bremby, Secretary

DEPARTMENT OF HEALTH
AND ENVIRONMENT

www.kdheks.gov

GALACTOSEMIA (GALT) NEWBORN SCREENING
PHYSICIAN REPORTING FORM

Return this form When Follow-Up is Complete

Date

Doctor's Name
Address Line 1
Address Line 2

If not a current patient of this practice, record name and
contact information for Primary Care Physician and return
form.

RE: Baby's Name
DOB: xx/xx/xxxx

Baby's name if different than listed

DIAGNOSIS EXCLUDED: Date Excluded:

Baby does NOT have Galactosemia or variant

DIAGNOSIS CONFIRMED: Date Diagnosis Confirmed:

Baby has Classical Galactosemia

Lab Results: (Please fill in and attach a copy of specialist's report)

enzyme level: GALT genotype:

Additional lab results:

Date treatment began:

Baby has variant Galactosemia

(Please circle one) Galactosemia carrier / Homozygous Duarte / Duarte carrier /
DG Galactosemia / Galactokinase deficiency / Galactose Epimerase deficiency

Date treatment began, if indicated:



Classical Galactosemia Information for Healthcare Professionals

Galactosemia is an inherited disorder resulting from the inability to digest galactose. Galactose-1-phosphate uridylyltransferase (GALT) deficiency is the cause of classical galactosemia and its milder variants. Infants with classical galactosemia, or severe GALT deficiency, generally present within the first weeks after birth with a life-threatening illness. Milder variants of galactosemia, such as Duarte, are not associated with such serious complications.

✓ Clinical Symptoms

The signs and symptoms of classical galactosemia typically appear within the first few days of life. Children with classical galactosemia may present with feeding problems, vomiting, diarrhea, jaundice and lethargy. Excessive bleeding may occur when blood is drawn. Hepatomegaly and cataracts are common findings on physical examination. Septicemia due to *E. coli* infection is a major cause of morbidity/mortality in infants with untreated galactosemia. A less common picture of classic galactosemia may be of an older infant with failure to thrive, developmental delay and feeding problems.

Variant galactosemia is a milder form of galactosemia. Many of these individuals will be asymptomatic, however, a metabolic disease specialist should evaluate all newborns with an abnormal screen.

✓ Incidence

Classical galactosemia occurs in approximately 1 in 50,000 newborns.

✓ Genetics of Galactosemia

Mutations in the *GALT* gene cause classical galactosemia and its variants. The *GALT* gene encodes galactose-1-phosphate uridylyltransferase. Galactose, as a component of lactose, is a sugar found in large quantities in milk. Because milk is the major source of nutrition for infants, babies with galactosemia must have all foods containing lactose and galactose removed from their diet immediately.

✓ Inheritance Pattern

Galactosemia is inherited in an autosomal recessive pattern. The parents of a child diagnosed with galactosemia deficiency are unaffected. Parents are carriers of the condition and have one normal *GALT* gene and one abnormal *GALT* gene. Each pregnancy between carrier parents has a 25% chance of producing a child affected with galactosemia, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

✓ Treatment

Treatment consists of complete removal of lactose-containing foods from the diet. Most newborns do well on a soy-based formula. Complete restriction of lactose-containing foods usually is required for at least the first year of life.

For those babies with variant galactosemia, a lactose “challenge” may be done (under the supervision of a metabolic specialist) at approximately one year of age, or when daily intake of lactose-containing foods diminishes. Because brain growth is greatest within the first year of life, many of these children can tolerate a limited amount of galactose in the diet once out of the infant stage, and can go on to eat a largely standard diet.

Children with classical galactosemia are at risk for brain damage and permanent neurological/developmental problems when dietary galactose is not strictly limited. The prognosis for individuals with galactosemia is very good when treatment is initiated before symptoms appear. However, even when treatment is promptly initiated, some individuals with classical galactosemia are at risk for

growth retardation and developmental delays (in particular, speech delays). Females with classical galactosemia are at risk for premature ovarian failure.

✓ **Screening Methodology**

Beutler assay of the specimen is performed to measure GALT enzyme activity. The enzyme evaluated in screening is prone to damage if the sample is delayed in the mail or exposed to high temperatures. Because GALT analysis is performed using red blood cells, there may be a false-negative result for up to 3 months if the infant has received a blood transfusion.

✓ **What to do After Receiving Presumptive Positive Galactosemia Screening Results**

- 1) The clinician should *immediately* check on the clinical status of the baby. Findings may include jaundice, vomiting, diarrhea, lethargy, excessive bleeding, hepatomegaly, and cataracts.
- 2) Refer the infant to a metabolic disease specialist.
- 3) ***Remove all breast milk and/or cow milk-based formula from newborn's diet. Initiate feedings with soy-based formula.***
- 4) **Molecular and/or biochemical testing to determine genotype (classic or variant) may be indicated.**
- 5) **Call KS Newborn Screening Program at 785-291-3363 with questions.**
- 6) **Report Clinical Findings to Newborn Screening Program at 785-291-3363.**

✓ **Confirmation of Diagnosis**

An abnormal newborn screening result requires immediate suspension of breast milk or cow milk-based formula. A metabolic specialist should promptly evaluate the newborn and initiate diagnostic testing.

✓ **Communication of Results to Parents**

If a baby has a presumptive positive galactosemia newborn screening result, additional testing needs to be performed to confirm a diagnosis. In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to initiate consultations or obtain repeat specimens when needed to complete the screening process.

If a baby is diagnosed with galactosemia, the following points should be conveyed to parents:

- ***Compliance with treatment is necessary for the best outcome.***
- ***Parents who have a child with galactosemia have a 25% chance with each pregnancy of having another affected child.***
- ***Prenatal diagnosis by molecular genetic testing may be available when parental mutations are known.***

For consultation, contact:

Dr. Majed Dasouki
KU Medical Center
Kansas City, KS
Office: 913-588-6326

Dr. Brenda Issa
KU School of Medicine - Wichita
Wichita, KS
Office: 316-962-7386

10/27/09



Classical Galactosemia Information for Parents

- **Overview**

Classical galactosemia is an inherited defect of galactose metabolism. It is caused by an enzyme deficiency that prevents the body from metabolizing galactose, or milk sugar, into glucose. The main dietary source of galactose is lactose, and is found in all forms of milk, except soy.

- **What is Classical Galactosemia?**

Classical galactosemia is a treatable disorder. It affects the way the body processes the sugar galactose, a component of milk and dairy products. Children with classical galactosemia cannot process galactose. As a result, galactose and other by-products build up in the bloodstream and cause physical and developmental damage. Poor mental and physical growth, cataracts, and serious liver and kidney problems are just a few of the possible effects of this disorder. In a child with classical galactosemia, galactose cannot be converted to glucose because the GALT enzyme does not work properly. Classical galactosemia can be life threatening in the first few weeks of life. Milder variants of galactosemia are not associated with serious complications. Galactosemia and lactose intolerance are totally separate medical conditions.

- **Why is newborn screening done for Classical Galactosemia?**

Newborn screening is done for classical galactosemia and other milder forms of galactosemia so babies with this condition can be diagnosed quickly. If babies are diagnosed quickly, treatment can begin immediately, reducing the chances of permanent damage from the accumulation of galactose and its by-products in the body.

- **Does a positive result from the Kansas Newborn Screening Lab mean that my baby has Classical Galactosemia?**

No, not necessarily. Newborn screening tests the baby's level of GALT or Galactose-1-phosphate uridyl transferase enzyme. The level was outside of the normal range in your baby. **Prompt follow up is important to help distinguish classical galactosemia from other benign forms.** Additional tests will need to be done to determine if your baby has classical galactosemia or not.

- **How common is Classical Galactosemia?**

Classical galactosemia occurs in approximately 1 in 50,000 births.

- **How is Classical Galactosemia inherited?**

Classical galactosemia is inherited in an autosomal recessive pattern. The parents of a child diagnosed with galactosemia do not have the condition. Parents are carriers of the condition. Carriers have one normal copy of the gene for the GALT enzyme and one abnormal copy. In order to have classical galactosemia, a child must inherit two abnormal copies of the gene, one from each parent. Each pregnancy between carrier parents has a 25% chance of producing a child affected with galactosemia, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

- **What are the signs and symptoms of Classical Galactosemia?**

Signs and symptoms of classical galactosemia appear in the newborn period. Symptoms may include:

- ✓ Problems with feeding
- ✓ Vomiting
- ✓ Diarrhea
- ✓ Yellowing of skin or whites of the eyes (jaundice)
- ✓ Over-sleepiness
- ✓ Excessive bleeding
- ✓ Swollen belly due to liver enlargement
- ✓ Cataracts (clouding of the eyes)

- **How is Classical Galactosemia diagnosed?**

Kansas newborn screening measures the activity of the GALT enzyme in your baby's blood. If your baby has diminished GALT activity and has no other symptoms, the newborn screening test should be repeated. **An absence of GALT activity in a newborn screen requires immediate suspension of breastfeeding or cow milk-based formula; soy-based formula feedings should be started.** The child should be evaluated as soon as possible for symptoms of galactosemia and referred to a metabolic specialist for diagnostic testing. The baby's enzyme levels will be re-measured and biochemical or genetic testing may be performed to confirm the diagnosis. Diminished GALT enzyme level means that your child most likely has a benign form of galactosemia that may not need treatment.

- **Is there a cure for Classical Galactosemia?**

No, there is no cure for classical galactosemia. Individuals who are diagnosed and treated before damage from galactose accumulation occurs can develop and grow normally. Even with early diagnosis and strict adherence to the diet, some children remain at risk for some problems, including growth retardation or developmental delays (particularly speech delays). Females with classical galactosemia are at risk for ovarian failure. The galactose free diet must be followed for life and requires close medical supervision.

- **How is Classical Galactosemia treated?**

Classical galactosemia is treated by removing all lactose and galactose from the diet.

- **Where can I get additional information?**

Genetic Fact Sheets for Parents:

<http://www.newbornscreening.info/Parents/otherdisorders/Galactosemia.html>

Parents of Galactosemic Children: <http://www.galactosemia.org/>

Children Living with Inherited Metabolic Disorders (CLIMB): <http://www.climb.org.uk>

11/24/08