Galactosemia Fact Sheet for Providers
Interpreting a Newborn Screen with an Elevated Total Galactose & Low Enzyme

What is the screening methodology utilized by the Public Health Laboratory Service, Oklahoma State Department of Health? Each filter paper is tested for a total galactose level. If the total galactose is elevated, reflex testing is performed to determine the presence or absence of galactose-1-phosphate uridyltransferase (GALT) activity using a qualitative methodology.

Special Note: If an infant was on a lactose-free formula (e.g. soy) at time of testing or transfused prior to specimen collection, the galactosemia screen results are invalid. For follow-up recommendations, please contact the Newborn Screening Program at (405) 271-6617, option 2 or 1-800-766-2223.

What is the normal range for galactosemia screening? A total galactose < 10 mg/dl or a total galactose < 12 mg/dl with Enzyme Present.

Is a screen result with low enzyme consistent with classic galactosemia? No, an infant with classic galactosemia usually will have an elevated total galactose (> 10mg/dl) with no enzyme activity. In our experience, a low enzyme result is consistent with a variety of genotypes including normal, variant (e.g. Duarte Galactosemia) or carrier.

Is further testing needed? Yes, at a minimum, a repeat screen is needed. If the repeat screen is abnormal, confirmatory testing will be requested by the metabolic specialist.

Special Note: Screening identifies infants at risk for classic galactosemia and in need of more definitive testing. As with any laboratory test, both false negative and false positive results are possible. Screening test results are insufficient information on which to base diagnosis or treatment.

Is treatment indicated? No. Based on the screen result of low enzyme, the infant is at low risk for classic galactosemia. However, if clinically indicated (i.e., a family history of classic or Duarte galactosemia and/or if infant is symptomatic) treatment with soy formula and confirmatory testing, not a repeat screen, are recommended.

What is Duarte galactosemia? Duarte galactosemia (DG) is a variant form of galactosemia. In this condition, the activity level of the enzyme required to break down galactose (a special sugar in milk) is reduced and functions at less than 25%. This means the enzyme works but does not function at a level that is needed, especially during the first year of life. Infants with DG are often treated with a lactose-free diet in infancy.

What is classic galactosemia? In classic galactosemia, the body lacks the GALT enzyme to break down galactose. Since the body cannot break down galactose, it builds up in the blood, tissues, and organs, which can cause serious complications and possible death. Treatment with a lactose-free diet is required for life.

Does galactosemia screening detect carriers? Yes, some carriers for the classic and Duarte galactosemia are detected. However, a normal screen does not indicate a normal genotype. Carriers do not require treatment.

Special Note: Classic and Duarte galactosemia are inherited in an autosomal recessive fashion (i.e., both parents are carriers). Genetic counseling is indicated for parents of any infant found to be a carrier or affected with classic or Duarte galactosemia.

What is my role in screening? You have been listed as the infant's planned Health Care Provider on the filter paper requisition and are required by Newborn Screening Program (NSP) *Rules* to arrange follow-up testing as recommended.

What if I have not seen this infant in my practice? The lab report lists the Mother's contact information (home phone number & address). If after notification of the Mother, you determine the infant is under the care of another physician or if you are unable to contact the mother after a reasonable search (i.e., phone call and letter), you should promptly contact the newborn screening follow-up program for assistance at (405) 271-6617.

For more information or assistance, call (405) 271-6617 or 1-800-766-2223