

Carbohydrate Metabolism Disorders

Galactosemia

Galactosemia is a condition of abnormal galactose metabolism caused by deficient functioning of any of three separate enzymes. These include galactose-1-P-uridyl transferase (GALT) deficiency or classical galactosemia; galactokinase deficiency (GALK); and UDP galactose-4-epimerase deficiency (GALE). Individuals with galactosemia are unable to break down and use the sugar galactose (a component of lactose found primarily in dairy products and human milk).

If undiagnosed, the affected infant with classical galactosemia may develop gastrointestinal disturbances, fail to gain weight and become jaundiced. Life-threatening infection can occur in the newborn period. Mental retardation and delayed physical growth occur in some untreated infants who survive. Some infants with low levels of GALT are subsequently diagnosed with a form of galactosemia called Duarte variant. Almost all cases of Duarte variant galactosemia are benign; however, most affected infants are treated during the first year of life as a precaution.

Infants with GALK deficiency only have cataracts. Infants with GALE deficiency will have varying outcomes. If the GALE deficiency is localized in the red blood cell, the infant does not have any symptoms of disease and no treatment is necessary. If the GALE deficiency involves other tissues, the clinical course is similar to that of GALT deficiency.

Inheritance: Autosomal recessive

Estimated Incidence: GALT (classical galactosemia)—1:60,000
Duarte variant galactosemia—1:16,000
GALK unknown, thought to be rare
GALE unknown, thought to be very rare

Abnormal Screen Result: Elevated total galactose with low GALT—at risk for classical galactosemia

Normal total galactose with low GALT—at risk for Duarte galactosemia or at risk for classical galactosemia if infant on non-lactose feeding

Elevated total galactose with normal GALT—at risk for GALK or GALE deficiency

Method of Notification: All results where the GALT is low are called to physician of record regardless of the total galactose level. Any other combinations of results are mailed to physician of record.

Next Steps if Abnormal: **Potential medical emergency when GALT is low and total galactose is elevated.** Regardless of total galactose result, see infant as soon as possible to ascertain health status if GALT is low. Change to soy based formula when GALT is low and total galactose is elevated. If total galactose is not elevated, consider change to soy based formula based upon clinical observation.

Repeat galactosemia screening as soon as possible. Consult pediatric metabolic specialist for further instructions and diagnostic evaluation.

If GALT is normal in the initial specimen, repeat galactosemia screening as soon as possible. **NO NEED TO STOP BREAST FEEDING OR CHANGE FORMULA TYPE** at this time. If total galactose remains elevated in the repeat specimen or if the GALT result is now low, refer to pediatric metabolic specialist for further diagnostic evaluation. Change to soy based formula.

Neonatal Presentation: GALT—hypoglycemia, jaundice, sepsis, failure to thrive
Duarte variant galactosemia—None
GALK—None
GALE—Usually none

Treatment: Galactose restricted diet for life.

Special Considerations

Reporting of Feeding Type--It is crucial that staff report whether the infant is on a lactose containing feeding (breast milk or cow's milk based infant formula), a soy based infant formula or any other non-lactose containing feeding (including IV fluids or total parenteral nutrition/hyperalimentation) so that the lab test can be interpreted appropriately. (A list of non-lactose containing feedings is included on page 28. This list is not all inclusive).

Exposure of the Specimen to Heat/Humidity--Both heat and humidity can affect the test for GALT. The enzyme activity can be diminished causing a false positive result for galactosemia. **NEVER put a newborn screening specimen in a plastic bag as this can increase exposure to both heat and humidity!**

Non-Lactose Containing Formulas/Feedings

Formula/Feeding Type	Product Name	Special Considerations
Soy-Based Formulas	Similac Isomil, Similac Isomil Advance, Similac Isomil DF, Similac Go and Grow Soy Enfamil Prosobee Lipil, Enfamil Next Step Prosobee Lipil Good Start Supreme Soy DHA & ARA, Good Start 2 Supreme Soy DHA & ARA Bright Beginnings Soy with Lipids, Bright Beginning Soy Pediatric Nutritional Drink Various Store Brand Soy Formulas	None
Free Amino Acid Formulas	Neocate Infant, Neocate Infant with DHA & ARA, Neocate One +, Neocate Junior E028 Splash Peptidite Junior Vivonex Pediatric Elecare	None
IV Fluids Total Parenteral Nutrition		None
Protein Hydrolysates	Nutramigen Lipil Pregestimil, Pregestimil Lipil Similac Alimentum Peptamen Jr, Peptamen Jr with Fiber, Peptamen Jr Prebio Vital Junior Pediatric Peptinex DT, Pediatric Peptinex DT with Fiber	Although considered non-lactose for the purposes of screening, these formulas contain some galactose and are not recommended for the routine management of infants and children with galactosemia.
Non-Lactose Formulas Containing Casein	Portagen Enfamil Lacto-Free Lipil Similac Sensitive, Similac Sensitive RS Pediasure, Pediasure with Fiber, Pediasure Enteral with Fiber & SoFOS Kindercal Nutren Junior, Nutren Junior with Fiber Resource Just for Kids, Resource Just for Kids with Fiber, Resource Just for Kids 1.5 Cal, Resource Just for Kids 1.5 Cal with Fiber Modulen IBD Compleat Pediatric Carnation Instant Breakfast Juice Drink	Although considered non-lactose for the purposes of screening, these formulas contain some galactose and are not recommended for the routine management of infants and children with galactosemia.

This list is not all-inclusive. Presence on this list does not constitute any endorsement by DHEC.