VDH VIRGINIA DEPARTMENT OF HEALTH

Division of Child and Family Health- Newborn Screening Follow-Up Program

Classic Galactosemia (GALT)

Health Care Professional Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An "abnormal" or "critical" result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is **vital** to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.

Disorder Indicated: Classic galactosemia (GALT) is an inherited condition in which the body is unable to properly digest galactose, a sugar found in all foods that contain milk. If a child with GALT eats galactose, undigested sugars build up in the blood rather than being used for energy. If GALT is left untreated, it can cause seizures, serious blood infections, liver damage, or even death. However, with early treatment, the child will most likely have a healthy life with avoiding developmental and intellectual disabilities. Some children with mild forms may not need any treatment.

Incidence	1 in every 30,000 to 60,000 newborns.
Analyte Measured	GALT enzyme (galactose-1-phosphate uridyltransferase) TGal (galactose and galactose-1-phosphate (G1P))
Normal Test Results	GALT > 5.5 U/dL TGal < 10mg mg/dL
Abnormal Test Results	GALT ≤5.5 U/dL TGal ≥ 10-15 mg/dL
Critical Test Results	GALT – 3 consecutive samples with abnormal results TGal ≥ 15 mg/dL ***(Critical results require immediate evaluation and follow-up)***
Signs and Symptoms *Please note: these findings may not be present in young infants or in milder forms of the disease*	When a child has classical galactosemia, you may see symptoms including: • Poor weight gain and growth (failure to thrive) • Poor feeding and sucking • Vomiting • Diarrhea • Sleeping longer or more often • Tiredness • Irritability • Low blood sugar (hypoglycemia)
Next Steps ***may*** include:	**Discuss the next steps of evaluation and *possible* treatment with the regional metabolic consultant** Provide parental education (see accompanying sheet) Clinical Assessment: Look for worsening jaundice and/or progressive emesis Quantitative RBC GALT Assay Urine reducing substance assay
Treatment (***if indicated***)	**Discuss with regional metabolic consultant before any diet changes**
Additional Resources	VDH Newborn Screening *http://vdhlivewell.com/newbornscreening* Baby's First Test *www.babysfirsttest.org* American College of Medical Genetics (ACMG) ACT Sheets *www.ACMG.net* Genetics Home Reference *https://ghr.nlm.nih.gov/* Galactosemia Foundation *http://www.galactosemia.org/*

Educational content adapted from www.babysfirsttest.org

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