VDH VIRGINIA DEPARTMENT OF HEALTH

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

Homocystinuria (HCY)

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: Homocystinuria (HCY) is a condition in which the body cannot break down methionine, an amino acid found in proteins. If left untreated, HCY could cause behavioral or learning disabilities, cardiac complications, strokes, brain damage or even death. However, if the condition is detected early and treatment is begun, individuals with HCY can lead healthy lives.

Incidence	1 in every 200,000 to 300,000 newborns.
Analyte Measured	Amino Acid: Methionine *Note: Total Parental Administration (TPN) could affect amino acid results*
Normal Test Results	Methionine < 70 µmol/L
Abnormal Test Results	Methionine ≥ 70 µmol/L to < 140 µmol/L
Critical Test Results	Methionine ≥ 140 µmol/L ***(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 Homocystinuria, you may see symptoms including: • Poor growth • Trouble gaining weight • Developmental delays • Behavioral difficulties • Weak muscle tone (hypotonia) • Pale skin and hair • Seizures (epilepsy) • Risk for stroke in infancy/childhood *Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.*
Next Steps ***may*** include:	**Discuss the next steps of evaluation and *possible* treatment with the regional Geneticist** Provide parental education (see accompanying sheet) Clinical Assessment Plasma Amino Acids Assay
Treatment **(*if indicated*)**	Restricted diet (discuss with the regional Geneticist)
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/* Homocystinuria Support Group *https://hcunetworkamerica.org/*

Educational content adapted from www.babysfirsttest.org

Virginia Newborn Screening Services
Keeping Virginia's Babies Healthy – One Foot at a Time

109 Governor St. 9th Floor, Richmond VA 23219
Phone: 804-864-7711 Fax: 804-864-7807