VDH VIRGINIA DEPARTMENT OF HEALTH

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

Homocystinuria (HCY)

Parent Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An "abnormal" result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is **very important** to determine if your baby has the disorder indicated.

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.
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:	**Follow up with your child's pediatric provider** Clinical assessment Laboratory Testing
Treatment ***(if indicated)***	Restricted diet (follow up with your child's pediatric provider)
Additional Resources	VDH Newborn Screening *http://vdhlivewell.com/newbornscreening* Baby's First Test *www.babysfirsttest.org* Genetics Home Reference *https://ghr.nlm.nih.gov/* Homocystinuria Support Group *https://hcunetworkamerica.org/*

Educational content adapted from www.babysfirsttest.org

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