VDH VIRGINIA DEPARTMENT OF HEALTH

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

Phenylketonuria (PKU)

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 Phenylketonuria (PKU) is a condition in which the body cannot break down phenylalanine, an amino acids found in proteins. If left untreated, PKU can cause developmental delays, brain damage or even death. However, if the condition is detected early and treatment is begun, individuals with PKU can lead healthy lives.

Incidence	1 in every 10,000 to 15,000 newborns.
Analyte Measured	Amino Acid: Phenylalanine *Note: Total Parental Administration (TPN) could affect amino acid results*
Normal Test Results	Phenylalanine < 140 µmol/L
Abnormal Test Results	Phenylalanine ≥ 140 µmol/L to < 280 µmol/L
Critical Test Results	Phenylalanine ≥ 280 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 classic Phenylketonuria, you may symptoms including: • Irritability • Seizures (epilepsy) • Dry, scaly skin (eczema) • "Musty" or "mouse-like" body odor • Pale hair and skin • Developmental delays *Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.* *Different forms of PKU can vary in severity of signs; however, classic PKU is the most severe.*
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 (low protein foods) including phenylalanine-free medical formula
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/* Children's PKU Network *www.pkunetwork.org/Childrens_PKU_Network/Home.html*

Educational content adapted from www.babysfirsttest.org

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

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