VDH VIRGINIA DEPARTMENT OF HEALTH

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

Carnitine Uptake Defect (CUD)

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: Carnitine Uptake Defect (CUD) is an inherited condition in which the body does not have enough carnitine in the cells to break down fats for energy. This results in an accumulation of fatty acids in the body. If left untreated, CUD could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals with MCAD can have healthy growth and development.

Incidence	Estimated 1 in every 100,000 newborns.
Analyte Measured	**Primary Marker**: C0 (*free carnitine*)
Normal Test Results	C0 >9.2 µmol/L
Abnormal Test Results	C0 ≤ 9.2 µmol/L to > 4.3 µmol/L
Critical Test Results	C0 ≤ 4.3 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 CUD, you may see symptoms including: • Poor appetite • Tiredness/Sleeping longer or more often • Vomiting/Diarrhea • Hypoglycemia (low blood sugar) • Behavior changes/irritability • Breathing difficulty • Seizures *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 Assay: Plasma free and total carnitine
Treatment **(*if indicated*)**	Restricted diet/Supplements (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/* Fatty Oxidation Disorder Family Support Group *http://www.fodsupport.org/*

Educational content adapted from www.babysfirsttest.org

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