VDH VIRGINIA DEPARTMENT OF HEALTH

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

Maple Syrup Urine Disease (MSUD)

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: Maple Syrup Urine Disease (MSUD) is a condition in which the body cannot break down certain parts of protein, including certain amino acids due to an enzyme deficiency. This results in an accumulation of the amino acids leucine, isoleucine and valine. If left untreated, MSUD could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals with MSUD can have healthy growth and development.

Incidence	1 in every 185,000 newborns.
Analyte Measured	Amino Acid: Leucine/Isoleucine *Note: Total Parental Administration (TPN) could affect amino acid results*
Normal Test Results	Leucine < 222 µmol/L
Abnormal Test Results	Leucine ≥ 222 µmol/L to < 300 µmol/L
Critical Test Results	Leucine ≥ 300 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 Maple Syrup Urine Disease, you may see symptoms including: • Urine that smells sweet like maple syrup • Poor appetite/ Weight loss/ Vomiting • Trouble sucking during feeding • High pitched cry • Sleeping longer or more often/Tiredness • Irritability • Developmental delays *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 Amino Acids, Urine Organic Acids
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/* MSUD Family Support Group *http://www.msud-support.org/*

Educational content adapted from www.babysfirsttest.org

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