

Isovaleric Acidemia (IVA)

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: Isovaleric Acidemia (IVA) is a condition in which the body cannot break down the amino acid leucine due to an inherited enzyme deficiency. This results in an accumulation of organic acids in the body. If left untreated, IVA could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals with IVA can have healthy growth and development.

Incidence	1 in every 230,000 newborns.
Analyte Measured	Organic Acid: C5 (<i>isovalerylcarnitine</i>)
Normal Test Results	C5 < 0.87 µmol/L
Abnormal Test Results	C5 ≥ 0.87 µmol/L to < 1.62 µmol/L
Critical Test Results	C5 ≥ 1.62 µmol/L <i>(Critical results require immediate evaluation and follow-up)</i>
Signs and Symptoms <i>Please note: these findings may not be present in young infants or in milder forms of the disease</i>	When a child has Isovaleric Acidemia, you may see symptoms including: <ul style="list-style-type: none"> • Poor appetite • Sleeping longer or more often • Tiredness • Vomiting • A “sweaty feet” smell • Seizures <i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i>
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: Urine Organic Acids, Plasma Acylcarnitine
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/ Organic Acidemia Association http://www.oaanswers.org/

Educational content adapted from www.babysfirsttest.org

