

Glutaric Acidemia Type I (GA I)

Health Care Professional Fact Sheet

<p>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.</p>	
<p>Disorder Indicated: Glutaric Acidemia Type I (GA I) is a condition in which the body cannot break down the amino acids lysine, hydroxylysine, and tryptophan due to an inherited enzyme deficiency. This results in an accumulation of organic acids in the body. If left untreated, GA I could cause brain damage or death. However, if the condition is detected early and treatment is begun, individuals with GA I can have healthy growth and development.</p>	
Incidence	1 in every 40,000 newborns (more common in Amish populations)
Analyte Measured	Organic Acid: C5-DC (<i>glutaryl</i> carnitine)
Normal Test Results	C5-DC < 0.25 µmol/L
Critical Test Results	C5-DC ≥ 0.25 µmol/L (Critical results require immediate evaluation and follow-up)
Signs and Symptoms	<p>When a child has untreated Glutaric Acidemia Type I, you may see symptoms including:</p> <ul style="list-style-type: none"> • Poor appetite/ Vomiting • Sleeping longer or more often/Tiredness • Irritability • Twitches/ Rigid muscles (spasticity) • Weak muscle tone (called hypotonia) • Delayed growth/Developmental delays • Excessive sweating/Fever <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i></p>
Next Steps may include:	<p>Discuss the next steps of evaluation and possible treatment with the regional Geneticist</p> <p>Provide parental education (see accompanying sheet)</p> <p>Clinical Assessment</p> <p>Assay: Urine Organic Acids, , Plasma Acylcarnitine</p>
Treatment (if indicated)	Restricted Diet, Supplements (Discuss with regional Geneticist)
Additional Resources	<p>VDH Newborn Screening http://vdhlivewell.com/newbornscreening</p> <p>Baby’s First Test www.babysfirsttest.org</p> <p>American College of Medical Genetics (ACMG) ACT Sheets www.ACMG.net</p> <p>Genetics Home Reference https://ghr.nlm.nih.gov/</p> <p>Organic Acidemia Association http://www.oaanswers.org/</p>

Educational content adapted from www.babysfirsttest.org

