

Elevated Organic Acid: C3 propionylcarnitine

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(s) Indicated: Multiple Carboxylase Deficiency (MCD), Methylmalonic Acidemia due to Mutase Deficiency (MUT), Methylmalonic Acidemia Cbl A and Cbl B forms (Cbl A,B), and Propionic Acidemia (PROP) are multiple organic acid disorders that could be identified through the elevation of the organic acid C3. These disorders are conditions in which the body does not have sufficient enzymes to remove ammonia from the body due to an inherited enzyme deficiency. Ammonia is accumulated through the breakdown of protein or amino acids. If left untreated, these disorders could cause brain damage, coma, or death. However, if the conditions are detected early and treatment is begun, individuals can have healthy growth and development.

Incidence	Varies by disorder, average 1 in every 50,000 newborns.
Analyte Measured	Primary Marker: C3 propionylcarnitine Secondary Markers*: May vary by condition (C4-DC, C5-OH, C3/C2) <i>* an abnormal secondary marker paired with an abnormal primary marker will result in critical group and require referral and diagnostic testing</i>
Normal Test Results	C3 < 7.3 µmol/L
Abnormal Test Results	C3 ≥ 7.3 µmol/L to < 9.7 µmol/L
Critical Test Results	C3 ≥ 9.7 µmol/L (Critical results require immediate evaluation and follow-up)
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 an organic acid disorder, you may see symptoms including: <ul style="list-style-type: none"> • Poor feeding/appetite • Vomiting • Hypotonia • Tiredness • Sleeping longer or more often • Difficulty breathing <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections</i></p>
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/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/ Organic Acidemia Association http://www.oaanews.org/

Educational content adapted from www.babysfirsttest.org

