VDH VIRGINIA DEPARTMENT OF HEALTH

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

Citrullinemia (CIT) / Argininosuccinic Aciduria (ASA)

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: Citrullinemia (CIT) and Argininosuccinic Aciduria (ASA) are conditions in which the body does not have sufficient enzymes to remove ammonia from the body. Ammonia is accumulated through the breakdown of protein or amino acids. If left untreated, CIT/ASA could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals can have healthy growth and development.

Incidence	CIT: 1 in every 57,000 newborns. ASA: 1 in every 70,000 newborns.	
Analyte Measured	Amino Acid: Citrulline	
Normal Test Results	Citrulline < 55 µmol/L	
Abnormal Test Results	Citrulline ≥ 55 µmol/L to < 100 µmol/L	
Critical Test Results	Citrulline ≥ 100 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 Citrullinemia, you may see symptoms including: • Sleeping longer or more often • Tiredness • Poor appetite • Vomiting • Seizures (epilepsy) • Irritability • Delayed growth.	When a child has Argininosuccinic Aciduria, you may see symptoms including: • Poor appetite • Sleeping longer or more often • Tiredness • Irritability • Vomiting • Trouble breathing • Seizures (epilepsy) • Involuntary body movements • Delayed growth
	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 Amino 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/* National Urea Cycle Disorders Foundation *http://www.nucdf.org/*	

Educational content adapted from www.babysfirsttest.org

Virginia Newborn Screening Services
Keeping Virginia's Babies Healthy – One Foot at a Time

109 Governor St. 9th Floor, Richmond VA 23219
Phone: 804-864-7711 Fax: 804-864-7807