

Citrullinemia (CIT) / Argininosuccinic Aciduria (ASA)

Parent Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An “abnormal” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is **very important** to determine if your baby has the disorder indicated.

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. Ammonia is a very toxic product which can cause irreversible harm to the brain within hours.

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.
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 Citrullinemia, you may see symptoms including: <ul style="list-style-type: none"> • 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: <ul style="list-style-type: none"> • Poor appetite • Sleeping longer or more often • Tiredness • Irritability • Vomiting • Trouble breathing • Seizures (epilepsy) • Involuntary body movements • Delayed growth
	<i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections</i>	
Next Steps <i>may</i> include:	Follow up with your child’s pediatric provider Clinical assessment Laboratory Testing	
Treatment <i>(if indicated)</i>	Restricted diet (follow up with your child’s pediatric provider)	
Additional Resources	VDH Newborn Screening http://vdhlivewell.com/newbornscreening Baby’s First Test www.babysfirsttest.org Genetics Home Reference https://ghr.nlm.nih.gov/ National Urea Cycle Disorders Foundation http://www.nucdf.org/	

Educational content adapted from www.babysfirsttest.org

