

Elevated Tyrosine

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 Indicated: There are several reasons why tyrosine might be elevated in the newborn screening sample. Some of these are temporary, while others may require treatment with diet or medication. Follow-up with a metabolic specialist is important to help distinguish those causes.

Incidence	1 in every 100,000 newborns.
Signs and Symptoms <i>Please note: these findings may not be present in young infants or in milder forms of the disease</i>	<p>A very rare disorder, Tyrosinemia I (TYR I) is a condition in which the body cannot break down the amino acid tyrosine due to an inherited enzyme deficiency. If left untreated, TYR I could cause potentially fatal kidney and liver issues. However, if the condition is detected early and treatment is begun, individuals with TYR I can have healthy growth and development.</p> <p>When a child has Tyrosinemia I (TYR I), you may see symptoms including:</p> <ul style="list-style-type: none"> • Diarrhea/Bloody stool • Vomiting/Poor weight gain • Sleeping longer and more often/Tiredness/Irritability • “Cabbage-like” odor • Yellowing skin (known as jaundice) • Increased bleeding or bruising/Swollen legs or abdomen • Developmental delays • Trouble breathing <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i></p>
Next Steps <i>may</i> include:	<p>Follow up with your child’s pediatric provider</p> <p>Clinical assessment Laboratory Testing</p>
Treatment <i>(if indicated)</i>	Restricted diet (follow up with your child’s pediatric provider)
Additional Resources	<p>VDH Newborn Screening http://vdhlivewell.com/newbornscreening Baby’s First Test www.babysfirsttest.org Genetics Home Reference https://ghr.nlm.nih.gov/ Network of Tyrosinemia Advocates http://notacares.org</p>

Educational content adapted from www.babysfirsttest.org

