VDH VIRGINIA DEPARTMENT OF HEALTH

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

Elevated Tyrosine

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 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.
Analyte Measured	Amino Acid: Tyrosine Ketone: Succinylacetone (*Note: Total Parental Administration (TPN) could affect amino acid results)*
Normal Test Results	Tyrosine < 355.0 µmol/L Succinylacetone < 2.00 µmol/L
Abnormal Test Results	Tyrosine ≥ 355.0 µmol/L to < 400.0 µmol/L Succinylacetone ≥ 2.00 µmol/L to < 4.99 µmol/L
Critical Test Results	Tyrosine ≥ 400.0 µmol/L Succinylacetone ≥ 5.0 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*	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. When a child has Tyrosinemia I (TYR I), you may see symptoms including: • Diarrhea/Bloody stool • Vomiting/Poor weight gain • Sleeping longer and more often/Tiredness/Irritability • "Cabbage-like" odor/Yellowing skin (jaundice) • Increased bleeding or bruising/Swollen legs or abdomen • Developmental delays
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 Organic Acids
Treatment **(*if indicated*)**	Restricted diet (consult 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/* Network of Tyrosinemia Advocates *http://notacares.org/*

Educational content adapted from www.babysfirsttest.org

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