

## Overview of Newborn Screening for Tyrosinemia – For Parents

### What is newborn screening?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to test for a group of conditions. One of these conditions is **tyrosinemia**. Babies who screen positive for tyrosinemia need follow-up tests done to confirm they have tyrosinemia. **Not all babies with a positive newborn screen will have tyrosinemia.**

### What is tyrosinemia?

When a person has tyrosinemia, his/her body is not able to break down **tyrosine**. Tyrosine is an **amino acid** (one of the “building blocks” used to make proteins). Tyrosine is found in many of the foods we eat.

Most people with tyrosinemia are missing an **enzyme** (a protein that helps our bodies function) called **fumarylacetoacetase** (also called FAH). When the FAH enzyme is missing, a person cannot break down tyrosine. People with tyrosinemia have high levels of tyrosine and other proteins in their bodies. One of these proteins is **succinylacetone**.

There are other reasons a person could have high levels of tyrosine in his/her blood. Follow-up testing is needed to identify the reason why a person has high levels of tyrosine.

### What causes tyrosinemia?

Tyrosinemia is an **inherited** (passed from parent to child) condition. Everyone inherits two copies of the gene for FAH. We inherit one copy of the FAH gene from our fathers and one copy from our mothers. Sometimes these genes have changes (also called mutations) that prevent the gene from working correctly. In order for a person to have tyrosinemia, he or she must have two FAH gene changes. People with one FAH gene change do not have tyrosinemia.

### What are the symptoms of tyrosinemia?

Every child with tyrosinemia is different. Most babies with tyrosinemia will look normal at birth. Symptoms of tyrosinemia can appear in the first months of life if a baby with tyrosinemia does not receive treatment.

Most of the symptoms of tyrosinemia are caused by the high level of succinylacetone in the blood of people with tyrosinemia. Some of the symptoms of untreated tyrosinemia include:

- Vomiting & diarrhea
- Feeding problems
- Poor weight gain
- Liver problems (including a large liver, **jaundice** or yellow skin, and bruising)
- Kidney problems
- Seizures

### What is the treatment for tyrosinemia?

There is no cure for tyrosinemia. However, there are treatments that can help with the symptoms. People with tyrosinemia usually need to follow a special diet that contains low levels of tyrosine and another amino acid called **phenylalanine**. Some people with tyrosinemia are given medicines or a special formula to drink. A person with tyrosinemia will need treatment for his/her entire life.

### What happens next?

Although there is no cure for tyrosinemia, good medical care makes a difference. Children with tyrosinemia should see a metabolic geneticist (a doctor who specializes in tyrosinemia and other related conditions) as well as their pediatrician. Your child’s doctor will work with the metabolic geneticist to coordinate any treatment, tests, or appointments that your child needs.

### Where are Indiana’s metabolic genetics clinics?

Indiana’s metabolic genetics clinics are located at Riley Hospital for Children at IU Health, Indianapolis, IN, (317) 274-3966 and The Community Health Clinic, Topeka, IN, (260) 593-0108.

### Where can I get more information about tyrosinemia?

- **STAR-G** – <http://newbornscreening.info/Parents/aminoaciddisorders/Tyrosinemia.html>
- **Region 4 Genetics Collaborative** – <https://www.region4genetics.org/education/families/>