TYROSINEMIA TYPE I (TYR I)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called tyrosinemia type I.

A positive newborn screen does not mean your baby has tyrosinemia type I, but it does mean your baby needs more testing to know for sure.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What is Tyrosinemia Type I?

Tyrosinemia Type I affects an enzyme needed to break down proteins from the food we eat. In tyrosinemia type I, the enzyme used to break down proteins is missing or not working properly.

A person who has tyrosinemia type I doesn't have enough enzyme to break down protein containing tyrosine. When the body can't break down tyrosine, it builds up in the body and causes health problems.

Tyrosinemia type I is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause tyrosinemia type I. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can Tyrosinemia Type I cause?

Tyrosinemia is different for each child. Some children with tyrosinemia have only a few health problems, while other children may have very serious complications.

If tyrosinemia is not treated, a child might develop:

- Vomiting
- Yellowing of skin (jaundice)
- Problems growing
- Liver problems
- Kidney problems
- Diarrhea
- Bone problems (rickets)

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for Tyrosinemia Type I?

Tyrosinemia Type I can be treated. Treatment is life-long and can include:

- Diet low in tyrosine —a dietitian will help you with the best diet for your child
- Medications to prevent liver and kidney problems

Children with tyrosinemia type I should see their regular doctor, a doctor who specializes in tyrosinemia type I, and a dietitian.

Prompt and careful treatment helps children with tyrosinemia type I live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening
Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Michigan Metabolic Coordinating Center
Michigan Medicine: 1-734-764-0579

Children's Special Health Care Services
Toll-free: 1-800-359-3722

MDHHS Michigan Department of Health & Human Services | Michigan Newborn Screening

Phone 1-866-673-9939
www.michigan.gov/newbornscreening
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