



Newborn Screening FACT Sheet

Tyrosinemia Type II (TYR II)

What is TYR II?

Tyrosinemia, type II (TYR II) is a condition that can affect the eyes, skin and intellectual development. It is considered an amino acid condition because people with TYR II are unable to break down an amino acid, a small molecule that makes up proteins, known as tyrosine. Detecting TYR II early and beginning treatment can prevent some of the severe outcomes of the condition.

What Causes TYR II?

When we eat food, enzymes help break it down. Some enzymes help break down proteins into their building blocks, called amino acids. Other enzymes break down these amino acids. In TYR II, the enzyme tyrosine aminotransferase (TAT) is not working correctly. TAT's job is to break down the amino acid tyrosine. Babies with TYR II either make non-working or do not make enough TAT. When TAT is not working correctly, the body cannot breakdown tyrosine. This causes tyrosine and other amino acids (methionine and phenylalanine) to build up in the body, which can be toxic.

What Symptoms or Problems Occur with TYR II?

[Symptoms are something out of the ordinary that a parent notices.]

There are three types of tyrosinemia (I, II, and III). TYR II tends to affect the skin and eyes. Signs of TYR II usually begin in the first year of life. These signs include:

- Sensitivity to light (called photophobia)
- Eye redness
- Skin lesions on the hands and feet
- Behavior changes
- Poor coordination

Many of these signs may occur when your baby eats food that his or her body cannot break down. They can be triggered by long periods of time without eating, illnesses, and infections.

If your baby shows any of these signs, be sure to contact your baby's doctor immediately.

What is the Treatment for TYR II?

Dietary Treatments

Your baby may need to be on a restricted diet in order to avoid certain proteins that his or her body cannot break down. Babies with TYR II may need to limit certain amino acids (phenylalanine, tyrosine, and methionine) in their diet. Amino acids are the building blocks of proteins. These are all found in many proteins, and phenylalanine is also found in artificial sweeteners.

Medication

Some medications may be available.

Things to Remember

Children who receive early and ongoing treatment for TYR II can have healthy growth and development. This is why newborn screening for TYR II is so important.

About half of the individuals who have been diagnosed with TYR II have intellectual disabilities. Early treatment can reduce the risk of developing intellectual disabilities.