



Newborn Screening FACT Sheet

Tyrosinemia (TYR I)

What is Tyrosinemia I?

Tyrosinemia I is an amino acid disorder. People with tyrosinemia I can't break down the amino acid tyrosine from the food they eat. This causes severe liver disease and other health problems if not treated.

What Causes Tyrosinemia I?

Enzymes help start chemical reactions in the body. Tyrosinemia I happens when the enzyme fumarylacetoacetase (FAH) is missing or not working. When FAH doesn't work, it can't break down tyrosine. Tyrosine and other harmful substances then collect in the blood. One of these is succinylacetone. This causes serious liver and kidney damage. It can also cause periods of weakness or pain.

What Symptoms or Problems Occur with Tyrosinemia I?

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

Babies usually show symptoms in the first few months of life. Some of the first signs are:

- diarrhea and bloody stools
- vomiting
- poor weight gain
- sleeping longer or more often
- irritable mood
- "cabbage-like" odor to the skin or urine

Liver problems are common. They can cause:

- enlarged liver
- yellow skin
- bleeding and bruising easily
- swollen legs and stomach

Kidney problems also happen and can cause:

- rickets, a bone thinning condition
- delays in walking

Babies with severe liver and kidney problems usually die without quick treatment.

What is the Treatment for Tyrosinemia I?

Doctors use these treatments for children with TYR I:

1. Medication – The doctor may prescribe Nitisinone (Orfadin®). This prevents liver and kidney damage. It also stops the weakness and pain. Your child should take Nitisinone right away.

2. Medical Formula – The doctor may prescribe a special formula with the right amount of amino acids. He or she will tell you what kind is best and how much to use.

3. Low-tyrosine diet – Most children with Tyrosinemia I need to limit foods such as cow's milk and regular formula. Your doctor and dietitian will decide on the best food plan for your child. Dietitians know what the right formulas are and foods to eat.

4. Blood, urine, and other tests – Regular blood and urine tests will show if your child's diet or medicine needs to be changed.

5. Liver transplantation – A liver transplant is one way to prevent liver cancer.

Things to Remember

Early treatment can prevent severe symptoms of liver and kidney problems and weakness and pain. Children who are treated early usually have normal growth and intelligence.

If treatment isn't started right away, children may have liver or kidney damage. Rickets might be a problem and need treatment. Delays in growth and development can also happen.