

Tyrosinemia Type I (TYR-I)

General Overview

Q. What is TYR-I?

A. TYR-I is a treatable disorder that affects the way the body processes protein. It is treatable, but can cause life-threatening illness.

Q. What happens to protein in a child with TYR-I?

A. Proteins are made up of amino acids. These amino acids are normally used to make new proteins or broken down further for energy. In a child with TYR-I, the enzyme needed to break down the amino acid tyrosine does not work properly. If left untreated, tyrosine and its byproducts build up in the bloodstream and cause serious health problems.

Q. What are the effects of having TYR-I if it is not treated?

A. Untreated TYR-I can result in heart, kidney and bone problems, brain damage, liver failure, and sometimes death.

Q. What is the treatment for TYR-I?

A. Tyrosinemia is treated with a drug called nitisinone and a special diet that carefully controls the amount of tyrosine that the body has to process. Treatment should begin shortly after birth. Since protein is essential for growth and development, the child must continue on a special formula that provides protein and essential nutrients, but contains little or no tyrosine. This special diet must be followed throughout life. Some doctors may also prescribe dietary supplements. People with TYR-I require specialized treatment through a specialty clinic with experience in treating this disorder.

Q. Why would a child have TYR-I?

A. Tyrosinemia is an inherited disorder. It results when a baby receives a double-dose of a non-working gene (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. How common is TYR-I?

A. About one in every 100,000 babies in Washington State is born with TYR-I. However, parents who have a child with TYR-I each carry one copy of the non-working gene. That means, with each pregnancy, there is a one in four chance of the child having TYR-I.

For more information about TYR-I, please see the Disorders section of our website: www.doh.wa.gov/nbs.



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