

## Tyrosinemia Type I (TYR I) (Ty-ro-seen-ee-me-a)

### What is TYR I?

Tyrosinemia I (TYR I) is a metabolic disorder. This means the body has a chemical imbalance. TYR I is a condition in which the body has a lack of the enzyme fumarylacetoacetate hydrolase (FAH), which is needed to break down the amino acid tyrosine. Amino acids are the building blocks of protein. When this occurs, it causes a build-up of tyrosine in the liver. TYR I is inherited. This means it is present at birth.

### What type of problems occurs with TYR I?

A baby who has this condition may have low weight, decreased growth rate, fever, diarrhea, vomiting, enlarged liver, and yellowing of the skin and the whites of the eyes. If untreated, this condition can lead to death. Early diagnosis and treatment will help prevent these problems.

### What is the chance my baby will have TYR I?

This condition occurs in less than 1 in every 100,000 births. It is more common in the French-Canadian or Scandinavian population. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

### What is the treatment of TYR I?

The treatment of this condition consists of following a special diet. Your baby's metabolic doctor will help you make sure that you baby gets the right diet and medical care. A liver transplant is sometimes necessary.

### Where in Virginia can I take my baby for care?

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is <http://www.vahealth.org/gns>.



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