AMINO ACID DISORDERS:

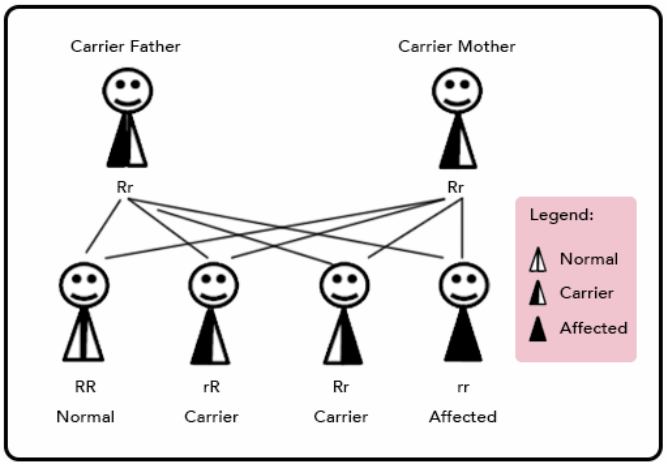
TYROSINEMIA TYPE I

What is Tyrosinemia Type I?

Tyrosinemia type I is a condition where there is accumulation of tyrosine (an amino acid which is a building block of protein) in the body. Due to this accumulation, other by-products of tyrosine, succinylacetone and succinylacetoacetate, also have increased levels in the blood. It is the rise of these chemicals that cause the signs and symptoms. Untreated children with this condition may have jaundice (yellowing of the skin), diarrhea and bloody stools, poor weight gain, irritability, drowsiness, enlarged liver and bleeding problems. Early detection can prevent liver damage.

What causes Tyrosinemia Type 1?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors (in this case, the fumarylacetoacetate hydrolase [FAH] enzyme), tyrosine (an amino acid) cannot be broken down. The accumulation of tyrosine, in turn, causes an increase in its by-products succinylacetone and succinylacetoacetate. It is the accumulation of these substances that causes the signs and symptoms of tyrosinemia.



Tryosinemia is an inherited condition. The FAH gene is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the FAH gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Tryosinemia have one working and one non-working gene coding for FAH. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.

If the child inherits the non-working gene from both parents, he or she will have Tyrosinemia Type I. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Tyrosinemia Type 1?

Children with Tyrosinemia Type 1 may present with jaundice (yellowing of the skin), diarrhea and bloody stools, poor weight gain, irritability, drowsiness, enlarged liver and bleeding problems. Without treatment, the liver can be damaged.

TYROSINEMIA TYPE I

WHAT IS THE TREATMENT OF TYROSINEMIA TYPE I?

The main treatment of Tyrosinemia Type 1 is through control of the diet. If the child is well or does not have any illness, he/she should continue taking the special milk formula and be on a low-protein diet as ordered by your pediatrician or metabolic specialist. It is important that the diet be followed to avoid any complications. Patients are also given nitisinone (NTBC), which helps to control tyrosine levels.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS UNWELL?

When a child is sick, has not eaten/ drank well during stressful events (such as surgery and severe infection) or has consumed a high amount of protein, there can be a build-up of toxic substances in the blood. Your child may present with yellowing of the skin, drowsiness, lethargy and bleeding. If not treated properly and immediately, it might lead to serious liver damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

Normal breakdown of protein vs Tyrosinemia Type 1 patient's breakdown of protein