Genetics Newborn Screening Program

Family Fact Sheet

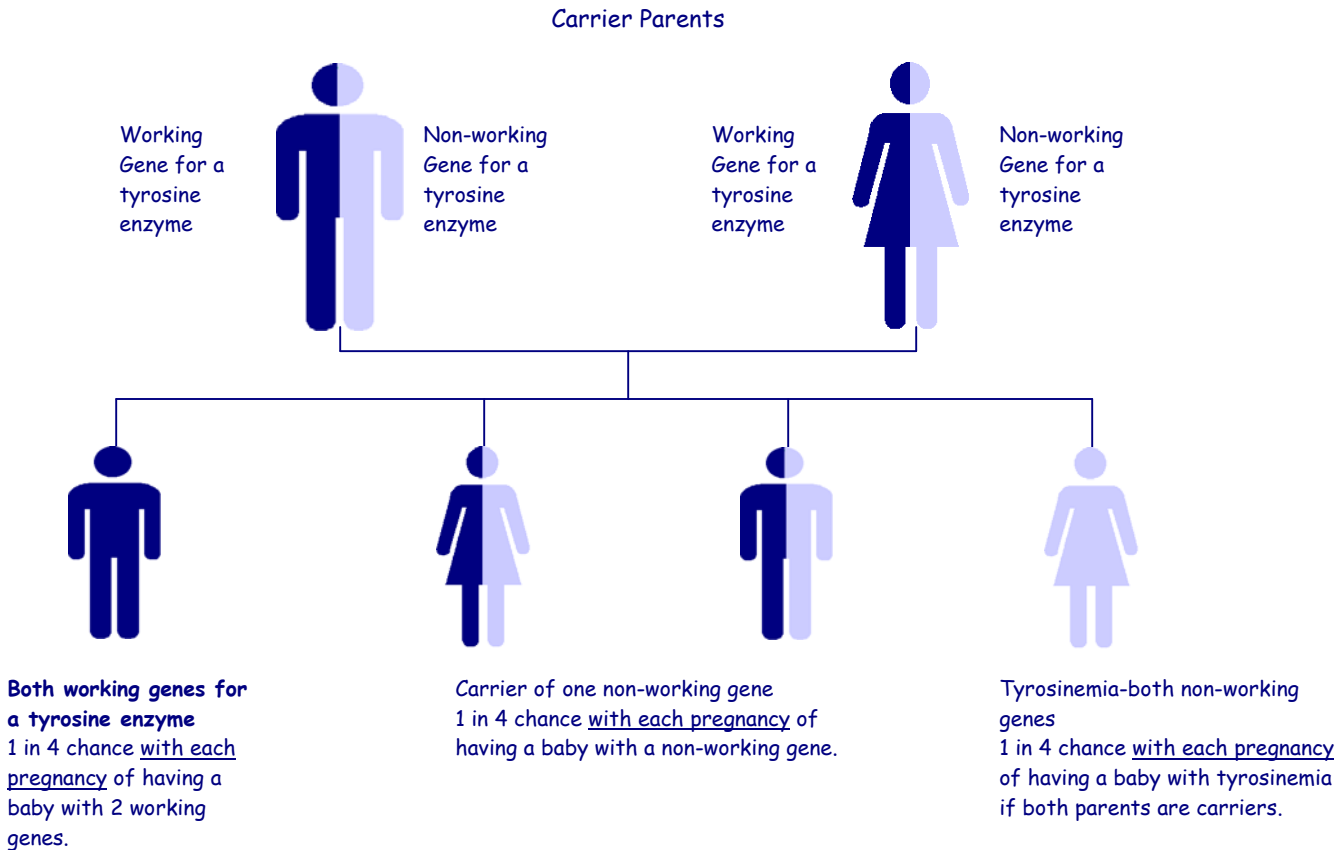
AMINO ACID DISORDER

Tyrosinemia

Protein is an important part of our diet. Amino acids are the building blocks of proteins. Babies who have the disorder called tyrosinemia cannot break down the amino acid tyrosine. Babies with tyrosinemia have high levels of tyrosine in their body. There are three known forms of tyrosinemia. Symptoms may occur at birth or later in infancy. Some of the more common symptoms include liver and kidney problems, vomiting, diarrhea, and poor bone development (Rickets).

How does a baby get tyrosinemia?

Tyrosinemia is an inherited condition. Parents pass the tyrosinemia genes to their children the same way they pass on other inherited traits. Tyrosinemia can occur if both the mother and the father have a non-working gene for an enzyme. As shown in the figure below, a baby can receive a non-working gene from both parents and have tyrosinemia. If both parents have a non-working gene for tyrosinemia, they have a one in four chance with each pregnancy of having a baby with tyrosinemia.



How is a baby tested for tyrosinemia?

Prior to discharge from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State Laboratory to screen for tyrosinemia. Abnormal screening results are reported to the Department of Public Health (DPH), Genetic Newborn Screening Tracking Unit nurse consultant. If the results are positive, the DPH nurse consultant will notify the baby's primary care physician of the need for a prompt referral to one of the Genetic Regional Treatment Centers at Yale or the UCONN Health Center for confirmation testing and follow-up treatment if necessary.

What is the treatment for tyrosinemia

Early diagnosis of tyrosinemia and ongoing treatment at a Metabolic Treatment Center is essential for the health of your baby. Treatment includes a special low protein diet that is restricted in tyrosine and phenylalanine, a special metabolic formula, and medications. Even with treatment, liver transplant in childhood may be a consideration for individuals with the most common form.

Regional Metabolic Treatment Centers

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Genetics Newborn Screening Program
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Newborn Screening Laboratory Services
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