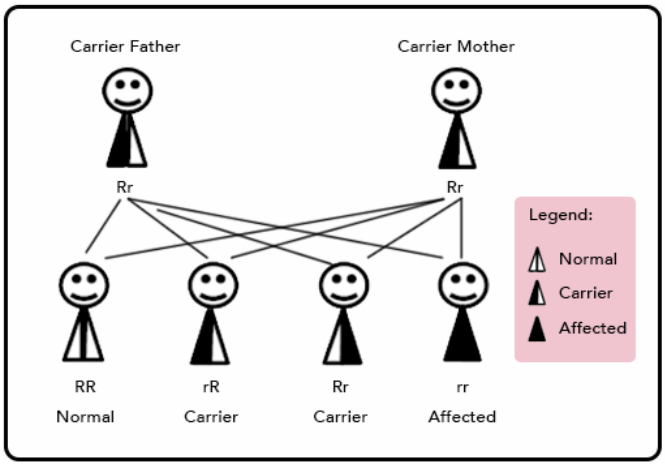
AMINO ACID DISORDERS:
HOMOCYSTINURIA

What is Homocystinuria?

Homocystinuria is a condition where there is accumulation of methionine (an amino acid which is a building block of protein). Children born with this condition appear normal at birth but during childhood untreated children may present with visual problems, mental retardation, scoliosis and a higher risk of blood clotting abnormalities. It is one of the conditions that can be detected through newborn screening. Early detection can prevent the complications of homocystinuria.

What causes Homocystinuria?

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 cystathionine B synthase enzyme), methionine (an amino acid) cannot be broken down. The accumulation of this amino acid causes the signs and symptoms of homocystinuria.



Homocystinuria is an inherited condition. The CBS 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 gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with HCY have one working and one non-working gene coding for CBS. 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 homocystinuria. 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 Homocystinuria?

Children with homocystinuria appear normal at birth but during childhood untreated children may present with visual problems, mental retardation, scoliosis and a higher risk of blood clotting abnormalities.

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WHAT IS THE TREATMENT OF HOMOCYSTINURIA?

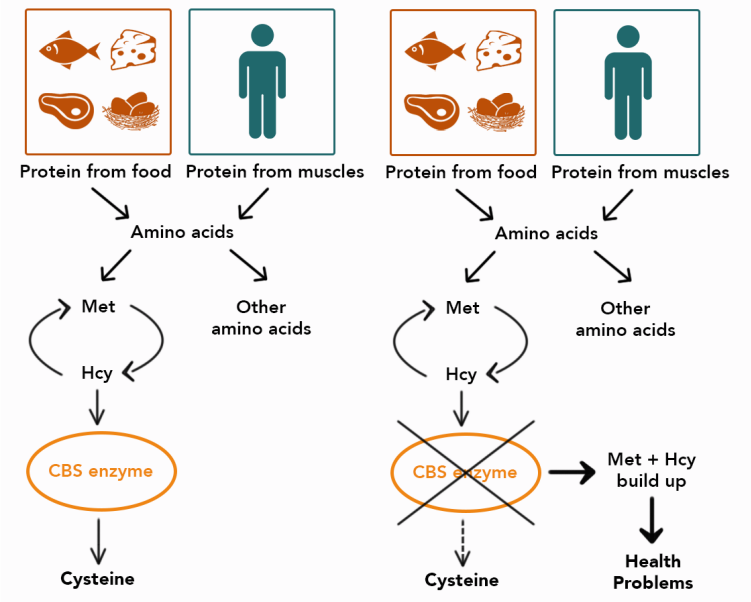
The main treatment of homocystinuria is through control of the diet and intake of some vitamins. If the child is well or does not have any illness, he/she should continue taking the special milk formula and be on a low protein diet as ordered by your pediatrician or metabolic specialist. It is important that the diet be followed to avoid any complications. In addition, vitamin B6, folic acid and a medication called betaine may help in breaking down excess methionine.

Please consult with your attending physician prior to starting treatment.

WHAT ARE THE PRECAUTIONS THAT I SHOULD TAKE?

Children with homocystinuria may have a **metabolic crisis** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not drank well or during stressful events such as surgery. Your child may present with blood clotting abnormalities such as stroke. If your child is unwell or will undergo surgery, please bring him/her to the hospital for management and alert your pediatrician or metabolic physician.

Normal breakdown of protein vs Homocystinuria patient's breakdown of protein



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