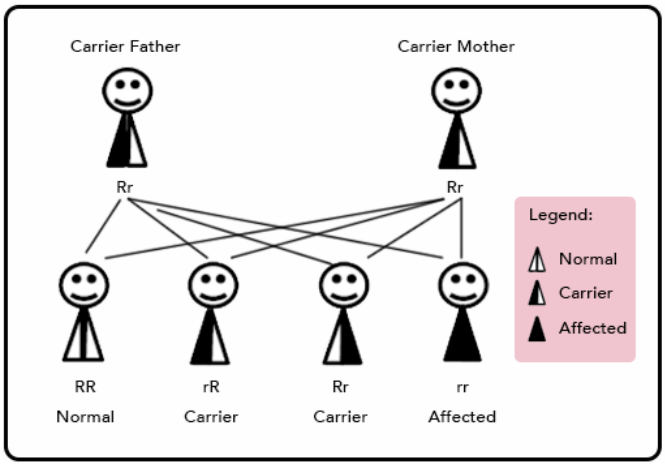
ORGANIC ACIDURIAS:
BETA-KETOTHIOLASE DEFICIENCY

What is Beta-ketothiolase Deficiency?

Beta-ketothiolase Deficiency is due to a deficiency in the mitochondrial acetoacetyl-CoA enzyme or chemical scissors that breaks down amino acids (or the building blocks of protein) in the body. Children with this condition cannot properly breakdown protein in their body or from the food they eat. Children born with this condition appear normal at birth but may present with severe vomiting and diarrhea, low blood sugar, seizures and coma. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, increased sleepiness, acidosis, coma and death.

What causes Beta-ketothiolase Deficiency?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissor, children with this condition cannot effectively utilize protein and its building block called amino acids (specifically isoleucine) from their body or from the food they eat.



Beta-ketothiolase is an inherited condition. The gene for the mitochondrial acetoacetyl-CoA enzyme 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 Beta-Ketothiolase deficiency have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat. 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 a Beta-Ketothiolase Deficiecy. 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 Beta-ketothiolase Deficiency?

Children born with this condition appear normal at birth but if treatment is not given early, they can present with low blood sugar which can lead to seizures, coma and death. They may also have acidosis, vomiting and diarrhea.

BETA-KETOTHIOLASE DEFICIENCY

WHAT IS THE TREATMENT OF BETA-KETOTHIOLASE DEFICIENCY?

The main treatment of Beta-Ketothiolase Deficiency is through a low protein diet. If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours.

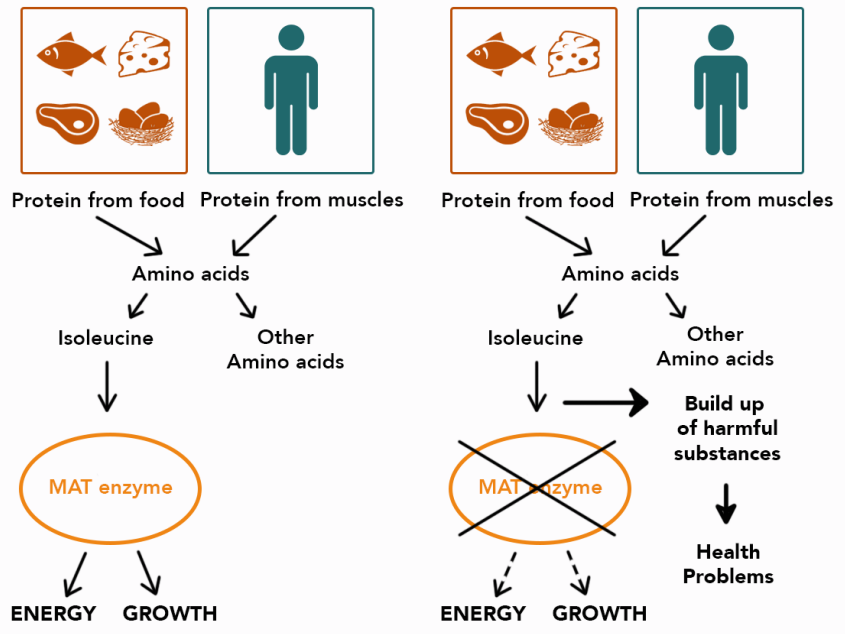
Carnitine, a supplemental medication essential for muscle energy production has been found to be of benefit for some patients.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS UNWELL?

Children with Beta-Ketothiolase Deficiency 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 eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting and acidosis. If not treated properly and immediately, it might lead to serious brain 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 Beta-ketothiolase Deficiency patient's breakdown of protein



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