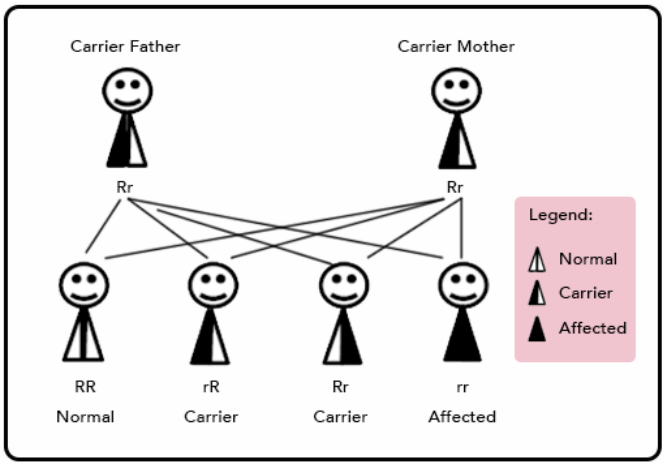
FATTY ACID OXIDATION DISORDERS:
CARNITINE PALMITOYL-TRANSFERASE DEFICIENCY TYPE 1 [CPT1 Deficiency]

What is Carnitine Palmitoyl-transferase Deficiency Type I?

Carnitine Palmitoyl-transferase Type I (CPT1) Deficiency is due to a deficiency in an enzyme or chemical scissors called Carnitine Palmitoyl-transferase Type I that breaks down fats in the body. Children with this condition cannot properly breakdown fat in their body or from the food they eat. Children born with this condition appear normal at birth but if treatment is not given early, they may present with low blood sugar and lead to seizures, coma and death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, liver disease, increased sleepiness and muscle weakness.

What causes CPT1 Deficiency?

To effciently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively utilize fat from their body or from the food they eat.



CPT1 Deficiency is an inherited condition. The gene for the different enzymes 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 CPT1 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 CPT1 Deficiency. 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 CPT1 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. Muscle weakness and liver disease may be among the complications.

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WHAT IS THE TREATMENT OF CPT1 DEFICIENCY?

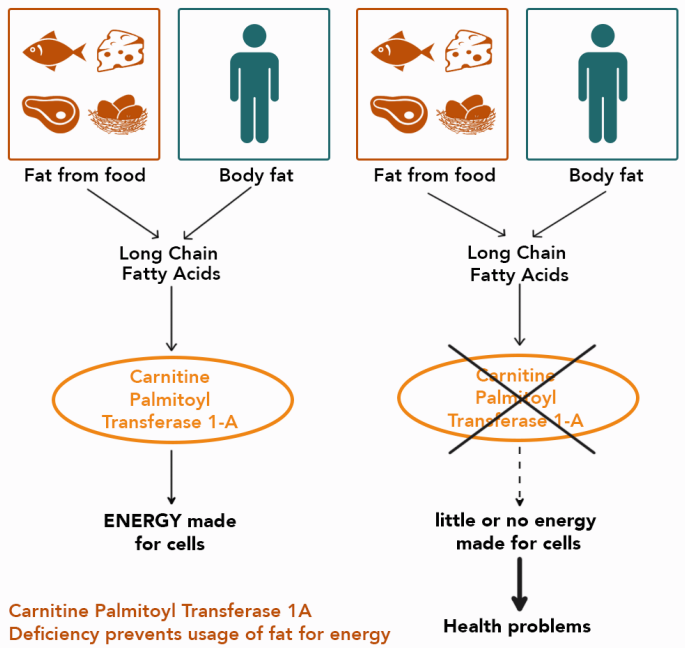
The main treatment of CPT1 Deficiency is through a low fat diet. Children may be given a special milk formula called MCT (medium chain triglycerides). If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS UNWELL?

Children with CPT1 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. 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 CPT1 Deficiency patient's breakdown of protein



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