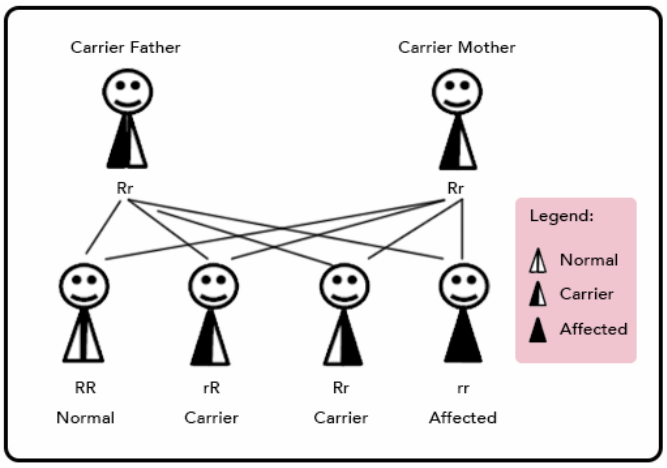
ORGANIC ACIDURIAS:
3– METHYLCROTONYL CoA CARBOXYLASE DEFICIENCY [3-MCC]

What is 3– Methylcrotonyl CoA Carboxylase Deficiency [3-MCC]?

3-MCC is a condition that is due to a deficiency in an enzyme or chemical scissors called 3-methylcrotonyl CoA carboxylase which is needed to breakdown an amino acid called leucine. Amino acids are the building blocks of protein. Children with this condition will look normal at birth. Untreated children may remain without symptoms while others may have seizures, drowsiness, low muscle tone, poor appetite and failure to thrive. This disorder can be detected through newborn screening. However, neonates who test positive for this condition in expanded newborn screening may not actually have the condition but instead reflect the increased levels of the metabolites of their mothers. Thus further investigation of the mother should be done as well.

What causes 3-MCC 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 scissors, children with this condition cannot effectively breakdown the amino acid leucine. The accumulation of leucine causes the signs and symptoms of this condition.



3-MCC is an inherited condition. The gene for the 3-methylcrotonyl CoA carboxylase 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 3-MCC 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 3-MCC. 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 3-MCC Deficiency?

Children with this condition will look normal at birth. Untreated children may remain without symptoms while others may have seizures, drowsiness, low muscle tone, failure to thrive and poor appetite.

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WHAT IS THE TREATMENT OF 3-MCC?

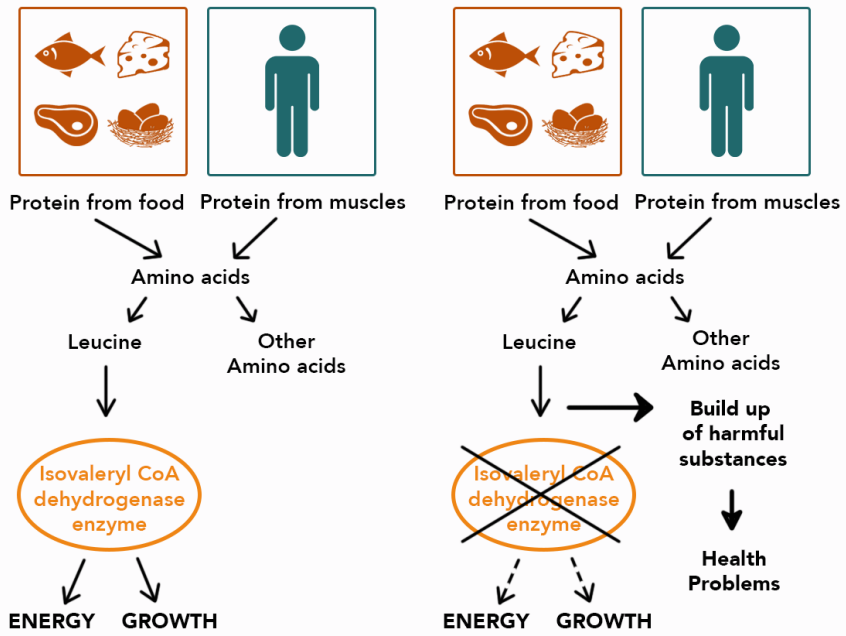
The main treatment of a 3-MCC is through a low protein diet. If the child is well or does not have any illness, he/she should eat a low protein diet. Glycine and/or Carnitine, a medication that helps boost energy, is also given.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS UNWELL?

Children with 3-MCC 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 3-MCC patient's breakdown of protein



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