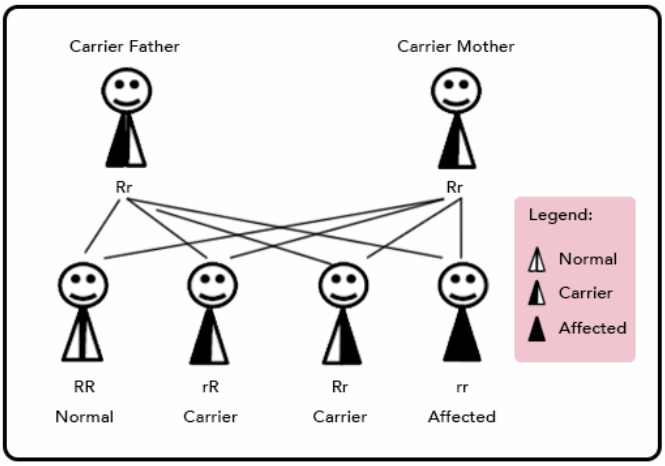
AMINO ACID DISORDERS:
METHIONINE ADENOSINE TRANSFERASE (MAT) DEFICIENCY

What is MAT deficiency?

Methionine Adenosine Transferase (MAT) Deficiency is due to a deficiency in an enzyme or chemical scissors that breaks down the amino acid (or building block of protein) called methionine. Children with this condition cannot properly breakdown methionine 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 tremors, movement disorders, seizures and a "boiled cabbage" odor in the urine or sweat. This condition can be detected through newborn screening. Early detection can prevent the complications of this condition such as mental retardation and seizures.

What causes MAT 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 methionine from their body or from the food they eat.



MAT 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 MAT 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 MAT 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 MAT deficiency?

Children born with this condition appear normal at birth but if treatment is not given early they can present with tremors, movement disorders and seizures. Mental retardation and death are among the complications.

METHIONINE ADENOSINE TRANSFERASE [MAT] DEFICIENCY

WHAT IS THE TREATMENT OF MAT DEFICIENCY?

The main treatment of a MAT is through a low protein diet, specifically low methionine diet. S-adenosylmethionine administration has been found to be effective in neurologic development. Please consult with your attending physician prior to starting treatment.

Normal breakdown of protein vs
MAT Deficiency patient's breakdown of protein