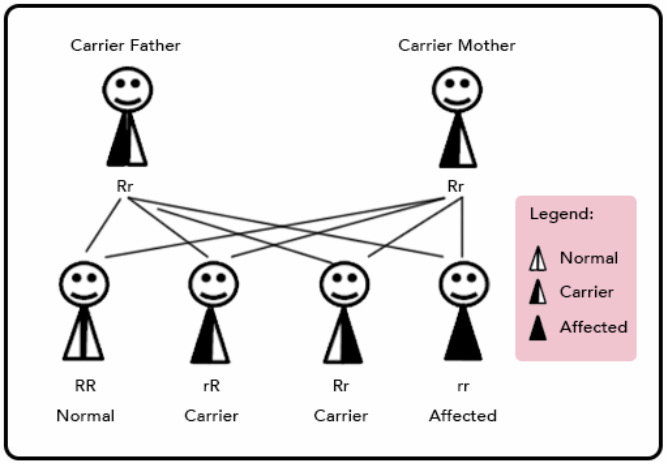
ORGANIC ACIDURIAS:
MULTIPLE CARBOXYLASE DEFICIENCY [MCD]

What is MCD?

Multiple Carboxylase Deficiency is due to a deficiency of an enzyme (or chemical scissors) called holocarboxylase synthetase. Holocarboxylase synthetase adds biotin (a vitamin) to carboxylases which are other enzymes needed to change the food we eat into energy. Untreated children born with this condition may present with drowsiness, poor appetite, poor weight gain, vomiting, alopecia and a skin rash. They may have mental retardation and brain damage as a complication. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition.

What causes MCD?

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 certain carbohydrates and fats which in turn are needed to breakdown protein.



The gene for the holocarboxylase synthetase 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 multiple carboxylase deficiency have one working and one non-working gene coding for this enzyme. 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 multiple carboxylase 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 MCD?

Untreated children born with this condition may present with drowsiness, poor appetite, poor weight gain, vomiting and a skin rash. They may have mental retardation and brain damage as a complication.

MULTIPLE CARBOXYLASE DEFICIENCY

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