

Multiple Carboxylase Deficiency (Biotinidase and Holocarboxylase Synthase) (MCD)

What is MCD?

Multiple carboxylase deficiency (MCD) is an organic acid disorder. People with MCD can't change protein and carbohydrates from the food they eat into energy for the body.

What Causes MCD?

Enzymes help start chemical reactions in the body. The enzyme holocarboxylase synthetase (HCS) adds the vitamin, biotin, to other enzymes called "carboxylases." These change the food we eat into energy. Harmful matter builds up in the blood and urine when the HCS enzyme doesn't work. This can cause serious health problems.

What Symptoms or Problems Occur with MCD?

[Symptoms are something out of the ordinary that a parent notices.]

MCD causes periods of illness called Metabolic Crises. Some of the first symptoms are:

- poor appetite
- vomiting
- too much sleepiness or lack of energy
- irritable behavior
- low muscle tone (floppy muscles and joints)

If a Metabolic Crisis is not treated, a child with MCD can develop:

- breathing problems
- seizures
- brain swelling
- coma, sometimes leading to death

Later problems can include:

- intellectual disability
- skin rash and hair loss
- unsteady movements
- rigid movement
- vision and hearing loss

What is the Treatment for MCD?

Biotin is usually the only treatment for MCD. Your doctor will prescribe the right amount. This will prevent symptoms in your child. It may rid your child of health problems when symptoms already exist. Early treatment may prevent intellectual disability and other serious medical problems. Treatment should start as soon as you know your child has MCD. Your child will need to take biotin for life.

Things to Remember

Babies who get treatment before they have a Metabolic Crisis usually have normal growth and development.

A few children develop learning problems even when treated. Children may also develop intellectual disability. If treated late, biotin may not reverse all existing symptoms.