PARENT FACT SHEET

DISORDER

Multiple carboxylase deficiency (MCD)

CAUSE

MCD occurs when an enzyme called "holocarboxylase synthetase" (HCS) is either missing or not working properly. This enzyme's job is to add a vitamin called "biotin" to other enzymes called "carboxylases" so that they can change the food we eat into energy for the body. When the HCS enzyme is not working certain harmful substances build up in the blood and urine. This can cause serious health problems.

IF NOT TREATED

Each child with MCD is likely to have slightly different effects. Many babies with this condition start to have symptoms within hours of birth or during the first few days or weeks of life. Other babies have their first symptoms sometime in infancy, usually before two years of age. Without treatment, brain damage can occur. This can result in mental retardation or, sometimes, even death.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist. Treatment is needed throughout life.

- The main treatment for MCD is a type of vitamin B called "biotin." In babies found to have MCD through newborn screening, biotin treatment can prevent symptoms from occurring. It can also reverse some of the health problems in children who have already shown symptoms.
- You will need a prescription from the metabolic specialist for biotin. They will determine the proper amount your child needs to take.
- Contact your child's doctor immediately at the start of any illness.

IF TREATED

Babies who receive prompt and ongoing treatment with biotin before they have a metabolic crisis are expected to have normal growth and development. Even with treatment, a few children have developed life-long learning problems or mental retardation. In children who have already show delays in learning, or a loss of hearing or eyesight, treatment can prevent additional effects. But it may not be able to correct the effects that are already present.

For more information go to the following website: http://www.newbornscreening.info