Parent Fact Sheet

3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)

(3-meth-ill-crow-ton-ill CO-a car-BOX-ill-aze De-FI-shen-see)

What is 3MCC?

3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC) is a metabolic disorder. This means the baby has a chemical imbalance. 3MCC is a condition in which the body is not able to process proteins properly. People with this condition do not have enough of a certain enzyme. The enzyme is needed to break down an amino acid called leucine. Leucine is a part of many proteins. The lack of the enzyme leads to a build up in the body of a chemical called 3–methylcrotonyl-CoA. 3MCC is inherited. This means it is present at birth.

What type of problems occurs with 3MCC?

A baby who has this condition may refuse to eat. The baby may develop vomiting and diarrhea. The baby may develop a lack of energy (lethargy) and poor muscle tone. If untreated, this condition can lead to delayed development, seizures, and coma. These problems might show up as early as 3 months of age. Sometimes these problems do not show up until the baby becomes an adult. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have 3MCC?

This condition occurs in less than 1 in every 75,000 births. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

What is the treatment of 3MCC?

The treatment for this condition is a low protein diet. A medical formula or food supplement, such as carnitine, might be needed. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care. Routine illness, like the common cold, can make this condition worse. So be sure to contact your baby's doctor whenever your baby gets sick.

Where in Virginia can I take my baby for care?

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is http://www.vahealth.org/gns

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www.vdh.virginia.gov

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