

Parent Fact Sheet

3-Hydroxy - 3- Methylglutaryl - CoA Lyase Deficiency (HMG) (3- High-drox-ee 3- Meth-ill-glue-tar-ill Co-A LIE-ace De-fish-n-see)

What is HMG?

3-hydroxy-3methylglutaryl-CoA lyase deficiency (HMG) is a metabolic disorder. This means the body has a chemical imbalance. HMG is caused by a lack of the enzyme 3-hydroxy-3-methylglutaryl-CoA lyase. Patients with this disorder are unable to break down leucine. HMG is an inherited condition. This means it is present at birth.

What type of problems occurs with HMG?

The baby who has this condition may have low blood sugar, vomiting, enlarged liver, and fever. People with HMG should not go long periods without eating. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have HMG?

This condition occurs in less than 1 in every 100,000 births. This condition is found in the Saudi Arabian population. 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 HMG?

The treatment for this condition is a restriction of leucine and fat in the diet. A regular feeding schedule is important. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care.

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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