TEXAS Department of State Health Services

Newborn Screening FACT Sheet

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)

What is HMG?

3-hydroxy-3-methylglutaric aciduria (HMG) is a condition in which the body is unable to break down certain proteins. People with the condition are also unable to produce ketone bodies, substances that help the body store energy. HMG is considered an organic acid condition because it can lead to a harmful amount of organic acids and toxins in the body.

Early detection and treatment can often prevent the serious outcomes of this condition.

What Causes HMG?

Enzymes help start chemical reactions in the body. HMG happens when an enzyme called "HMG CoA lyase" is either missing or not working well. This enzyme has two jobs. The first is to help break down leucine. All foods with protein contain leucine. The second job is to help the body make something called "ketone bodies" from stored fat.

What Symptoms or Problems Occur with HMG?

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

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

- poor appetite
- sleeping longer or more often
- irritable mood/behavior changes (such as crying for no reason)
- muscle weakness

Other problems then follow:

- fever
- diarrhea
- vomiting
- hypoglycemia (low blood sugar)
- increased levels of acid matter in the blood, called metabolic acidosis
- high levels of ammonia in the blood
- enlarged liver

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

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

When not treated, many babies die during their first Metabolic Crisis. Babies who live may have repeated Metabolic Crises that can cause brain damage.

This can result in life-long learning problems or intellectual disabilities and a seizure disorder.
Other problems can include:

- heart enlargement
- pancreatitis (inflamed pancreas gland)
- hearing loss
- vision problems

What is the Treatment for HMG?

The following treatments are often used for babies and children with HMG:

1. Do not go a long time without food – Babies and young children need to eat often to avoid a Metabolic Crisis. Most babies should not go without food for more than 4 to 6 hours. Some babies may need to eat more often. It is important to feed babies during the night. They may need to be woken to eat if they don't wake up on their own.

2. Low-leucine diet, including medical foods and formula – A food plan low in leucine that limits fat and protein is often part of the treatment. High-protein and high-fat foods that your child may need to limit or not eat at all include:

- milk and milk products
- meat and poultry
- fish
- eggs
- dried beans and peas
- nuts and peanut butter
- butter, margarine, oil, lard, and foods made with these fats

3. Medications – Taking L-carnitine may help some children. This is safe and natural and helps the body make energy. Use only the form prescribed by your doctor.

Things to Remember

Even minor illnesses such as a cold or the flu can cause a Metabolic Crisis. Call your doctor right away when your child has any of the symptoms or problems mentioned above.

Children who are sick often don't want to eat. If they can't eat, or if they show signs of a Metabolic Crisis, they may need to be treated in the hospital.