

3-Hydroxy-3-Methyl Glutaric Aciduria (HMG)

General Overview

Q. What is HMG?

A. HMG is a disorder that affects the way the body processes protein and fats. Children with HMG cannot properly use the amino acid leucine found in the protein they eat, and their bodies cannot properly break down fats. It is treatable, but can cause life-threatening illness.

Q. What happens to proteins and fats in a child with HMG?

A. Proteins are made up of amino acids. These amino acids are normally used to make new proteins or are broken down further for energy. In a child with HMG, the enzyme that normally helps break down leucine and fats does not work properly. This causes a buildup of harmful substances that are toxic to the brain and the rest of the nervous system.

Q. What are the effects of having HMG if it is not treated?

A. Untreated HMG can result in coma and death. Other effects may include brain damage, heart and pancreas problems, and possibly deafness.

Q. What is the treatment for HMG?

A. HMG is treated with a special diet that is low in leucine and fat but high in carbohydrates. Fasting must be avoided. Some doctors may also prescribe dietary supplements. To prevent mental retardation, treatment must begin shortly after birth. The child must continue on a medical formula that provides protein and essential nutrients but contains little or no leucine. People with HMG require treatment through a specialty clinic with experience in treating this disorder.

Q. Why would a child have HMG?

A. HMG is an inherited disorder. It results when a baby receives a double-dose of a specific non-working gene involved in breaking down leucine (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. How common is HMG?

A. The prevalence of HMG is unknown, but very rare. However, parents who have a child with HMG each carry one copy of the non-working gene. That means, with each pregnancy, there is a one in four chance of the child having HMG.

For more information about HMG, please see the Disorders section of our website:
www.doh.wa.gov/nbs.

