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

(Sounds like 3-HIGH-DROX-Y-3-METH-ILL-GLUE-TAR-ILL-CO-A LIE-ASE DE-FISH-IN-SEA)

What is newborn blood spot screening?

This is a blood test, from a heel poke, that is done a day or 2 after babies are born to test for treatable conditions including 3-hydroxy-3-methylglutaryl-CoA lyase (HMG) deficiency that you, your doctor, or midwife can't see by just looking at your baby. This screening test is different from those done during pregnancy.

What does it mean if my baby needs more tests for HMG deficiency?

A baby needs to have more testing for HMG deficiency when their screening test result is abnormal or positive. This doesn't mean your baby has HMG deficiency. It means your baby has a higher chance of having the condition and will need more tests to find out if they have HMG deficiency or not.

It's normal to feel worried if your baby needs more testing. Your specialists and other healthcare providers are there to support you. They'll explain what will happen and answer your questions.

What is HMG deficiency?

HMG deficiency is a metabolic condition that affects how your body uses protein and fat from the food you eat to make and store energy. This includes breastmilk and regular formula. With a deficiency, or not enough HMG, your body can't break down a building block of protein called leucine to make energy. Leucine and other substances then build up in the body and can cause health problems.

What causes HMG deficiency?

HMG deficiency is genetic. This means babies with HMG deficiency are born with a gene from both parents, which has a change in it. This change stops the gene from working properly. HMG deficiency isn't caused by anything that happened during pregnancy.

There may be no signs of HMG deficiency at birth. It's important to remember that this condition can't be seen by looking at your baby.

What can I expect with more testing?

Everything that will happen with more testing will be explained to you. Your baby may need to have their blood and urine tested to check for HMG deficiency.

This testing may be done at a clinic in Edmonton or Calgary that specializes in HMG deficiency.

What if more testing shows my baby has HMG deficiency?

If testing shows that your baby has HMG deficiency a specialized healthcare team will work with you to make a care plan for your baby. You and your baby will get the best care and support possible.

How is HMG deficiency treated?

Babies with HMG deficiency usually go on a low-protein diet and drink a special formula. Babies need to be fed often. They usually also need special medicine. The specialized healthcare team will give you detailed instructions about how to manage your baby's health.

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

Be careful when you search for information on the Internet. Get information from trusted sources and learn where to find reliable information.

If you're ever concerned about your baby's health or need support for you or your family, speak with your family doctor, the specialized healthcare team, or call Health Link at 811.

To learn more about HMG deficiency visit babysfirsttest.org and enter HMG into Find a Condition, or scan this QR code.

Newborn blood spot screening is provided through the Alberta Newborn Screening Program (ANSP) within Alberta Health Services (AHS). The program helps your baby have the best start in life. Early screening for certain conditions makes sure your baby gets treatment when it can help the most.

To learn more about newborn screening visit ahs.ca/newbornscreening or scan this QR code.

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