NEWBORN SCREENING BC

Information sheet

Hemoglobin Disorders:

Homozygous Hemoglobin D and Hemoglobin D Beta Thalassemia

Your baby had a newborn screen. This test was done on a blood sample collected by heel prick shortly after birth. This is done to identify a number of rare but treatable conditions which usually do not have any symptoms early on. Knowing if your child has one of these conditions early in life is important, because early diagnosis allows for early treatment and better health outcomes.

Your baby had a positive newborn screen result for possible homozygous hemoglobin D or hemoglobin D beta thalassemia. This result means the chance of a hemoglobin variant is increased and more testing is needed to find out for sure.

What causes homozygous hemoglobin D and hemoglobin D beta thalassemia?

Hemoglobin is an important protein found in our red blood cells. It picks up oxygen from our lungs and carries it to every part of our body. Most people make a single type of hemoglobin called hemoglobin A. Hemoglobin is made in our cells by following a set of instructions (like a recipe) called a gene. This gene is called the beta globin gene. Normally we have two copies, one inherited from each parent. Changes in the beta globin gene cause hemoglobin variants like hemoglobin D and beta thalassemia.

What is homozygous hemoglobin D?

People with homozygous hemoglobin D inherit two copies of the beta globin gene with the hemoglobin D variant, one from each parent. This can cause a mild anemia and a slightly enlarged spleen, but otherwise does not cause health problems in most people.

What is hemoglobin D beta thalassemia?

People with hemoglobin D beta thalassemia inherit one beta globin gene with the hemoglobin D variant and one with a beta thalassemia variant. This combination can cause the body to make fewer red blood cells than normal. These red blood cells are more fragile and break apart more easily. This can cause a more significant anemia that can lead to health problems. It is important for babies with hemoglobin D beta thalassemia to receive regular medical care.

What happens next?

More testing is needed to confirm whether your baby has a homozygous hemoglobin D or hemoglobin D beta thalassemia. Your primary care provider will arrange another blood test for genetic testing of the beta globin gene. This test will differentiate between homozygous hemoglobin D and hemoglobin D beta thalassemia.

If this follow up testing confirms a diagnosis of hemoglobin D beta thalassemia, your primary care provider will refer you to the Hematology Clinic at BC Children's Hospital. This clinic specializes in caring for and treating children with hemoglobin disorders. They will review the condition and treatment options, and answer your questions.

If this follow up testing confirms your baby has homozygous hemoglobin D, then a referral to hematology is not needed.

It is important to have the follow up testing as soon as possible because early diagnosis and treatment improves the health of children with hemoglobin disorders.

How are these disorders treated?

Generally, people with homozygous hemoglobin D do not need any medical treatment or special precautions. However, people with hemoglobin D beta thalassemia do benefit from regular medical care, and different treatments are available to relieve symptoms should they experience them.

Provincial Health Services Authority

Newborn Screening BC is a collaboration of Provincial Lab Medicine Services, BC Children's Hospital and BC Women's Hospital and Health Centre, and Perinatal Services BC, all part of the Provincial Health Services Authority.

What does having a baby with homozygous hemoglobin D or hemoglobin D beta thalassemia mean for me, my partner and for future pregnancies?

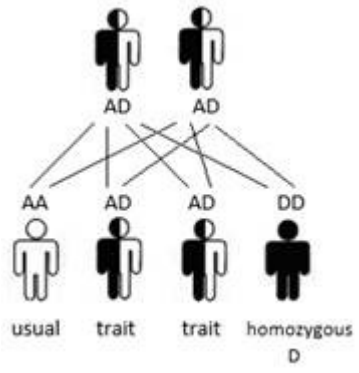
Most people do not know that they carry a hemoglobin variant because it does not typically affect their health. But now that your baby is known to have homozygous hemoglobin D or hemoglobin D beta thalassemia, this means that both you and your partner carry one of these hemoglobin variants (hemoglobin D trait or beta thalassemia trait). A carrier of a hemoglobin variant (also known as trait) is unaffected. Carriers have one beta globin gene which makes hemoglobin A and one which makes a variant hemoglobin like hemoglobin D or beta thalassemia. Many healthy people carry hemoglobin D or beta thalassemia trait.

To confirm your carrier status, you and your partner have the option of being tested. Testing involves a blood test called "hemoglobin investigation". Ask your primary care provider to arrange this test for you if you want to know your carrier status. This would be especially important to do if you plan on having more children.

If both of you have hemoglobin D trait

In every pregnancy, there will be a:

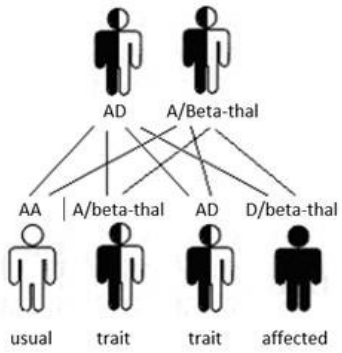
- 1 in 4 (25%) chance to have a baby with only hemoglobin A. This is normal.
- 1 in 2 (50%) chance to have a baby with hemoglobin D trait. This is harmless.
- 1 in 4 (25%) chance to have a baby with only hemoglobin D, so the baby does not make any hemoglobin A. This is called homozygous hemoglobin D.



If one of you has hemoglobin D trait and the other has beta thalassemia trait

In every pregnancy there will be a:

- 1 in 4 (25%) chance to have a baby with only hemoglobin A. This is normal.
- 1 in 4 (25%) chance to have a baby with hemoglobin D trait. This is harmless.
- 1 in 4 (25%) chance to have a baby with beta thalassemia trait. This is harmless.
- 1 in 4 (25%) chance to have a baby with hemoglobin D beta thalassemia, so the baby does not make any hemoglobin A. This is a hemoglobin disorder that requires medical care.



Should my relatives be tested?

Any blood relative (for example, brother, sister, aunt, uncle or cousin) of a person with hemoglobin D trait or beta thalassemia trait may also carry this trait. Your relatives may have questions about their chance of having a baby with a hemoglobin disease. We suggest they talk to their primary care provider who may offer them testing (hemoglobin investigation).

How common are hemoglobin variants?

There are many different types of hemoglobin variants. Although anyone can be a carrier of a hemoglobin variant, it is more common among people of African, Caribbean, Middle Easter, Asian, Mediterranean, Indian and Central and South American descent.

How do you feel?

For most parents, this is an unexpected result. You may feel scared and upset which are normal feelings. If you are feeling

overwhelmed or have questions, you can talk with your health care provider to discuss supports available.

How can I get more information?

Talk to your primary care provider.

If you and your partner have a chance of having a baby with hemoglobin D beta thalassemia, then genetic counselling is an option. Ask your primary care provider to refer you to the Department of Medical Genetics at BC Children's and Women's Hospital or Vancouver Island Medical Genetics (at Victoria General Hospital) for genetic counselling.

Information about the BC Newborn Screening Program can be found at
www.newbornscreeningbc.ca

This fact sheet provides basic information only. It does not take the place of medical advice, diagnosis or treatment. Always talk to a healthcare provider about specific health concerns.