NEWBORN SCREENING BC

Information sheet

Hemoglobin Disorders: Hemoglobin H Disorder

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 hemoglobin H. This result means the chance of a hemoglobin variant is increased and more testing is needed to find out for sure.

What causes hemoglobin H?

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 sets of instructions (like a recipe) called a gene. This gene is called the alpha globin gene. Normally we have four alpha globin genes, two inherited from each parent. Changes in the alpha globin gene can cause hemoglobin variants like hemoglobin H.

People with hemoglobin H only have one working alpha globin gene. For most this is because three copies of the gene are deleted (missing). This is called ***deletional hemoglobin H***. Less often, people with hemoglobin H can have two alpha globin genes but one of them does not work properly because of a spelling mistake within the gene. This is called ***non-deletional hemoglobin H***.

What is hemoglobin H?

Hemoglobin H (also called hemoglobin H disease) is a type of *alpha thalassemia*. Babies born with hemoglobin H are generally healthy. But it is important to know about hemoglobin H because it can cause moderate to severe anemia (having fewer red blood cells). At times, especially during an illness (like a cold or flu), the anemia can get worse causing fatigue and jaundice. How hemoglobin H presents can vary among different people with some having few to no symptoms and others requiring more regular medical care.

What happens next?

More testing is needed to confirm whether your baby has hemoglobin H. Your primary care provider will arrange another blood test for genetic testing. This test will look for common genetic variants in the alpha globin gene that cause hemoglobin H. Genetic testing can differentiate between deletional and non-deletional hemoglobin H.

If this follow up testing confirms a diagnosis of hemoglobin H, 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, follow up plan, and answer your questions.

How is hemoglobin H treated?

During times of illness, surgery or other physical stress, people with deletional hemoglobin H may need a blood transfusion. They may also need supportive treatments later in life. People with non-deletional hemoglobin H tend to have a more severe anemia, and they may require more frequent treatment. Both people with deletional and non-deletional hemoglobin H benefit from receiving regular medical follow up.

Can a family have more than one child with hemoglobin H?

Yes, because hemoglobin H is an inherited condition. Both parents of a child affected with hemoglobin H are almost always carriers of an alpha globin gene variant. Carriers are unaffected. When both members of a couple are carriers, they have a 1 in 4 chance in every pregnancy of having a baby with hemoglobin H. If you are interested in carrier testing, then talk to your primary care provider.

If your baby is confirmed to have hemoglobin H and you are worried about the possibility of having another baby with hemoglobin H, then ask your primary care provider for a

referral to Medical Genetics to meet with a genetic counsellor. The genetic counsellor will provide you with more information regarding how hemoglobin H is inherited and discuss your testing options for a future pregnancy.

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. You may also call the BC Children's Hospital Hemoglobinopathy Nurse Clinician at 604-875-2345 x7103.

If you and your partner have a chance of having a baby with hemoglobin H and you are worried about this possibility, 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.