NEWBORN SCREENING BC

Information sheet

Hemoglobinopathies:

Sickle Cell Disease (HbSS, HbSC or HbS/ß-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 sickle cell disease. This result does **not** mean that your baby has sickle cell disease but means the chance is increased and more testing is needed.

What is Sickle Cell Disease?

Sickle cell disease is an inherited condition that affects the shape of red blood cells. Normally red blood cells are shaped like a doughnut and can easily squeeze through small blood vessels. In sickle cell disease under certain conditions, the red blood cell's shape changes to a 'sickle' (crescent moon) shape. These sickle-shaped red blood cells do not squeeze through small blood vessels very well. As a result, they become stuck in the vessels and cause pain and damage to the surrounding tissue. Sickled cells also break down (hemolyse) more quickly than usual, causing anemia. Other issues may include jaundice, an increased chance of certain infections, and delayed growth. However, there is a lot of variability in how sickle cell disease can present and not every person experiences every symptom.

What causes the disease?

Sickling of red blood cells is caused by an abnormal form of hemoglobin called Hb S (hemoglobin S).

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. 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 S. Other hemoglobin variants caused by different beta globin gene variants include Hb C (hemoglobin C), Hb E (hemoglobin E), and Hb Beta thal (hemoglobin beta-thalassemia).

People with sickle cell disease inherit a hemoglobin S variant from each parent, or a hemoglobin S variant from one parent and a second hemoglobin variant listed above from the other parent.

What happens next?

More testing is needed to confirm whether your baby has sickle cell disease or not. Your primary care provider will arrange another blood test which will look at your baby's hemoglobin pattern using specialized testing methods. If needed, genetic testing of the beta globin gene will also be done.

If this follow up testing confirms a diagnosis of sickle cell disease, then 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 sickle cell disease and other blood disorders. They will review the condition, treatment options, and answer your questions.

It is important to have the follow up testing as soon as possible because early diagnosis and treatment greatly improves the health of a child with sickle cell disease.

How is sickle cell disease treated?

Infants who have sickle cell disease can benefit significantly from early medical care. Depending on your baby's specific diagnosis, treatments may include medications to help prevent infections and reduce the effect of sickle cells on the body, and special treatments such as immunizations and blood transfusions. Extra medical care may be needed around times

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.

of sickness and before and after surgical procedures. For some patients, curative therapy may be an option.

What is its incidence?

Although sickle cell disease occurs in all ethnic groups, it is more common in certain populations, such as the African, Mediterranean, Middle Eastern, and Asian communities – in parts of the world in which malaria is endemic. Carriers for sickle cell disease and thalassemia are less susceptible to malarial infection, which explains their prevalence in these parts of the world.

Can a family have more than one child with sickle cell disease?

Yes because sickle cell disease is an inherited condition. Both parents of a child affected with sickle cell disease are almost always carriers for a hemoglobin variant. Carriers are unaffected because they have one normal copy of the beta globin gene and one with a hemoglobin variant. When both members of a couple are carriers, they have a 1 in 4 chance in every pregnancy of having a baby with sickle cell disease.

If your baby is confirmed to have sickle cell disease, 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 sickle cell disease is inherited and discuss your testing options for a future pregnancy.

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.

Where 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.

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

Information about sickle cell disease can be found at:

- **www.sicklecelldiseasecanada.com**
- **www.kidshealth.org/en/parents/sickle-cell-anemia.html**
- **www.aboutkidshealth.ca/SCD**

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