

NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Spinal Muscular Atrophy (SMA)



What is newborn blood spot screening?

This is a simple blood test that is done shortly after a baby is born to test for conditions that could affect your child's health. A baby is tested at birth because it is important to start treatment early for best health outcomes.

There may be no signs of these conditions at birth. It is important to remember that many treatable conditions can not be seen by looking at your baby.

What is SMA?

Spinal muscular atrophy (SMA) is a disorder that affects the nerves in the spinal cord that send signals to the muscles. When these nerves do not work, muscles can not do their job and become very weak. People with SMA may have difficulty walking, eating and breathing because of muscle weakness.

SMA is caused by a missing or faulty gene known as the SMN1 gene. Babies usually receive 2 copies of this gene — one from each parent. A person with only one functioning SMN1 gene (a carrier) is healthy, so parents may pass down a missing or faulty SMN1 gene copy without knowing. A baby born with SMA has received a missing or faulty SMN1 gene copy from both parents.

Why does my baby have to take more tests for SMA?

It is normal that babies may need to have more tests, but it does not mean that your baby has SMA. More testing is needed to find out if they do or do not have SMA. You will be referred to a specialist who will examine your baby and arrange more tests. They will support you and answer any questions you have.

What health problems can occur if a baby has SMA?

There are several different types of SMA. SMA type I, appears within the first 6 months of life. These babies may have breathing problems and difficulty feeding and swallowing. Children with SMA type II and type III usually present after 6 months of age with low muscle tone and weakness which may affect the ability to sit or walk.

How can SMA be treated?

Treatment can slow or even stop the progression of SMA. Depending on the type, your specialist will give you detailed information about the SMA treatment available for your baby. Treatment may be recommended right away. Infants with less severe forms of SMA will be checked regularly for symptoms. If symptoms develop, treatment options will be discussed with you. Regular follow-up in a pediatric neurology clinic is important to ensure best outcomes.

Additional resources

Be careful when you search for information about SMA on the internet. There is a lot of outdated and unreliable information online that may not be true today.

curesma.ca/resources-spinal-muscular-atrophy-canada

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