Spinal muscular atrophy (SMA) and your baby

You have just learned that your baby might have Spinal Muscular Atrophy (SMA). The information in this leaflet will help you understand more about this condition and answer some of your questions.

Newborn screening

A short time after birth some blood was collected from your baby's heel to screen for some rare disorders, including SMA. The goal of newborn screening is to identify conditions that affect babies and young children. Babies are tested at birth because it is important to start treatment early for the best health outcomes.

Your baby's newborn screening result

The result for your baby suggests they might have a condition called SMA. This is a neurological condition which affects the motor nerves. Babies and children with this condition are at risk of muscle weakness. Without treatment the muscles become weaker over time resulting in very serious health conditions, and potentially death. There may be no signs of SMA at birth. Starting treatment before a baby has symptoms offers the best response to treatment.

Newborn screening tests alone cannot confirm or rule out disorders. A further blood test is needed to determine whether or not your baby has SMA. Your LMC (midwife or specialist) will work with you to help arrange for your baby to have this blood test and be seen by a paediatrician or neurologist within a few days of the newborn screening result.

About SMA

SMA is a disorder that affects the nerves in the spinal cord (motor neurons) that send signals to the muscles to control muscle movement. Over time, as more motor nerves are lost, the muscles become weak. People with SMA may have difficulty crawling, walking, eating and breathing because of muscle weakness.

There are several different types of SMA. SMA type 1 develops in the first 6 months of life. These babies may have life threating breathing problems and difficulty feeding and swallowing. Children with type 2 and type 3 SMA usually present after 6 months of age with low muscle tone and weakness, which affects the ability to sit or walk.

What causes SMA?

SMA is a genetic condition. It is caused by a missing or faulty gene called the SMN1 gene. Babies usually receive two copies of this gene – one from each parent. A person with only one functioning SMN1 gene, a carrier, is healthy. So healthy parents may pass down the missing or faulty copy without knowing. A baby born with SMA has received the missing or faulty SMN1 gene copy from both parents.

SMA isn't caused by anything that happened during pregnancy.

Treatment for SMA

Treatment can slow or even stop the progression of SMA. Depending on the type of SMA, a neurologist will give you detailed information about the SMA treatment available for your baby. This is medication given either orally or via or spinal infusion. Treatment may be recommended immediately. Regular follow up in a neurology clinic is important to ensure the best outcomes for your baby.

Is SMA common?

SMA is a rare condition. We expect that around 6 babies will be born with SMA each year in New Zealand.

How will I know the results of my baby's further testing?

The results of further testing will be available in 7 to 10 days. A paediatrician or neurologist will meet with you to explain the results of this test.

What happens if further testing shows that my baby has SMA?

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

What should I do right now?

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

Further information

Talk with your paediatrician or midwife.

For more information about newborn screening, go to: www.tewhatuora.govt.nz/health-services-and-programmes/newborn-metabolic-screening-programme/

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