Information on a Positive Newborn Screen Result for

Spinal Muscular Atrophy (SMA)

MARITIME Newborn Screening
Dépistage néonatal DES MARITIMES

What is newborn screening?

These are routine tests done soon after birth. A few drops of blood are taken from a baby's heel and put onto a piece of absorbent paper (blotter). The blood is tested for rare, treatable conditions. These tests are done because a newborn can look healthy but have one of these conditions and need treatment. We want to find babies with these conditions so that we can start treatment to keep them as healthy as possible.

How is a positive newborn screen result reported?

A healthcare provider from Maritime Newborn Screening will contact the primary care provider and the family to discuss the result. They will connect the baby to the clinical team providing the follow-up. The healthcare provider and/or the clinical team will arrange follow-up appointments and testing to happen as soon as possible. Families may feel worried about the baby's newborn screen result. Many families in this situation feel this way. The healthcare teams are here to support families during the next steps.

What does it mean if a baby has a positive screen result for Spinal Muscular Atrophy (SMA)?

This result means that the baby likely has a type of SMA. Further testing is needed to confirm a diagnosis of SMA.

What is SMA?

SMA is a genetic neuromuscular condition, meaning the baby is born with it. SMA affects the motor neurons, which are the neurons that control our muscles and movement. In SMA the motor neurons are lost over time, which causes the muscles to weaken.

There are four main types of SMA. The types are categorized based on the age when symptoms begin and the most advanced motor milestone (e.g., sitting, walking) achieved. Type 1 is the most severe of the four types and is the most common. With Type 1 SMA symptoms usually appear in the first few weeks or months of life and babies typically cannot control head movements or sit unassisted. There can also be swallowing difficulties and breathing problems, and without treatment most infants with Type 1 SMA do not live past two years of age. SMA Types 2 and 3 are less common and have symptoms appearing after 6 months or in the first few years of life. Someone with SMA Type 2 or 3 may not be able to stand or walk. Type 4 SMA is rare, and symptoms start in adulthood. There are effective treatments for babies with severe types of SMA that can slow or stop the progression of SMA.

How many babies have SMA?

SMA is a rare disease that affects approximately 1 out of every 10,000 babies.

Why screen for SMA?

Babies with SMA usually look normal at birth and may not show any health problems related to SMA until a few weeks or months after birth. If they are not screened by newborn screening they might not be diagnosed right away. There are treatments for SMA, and these treatments are most effective when they are given before or soon after symptoms start. Newborn screening allows us to find babies with SMA so they can have treatment as soon as possible.

How is a baby diagnosed with SMA?

To find out if a baby has SMA, they are seen by a doctor in the IWK Kids' Rehab Clinic to assess their nerves and muscles. Genetic testing (bloodwork) also helps confirm a diagnosis of SMA. Families will be connected with a genetic counsellor from Maritime Medical Genetics Service. It can take a few days to confirm that a baby has SMA. This waiting period can be hard for families. The healthcare teams are here to support families during this time.

How is SMA treated?

SMA is treated with special medications that target the cause of SMA but are not a cure. These treatments are offered based on the type of SMA and the genetic diagnosis. The IWK Kid's Rehab clinic will talk with families about the treatment options. The two approved therapies are a targeted treatment called nusinersen (Spinraza®) and a gene therapy called onasemnogene abeparvovec (Zolgensma®). These treatments are most effective when given before SMA symptoms start.

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How does a baby get SMA?

SMA is an **inherited (genetic) condition**. This means the baby is born with SMA. When a baby has SMA, we know that the biological parents are likely carriers of the condition. Carriers do not have the condition but can have a baby with the condition if the baby inherits a non-working copy of the gene that causes SMA from each parent. This is called autosomal recessive inheritance (see Figure 1).

Could a family have another baby with SMA?

Yes, if the biological parents are carriers there is a 25% (1 in 4) chance in each future pregnancy to have a child with SMA. We offer genetic counselling through **Maritime Medical Genetics Service at the IWK** to talk about the following:

- Inheritance of SMA
- Chance of recurrence of SMA in future pregnancies
- Testing options for other family members and in future pregnancies

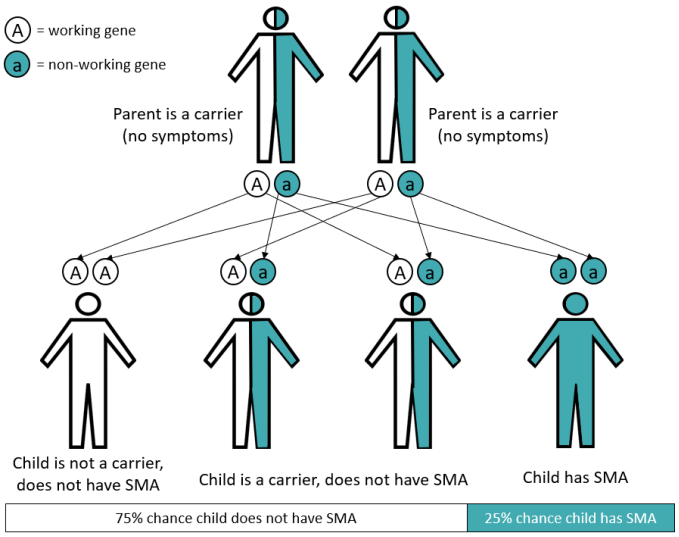

Figure 1. Autosomal recessive inheritance when both parents are carriers of spinal muscular atrophy (SMA).

Where can I get more information?

- For more information on newborn screening, please visit our website at www.maritimenewbornscreening.ca or call the newborn screening genetic counsellor at 902-470-2783

- Websites for families:
  - KidsHealth: https://kidshealth.org/en/parents/sma.html
  - Muscular Dystrophy Canada: https://muscle.ca/services-support/newborn-screening/
  - Cure SMA: https://www.curesma.org/

- Websites for healthcare providers:
  - MedlinePlus Genetics: https://medlineplus.gov/genetics/condition/spinal-muscular-atrophy/
  - UpToDate: https://www.uptodate.com/contents/spinal-muscular-atrophy
  - GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1352/

NOTE TO PARENTS/GUARDIANS: *This material is provided for informational purposes and provides basic information only. This material is not intended to be and does not take the place of medical advice, diagnosis, or treatment. Please talk to your health care provider if you have any questions or concerns.*

IWK Health

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