

# Alberta's NMS Program Annual Highlights: 2021 - 2022

## Spinal muscular atrophy screening comes to Alberta

Spinal muscular atrophy (SMA) is a genetic condition that affects the motor neurons of the brainstem and spinal cord resulting in muscle weakness and muscle wasting (atrophy) throughout the body. Early diagnosis and treatment leads to better health outcomes for infants with SMA.

Alberta's Newborn Metabolic Screening (NMS) Program has achieved another milestone with the launch of a SMA pilot program in February 2022. Intensive collaboration and consultation between all stakeholders and partners, including the NMS Program coordination team, the NMS Lab and Molecular Genetics Laboratory within Alberta Precision Laboratories, and Alberta Health made the launch successful.

- The Alberta Children's Hospital Foundation funded the development of SMN1 genetic testing to screen for SMA and a grant from Muscular Dystrophy Canada funded the pilot program in Alberta.
- New NMS Program resources were developed, including SMA-specific resources for [providers](#) and [parents](#), to support care provided to families who receive an abnormal SMA screen result (see below).
- The NMS Lab and Molecular Genetics laboratory will use the pilot period to work on validation of a combined test for both SMN1 and SCID to help reduce labour and consumables costs.

Following the completion of the SMA pilot program, SMA will officially become part of Alberta's publicly funded newborn screening program.

In the period of February 28<sup>th</sup> to March 31<sup>st</sup>, no infants received an abnormal screen result for SMA. We look forward to reporting on screening and detection for SMA in next year's annual report.

**Information for Health Professionals**

### Spinal Muscular Atrophy (SMA)

**What is SMA?**  
Spinal muscular atrophy (SMA) is a genetic disorder that affects the motor neurons of the brainstem and spinal cord, resulting in muscle weakness and muscle wasting (atrophy) throughout the body. Early diagnosis and treatment leads to better health outcomes for infants with SMA.

**What causes SMA?**  
SMA is a rare, progressive and irreversible neurodegenerative condition caused by a deficiency of the motor neuron protein SMN2 protein. The most common form is caused by a deletion of all copies of the SMN1 gene, which encodes the SMN protein. Other forms are caused by mutations in the SMN1 gene that affect its function, such as duplications, deletions, and point mutations. Other forms of SMA are caused by mutations in the SMN2 gene, which encodes the SMN2 protein. SMN2 copies are present in all cells, but their function is reduced in the presence of a SMN1 mutation.

**How common is SMA?**  
The incidence of SMA is about 1 in every 10,000 births.

**What are the clinical features of SMA?**  
SMA is a sub-classified into several types based on the age of symptom onset and the maximum age when a child can walk. There are four types of SMA (Type I, II, III, and IV) that vary in severity. Type I is the most common and severe form, with onset in infancy and a life expectancy of less than 2 years. Type II is the second most common and severe form, with onset in infancy and a life expectancy of less than 20 years. Type III is the third most common and severe form, with onset in childhood and a life expectancy of less than 40 years. Type IV is the least common and severe form, with onset in adulthood and a life expectancy of more than 40 years.

**How is SMA treated?**  
Treatment is currently available for some infants with SMA. Treatment can slow or even stop the progression of SMA, provided it begins before the onset of symptoms. Without treatment, those with more severe forms of SMA may die within the first few years of life. Infants with less severe forms of SMA will be checked regularly for symptoms of SMA. Some genetic counseling, treatment options, and/or educational support may be available to ensure optimal outcomes.

**Is SMA inherited?**  
SMA is inherited as an autosomal recessive disorder. Parents of a child with SMA are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. SMA carriers are healthy. Genetic counseling is available to families with SMA.

**Additional resources are available through:**  
Pediatric Neurology Clinic (Edmonton)  
5015 - 114 Street  
Edmonton, AB T6C 2G4  
Tel: 780-621-0700 (extension)  
Fax: 780-621-1185  
Emergency consultations:  
Phone: 403-495-3000 ext 461 or for pediatric neurology consult  
Alberta Children's Hospital  
2510 Chinnock Street  
Edmonton, AB T6C 2G4  
Tel: 403-495-3000  
Fax: 403-557-7500  
Emergency Consultations:  
Phone: 403-495-7211 ext 461 or for pediatric neurology consult  
Conditions Information for Parents:  
Visit us online and under "Child Reference" on our website or contact us at:  
www.albertahealthservices.ca

**Early screening and follow-up care – every baby, every time**  
For more information about the NMS Program, visit [www.albertahealthservices.ca](http://www.albertahealthservices.ca)  
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Resource for providers

**Information for Parents and Families**

### Spinal Muscular Atrophy (SMA)

*(Sounds like SPAI-NUHL, MUH-SKYUH-LR A-TRUH-FEE)*

**What is newborn blood spot screening?**  
This is a blood test that is done a day or 2 after babies are born to look for treatable conditions, including spinal muscular atrophy (SMA) that you, your doctor, or pediatric care team by just looking at your baby. This screening test is different from those done during pregnancy.

**What does it mean if my baby needs more tests for SMA?**  
Sometimes a baby needs to have more tests. This doesn't mean your baby has SMA. It means your baby will need more tests to find out if they have SMA or not.  
It's normal to feel worried if your baby needs more testing. Your specialist and other healthcare providers are there to support you. They'll explain what will happen and answer your questions.

**What is SMA?**  
SMA is a condition that affects the nerve cells that control your muscle movement. It causes muscle weakness, including weakness that gets worse over time (wasting). SMA affects your ability to crawl and walk. Over time, it may affect breathing and swallowing.

**What causes SMA?**  
SMA is genetic. This means babies with SMA are born with a gene from both parents, which has a change in it. This change affects the gene from working properly. SMA isn't caused by anything that happened during pregnancy.  
There may be no signs of SMA at birth. It's important to remember that this condition can't be seen by looking at your baby.

**What can I expect with more testing?**  
Everything that will happen with more testing will be explained to you. Your baby may need to have a physical examination and their blood may be tested to check for SMA.  
This testing may be done at a clinic in Edmonton or Calgary that specializes in SMA.

**What if more testing shows my baby has SMA?**  
If testing shows that your baby has SMA, you and your baby will get the best care available. Your healthcare team will work with you to make a care plan for your baby.

**How is SMA treated?**  
A team of SMA specialists will give you detailed instructions about the SMA treatment available for your baby. This treatment will be specific to your baby's needs.

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NMS Program Parent Conditions Information

Resource for parents

Visit [AHS.ca/newbornscreening](http://AHS.ca/newbornscreening) to learn more about Alberta's NMS Program