Alberta Health Services
Alberta Newborn Screening

Talking with Parents > When talking to a parent whose baby has received an abnormal screen result, let them know that an abnormal screen result doesn't mean that the infant has the condition. It means that they have a higher chance of having the condition, and they need more tests to find out for sure. A printable sheet for parents with more condition specific information can be used to support your conversation, visit ahs.ca/nms and under Quick Reference click on *What conditions are screened for?*

Spinal Muscular Atrophy (SMA) *Information for Health Professionals*

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. Motor neurons are specialized nerve cells that control the muscles used for activities such as breathing, crawling and walking.

What causes SMA?

SMA is a rare, progressive and irreversible neuromuscular condition caused by deficiency of the survival motor neuron (SMN) protein. It is most commonly caused by a deletion of both copies of the SMN1 gene, which encodes the SMN protein. Motor neurons fail in the absence of this protein affecting muscle functions such as breathing, swallowing and body movement. Differing amounts of this protein, in combination with SMN2 gene copy numbers and other modifying factors, determine the severity of the condition.

How common is SMA?

The incidence of SMA is about 1 in every 10,000 infants.

What are the clinical features of SMA?

SMA is sub-divided into several clinical subtypes based on the age of symptom onset and the maximum attainable gross motor function. Infants with early onset SMA (type 1) may appear normal at birth. They present with poor muscle tone, severe muscle weakness and muscle wasting which gets worse over time (e.g. unable to support their head or sit unassisted), breathing problems, difficulty feeding and swallowing, absent reflexes, and failure to thrive during the first six months of life. Children with type 2 (sitters, non-walkers) and type 3 (walkers) SMA usually present after 6 months of age, with low muscle tone (hypotonia), reduced or absent reflexes, and proximal muscle weakness leading to motor developmental delay or regression.

What is the screening test for SMA?

Screening for SMA is performed by looking for the common SMN1 gene deletion. If no copies of the SMN1 gene are detected, the infant has a positive screen result for SMA. Newborn screening may not detect all infants with SMA. Infants with symptoms or signs consistent with SMA require timely assessment and diagnostic testing even if their newborn screen result is normal.

How is the diagnosis of SMA confirmed?

The diagnosis of SMA is confirmed by a physical assessment/ examination by a pediatric neurologist and blood test for genetic testing. The Pediatric Neurology Clinic will arrange for diagnostic genetic testing and other tests as needed.

How is SMA treated?

Treatment is currently available for some infants with SMA. Treatment can slow or even stop the progression of SMA, especially if given before the onset of symptoms. Without treatment, those with more severe forms of SMA may die within the first two years of life.

Infants with less severe forms of SMA will be checked regularly for symptoms of SMA. If they develop symptoms, treatment options will be considered. Regular follow-up in a multidisciplinary pediatric neuromuscular clinic is important 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 counselling is available to families with SMA.

Additional resources are available through:

Pediatric Neurology Clinic (Edmonton)

Stollery Children's Hospital
8440 – 112 Street
Edmonton, AB T6G 2B7
Tel: 780-407-6108 (voicemail)
Fax: 1-888-353-1163

Emergency consultations:
Phone 780-407-8822 and ask for the pediatric neurologist on call.

Pediatric Neurology Clinic (Calgary)

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Tel: 403-955-5437
Fax: 403-955-7609

Emergency Consultations:
Phone 403-955-7211 and ask for the pediatric neurologist on call.

Early screening and follow-up care – every baby, every time

For more information about the Alberta Newborn Screening Program, visit www.ahs.ca/newbornscreening