INFORMATION FOR PARENTS/GAURDIANS:
My Baby Has a Positive Spinal Muscular Atrophy Newborn Screening Result

What is newborn screening?
These are routine tests done shortly after birth on every baby born in Ontario. A small sample of blood is taken from the heel of your baby and tested for rare, treatable diseases, including Spinal Muscular Atrophy (SMA).

What is Spinal Muscular Atrophy (SMA)?
Spinal muscular atrophy is a rare inherited (genetic) disease that causes muscle weakness and muscle wasting (atrophy). There are 4 main types of SMA. The type of SMA depends on the age that symptoms start to develop and by the highest level of motor skills (e.g. sitting, crawling, walking) that a baby or child is able to achieve:

- Type 1: babies are weak and not able to sit up by themselves. Symptoms start to appear in the first 6 months of life
- Type 2: children can sit by themselves but do not walk. Symptoms start to appear between 6-18 months of age
- Type 3: children can walk and sit by themselves but start having muscle weakness and muscle wasting in childhood, after 18 months of age
- Type 4: symptoms of this type of SMA appear in adulthood

Newborn screening for SMA detects SMA type 1 and 2, most cases of type 3, and some cases of type 4.

What are the symptoms of SMA?
Babies with SMA often have no signs or symptoms at birth. Symptoms appear typically in the first few weeks of life for SMA type 1. Early signs may include:

- Low muscle tone (hypotonia)
- Muscle weakness and wasting that gets worse as time goes on
- Loss of reflexes
- Uncontrolled movements of the tongue (fasciculations)
- Tremor
- Feeding and swallowing difficulties, poor weight gain (failure to thrive)
- Abnormal/rapid breathing patterns

How does a baby get SMA?
SMA is an inherited (genetic) disease. Most of the time (~97%), SMA is caused by deletions in the SMN1 (survival motor neuron 1) gene. Genes provide instructions for our bodies to make proteins. A deletion means that all or part of the SMN1 gene is missing and the body cannot make any working SMN1 protein. Newborn screening only detects cases of SMA caused by deletions in the SMN1 gene; it cannot detect SMA caused by other types of genetic changes in the SMN1 gene.

The type of SMA a person has depends on the number of copies of another gene, SMN2. People can have 1 to 6 copies of SMN2. The more SMN2 gene copies that a person with SMA has, the less severe their symptoms are anticipated to be.

Each parent of a child with SMA usually has one non-working SMN1 gene and one working copy and is considered a "carrier". Carriers are healthy and often unaware they carry a non-working SMN1 gene.

What does it mean if my baby has a positive SMA newborn screening result?
A positive SMA newborn screening result means that deletions in the SMN1 gene and 1, 2, 3 or 4 copies of the SMN2 gene were found in your baby's sample. There is a high chance that your baby has a form of SMA and follow-up with a pediatric neurologist (a doctor who specializes in disorders that affect the nerves, spinal cord, and brain) is needed to know for sure.

What tests will help determine if my baby has SMA?
Blood tests are usually done to confirm a baby has SMA. Your baby may also have a physical exam and other tests to look for signs of SMA.

When can my baby have these tests?
Your baby's doctor or a health care professional at a Newborn Screening Regional Treatment Centre will call you to talk about the results of your baby's positive newborn screen for SMA and arrange follow-up testing as soon as possible.

How is SMA treated?
Babies with SMA are followed closely by a team of doctors and healthcare providers who will discuss the management and treatment options available. In Ontario, a medication called Nusinersen (Spinraza) is approved for the treatment of patients who have SMA with 1, 2, or 3 copies of the SMN2 gene. Nusinersen is given through a spinal tap (lumbar puncture) in a hospital by a doctor and treatment is lifelong. Other treatments for SMA (e.g. gene therapy) are being developed but are not yet approved for use in Canada. Babies with 4 copies of SMN2 will be monitored regularly for symptoms of SMA.

Why screen for SMA?
Screening helps find babies with SMA as early as possible so that treatment and monitoring can be started sooner.

What happens when SMA is treated?
When treatment with Nusinersen is started early, it can stop the symptoms of SMA from getting worse and helps to give a better chance to achieve the highest level of motor skills possible.

Where can I get more information?
For more information on newborn screening, please talk to your local health care provider or visit our website at www.newbornscreening.on.ca

For more information about SMA, please visit the Families of SMA Canada Society at www.curesma.ca

NOTE TO PARENTS/GUARDIANS: This information is intended for parents whose baby has had a positive newborn screening result for Spinal Muscular Atrophy (SMA). This fact sheet was written for information purposes only and should not replace professional medical advice, diagnosis, or treatment.