

Is there a cure?

At present there is no cure for Spinal Muscular Atrophy, and there is no treatment that can repair the motor neurons of the anterior horn region of the spinal cord or reverse the progressive weakening of the muscles caused by SMA.

As a result, treatment for SMA focuses on the symptoms and on supportive care.

Carrier testing

Testing can be done to determine if someone is a carrier for SMA by a blood test. This is done by testing the number of copies of the SMA gene. Normally there are 2 copies (one copy on each chromosome 5).

Carriers can be determined if they are found to have only one copy of the SMA gene by this test. However, a small percentage of carriers are missed using this test. This is because they have 2 copies of the SMA gene on one chromosome 5. They are missing a normal SMA gene on the other chromosome 5 and are therefore carriers. This only occurs in 4% of carrier tests.

Pre-natal test

Pre-natal testing is available for couples who may wish to know if their baby may be affected with SMA.

A very early test at 11 weeks called the chorionic villus sampling test also known as CVS can be performed during pregnancy to test the baby.

A test called amniocentesis is also available later in pregnancy.

If you wish to consider any of the tests above it is vital to contact your local Genetics Centre before nine weeks of pregnancy to make arrangements for the test.

For more information

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Birmingham Women's 
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Spinal Muscular Atrophy Type 1

An information leaflet for
patients and families

If you need more advice about any aspect
of SMA please contact:

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What is SMA?

Spinal Muscular Atrophy (SMA) is a genetic disease that affects muscle movement. It causes the motor neurons in an area of the spinal cord called the anterior horn to deteriorate.

Motor neurons are nerve cells in the spinal cord that send impulses to the muscles, telling them to relax or contract. The deterioration of the motor neurons gradually breaks the link between the brain and the muscles that this part of the spinal cord controls. As the link is broken, the muscles used for activities such as crawling, walking, sitting up and moving the head are used less and less and so become weaker, or shrink (atrophy).

Symptoms

SMA affects muscles throughout the body, although the proximal (those closest to the shoulders, hips and back) are usually most seriously affected. They are also affected much sooner than the muscles further away from the centre of the body. For example, the muscles of the thighs become weaker than the muscles of the lower legs and feet.

How many types of SMA are there?

SMA is classified into four types, based on the age at which it develops and the severity of the symptoms.

Type I, II and III develop in childhood. Type IV is SMA that starts in adulthood, with symptoms usually beginning over the age of 35.

SMA Type I

Also known as Werdnig-Hoffman disease, Type I is the most severe form. It can develop before birth (some mothers notice decreased movements of the baby in the final months of their pregnancy), up to six months of age.

Babies with Type I have weak, thin muscles that make them limp or floppy, unable to sit without support or to raise their head. They usually have breathing and swallowing problems due to the weakness of their chest muscles, and their tongue may shrink. Affected children are unlikely to live past their second birthday.

What is the cause?

A person with SMA has a deleted or altered gene (the Survival Motor Neuron 1 gene), making them unable to produce Survival Motor Neuron protein. Without this protein, motor neuron cells in part of the spinal cord deteriorate and die. As a result, nerve impulses are unable to get through to the muscles that the motor neurons control, which become weaker and shrink due to lack of use.

Genes and chromosomes

Genes are the unique set of instructions in every cell which make each of us an individual. There are many thousands of genes, each carrying a different instruction. If a gene is altered, it can cause a genetic condition or disease.

We have two copies of each gene and they are stored on chromosomes which

also come in pairs.

One copy is inherited from our mother and one copy from our father. When we have children, we pass on only one copy of each of our genes by passing one of each pair of chromosomes.

Inheritance

SMA Type 1 is an autosomal recessive disease. This means that in order for a child to be affected by SMA the deleted or altered gene must be passed on from both parents.

The parents of an affected child have one normal copy of the gene and one deleted/altered copy. This is known as being a carrier. Carriers are unaffected. If two carriers have a child, the baby has a 1 in 4 (25%) chance of having SMA (see diagram below). About 1 in 50 people carry the deleted/altered gene.

