Sonic Genetics
Quality is in our DNA

Genetic carriers of spinal muscular atrophy

Information for patients

The healthcare professional responsible for your care has given you this leaflet because you have been identified by Preconception Carrier Screening as a carrier of a change in the gene that causes spinal muscular atrophy. This fact sheet contains more information about the particular genetic disorder mentioned in your report. We recommend that you also discuss your result with an experienced doctor or genetic counsellor.

Each year, about one in 6,000 Australian babies, boys and girls, are born with spinal muscular atrophy (SMA). This inherited genetic condition affects the nerve cells in the spine so that the muscles in the body become weak and waste away. New medication for SMA is becoming available, but there is no cure for the condition.

There are four different types of SMA, with a wide range of symptoms and age of onset. Babies born with SMA type 1 usually show muscle weakness at birth or soon after. The weakness progresses so that they rarely survive more than two years.

Usually the symptoms of SMA type 2 start in early childhood but are less severe. The onset of symptoms of type 3 is from childhood to adolescence and it is the mildest form. The muscle weakness in type 4 does not start until adulthood and life expectancy is unaffected.

Genetic conditions are caused by inheriting faulty genes. Our genes are made of DNA and tell the body how to function, grow and develop. The SMA gene (its scientific gene name is SMN1) contains the information that the nerve cells in the spinal cord use to make our muscles work. Everyone has two copies of the SMA gene. Most people will have both of their SMA gene copies working as they should. It is only when both of their SMA genes are faulty that the person has one of the types of SMA. The type of SMA they have depends on the particular type and location of the faults in the person's SMA genes.

About one in 40 Australians will be born with one SMA gene copy that is working normally and one copy that is faulty. These people are genetic carriers of SMA. However, the cells need only one SMA gene copy to be working normally to stop the muscles becoming weak and wasted. Genetic carriers of SMA do not have any symptoms of the condition and will not develop them.

How to interpret a carrier result for spinal muscular atrophy

Hearing about the carrier result will usually be unexpected. It may lead to a range of physical and emotional reactions. In the midst of this emotional confusion, there are some decisions that need to be made. You need as much information and support as possible when making these decisions.

Genetic counsellors are experienced in providing support and information that will help you and your partner make an informed decision. The only right decision is the one that is best for you and your family.

If you are not pregnant, you have time to think about the carrier test result when discussing your family planning options. Among these options is testing the baby in pregnancy for SMA (prenatal testing). Another is using IVF and then testing the embryo for SMA before implantation. However, having no testing for SMA at all is also an option.

If you are pregnant, your options include prenatal testing or no testing for SMA. Again, seeking support and information when making these decisions is important.

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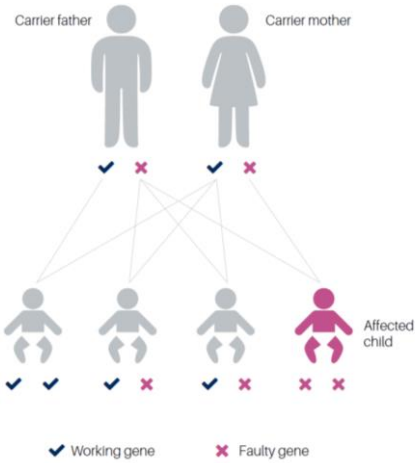
How does a person inherit SMA?

For a baby to have SMA, it must inherit the faulty gene from both their mother and father. Where both parents are genetic carriers of SMA, there is a one in four chance (25%) in **every pregnancy** that they will each pass on the faulty SMA gene copy to their baby who will then have SMA at birth or later.

In the Figure shown, both parents are carriers of SMA, having one normal working SMA gene and one faulty SMA gene each. Their child will inherit one SMA gene from each parent. There are four possible combinations of SMA genes that the child could have, i.e. the child has two normal genes (one in four chance), or is a carrier (two in four chance), or has two faulty genes (one in four chance).



What does being a SMA genetic carrier mean for me?

A genetic carrier of SMA has one faulty SMA gene and one normal SMA gene which is working normally. The presence of the normal SMA gene is sufficient to ensure that a carrier will not develop SMA. There is a one in two chance (50%) of passing the faulty gene onto your child, but the child will not be affected if the child has inherited a normal SMA gene from the other parent.

A child will develop SMA only if the child has inherited a faulty gene from both parents. You have been identified as a carrier of SMA. It is important to know the result of your partner's SMA carrier test.

What if one parent is a carrier and the other parent's carrier test is normal?

There are many variations in the SMA gene that make it faulty. The carrier test looks for the most common variations. There is a small chance that the test has not found one of the rare variations that make the SMA gene faulty. The possibility of having a baby with SMA is still low (about one in 2,000), but the chance is higher than in the general population (one in 6,000). You should discuss this result with your doctor. Genetic counsellors are also experts in providing this information.

Your test result is important for your relatives

If you are a carrier of the faulty gene, you very likely inherited it from one of your parents. That means that your brothers, sisters and cousins, if you have any, also have a chance of being a SMA genetic carrier. Telling them about the availability of the carrier test may help them make informed family planning decisions as well.

Sonic Genetics offers free genetic counselling for couples where both partners are identified as carriers for spinal muscular atrophy. If you are eligible for this service, your doctor may refer you for genetic counselling. Conditions apply and can be discussed with your doctor.

Further information and contacts

The Centre for Genetics Education www.genetics.edu.au