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Sonic Genetics

Information for patients

Carriers of spinal muscular atrophy

The healthcare professional responsible for your care has given you this leaflet because you have been identified by reproductive carrier screening as a carrier of spinal muscular atrophy. This fact sheet contains some information about this genetic disorder. We recommend that you also discuss your test result with your doctor or genetic counsellor.

What is spinal muscular atrophy?

Each year, about one in 10,000[1] babies in Australia is born with spinal muscular atrophy (SMA). This inherited genetic disorder affects the nerve cells in the spine so that the muscles in the body become weak and wasted.

SMA is classified into four types, according to the severity of the disorder. 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. SMA type 2 starts in early childhood and is less severe, but these children never stand without support. The onset of SMA type 3 is from childhood to adolescence; these children may initially walk but become wheelchair-dependent. The muscle weakness in SMA type 4 does not start until adulthood and life expectancy is unaffected. New medication for SMA is becoming available, but there is no cure for the disorder.

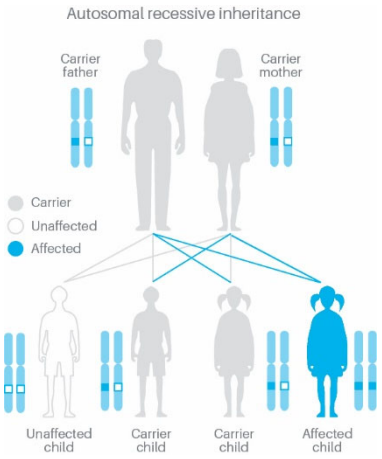
Genetic disorders can be caused by inheriting faulty genes. Our genes are made of DNA and tell the body how to function, grow and develop. The SMA gene (its scientific gene name is SMN1) contains the information that the nerve cells in the spinal cord use to make our muscles work. Everyone has two copies of the SMA gene. In most people, both copies of the SMA gene are working as they should.

About one in 50[1] Australians will be born with one SMA gene that is working normally and one that is faulty. These people are carriers of SMA. The cells need only one copy of the SMA gene to be working normally to stop the muscles becoming weak and wasted. Carriers of SMA do not have any symptoms of SMA. A person will develop SMA only when both of their SMA genes are faulty. The variation in the severity of SMA is due to variations in a second, related gene called SMN2.

How does a child inherit SMA?

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

In the Figure shown, both parents are carriers of SMA, having one normal 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, that is, 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). The sex of the child bears no relevance to the inheritance of this disease.



Information for patients

What does being an SMA genetic carrier mean for me and my child?

A genetic carrier of SMA has one faulty SMA gene and one normal SMA gene. The presence of the normal SMA gene is sufficient to ensure that a carrier will not develop SMA.

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

What if both parents are carriers of SMA?

If you are not pregnant, you and your partner have time to think about the carrier test result when discussing your options for family planning. These options include testing the baby in early pregnancy for SMA (prenatal testing). Another option is using IVF and then testing the embryo for SMA before implantation. There are other options, such as using a donor egg or sperm, or having no testing for SMA at all. You should discuss these options with your doctor.

If you are pregnant, your options include prenatal testing, or no testing for SMA. You should discuss these options with your doctor.

Hearing about your carrier result will usually be unexpected. Some people find this information confronting or confusing at the very time that decisions need to be made. You need information and support when making these decisions.

Your doctor may suggest that you discuss these issues with a genetic counsellor, a healthcare professional who can help you and your partner make an informed decision. Sonic Genetics offers free genetic counselling for couples where both partners are identified as carriers. Your doctor can refer you for this service; conditions apply and can be discussed with your doctor.

Your doctor or genetic counsellor will not tell you what to do: the right decision is what you decide is best for you and your family.

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

If one parent is a carrier there is a one in two chance (50%) of passing the faulty gene to your child. However, the child is very unlikely to be affected as the child will almost certainly inherit a normal SMA gene from the other parent.

There are rare situations in which a carrier test does not detect a person's faulty SMA gene. This means that the carrier test result is 'normal' even though the person is actually a carrier. The reason for this is that there are a number of variations in the SMA gene that can make it faulty. Our reproductive carrier screen looks for the commonest variations that occur in Australia. There is a small chance that the test has not found one of the rare variations that can make the SMA gene faulty. If one parent is known to be a carrier, and no SMA variant has been detected in the other parent, the possibility of having a child with SMA is very low (about one in 2,000), but the chance is higher than in the general population (one in 10,000[1]). You should discuss this result with your doctor or genetic counsellor.

Your test result is important for your relatives

If you are a carrier, you probably inherited the faulty gene from one of your parents. That means that close family members, such as brothers, sisters and cousins, may also be carriers. We recommend that you tell them about your result and the availability of reproductive carrier screening, so that they can make their own informed decisions about family planning.

Further information is available at The Centre for Genetics Education, genetics.edu.au

[1] Archibald A, Smith M, Burgess T, et al. Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. *Genet Med*. 2018; **20**(5):513-526

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