Victorian Clinical Genetics Services
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prepair™ fact sheet

Spinal muscular atrophy

What does this result mean?

The results of your test show that you are a carrier of SMA. Most people have two copies of the SMA gene. Carriers of SMA have one copy. Carriers of SMA are healthy and will not develop symptoms of SMA. About one in every 40 people are carriers of SMA.

SMA occurs when a baby inherits two faulty copies of the SMA gene. When both parents are carriers, the chance of a baby having SMA is 1 in 4.

What happens next?

Given your results, your partner will be offered SMA carrier testing. There are two possible outcomes for your partner's test:

- Partner receives a low risk result: There is a low chance (about 1 in 2000) that you and your partner will have a baby with SMA. No further testing is recommended but it is important that you discuss this result with your doctor or a genetic counsellor.
- Partner receives a carrier result: Your chance of having a baby with SMA is 1 in 4 for each pregnancy that you have. It is recommended that you see a genetic counsellor who will discuss your results with you.

If you and your partner are both carriers and you are not pregnant there are a number of family planning options available which a genetic counsellor will discuss with you. This includes pre-implantation genetic diagnosis using in vitro fertilisation (IVF).

If you and your partner are both carriers and you are pregnant then you might consider prenatal testing for SMA. Prenatal testing can confirm if the pregnancy is affected. It is your choice whether or not to have further testing.

What does prenatal testing involve? A prenatal diagnostic test will tell you if your baby has inherited two faulty copies of the SMA gene. Diagnostic testing involves passing a fine needle into the developing placenta or the amniotic fluid and drawing a small amount of sample into a syringe under ultrasound guidance. The sample is sent to the laboratory for testing. SMA testing results (often including full chromosome analysis) are ready in approximately 2 weeks. Diagnostic tests carry a small risk of miscarriage.

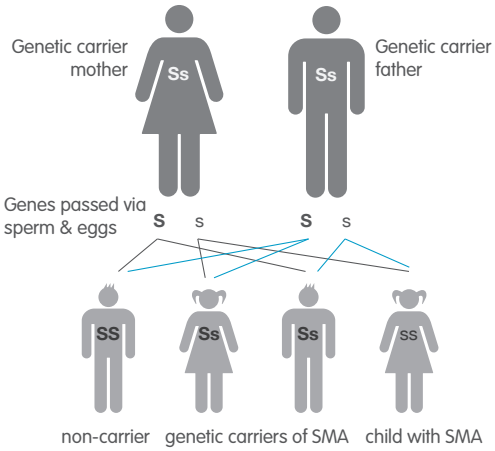
If your baby has spinal muscular atrophy: If SMA is detected, you and your partner will be offered genetic counselling to discuss the options available to you. You may need to face the difficult decision of whether to continue your pregnancy.

With prepair™, genetic counselling is available to you through the Victorian Clinical Genetics Services (VCGS). A genetic counsellor can talk with you about what your result means for you and your family. You can contact a genetic counsellor directly on 03 9936 6402. It is also advisable to discuss your results with your doctor.

What is spinal muscular atrophy?

SMA is a condition that affects nerves in the spinal cord and causes muscles to get weaker. There are four types of SMA with SMA type 1 being the most common and the most severe. Babies with SMA type 1 have weak muscles from birth and usually do not live past two years of age.

There is no cure for SMA, however treatment is aimed at managing symptoms and improving quality of life.



Inheritance of spinal muscular atrophy.
S = normal/working SMA gene
s = faulty SMA gene

*Males & females can be affected equally

What does my carrier result mean for my relatives?

SMA is an inherited condition which means it is likely that some of your relatives may also be carriers of SMA. Carrier testing is available to your relatives. Family members can access testing through VCGS or through their local clinical genetics service. Details of genetics services in Australia and New Zealand are available at:
www.genetics.edu.au/Genetics-Services/genetic-counselling-services

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prepair™ genetic carrier screening