Carrier Screening:

If you find out you are a carrier...

Have you just had a carrier screening test and found out you are a carrier of cystic fibrosis, fragile X syndrome, spinal muscular atrophy, or another inherited condition?

Are you wondering what to do next?

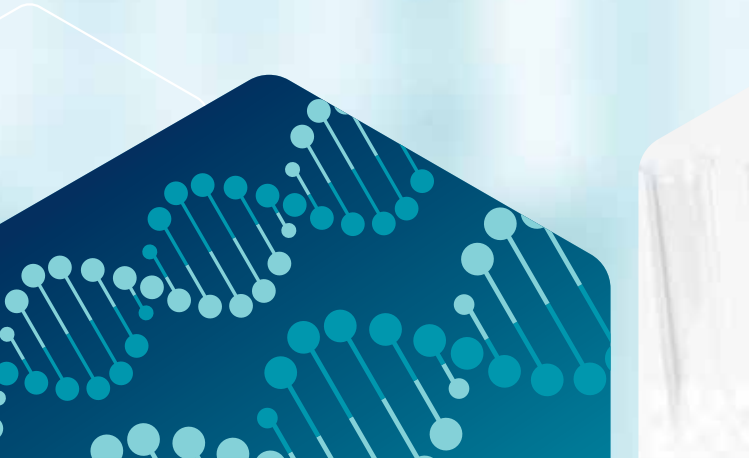
If your results show that you are a carrier of:

Cystic fibrosis (CF), spinal muscular atrophy (SMA), or any other 'autosomal recessive' condition:

- A couple can only have a child with the condition if both parents are carriers.
- When both parents are carriers for the same condition, the risk of having a child with that condition is 1 in 4 (or 25%) for each pregnancy you have.
- If you are a carrier, your partner should consider a carrier screening test.
- This test will determine if you have an increased chance of having a child with that condition. Carrier screening can be arranged by your healthcare provider or a genetic counsellor.

Fragile X syndrome (FXS) or an 'X-linked' condition (and you are female):

- You have an increased chance of having a child with that condition.
- Testing for your partner is not necessary.
- If you are a carrier of FXS you should speak to a genetic counsellor, as there are some considerations for your own health.



If you have an increased risk of having a child with an inherited condition

If results show that you and your partner are both carriers of an 'autosomal recessive' condition or if the female partner in the couple is a carrier of an 'X-linked' condition, it is recommended that you discuss your results with a genetic counsellor, who will help you to consider your options.

Options for further testing include:

- If you are not pregnant: pre-implantation genetic diagnosis using in vitro fertilisation (IVF).
- If you are pregnant: testing in early pregnancy through chronic villus sampling (CVS) or amniocentesis.

Some people may choose to avoid having a child with the condition whereas others may choose not to alter their path. These choices are entirely up to you, and genetic counselling services can support you in making these decisions.

Genetic carrier screening is available for your blood relatives

It is important to share your carrier status with blood relatives, and inform them on the availability of screening.

Having a family member with a genetic condition, or who is a carrier, increases your chance of being a carrier.

Carrier screening is recommended either before pregnancy or early in pregnancy (during the first 12 weeks)

Screening is relevant to everyone - most people who are carriers do not have a known family history of the condition. Making a decision about screening is a choice only you can make.

Recommended carrier screening provider

Victorian Clinical Genetics Services (VCGS)

VCGS provides carrier screening services including genetic counselling throughout Australia and New Zealand.

w. vcgs.org.au/tests/prepair
e. vcgs@vcgs.org.au
p. 1300 118 247

For more information

Cystic Fibrosis Community Care (CFCC)

w. cfcc.org.au
p. (03) 9686 1811

CYSTIC FIBROSIS Community Care

Fragile X Association of Australia (FXAA)

w. fragilex.org.au
p. 1300 394 636

Fragile X Association of Australia

Spinal Muscular Atrophy Australia Inc. (SMA Australia)

w. smaaustralia.org.au
p. (03) 9796 5744

Spinal Muscular Atrophy AUSTRALIA INC.

This brochure was produced by the Carrier Screening Community Education Campaign Reference Group which includes representatives from CFCC, FXAA, Genetic Support Network of Victoria, Royal Children's Hospital Melbourne, SMA Australia, and VCGS.VCGS has no financial affiliation with CFCC, FXAA, or SMA Australia.