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 genetic condition?

Are you wondering what to do next?

If your results show that you are a carrier of:

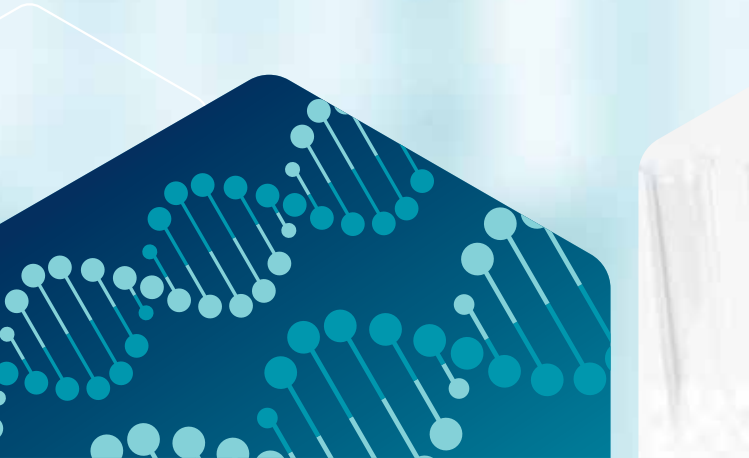
Cystic fibrosis (CF), spinal muscular atrophy (SMA), or any other 'autosomal recessive' condition:

- A couple can only have a child with the condition if both parents are carriers.
- If you are a carrier, your partner can consider a carrier screening test.
- When both parents are carriers for the same condition, the chance of having a child with that condition is 1 in 4 (or 25%) for each pregnancy you have.
- Being a carrier does not impact on your own health.

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 fragile X you should speak to a genetic counsellor, as there are some considerations for your own health.

It is recommended you discuss your results with a genetic counsellor. This can arranged by your healthcare provider.



If you have an increased chance of having a child with a genetic condition...

If both 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. They will help you consider your options and support you in making decisions about further testing.

People who have an increased chance of having children with a genetic condition have a range of options available to them. Some will choose to avoid having a child with the genetic condition and others will choose to use the information to plan and prepare. These choices are entirely up to you, and genetic counselling services can support you in making these decisions.

- If you are not pregnant, options you may consider include the use of in vitro fertilisation (IVF) with genetic testing of the embryos, IVF with the use of donor eggs, sperm or embryos, adoption or choosing not to have children.
- If you are pregnant, testing can be done in early pregnancy through chorionic villus sampling (CVS) or amniocentesis. This will tell you whether the baby has or will develop the condition.

Genetic carrier screening is available for biological relatives

If you are a carrier for a genetic condition, there is an increased chance that your biological relatives may also be carriers. It is important to consider sharing this information with your biological relatives, and to tell them that they can access carrier screening should they wish.

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.

Where can I go for more information?

carrierscreening.org.au

Spinal Muscular Atrophy Australia Inc. (SMA Australia)

w. smaaustralia.org.au
p. (03) 9796 5744
e. smaaa@smaaustralia.org.au

ABN 82 885 991 569 A004766OD

Spinal Muscular Atrophy AUSTRALIA INC.

Fragile X Association of Australia (FXAA)

w. fragilex.org.au
p. 1300 394 636
e. support@fragilex.org.au

ABN 18 655 264 477 ARBN 626 478 966

Fragile X Association of Australia

Cystic Fibrosis Community Care (CFCC)

w. cfcc.org.au
p. (03) 9686 1811
e. admin@cfcc.org.au

ABN 251 26 031 536

CYSTIC FIBROSIS Community Care

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