



prepair™

genetic carrier screening

Reproductive genetic carrier screening

Genetic carrier screening gives individuals and couples information about their chance of having a child with a genetic condition. prepair™ by VCGS can be arranged through your doctor. It will tell you if you are a carrier of three common inherited conditions: cystic fibrosis (CF), fragile X syndrome (FXS) or spinal muscular atrophy (SMA).

Many people are carriers of CF, FXS or SMA even though no one in their family has the condition.

prepair™ looks for the most common gene changes associated with CF, FXS and SMA in children. It will identify about 90% of people who are carriers of CF, 95% of people who are carriers of SMA and over 99% of people who are carriers of FXS. However, the test will not detect every person who is a carrier.

Could I be a carrier?

	Number of people who are carriers of the condition	Number of people with the condition
Cystic fibrosis	1 in 25	1 in 2,500
Fragile X syndrome	1 in 150	1 in 4,000
Spinal muscular atrophy	1 in 40	1 in 6,000 - 1 in 10,000

What causes these conditions?

Genetic conditions are caused by changes in genes, which provide the instructions for our bodies. Babies inherit one copy of each gene from each parent. Some people carry certain gene changes which mean there is a chance they may have a child with a genetic condition:

- CF and SMA occur when a baby inherits a gene change from both parents.
- FXS is caused by an increase in the size of the *FMR1* gene. prepair™ will tell you whether you carry a gene change that increases your chance of having a child with FXS.

How the test process works

1. Decide whether or not to have the test:

It is your choice whether or not to have the test. Read through the information provided and discuss the test with your doctor or genetic counsellor.

It is recommended that the female member of the couple is screened first as FXS testing is not necessary for the male partner.

2. Consider whether it is the right time to have the test:

To ensure there is time to make decisions it is best to have this test either before pregnancy or early in pregnancy. If you are pregnant, we recommend having screening before 12 weeks of pregnancy.

3. Request the test:

If you would like to have the test, your doctor or genetic counsellor will need to complete a test request slip. This test is not covered by private health insurance. Contact your health professional for information about cost.

4. Sample collection:

Your doctor will arrange for you to have a blood or saliva sample taken. Your sample will be sent to VCGS where it will be processed.

5. Receive your results:

It will take approximately 10 working days (from receipt of sample) for your test to be processed and your results to be sent to your doctor or genetic counsellor. They will discuss your results with you.

6. If the test shows you are a carrier, talk to your doctor or genetic counsellor:

Your result will tell you if you are a carrier of CF, FXS or SMA. If you are a carrier, your partner may need to have carrier testing and it is recommended that you speak to your doctor or genetic counsellor.

What is cystic fibrosis?

CF is an inherited condition affecting breathing and digestion. CF causes thick mucus which traps bacteria, resulting in recurrent infections that damage the lungs. Thick mucus in the gut also makes digestion of food difficult. Infants, children and adults with CF require daily chest physiotherapy to clear mucus from their lungs, frequent courses of antibiotics, and need to take medicine to aid digestion. There is no cure for CF but better treatments are under research and development.

What is fragile X syndrome?

FXS is the most common cause of inherited intellectual disability. People with FXS can have developmental delay, learning difficulties, anxiety, autism and epilepsy. The features of FXS vary from mild to severe with males more likely to be severely affected than females. There is no cure for FXS although some educational, behavioural and medical interventions can improve outcomes for people with FXS. Some females who are carriers of FXS may have early menopause.

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 there are treatments and interventions available aimed at managing symptoms and improving quality of life.

If I am a carrier, what is the chance I will have a child with the condition?

If your test shows that you have one copy of the gene change for CF or SMA, you are a carrier of this condition. However, a couple can only have a child with CF or SMA if both parents are carriers of that condition. If you are a carrier, your partner will be offered testing. Two people who are carriers of the same condition have a 1 in 4 (25%) chance of having a child with the condition for each pregnancy.

For FXS, only women who carry a gene of increased size have a greater chance of having a child with FXS. This means that your partner will not need to be tested for FXS. Female carriers of FXS have a 1 in 2 (50%) chance of passing that gene onto each child they have.

If I am a carrier, can I develop symptoms?

If you are a carrier of CF or SMA you have a change in one of your two genes associated with these conditions. Carriers do not develop symptoms. Some female carriers of FXS may develop fertility problems and go through menopause early (before 40).

Some male and a small number of female carriers of FXS may develop a late-onset neurological condition which causes tremors and balance problems which worsen with age.

My partner and I are both carriers. What happens now?

If you and your partner are carriers of CF, FXS or SMA your doctor or genetic counsellor will discuss your results with you, including options for further testing. A number of options are available to women who are carriers of FXS and couples who are both carriers of CF or SMA.

Genetic testing for these conditions in the fetus can be done during pregnancy. If the tests diagnose CF, FXS or SMA, you have a choice about whether to continue or terminate the pregnancy.

If you are identified as a carrier prior to pregnancy, there are family planning options available to you including, pre-implantation genetic diagnosis using in vitro fertilisation. If you are a carrier, genetic carrier testing is also available to your blood relatives.

Do I need to have carrier testing every time I have a baby?

No, you only need to have this test once in your lifetime. If you are not a carrier, you have a very low chance of having a child with the condition. If you are a carrier and you have a new partner, your new partner may need to be tested.

If no gene change is found, could I still be a carrier?

Yes. The current carrier test detects the majority of carriers but it cannot detect every gene change that causes CF, FXS and SMA. If no gene change is found, you are not a carrier of the most common gene changes, but there is still a small chance for each condition that you are a carrier of rarer gene changes.

What if I have a relative who is a carrier or has CF, FXS or SMA?

If you have a family history of one or more of these conditions, your chance of being a carrier is greater than most people and you and your partner should consider testing. If you or your partner has a family history of any of these conditions, you should discuss this with your doctor or genetic counsellor.

Testing laboratory:

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