



## What is prepair carrier screening?

prepair carrier screening will tell you if you are a carrier for three common inherited conditions: cystic fibrosis (CF), fragile X syndrome (FXS) and spinal muscular atrophy (SMA).

Being a carrier usually does not affect your own health.

Anyone can be a carrier of a genetic condition, even if no one in their family has that condition. <sup>1</sup>

## Why might I have carrier screening?

prepair carrier screening helps you understand your chance of having a baby with an inherited genetic condition.

# When can I have carrier screening?

Genetic carrier screening is ideally done before you get pregnant. Having this information before pregnancy allows you to consider your reproductive options (e.g. IVF and pre-implantation genetic diagnosis).

Carrier screening can also be done during early pregnancy. Couples with an increased chance of having a child with a genetic condition can then choose to have diagnostic testing. This can tell you if your pregnancy has the genetic condition.

<sup>1</sup>Archibald, A. D., et al (2018). Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. Genetics in Medicine, 20(5), 513-523.

## How does carrier screening work?

Carrier screening can be done using blood or a saliva sample. Testing will identify whether you are a carrier for a genetic condition (your carrier status).

There are two types of *prepair* carrier screening available.

## prepair carrier screening

This test looks at three commonly inherited genetic conditions in our population.

- Cystic fibrosis (CF)
- Fragile X syndrome (FXS)
- Spinal muscular atrophy (SMA)

## prepair+ (expanded carrier screening)

This test looks at over 250 different genetic conditions (including CF, FXS, SMA).

## How do I arrange testing?



#### Step 1

Talk to your doctor about genetic carrier screening. If this test is appropriate, they will complete a test request form.

# Step 2 For prepair

Testing can be performed using blood or saliva. Visit our website to find blood collection sites OR to arrange a saliva kit to be sent to you.

#### For **prepair+**

Email a copy of your request form to carrierscreening@vcgs.org.au and we will send you the specific saliva kits. If you're already pregnant, we may organise blood samples from both partners instead.





#### Step 3

Results will be available from your doctor. In some cases, your doctor might refer you to one of our genetic counsellors for your results.



## **Compare test options**

	prepair	prepair+
What conditions are included?	Screens for 3 conditions - CF, FXS, SMA.	Screens for hundreds of conditions.
Who is tested?	Biological mother usually screened first. If a carrier of CF or SMA, partner will be offered testing.	Both biological parents screened together.
How long does it take?	Takes 2 weeks*	Takes 5-6 weeks*
How much does it cost?	Costs \$389^ (partner testing is free if required)	Costs about \$900 / couple
When is testing done?	Ideally performed pre-conception, or before 12 weeks in pregnancy.	Ideally performed pre-conception, or before 8 weeks in pregnancy.
Who performs the test?	Tested by VCGS in Melbourne, Australia.	Tested by overseas partner in USA#
Why choose this option?	Suitable for all individuals.	Suitable for any couple.
	Useful if you just want to know your carrier status for the three most common genetic conditions.	May be helpful for couples with shared ancestry, from certain ethnic groups or for those that want as much information as

## Key things to know

- You can be a carrier of a genetic condition even if no one in your family has that condition.
- Carrier screening looks for the most common gene changes that cause genetic conditions.
   There may be rarer gene changes that are not identified.
- Most people get reassuring results. This means there is a low chance of having a child with one of these conditions. But low risk does not mean no risk. There is always a small chance of having a child with a genetic condition.
- If you and your partner are found to be carriers for the same condition, our genetic counselling team will guide you through your options including further testing.

Pregnancy screening team: (03) 9936 6402

possible.

### **Blood collection site**

To find a blood collection site please vist https://collection-sites.vcgs.org.au/



Murdoch Children's Research Institute

<sup>\*</sup> From when the laboratory receives your sample(s).
^ In some cases, there may be a Medicare rebate available.
# prepair+ is a package offered by VCGS. Testing is performed by an overseas partner, with additional genetic interpretation, clinical support and genetic counselling provided by VCGS in Melbourne