

# prepair™ fact sheet

## Cystic fibrosis

### What does this result mean?

The results of your test show that you are a carrier of a cystic fibrosis (CF) gene change (or mutation). We all have two copies of the gene associated with CF. Carriers have a change in one of their genes. Carriers are healthy and will not develop symptoms of CF. One in 25 people in the Caucasian population are carriers of CF.

A couple has a chance of having a child with CF if they are both carriers of a CF gene change. If both parents are carriers there is a 1 in 4 chance they will both pass on their CF gene changes to their child. If the child inherits two copies of the CF gene change he/she will have CF.

### What happens next?

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

- Partner receives a low risk result: There is a low risk (approx. 1 in 960) that you and your partner will have a child with CF. 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 child with CF is 1 in 4 for each pregnancy 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.

If you and your partner are both carriers and you are pregnant then you might consider prenatal testing for CF. 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 copies of the CF gene change. 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. CF testing results (often including full chromosome analysis) are ready in approximately 2 weeks. Diagnostic tests carry a small risk of miscarriage.

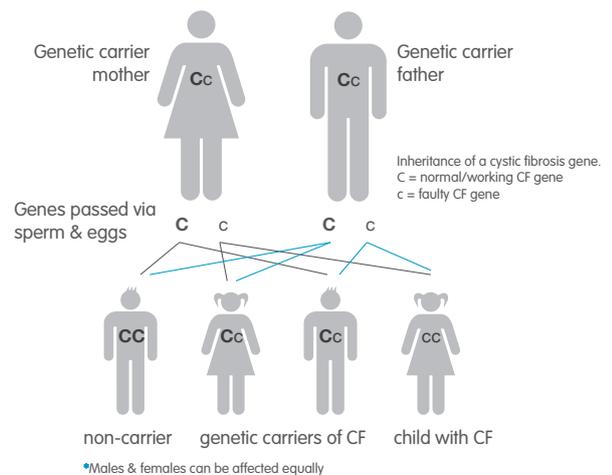
**If your baby has cystic fibrosis:** If cystic fibrosis is detected, you and your partner will be offered genetic counselling to discuss the options available to you. You may need to face the difficult decision of whether to continue your pregnancy.

*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 cystic fibrosis?

Cystic fibrosis is an inherited condition affecting breathing and digestion. CF causes the build-up of thick mucus which traps bacteria, resulting in recurrent infections that damage the lungs. Thick mucus in the gut also makes digestion of food difficult.

People with CF require daily physiotherapy to clear mucus from their lungs, frequent courses of antibiotics to clear lung infections and medication to aid digestion. There is no cure for CF but better treatments are under research and development. People with CF have a reduced life expectancy.



### What does my carrier result mean for my relatives?

CF is an inherited condition which means it is likely that some of your relatives may also be carriers of CF. 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](http://www.genetics.edu.au/Genetics-Services/genetic-counselling-services)

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