

Cystic Fibrosis (CF) Carrier Screening

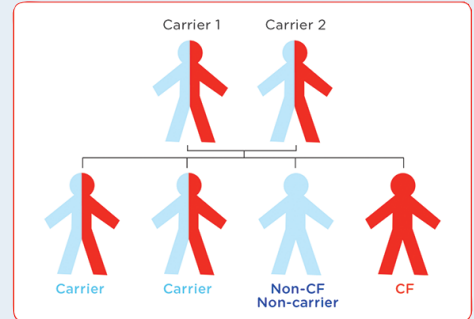
Frequently asked questions

What is a cystic fibrosis (CF) carrier?

- About 1 in 25 people in Australia carry a gene change (or gene mutation) that can cause CF. However, carriers do not have CF and related symptoms.

- Carrier status can be passed from parent to child, but it is only when both parents are carriers that there is a 25% chance with each pregnancy that the child will be born with CF.

- Carrier screening is the only way to find out if you are a CF carrier prior to pregnancy.



What is cystic fibrosis carrier screening?

- CF carrier screening is a genetic test that can detect if people have (or 'carry') changes to the gene that can cause CF. This test can be done on cells taken from a swab inside the cheek or a blood sample.
- There are over 2000 different gene changes that can cause CF. Most laboratories test for the most common 10-40 changes, which covers about 85% of the possible gene changes. This may mean that a very small number of people, who are not identified as a general CF carrier, could still be a carrier for a rare CF-related gene change.
- Important: If you have a family history of CF or know of a relative who is a carrier, try to find out the specific gene change (or mutation) in your family. If you know the gene change in your family ensure your doctor provides the details on the carrier screening request form. This will make sure that the known gene change is included in the screening process.

Should I have carrier screening?

- Making the decision to know your carrier status is a choice only you can make.
- Be sure to ask questions and discuss in detail what the options and potential impacts are with a genetic counsellor or your doctor.

There has never been CF in my family. Could I still have a child with CF?

- Approximately 95% of children born with CF have no family history of CF.
- Cystic fibrosis is a recessive condition. This means that both parents have to be CF carriers for their child to have CF (see diagram).
- As carriers do not have CF, it is possible that there are CF carriers in a family but no-one has ever been diagnosed with CF.
- If you want to understand more about your risks of having a child with CF it is recommended that you talk to a genetic counsellor and consider whether you and/or your partner want to do CF carrier screening.

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Is it possible for someone to have CF even though only one parent is a carrier?

- No: Both parents must be carriers of the gene changes that can cause CF in order for a child to be born with CF.
- If only one parent is a carrier, while their child cannot be born with CF, there is a 50% chance with each pregnancy that their child will also be a CF carrier.

Where can I do CF carrier screening?

- Your General Practitioner (GP) or Obstetrician can request a carrier screen for you.
- Screening is available to anyone in Victoria as long as you have access to a pathology collection point. If you don't have access to a collection point you can request a saliva swab kit directly from Healthscope Pathology.
- Be sure to talk to your health professional about the options for carrier screening. It is possible to have screening for CF alone, or in combination with screening for other conditions. For example Victorian Clinical Genetics Services (VCGS) www.vcgs.org.au offer CF carrier testing in conjunction with carrier testing for Fragile X syndrome and Spinal muscular atrophy (SMA).
- For a list of locations in Victoria, please refer to the contact list <http://www.cysticfibrosis.org.au/vic//info-resources-kit>
- For information on carrier screening services in all other states and territories please go to the NSW Centre for Genetics Education. www.genetics.edu.au/Genetics-Services/genetic-testing-services
- Cystic Fibrosis Victoria (CFV) recommends that you take the time to talk to a genetic counsellor as well. Genetic counsellors are available all over Victoria and can also provide information to you over the phone.
- VCGS, Austin Hospital, and Monash Health all have genetic counselling services that you can contact by phone to get further information about carrier screening. Click here for the contact list <http://www.cysticfibrosis.org.au/vic//info-resources-kit>
- Healthscope Pathology also has genetic counsellors available should your health professional refer you to them.

Is newborn screening the same as carrier screening?

- Newborn screening is not the same as carrier screening.
- Newborn screening is conducted on all babies born in Australia to identify certain diseases, including CF.
- Only a very small proportion of all CF carriers are detected by newborn screening. If you do not receive results from the newborn screen it only means that your child is very unlikely to have CF, but they may still be a CF carrier.
- For further information on newborn screening visit the VCGS website www.vcgs.org.au

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Should siblings and family members of a person with CF be screened?

- It is important that siblings of children with CF and their extended families know they have an increased risk of being CF carriers. However, it is also important that they make an informed choice on whether they want to be screened.

- Currently the Human Genetics Society of Australasia (HGSA) guidelines for genetic counsellors recommend that carrier screening only be conducted on children if there is an immediate health management need for the genetic information.

www.hgsa.org.au/resources/hgsa-policies-and-position-statements

Can doctors share information about my carrier status with others?

- In Victoria doctors are required by law to have your permission before informing others of your medical information. This means that in Victoria a doctor cannot inform your family of your carrier status without your permission.

- Please note each state and territory has different regulations around confidentiality.

Is carrier screening covered by Medicare?

- CF carrier screening is not covered by Medicare so there is a cost.
- Different services have different products and prices. You should contact each service to find out their current prices but expect to pay no less than \$150 to do a CF carrier screening test.

What are my reproductive options if my partner and I are both carriers?

Only you can decide the best reproductive choice for you. Some of the reproductive options include:

- Natural pregnancy with no testing accepting the 25% risk that your child may have CF.
- Natural pregnancy followed by pre-natal testing of the foetus to decide to continue or terminate the pregnancy.
- IVF with pre-implantation genetic diagnosis to detect which embryos do not have CF prior to implantation.
- IVF with non-CF carrier donor eggs or sperm.
- Adoption/Fostering.
- Choosing to not have children.

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Steps to having CF carrier screening:

Step 1 - Get Informed: Gather as much information to help you make a decision about how knowing your CF carrier status will inform your reproductive and life choices. CFV recommends talking to a genetic counsellor at a clinical genetics service or your health professional as part of this process.

Step 2 - Contact a service: Contact a carrier screening service directly to ask about their pathology referral/collection process. Refer to the contact list on the CFV website <http://www.cysticfibrosis.org.au/vic//info-resources-kit>

Step 3 - Provide your sample: If you are doing a blood sample test go to the pathology collection point that is most convenient to you. If you have requested a check/saliva swab kit follow the instructions and return your sample to the address provided

Step 4 - Getting the results: Different services have different ways of informing you of the results. Some will contact you directly through a genetic counsellor others will send the results to your health professional. It is recommended you are clear about how you will receive your result before sending your sample for CF carrier screening.

Step 5 - Understanding the results: CFV recommends talking to a genetic counsellor about your results. Remember the results will tell you if you have a confirmed gene change and are a carrier, or give you a negative result. A negative result means you have less risk of being a carrier, but that there is still a possibility you could be a carrier of a rare or as yet unidentified CF-causing gene change.

Step 6 - Informing others: Even if you are test negative, your relatives could still be carriers and may not have heard of carrier screening. If you are found to be a carrier, your siblings have a 50% chance of also being carriers and may wish to know this increased risk as they plan their own family. If you find starting the conversation about carrier status difficult genetic counsellors can offer anonymous options to help you inform your relatives. You may also like to refer to the information on the CFV website <http://www.cysticfibrosis.org.au/vic//info-resources-kit>

Useful weblinks - CF carrier status and screening

- Knowing Your Options - CF carrier screening brochure: <http://www.cysticfibrosis.org.au/vic/carrier-screening>
- Cystic Fibrosis Victoria: <http://www.cysticfibrosis.org.au/vic>
- Genetic testing services in Australia: <http://www.genetics.edu.au/Genetics-Services/genetic-testing-services>
- Thinking about collecting family health history: <https://www.nhmrc.gov.au/your-health/egenetics/health-practitioners/genetics-family-medicine-australian-handbook-general-prac>
- Victorian Clinical Genetics Service: www.vcgs.org.au
- Genetic Support Network Victoria: www.gsnv.org.au
- Human Genetics Society of Australasia: www.hgsa.org.au

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- Archibald A D, Massie J, Smith M J, Dalton D G, Du Sart D, Amor D J. *Population-based genetic carrier screening for cystic fibrosis in Victoria*. MJA 2014; 200 (4): 205-6
- Cunningham F, Lewis S, Curnow L, Glazner J, Massie J. *Respiratory physicians and clinic coordinators' attitudes to population-based cystic fibrosis carrier screening*. Journal of Cystic Fibrosis 2014; 13: 99-105
- Human Genetics Society of Australasia. *Population Based Carrier Screening for Cystic Fibrosis Position Paper (2013)* <http://www.hgsa.org.au/>
- NSW Government Centre for Genetics Education <http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/cystic-fibrosis>

The content of this information sheet was checked by representatives of the Victorian Clinical Genetic Services (VCGS), RCH Respiratory unit and Genetic Support Network of Victoria (GSNV).

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