Cystic Fibrosis (CF) Carrier Screening

Discussing cystic fibrosis carrier testing with adult family members

When an inherited condition is diagnosed in a family, people often have questions about how and when to talk about it with other members of their family. If a family member has cystic fibrosis (CF) or is a CF carrier, their relatives have an increased chance of also being CF carriers.

Discussing CF with family members, in particular whether relatives are carriers, gives people the opportunity to decide for themselves whether or not to have genetic carrier testing. Here are some points that may be considered when thinking about telling your relatives about CF and genetic carrier testing.

What is the chance that my relatives could be carriers?

If somebody in your family has CF or is a carrier, other relatives will be carriers. The chance of relatives being CF carriers depends on their relationship to the person who is a carrier or who has CF.

The chance of anybody in the population being a carrier of CF (even if they have no family history of CF) is 1 in 25.

For a person who has CF the chances of their relatives being a carrier are:

Relative of person with CF	Chance of being a carrier
Parent	100%
Siblings (who do not have CF)	66%
Aunts and uncles	50%
Grandparents	50%
First cousins	25%

For a person who is a carrier of CF, the chances of a relative being a carrier are:

Relative of person who is a carrier CF	Chance of being a carrier
Parent	50%
Siblings	50%
Aunts and uncles	25%
Grandparents	25%
First cousins	13%

Who should be informed?

• **Relatives thinking of having children:** Carrier testing information can be helpful to all of your relatives, but especially those who may be thinking of having children.

• **Family members with children:** It may also be important to talk about being a carrier with family members who already have children. Although babies are screened for CF at birth, newborn screening is designed to diagnose babies with CF and will not identify the majority of carriers. Additionally, grandparents of a child with CF may want to consider carrier testing. This can determine which side of the family the gene change is on and helps other relatives know their chance of being a CF carrier.

• **Different people at different times:** It is important to remember that relatives who may not have been thinking of having children when their family member was diagnosed with CF or identified as a carrier of CF could have altered their plans over the years and the information may become more relevant to them. Talking about carrier testing with relatives may need to be an ongoing conversation that is re-visited to meet the changing life stages of relatives.

> *"...[we were] given the usual handouts...and we just photocopied the whole lot and sent it out to extended family...because a lot of them, like their children are my age now and started their own families."*
>
> **Mother of child with CF**

What do your relatives need to know?

• **What CF is:** Giving your relatives some information about CF can help them to understand and process the information.

• **There is a chance they may be a carrier:** Explain that there is an increased chance that they could be a carrier of CF compared to anyone in the population, and that for couples in which both partners are carriers, they have a 25% chance of having a child with CF.

• **They can choose to know or not know:** Finding out your carrier status is a personal choice. The aim is for everyone to know their options and decide for themselves. Carrier testing is available to anyone.

• **Carriers of CF are healthy:** It is important that your relatives are made aware that even if they are carriers of the CF gene, there are no known implications for their own health.

• **Genetic counselling is available:** Let your relative know that they can speak with a genetic counsellor if they are thinking about testing, or just want more information.

• **The specific CF gene change your family has:** If your relatives would like to have genetic testing done to find out if they are carriers of CF, it is helpful to know the name of the CF gene change in the family, so it can be tested for specifically. They will need to let the person who is ordering the testing (doctor or genetic counsellor) know this information.

When to start the conversation?

For a lot of families, the first discussion about genetic information occurs simultaneously with the sharing of the diagnosis and the details of CF. Research shows that in the immediate period following a diagnosis, relatives seek testing.

How can I start the conversation?

• **When the opportunity arises:** When the topic of CF comes up in conversation with relatives, such as talking about current health status or planned hospital admissions, a discussion of the genetic aspect of CF can be initiated. This can be a very natural and supportive way to talk to relatives about the genetic information, however a consequence is that only relatives with whom you have a close social relationship may be informed, while others may not become aware of the information.

• **Deliberately contacting relatives:** Some relatives may be more distant, whether this is because they live far away or there is limited social contact. Some families do this by sending letters/emails to relatives they have contact with and asking that it be passed on through family networks. A genetic counsellor can assist you with writing a letter to relatives and making sure it contains all the relevant genetic and contact information.

• **Nominating a family communicator:** In some families, the most logical person to pass on the information to other relatives is the person who has the greatest contact with the wider family. You may wish to ask a relative, such as your parent, to take on the role of communicator when informing your relatives.

> *"I didn't really speak to my aunties and uncles in regard to the testing side of it, I have spoken to them of the illness but I don't think I was the one that discussed the testing with them. Not for any reason, it would have just been that mum was talking to her brothers and sisters"*
>
> **Person with CF**

Further Info:

If you need more information on cystic fibrosis and carrier testing, you can find more information on the following websites:

www.cfscreening.com.au
www.cysticfibrosis.org.au/vic/info-resources-kit
www.gsnv.org.au/individuals-and-families/planning-a-pregnancy.aspx
www.vcgs.org.au/

References:

VCGS Cystic Fibrosis Carrier Fact Sheet – talking to your family about CF genetic testing

'Cystic fibrosis cascade carrier testing in Victoria, Australia' – Thesis, Doctor of Philosophy, The University of Melbourne, 2010. Belinda McClaren

'Cascade carrier testing after a child is diagnosed with cystic fibrosis through newborn screening: investigating why most relatives do not have testing.' 2013. Genetics in Medicine 15(7), pg 533-540. Belinda McClaren, MaryAnne Aitken, John Massie, David Amor, Obioha Ukoumunne, Sylvia Metcalfe.

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).