

Screening Relatives of People with Cystic Fibrosis

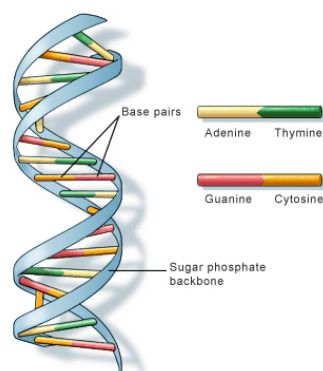
By Bob Williamson

Every doctor and scientist who works with people who live with cystic fibrosis is familiar with many parents who say: "How can I have a child with CF, when there is no CF in my family? Didn't you say it is a genetic disease?"

This little puzzle is easily explained. Every person has two copies of each of the 30,000 or so human genes, one inherited from mum and one from dad. For the CF gene (named CFTR), one gene in twenty does not function properly because it is mutated. CF is "recessive" genetically. People who have one normal copy of the CFTR gene are completely healthy, even though they carry one copy of the wonky CFTR gene that does not work properly. (These people are called "carriers" of CF.) It is only if you have two copies of the mutated gene, one from dad and one from mum, that you have CF. In Australia, where I live, and in the United States and Britain and France and many "European" countries, about one person in 20 (in the "white" population) is a carrier. Australia has 20 million people, so there are about one million carriers.

You can only have a child with CF if both parents are carriers, and the chance of that happening is 1/400. There is a one in four chance that a baby will get the CF mutation from both parents, making the total chance 1/1600 or so. Since there are about 160,000 new births each year in Australia, that means there are about 100 children born each year with cystic fibrosis. One million CF carriers, but only one hundred affected children, because most carriers marry someone who is not a carrier, and never know they "carry" the CF gene.

It is possible to do a lab test on DNA from a teaspoon of blood or (at some centres) from a cheek scrape (just like in the television show "CSI") and determine if someone is a carrier. In many countries, this is on offer (for a fee), and in some places it is more widely available to any couple. However, it is not clear if this is the most effective way to offer screening. First, only one couple in about 400 will be identified. Second, since screening is usually performed during pregnancy when there is already a fetus on board, the "at risk couple" will have a roller-coaster ride of information and ethical issues to deal with.



The double helix: the structure of DNA



Professor Bob Williamson

The group for whom screening is highly recommended, and usually offered free of charge, are relatives of those who are living with cystic fibrosis. Each is at an increased risk of being a carrier. Unaffected brothers or sisters of those with CF have a two in three chance of being a carrier, and first cousins have greater than a one in four chance. If a relative is shown to be a carrier, their partner can be tested. If a relative would rather not be tested, that is fine too; no one should

One of the best aspects of "cascade testing", as testing of relatives is called, is that people can find out whether they are carriers before they are pregnant. This means they have more options. Some couples who would not have an abortion at ten weeks of a pregnancy nonetheless would choose to avoid having a child with CF by pre-implantation diagnosis (using IVF), or adoption. Others might choose not to have children, while some would "take a chance".

Those of you who have read my articles before know that I believe in offering knowledge to people, because I think that knowledge empowers people to take control of their own lives and choices. The choices will be yours, but the knowledge gives you the chance to think through what is right for you and your partner and family.

Bob Williamson AO, FRS is a Professor of Medical Genetics at the University of Melbourne, Australia. He Chairs the National Committee for Medicine of the Australian Academy of Science, is Co-Chair of the OECD Committee on Regulation of Human Molecular Genetic Testing, and has worked extensively for the World Health Organisation. Since he retired he has become active in CF charities in Australia. He is a Fellow of the Australian Academy of Science, a Fellow of the Royal Society, and an Officer of the Order of Australia.