



Each of us carries two copies of every gene, one inherited from our mother and one inherited from our father. The following information explains more about genetic testing for CF, where the test takes place and how to go about finding out if you carry an altered CF gene.

Approximately 1 in 19 Irish people are said to 'carry' one copy of the altered gene that causes Cystic Fibrosis. A person who carries CF has one copy of the altered CF gene and one copy of the normal CF gene, which compensates for the loss of function of the altered gene – a person who carries CF therefore does not have cystic fibrosis or any of its disease symptoms.

To be born with the disease a child must inherit 2 copies of the altered CF gene – one from each parent. If both parents 'carry' the faulty or altered gene, each child would have a:

- 25% chance of being born with Cystic Fibrosis
- 50% chance of being a healthy and carrying the altered gene, but not having the disease
- 25% chance of not having Cystic Fibrosis and not carrying of the altered gene

If you have a relative who has CF or carries an altered CF gene, your chances of also carrying a CF gene alteration are greater because of your family history.



What does the test involve?

Genetic testing for CF involves taking a blood sample of the individual and examining the DNA for common CF gene alterations (mutations). More than 1,500 CF alterations have been identified to date. The 39 most common Irish mutations are screened for during carrier status determination. These account for 93.5% of Irish CF mutations. If you have a relative who has CF or carries an altered CF gene it really helps if this information is included along with the blood sample as the lab will be better able to interpret the results.

Where does the test take place?

Testing is available through the Molecular Genetics Department at the National Centre for Medical Genetics, Our Lady's Children's Hospital Crumlin, Dublin 12.

Note: Carrier testing is available to adults over the age of 16 where there is a family history of CF, or where a family member/partner has been found to carry a CF mutation.

How do I arrange to have the test?

Your GP can take a blood sample, fill in the relevant documentation and send everything directly to the NCMG for testing. Essential information and sample/packaging specifications can be found on the NCMG website. The results will be sent back to your GP.

If you would like to see a Genetic Counsellor prior to carrier testing your GP can send a written referral letter to the NCMG. If you would like to see a Genetic Counsellor prior to carrier testing and your relative has been seen previously in Genetics you can contact the NCMG directly to make an appointment quoting the family reference number. Genetics clinics are held in Dublin, Cork, Limerick and Galway.



Will I have access to genetic counselling once I get the results?

If, after receiving your test results, you feel you would benefit from genetic counselling this can be arranged by asking your GP to send a written letter of referral to the NCMG.

How long do I have to wait to receive results?

Waiting times vary, but a general estimate is 6-8 weeks after the blood sample is taken. If you or your partner is pregnant it is important that this information is noted on the blood form as the sample(s) will be prioritised and results are usually available within 2 weeks.

How much does the test cost?

GP fees will apply for arranging the blood test but molecular genetic testing at NCMG and any genetic counselling you may have is a public service and therefore free of charge.

Further Information:

- National Centre for Medical Genetics: www.genetics.ie/documents/cystic-fibrosis.pdf
Tel: 01 4096739; Fax: 01 456 0953
- Standard referral information/NCMG request form: www.genetics.ie/pir/2006_NCMG_Referral_Form.pdf
- Sample/Patient identification policy: www.genetics.ie/pir/SampleIdentificationPolicyWeb.pdf
- Packaging of specimens for transport: www.genetics.ie/pir/sending_samples.pdf
- Cystic Fibrosis PIR form: www.genetics.ie/pir/CF_pir.pdf
- Genetics Factsheet, CFAI
- Cystic Fibrosis Foundation – Genetic Carrier Testing: www.cff.org/AboutCF/Testing/GeneticCarrierTest/

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