



The Heart Rhythm Charity

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A Guide to Genetic Testing and your Genetic Counselling appointment

What is Genetic Testing?

A genetic test is a type of medical test that is used to try and identify the change in a gene that can cause an Inherited Heart Condition (IHC). Within the genes that are associated with IHCs, there are a great number of gene changes that can cause the condition.

Genetic testing, may be either 'diagnostic' or 'predictive', depending on whether you have already been diagnosed with an IHC or not.

Diagnostic Genetic Testing

Diagnostic genetic testing can be offered to someone who has been diagnosed with an IHC. If a change in a gene is found, it may be that this is the genetic cause for their IHC.

The aim of diagnostic genetic testing is to try and find the genetic cause of an IHC, not to actually establish a diagnosis.

There are several possible results from diagnostic genetic testing:

- ♥ The change in a gene is found. This means a genetic cause of your IHC has been identified.

- ♥ The causative gene change has not been found. This does not mean you do not have the IHC you were clinically diagnosed with, just that a genetic cause has not been identified.
- ♥ A gene change has been found but it is not yet certain whether this is what is causing your IHC.

Predictive Genetic Testing

If an IHC causing gene change is found in a family member who has already been diagnosed with an IHC, genetic testing can be offered to other members of their family who may be at risk of this same IHC.

The aim of predictive testing is to predict your future risk of developing the IHC that is already known to run in your family. There are only two possible results from predictive genetic testing. Either you have the gene change already found in one or more of your relatives and therefore you are at increased risk of developing that particular IHC; or you do not have this same gene change and are at no greater risk of developing this IHC than anyone in the general population. In this instance genetic testing will only be able to tell you whether you are at an increased risk of developing an IHC. It will not be able to predict if you will get symptoms, when you will get them and how severe they might be.

For further information contact Arrhythmia Alliance



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President: Prof A John Camm Trustees: Dr Derek Connelly,
Mr Nigel Farrell, Dr Adam Fitzpatrick, Mrs Trudie Lobban





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A genetic counselling appointment will typically last about one hour and can be used to discuss a wide variety of issues, as well as any other questions you may have. It is a chance to learn about the genetics behind your IHC and to discuss what having the condition may mean for you and your family. It can also provide a space to discuss the pros and cons of genetic testing in reference to your own personal situation, so you can work out whether having a genetic test really is the right choice for you.

How is the Genetic test done?

The genetic test itself involves you giving a blood sample or cheek swab. Scientists in a specialist genetics laboratory will look at the genes in your blood sample, or cheek cells, to see if they can find the change that is causing your IHC. Results will be sent back to the genetic counsellor or doctor who you saw originally and they will then let you know by whatever means you arranged e.g. another appointment, telephone, letter or email. When your genetic counsellor or doctor informs you of your results, you can have another discussion as to what are the appropriate next steps to take in light of your results.

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