Genetic Testing for Inherited Heart Conditions

Arrhythmia Alliance

The Heart Rhythm Charity

Promoting better understanding, diagnosis, treatment and quality of life for individuals with cardiac arrhythmias

A Guide to Genetic Testing and your Genetic Counselling appointment

www.heartrhythmcharity.org.uk

A Guide to Genetic Testing and what to expect from your appointment

This information leaflet has been produced for people who have been diagnosed with an Inherited Heart Condition and their families. Its aim is to outline the process and implications of genetic testing and to give you an idea of what to expect from your genetic counselling appointment.

Arrhythmia Alliance (**A-A**) is a coalition of charities, patient groups, patients, carers, medical groups and allied professionals.

These groups remain independent, however, work together under the **A-A** umbrella to promote timely and effective diagnosis and treatment of arrhythmias.

A-A supports and promotes the aims and objectives of the individual groups.

Contents

Arrhythmia Alliance patient booklets are reviewed annually.
This booklet will be next updated March 2010,
if you have any comments or suggestions
please contact A-A.

Glossary of terms

IHCs
Inherited Heart Conditions

Hypertrophic Cardiomyopathy
A disease of the heart muscle

Long QT
A disturbance of the heart's electrical system

ECG
An electrocardiogram (ECG) records the electrical activity within the heart. It is a simple procedure which involves applying small stickers to the patients' arms, legs and chest. The patient is then connected to an ECG machine via leads that are attached to the stickers and then connected to the machine.

What is an Inherited Heart Condition?

Inherited Heart Conditions (IHCs), for example Hypertrophic Cardiomyopathy and Long QT Syndrome, are conditions that can be passed down through families via your genes. Genes act as instructions telling our bodies how to grow, develop and function. There are many genes known to be involved with IHCs but what actually causes someone to be affected by or be at risk of an IHC is a change, or 'spelling mistake' in one of those genes.

What is Genetic Testing?

A genetic test is a type of medical test that is used to try and identify the change, or 'spelling mistake' that can cause an IHC. For example:

A correctly 'spelt' gene: THE CAT SAT MAT

A gene with a 'spelling mistake': THE CAT TSA TMA T

Within the genes that are associated with IHCs, there are a great number of gene changes that can cause the condition. The likelihood of finding the spelling mistake that causes the IHC within one of these genes varies from one condition to the next.

Are there different types of Genetic Tests?

Genetic testing may be called either 'diagnostic' or 'predictive', depending on whether you have already been diagnosed with an IHC or not. The process involved and implications to consider are quite different for each type of test.

Diagnostic Genetic Testing

Diagnostic genetic testing can be offered to someone who has been diagnosed with an IHC by their doctor based on the results of clinical tests (like ECG and Echo) and examination. If a spelling mistake (like the one illustrated above) is found in one of their genes, we may then be able to say this is the genetic cause for their IHC. This type of test is not available to someone who does not have a clinical diagnosis of an IHC. An unaffected person may be able to have predictive testing (see section below) if a diagnostic test can be done in an affected relative first.

The aim of diagnostic genetic testing is to try and find the genetic cause of your IHC, not to actually establish your diagnosis. The results of the genetic test may not change anything for you in terms of your diagnosis and management as decisions related to these issues are usually based on the results of clinical tests and examination. However, if the causative spelling mistake is found, it can be a very useful tool for finding out who else in your family may be at risk of developing the same IHC as you.

There are several possible results from 'diagnostic' genetic testing:

1) The spelling mistake is found. This means we have been able to identify the genetic cause of your IHC. For many IHCs this will not necessarily change your management, although sometimes, for example in Long QT Syndrome, it can direct lifestyle advice and treatment. However it does mean that we can offer predictive genetic testing to other at-risk family members.

2) 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 we have not been able to identify the genetic cause for it. We will not now be able to offer predictive genetic testing to other at risk family members.

3) A gene change has been found but it is not yet certain whether this is what is causing your IHC. In this instance genetic testing may be offered to other family members who are also affected by the same IHC to see if they have this same gene change as well. If they do, we can be more certain it is the causative gene change in your family. If they do not, or if there are no other members of your family also affected by the IHC, then we may not be able to offer you anything further at this time.

If you would like to find out more about genetic testing and how you can get it, you should ask your GP or cardiologist to refer you to a specialist cardiac genetics clinic or your local clinical genetics department. There you will see either a genetic counsellor or a genetic doctor to discuss the possibility of genetic testing in greater depth. This may be called a 'Genetic Counselling' appointment.

Predictive Genetic testing

If an IHC causing gene change is found in a family member who has already been diagnosed with an IHC (see Diagnostic testing section), we can then offer genetic testing to other members of their family who may be at risk of this same IHC. In the first instance this would usually mean offering testing to first degree relatives such as parents, siblings and children. If they choose to have predictive testing and the causative gene change is also found in them, they will be at increased risk of developing the IHC and will need to be assessed and followed up by a cardiologist. If a family member has not inherited the causative gene change then they, and their children, will not be at any increased risk of developing the IHC and so should not need further follow up. This approach of finding out which branches of a family may have inherited the IHC may also be called 'cascade screening'.

The aim of predictive testing is to predict your future risk of developing the IHC that is already known to run in your family. From a medical point of view, we hope that by identifying those family members who are at risk of these conditions, we can reduce your risk of ill health and sudden death through targeted screening, surveillance and appropriate medical and/or surgical therapy. 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. However, before you decide to go ahead with predictive genetic testing, a genetic counselling appointment can help you think about how the results of this test may affect you and whether it really is the right choice for you at this time in your life.

If you would like to find out more about the process and implications of predictive genetic testing, you should ask your GP to refer you to either a specialist cardiac genetics clinic or your local clinical genetics department.

What is Genetic Counselling?

Your genetic counselling appointment 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. You may decide you do not want to have genetic testing after all. This will not affect your cardiac care in any way and at-risk family members can still have their hearts checked by clinical tests (e.g. ECG and Echocardiogram etc) if they so wish.

How is the Genetic test done?

The genetic test itself involves you giving a blood sample or cheek swab. This will then be sent away to a specialist Genetics Laboratory. Scientists there 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.
They will only look in the genes they know are involved in your IHC, not in any other genes, so you will not be told you are also at increased risk of other diseases like bowel cancer for example.

The results for diagnostic genetic tests can take several months (sometimes longer) to come through as looking for genetic spelling mistakes is complex and often time-consuming. 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.

The results for predictive genetic tests take approximately two to four weeks to come back. 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.

Where can I get more information?

If you would like more information about testing then please ask your GP to refer you to either a specialist cardiac genetics clinic or your local clinical genetics department.

Useful websites

A list of useful sites can be found at:- www.heartrhythmcharity.org.uk This list is not exhaustive and it is constantly evolving. If we have excluded anyone, please accept our sincerest apologies and be assured that as soon as the matter is brought to the attention of the Arrhythmia Alliance, we will quickly act to ensure maximum inclusiveness in our endeavours.

If you wish to contact us direct please phone on 01789 450 787 or email info@heartrhythmcharity.org.uk

Further reading

The following list of Arrhythmia Alliance Patient booklets are available to download from our website or to order please call 01789 450 787.

- Arrhythmia Checklist - Could your heart rhythm problem be dangerous?
- Atrial Fibrillation (AF)
- AF Checklist
- Blackouts Checklist
- Bradycardia (Slow Heart Rhythm)
- CRT/ICD
- Catheter Ablation
- Catheter Ablation for Atrial Fibrillation
- Drug Treatment for Heart Rhythm Disorders (Arrhythmias)
- Electrophysiology Studies
- Exercising with an ICD
- FAQs
- Highlighting the Work of Arrhythmia Alliance
- ICD
- Implantable Loop Recorder
- Long QT Syndome
- National Service Framework Chapter 8
- New Genetics
- CRT/Pacemaker
- Pacemaker
- Palpitation Checklist
- Remote Monitoring for ICDs
- Sudden Cardiac Arrest
- Supraventricular Tachycardia (SVT)
- Tachycardia (Fast Heart Rhythm)
- Testing Using Drug Injections to Investigate the Possibility of a Risk of Sudden Cardiac Death
- Tilt Test

Please feel free to discuss any concerns with your doctor, physiologist or specialist nurse, at any time.

Please help us to improve services for all those affected by arrhythmias and to save lives by making a donation today. Please complete the donation form below and return to P.O Box 3697, Stratford upon Avon, CV37 8YL or visit www.heartrhythmcharity.org.uk and click the donate icon.

Membership is free to individuals, however, if you would like to make a DONATION please complete and return.

I would like to make a donation to A-A and enclose:	£
I have made a donation to A-A via PAYPAL at www.arrhythmiaalliance.org.uk to the sum of:	£
I have arranged a standing order from my Bank/ Building Society Account to A-A, (min amount £10p.a.)	£
Please tick here if you agree to Gift Aid your subscription/donation	☐ Tick here

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Standing Order Authority

My Bank:

Bank Address:

Please Pay: A-A, Account: 02685818 Sort Code: 30-98-26, Lloyds TSB Plc, 22 Bridge St, Stratford upon Avon, CV37 6AG

The Sum of £/€/$:	On (1st Date): / / 20.....
And after this, every: Month / Year (delete)	Account No.:
Sort Code:	Signature:
Date:	Please hand this form in to your Bank

Credit Card Payment

Card Type:	Expiry Date:
Card Number:	Amount of £/€/$:
Name on Card:	Address:

Arrhythmia Alliance

The Heart Rhythm Charity

Executive Committee

This booklets has been written with the help of Miss Stephanie Oates and Colleagues from the Oxford Cardiomyopathy and Inherited Heart Disease Clinic.

endorsed by DH *Department of Health*

Arrhythmia Alliance
PO Box 3697 Stratford upon Avon
Warwickshire CV37 8YL
Tel: 01789 450 787
e-mail: info@heartrhythmcharity.org.uk
www.heartrhythmcharity.org.uk

Please remember these are general guidelines and individuals should always discuss their condition with their own doctor.

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