Arrhythmia Alliance

The Heart Rhythm Charity

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

Genetic Testing for Inherited Heart Conditions

www.heartrhythmcharity.org.uk

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A Guide to Genetic Testing for Inherited Heart Conditions

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 January 2012,
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 in 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. A change, or 'spelling mistake,' in one of these genes can cause someone to be affected by, or be at risk of, an IHC. The way these 'spelling mistakes' are inherited can vary but there is usually a 50:50 or 1 in 2 chance that a close blood relative (child, parent, sister or brother) also has the spelling mistake. Most people with IHCs do not suffer troublesome ongoing symptoms. However a small number of people are at risk of dangerous heart problems and it is important to have regular heart checks, as many of these problems are treatable or preventable.

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 large number of gene changes that can cause the condition. It is not always possible, using current technology, to find the spelling mistake. If this is the case, our practice is to store your sample as we may be able to do additional testing in the future. In fact, even if we do find the causative spelling mistake, we may still store your sample for further testing if and when our knowledge increases or new technology becomes available.

Your written consent will be asked for before you give a sample and we will ask for your permission to share any results with your family members if they attend for testing in the future.

Are there different types of Genetic Tests?

There are two different types of genetic tests: 'genetic screening' and 'pre-symptomatic' or 'familial' testing. The first affected person in the family to come forward for testing will be offered a genetic screen; if a causative spelling mistake is found, then their relatives may be offered a familial test. The process involved and implications to consider are quite different for each type of test.

Genetic Screening

Genetic screening can be offered to someone who has been diagnosed with an IHC by their specialist heart doctor (cardiologist) based on the results of clinical tests (like ECG and Echocardiography) and examination.

The aim of genetic screening is to try and identify the genetic cause of your IHC. The results of a genetic screen may not change anything for you in terms of your diagnosis and management - these decisions 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 and efficient 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 'genetic screening':

1) A spelling mistake is found that we know is the genetic cause of your IHC. This will allow us to offer pre-symptomatic genetic testing to other at-risk family members.

2) The responsible spelling mistake has not been found. This does not mean you do not have the IHC you were clinically diagnosed with, or that it is not an inherited condition, just that we have not been able to identify the precise genetic cause of it in you. This may be because the testing procedure was unable to identify your particular genetic alteration or because the particular genetic alteration is in genes the laboratory does not test or have not been discovered yet. In this situation, we cannot offer pre-symptomatic genetic testing to other family members. However, our usual practice is to store your sample, in case we may be able to do additional testing in the future.

3) A gene change has been found but it is not yet certain whether this is what is causing your IHC. In this situation we might want to use the genetic test as well as heart checks, such as echocardiography and ECG, in your family members if they wish, until we learn more about the gene change.

Pre-symptomatic Genetic Testing

If an IHC causing gene change is found in a family member who has already been diagnosed with an IHC (see genetic screening section), we can then offer pre-symptomatic genetic testing to blood relatives who are not already known to have the IHC. In the first instance this would usually mean offering testing to close relatives such as parents, brothers, sisters and children, depending on their age.

If a relative chooses to have pre-symptomatic testing, there are two possible outcomes:

1) The causative gene change is found. That person is at increased risk of developing the IHC and should have their heart checked by a cardiologist. If they have children, they will also be at risk of the familial IHC and we would therefore recommend genetic testing and/or screening, depending on their age.

2) The causative gene change is not found. That person is not at any increased risk of developing the IHC and does not need heart checks. If they have children, then they do not need genetic testing or heart checks either.

This approach of finding out who else in the family may have inherited the IHC is sometimes called 'cascade screening'.

The aim of pre-symptomatic testing is to predict someone's future risk of developing the IHC that is known to run in the family. By identifying family members who are at risk, we hope to reduce their risk of ill health and dangerous heart problems through screening, surveillance and appropriate medical and/or surgical therapy.

If you would like to find out more about genetic testing, you should ask your GP or cardiologist to refer you to a specialist cardiac genetics clinic or your local clinical genetics department. You should be offered a 'Genetic Counselling' appointment with a specialist health professional (usually a genetic counsellor or doctor) to discuss genetic testing in greater depth.

If for any reason, you do not want to have pre-symptomatic genetic testing you can still have clinical tests such as ECG and Echocardiography. Your GP should refer you to a specialist cardiac genetics clinic for this.

What is Genetic Counselling?

Your genetic counselling appointment is a chance to learn more about the genetic basis of your IHC and to discuss what having the condition may mean for you and your family. You will be able 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.

Genetic counselling has an especially important role when you are considering pre-symptomatic genetic testing. Discovering you are at increased risk of an IHC, when you feel well can have a significant impact on your life, and we know that people can react to this in different ways. It is worth thinking about your motivation for having genetic testing and finding out what genetic testing can actually tell you. For example, pre-symptomatic genetic testing will only be able to tell you whether you are at an increased risk of developing the IHC. It will not be able to tell you if you will ever develop symptoms, when you will develop them and how severe they may be.

If your pre-symptomatic genetic test is positive, an outpatient appointment with a heart specialist (cardiologist or specialist nurse) is recommended and this can have implications for insurance, exercise, lifestyle and career choice. So it is worth thinking about how it may affect you and your lifestyle, before and during your appointment.

A genetic counselling appointment will typically last about one hour and can be used to discuss a variety of issues, as mentioned above, as well as any other questions you may have. The genetic counsellor or doctor will draw a family tree and discuss any health issues in the family. You should therefore come prepared with as much relevant personal and family information (including documents where available) as possible.

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 sample to see if they can find a change or 'spelling mistake' that might be responsible for causing your IHC. They will only look in the genes they know are involved in your IHC, not in any other genes, so it will not be possible to tell whether you are also at increased risk of other inherited conditions.

The results of genetic screening can take several months (sometimes longer) to come through as this is a complex process. The results for pre-symptomatic genetic tests take approximately two to four weeks to come back. Results will be sent back to the genetic counsellor or doctor 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 and ask any more questions that you may have.

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, a specialist cardiologist 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 +44 (0) 1789 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 +44 (0) 1789 450 787.

- Arrhythmia Checklist - Could your heart rhythm problem be dangerous?
- Atrial Fibrillation (AF)
- AF Checklist
- Blackouts Checklist
- Bradycardia (Slow Heart Rhythm)
- CRT/ICD
- CRT Patient Information
- Catheter Ablation
- Drug Treatment for Heart Rhythm Disorders (Arrhythmias)
- Electrophysiology Studies
- Exercising with an ICD
- FAQs
- Genetic Testing for Inherited Heart Conditions
- Highlighting the Work of Arrhythmia Alliance
- ICD
- Implantable Device Recall
- Implantable Loop Recorder
- Long QT Syndrome
- National Service Framework Chapter 8
- CRT/Pacemaker
- Pacemaker
- Palpitation Checklist
- Remote Monitoring for ICDs
- Sudden Cardiac Arrest
- Supraventricular Tachycardia (SVT)
- Tachycardia (Fast Heart Rhythm)

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 PO 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

Gift Aid

Name of taxpayer:.....

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Please allow Arrhythmia Alliance to claim an extra 28p for every £1 you donate at no cost to you. I want Arrhythmia Alliance to treat all donations I have made since 6 April 2000, and all donations I make from the date of this declaration until I notify you otherwise, as Gift Aid donations. I currently pay an amount of income tax and/or capital gains tax at least equal to the tax that Arrhythmia Alliance reclaims on my donations in the tax year. I may cancel this declaration at any time by notifying A-A. I will notify A-A if I change my address. Please note full details of Gift Aid tax relief are available from your local tax office in leaflet IR 65. If you pay tax at a higher rate you can claim further tax relief in your Self-Assessment tax return.

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

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

Arrhythmia Alliance
The Heart Rhythm Charity

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

Published 2005 revised January 2011