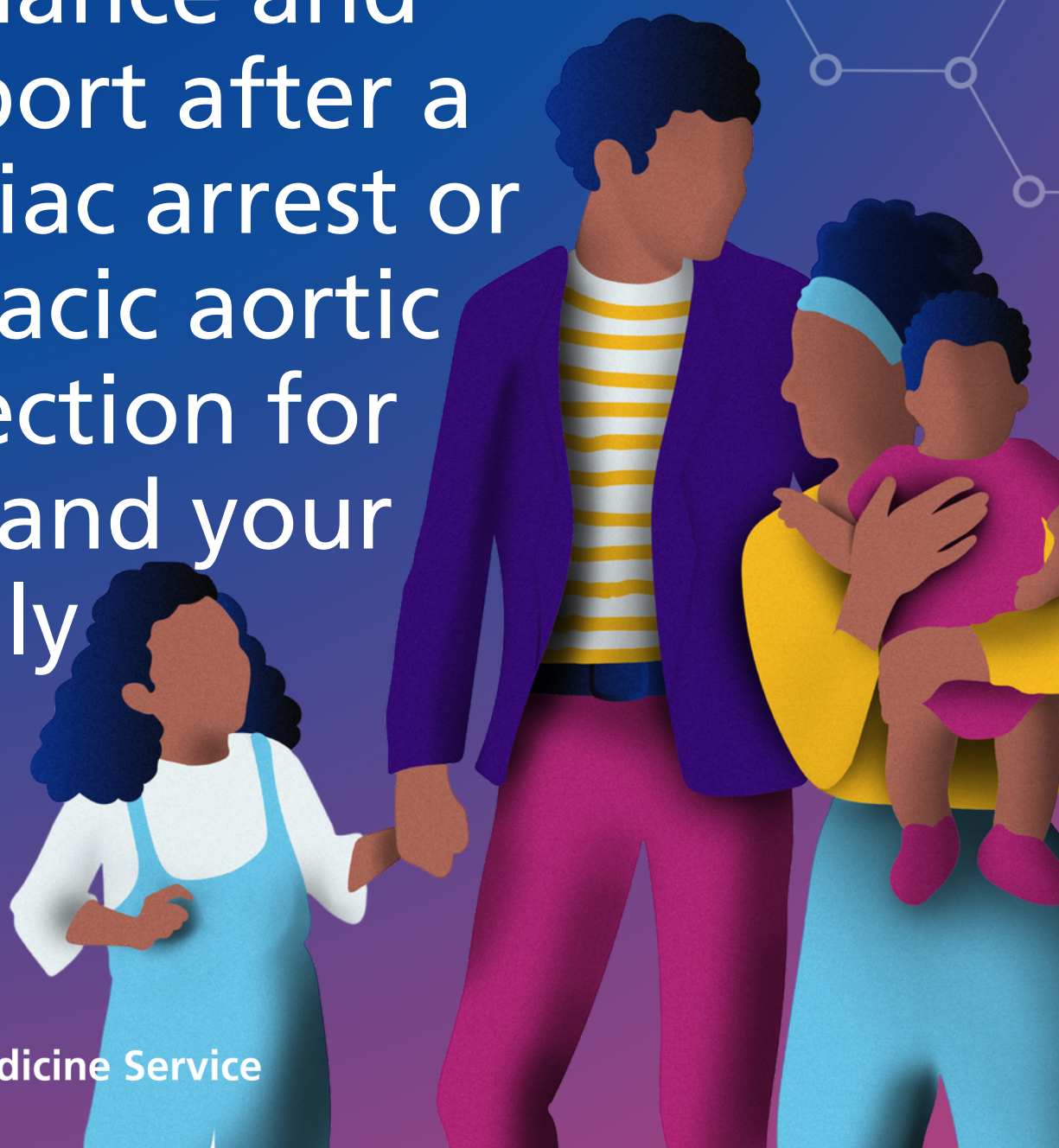




Understanding genetic testing for inherited cardiac conditions

Guidance and support after a cardiac arrest or thoracic aortic dissection for you and your family



South East
Genomic Medicine Service

Contents

2 Introduction

3 Genetic testing explained

What is a gene?

How do we inherit genes?

What is genetic testing?

4 Which Inherited Cardiac Conditions (ICC) can cause cardiac arrest or thoracic aortic dissection?

5 What to expect from a conversation about genetic testing?

7 Glossary

8 More support



Introduction

Cardiac arrest or thoracic aortic dissection (TAD) is a serious and unexpected medical event. Surviving this often brings many questions about what happened and what it means for the future. It is common to feel like having been through a major event in life, with little understanding of what has happened and what to do next. Sometimes, these cases are due to Inherited Cardiac Conditions (ICC) that can run in the family.

Genetic testing will help identify if there is an underlying inherited condition. This leaflet explains the role of genetic testing in your care, what the process involves, and what the next steps are for you and your close relatives.

Extra info on any terms that look like this can be found in the glossary on page 7

Genetic testing for you and your family members, has three main purposes:

- 1** Identifying genetic changes that may have caused the cardiac arrest or thoracic aortic dissection.
- 2** Informing first-degree family members (parents, siblings and children) of their risk of developing the same condition.
- 3** Guiding early detection, monitoring and treatment to reduce the risk for yourself and your relatives.

Genetic testing explained

What is a gene?

Genes are the instructions that tell the body how to develop and work properly. Genes are made up of DNA. Most differences in our DNA have no impact on our health or wellbeing, but some changes can cause genetic conditions or increase your risk of developing specific diseases—including heart conditions.

How do we inherit genes?

We inherit one copy of each gene from our mother and one from our father. A gene change may be passed down from parents to children (inherited) and others can occur for the first time in a child (new). Inheriting most gene differences happens completely by chance, but can increase the risk of heart conditions.

What is genetic testing?

If a cardiac arrest or thoracic aortic dissection occurs and a genetic cause is suspected, the ICU or CCU nurse may keep a small DNA sample, from the blood, and send it to a lab to be stored. This is because the blood is rich in the DNA that makes up genes. Later, your healthcare team will have a conversation with you about genetic testing.

Your DNA is like a library and your genes are like the books. The lab 'reads' some of the books, looking for spelling mistakes known to cause heart conditions. Understanding any changes in the genes helps doctors to diagnose genetic conditions, and assess the risk of family members developing a heart condition in the future.



Which Inherited Cardiac Conditions can cause cardiac arrest or thoracic aortic dissection?

Some heart conditions are known to have genetic causes. For diseases that can be inherited, testing can be offered to family members, which may help them know their risk of developing the same condition and take steps to reduce their risk.

These conditions fall into three main categories:

Aortopathies (aorta disease):

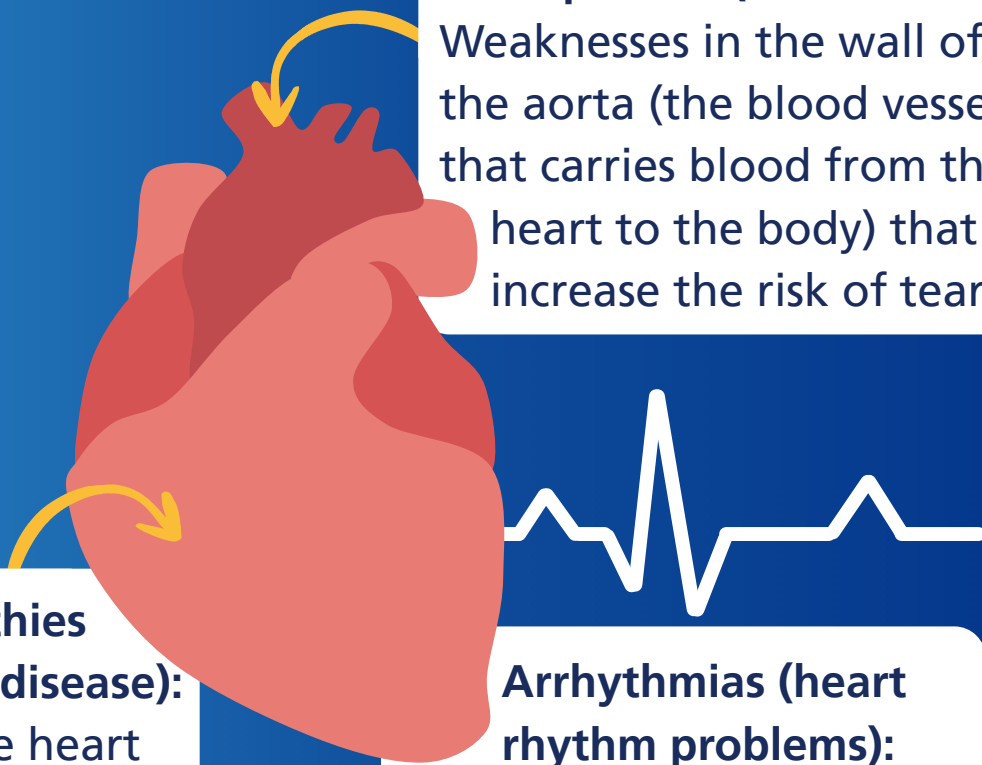
Weaknesses in the wall of the aorta (the blood vessel that carries blood from the heart to the body) that increase the risk of tearing.

Cardiomyopathies (heart muscle disease):

Changes in the heart muscle affecting its ability to pump blood.

Arrhythmias (heart rhythm problems):

A disruption of the heart's electrical system, causing changes in the heart beat.



What to expect from a conversation about genetic testing

After surviving a cardiac arrest or thoracic aortic dissection you may be referred to an Inherited Cardiac Condition (ICC) Centre to explore whether an underlying genetic condition could have contributed to what happened.

You will meet members of the ICC healthcare team. The conversation usually lasts around 45 minutes and is a chance to talk about whether genetic testing might be helpful for you and your family.

What normally happens in this conversation?

- We discuss if a genetic test may be right for you and your family
- We will have a conversation about consenting to genetic testing
- We will take your family history to learn more about you and your family's health
- We will discuss the potential outcomes of the test
- You will have a chance to ask any questions

What happens after this conversation?

If you decide to have the genetic test.....

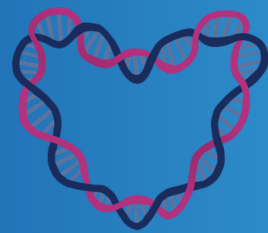
- If a DNA sample was taken during your medical care it may be sent to the genetics laboratory for testing.
- The testing process can take 4-6 months
- When the results are ready, they will be discussed with you by a member of your ICC healthcare team. They will explain what the result means for you and whether your relatives may need further assessment.

If you decide not to carry out the genetic test at this point...

- You will still be offered specialist cardiac screening at the ICC Centre.
- If a DNA sample has been stored you can choose to have genetic testing at a later date.
- Whether or not you have genetic testing, the ICC team will still offer ongoing care and advice



Information for first degree relatives



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If someone in your family has had a cardiac arrest or thoracic aortic dissection, relatives may also be invited to the ICC Centre. This is to help understand whether there could be an inherited cause and whether other family members may be at risk.

If a positive genetic result is found in your relative

If the genetic test in your relative identifies a clear, disease-causing change in a gene:

- Other family members may be offered targeted genetic testing to see if they carry the same change.
- This testing is usually quick and focused, because the specific genetic change is already known.
- If you do carry the same change, the ICC team will discuss what this means for your heart health and what monitoring or treatment may be helpful.
- If you do not carry the change, you may not need ongoing cardiac screening.

Screening for family members

Even when a genetic test on your family member does not find a clear cause or finds a variant of uncertain significance (VUS), first degree relatives are still recommended to have full cardiac screening at a specialist ICC clinic. This is because:

- Heart conditions can sometimes be present even without symptoms
- Some genetic changes become clearer as scientific knowledge grows

For more info or to signpost, speak to your local Pathway Coordinator

GSTT

London Inner South
Kent Central and South East
Kent Mid and Medway
Kent North East
Kent North West
East Sussex



St George's

London Inner West
London South
Surrey
West Sussex, Brighton and Hove



gstt.scdreferrals@nhs.net



SCD.referrals@stgeorges.nhs.uk

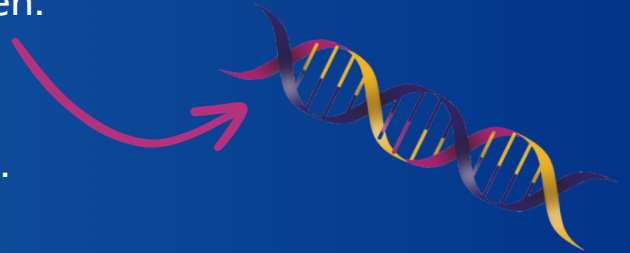
Glossary



CCU - Cardiac Care Unit, a specialised hospital ward for patients with serious heart conditions

DNA - DNA is made of proteins. DNA forms a special code (the genetic code) that tells body cells how to grow and work. DNA is passed down from parents to children.

ECG - Electrocardiogram. A test to record your heart's electrical activity.



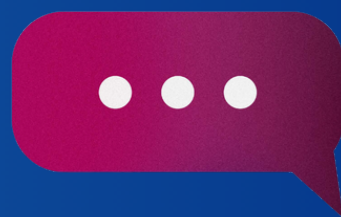
Genes - Genes are made of DNA.

ICC - Inherited Cardiac Conditions are heart conditions that can run in the family and be passed down to children.

ICC Centre - A specialist centre for diagnosis, treatment, and genetic counselling for patients and families managing inherited cardiac conditions. Teams will include doctors, genetic counsellors, clinical nurse specialists, and imaging experts. A link to a directory of centres can be found on the back of this leaflet.

ICU - Intensive care units are specialist hospital wards that provide treatment and monitoring for people who are very ill.

VUS - Variant of uncertain significance is a genomic variant about which there is insufficient data to conclude that it is either benign or pathogenic.



More support

You are not alone. These UK organisations offer support, information and counselling:

British Heart Foundation Genetic Information Service (GIS)

Information on heart conditions and family health

Tel: 0808 802 1234

Web: www.bhf.org.uk



Cardiac Risk in the Young (CRY)

Support, screening, and bereavement counselling

Tel: 01737 363222

Web: www.c-r-y.org.uk



Sudden Cardiac Arrest UK

Information and support for those who experience a sudden cardiac arrest

Tel: 0794 2288111

www.suddencardiacarrestuk.org



Cardiomyopathy UK

Support, information, and advocacy for people affected by cardiomyopathy

Tel: 01494 791224

contact@cardiomyopathy.org

Genetic Alliance UK

Advocacy and resources for families with genetic disorders

Tel: 0300 124 0441

Web: www.geneticalliance.org.uk



Aortic Dissection Awareness UK and Ireland

Information and support for those affected by Aortic Dissection

www.aorticdissectionawareness.org



SADS UK

Information and support for those affected by Sudden Arrhythmic Death

Tel: 01277 811 215

www.sadsuk.org.uk

Inherited Cardiac Centres

There are two major ICCs covering the South East England region

King's Health Partners, London

✉ gstt.cardiology@nhs.net

☎ 020 7188 8880

St George's Hospital, London

✉ icc.admin@stgeorges.nhs.uk

☎ 020 8725 5526