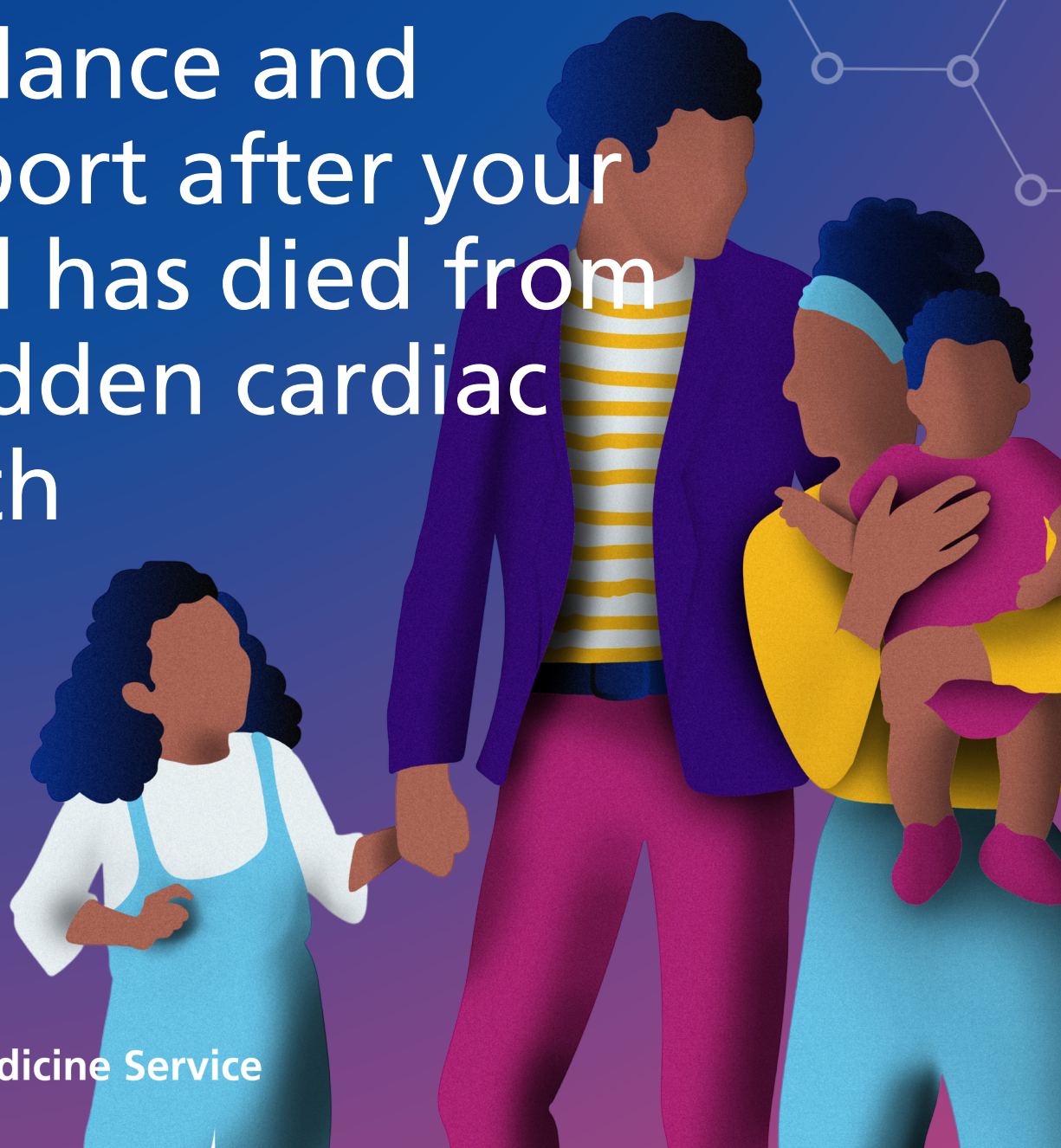




Understanding genetic testing for inherited cardiac conditions

Guidance and support after your child has died from a sudden cardiac death



South East
Genomic Medicine Service

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Introduction

Why do we offer genetic testing after your child died suddenly from a heart-related condition?

You may have already had a conversation with a healthcare professional about consenting to genetic testing. The sudden and unexpected death of a child is a distressing and overwhelming experience—especially when they were previously healthy. In some cases, these deaths are due to **Inherited Cardiac Conditions (ICC)** that can run in the family, even if no one was previously affected.

This leaflet will help explain the testing process and how it may benefit your family. You will find some useful websites on the back of this leaflet with extra resources to support you through this difficult time.

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

Post-Mortem Genetic Testing, sometimes called 'Molecular Autopsy', has three main purposes:

- 1** Identifying genetic changes that may have caused your child's sudden death.
- 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 in 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?

When a child's death is unexplained and a possible genetic cause is considered, the pathologist may keep a small sample of tissue - usually from the spleen - so that DNA can be stored safely. This is because the spleen 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 sudden unexplained death?

Some heart conditions are known to have genetic causes. When a child dies suddenly, identifying whether an inherited condition may have been involved can help parents and siblings understand whether they may also be at risk and whether monitoring or treatment could help keep them safe.

These conditions fall into three main categories:

Aortopathies (aorta disease):

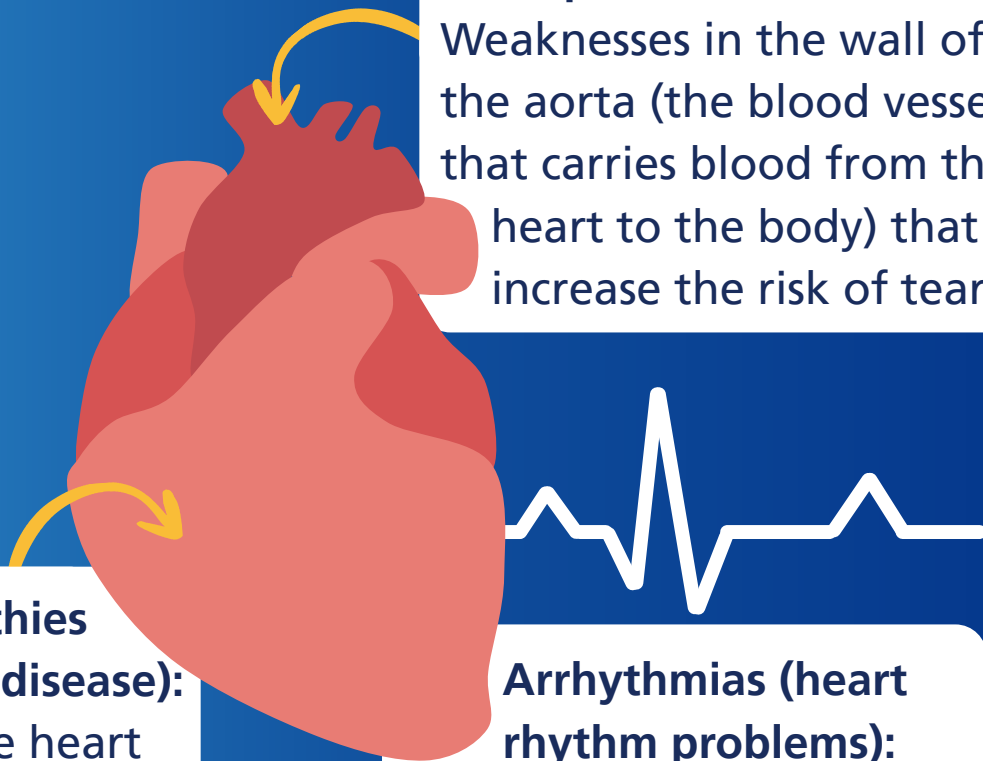
Weaknesses in the wall of the aorta (the blood vessel that carries blood from the heart to the body) that increase the risk of tearing.

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



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If it is believed that an inherited heart condition is the cause of what happened to your child, the coroner will pass on your contact details to the Inherited Cardiac Condition (**ICC**) **Centre** with your consent. The ICC coordinator will contact you by letter, email or phone to set up an appointment when you are ready.

At this appointment, a conversation usually takes place with the healthcare team at the ICC service to help you make informed decisions around genetic testing. This conversation can take around 45 minutes.

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 you consented to retain a small sample of your child's spleen for genetic testing, this will be sent and stored at the genetic laboratory.
- Once you have signed the consent form, the genetic laboratory will begin testing, which could take around 4–6 months.
- Results will be discussed with you by a member of your healthcare team.

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

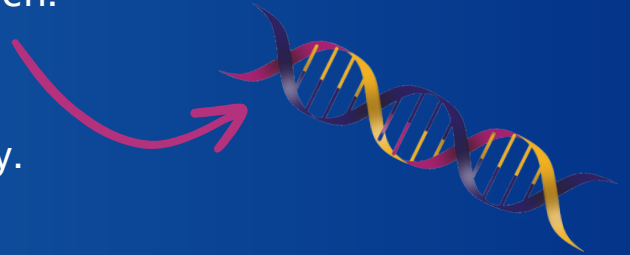
- You will still be recommended specialist heart screening at the ICC Centre, which may include looking at the electrical activity of the heart (**ECG**), ultrasounds to look at your heart's structure and blood vessels, and an exercise test to look at your heart's electrical activity during exercise.
- If you consented to store your relatives DNA sample long-term, you will still be able to consider genetic testing at a later date.

Glossary



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.

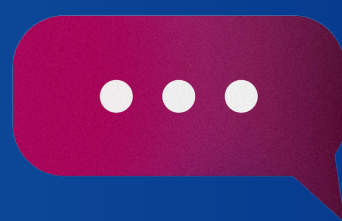
ICC - Inherited Cardiac Conditions are heart conditions that can run in the family and be passed down to children.

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.

Pathologist - A pathologist is a medical expert who carries out a detailed examination to find out a person's cause of death.

Post-Mortem Genetic Testing - A post-mortem is a detailed examination to find out a person's cause of death, otherwise known as an 'autopsy'. Post-mortem genetic testing involves doing tests to look for genetic causes.

Spleen - The spleen is a small organ in the upper-left of your abdomen, next to your stomach and behind your left ribs. It filters the blood and plays a role in the immune system that defends the body from infection.



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



The Compassionate Friend

Supporting bereaved parents and their families

Tel: 0345 123 2304

<https://www.tcf.org.uk>



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