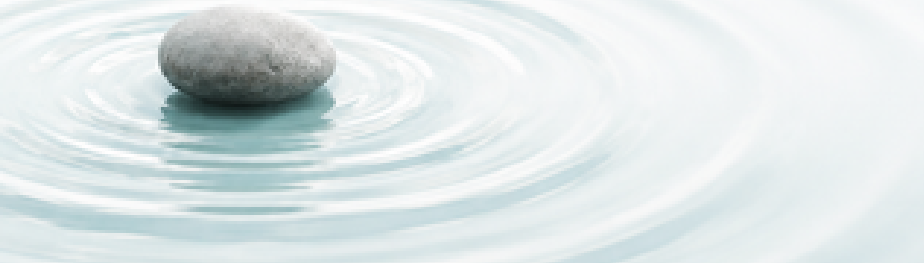
North Thames Family Support Guides

Diagnostic genetic testing

Supporting you and your family through a diagnostic genetic test



Before you begin

You may have been given this guide because diagnostic genetic testing is being considered for you or your family.

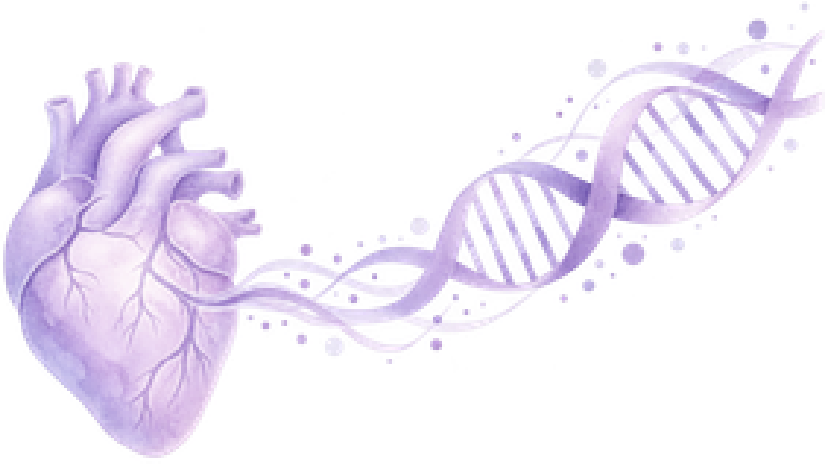
Some people have lots of questions about genetic testing and want time to think. Others make a decision quite quickly. There is no right or wrong way to approach it.

This guide explains what genetic testing is, what the results may mean, and what may happen next.

What is genetic testing?

Our genes are the instructions that help our bodies grow and work. We inherit them from our parents.

Genetic testing looks at your genes to see if there is a change that may help explain a heart or aortic condition.



• Why might genetic testing be helpful?

To help explain what has happened
Finding a genetic cause may help explain why a heart or aortic condition has happened.

To understand what it may mean for your family
It may help us to understand whether other people in the family could have the same condition.

To help guide screening
The result may help guide cardiac or aortic screening for you and your family.

To provide information for the future
For some families, a result may provide useful information for children, younger relatives or when thinking about future pregnancies.

• How is genetic testing carried out?

If you are having genetic testing
Genetic testing is usually carried out using a blood sample. The DNA from the sample is then tested in a specialist laboratory.

If DNA has already been stored
Sometimes DNA has already been stored, for example after someone has died. This DNA may be used for genetic testing if testing is recommended.

Your healthcare team will explain which applies to you or your family.

• What results might I get?

Genetic testing can give different types of results. Your healthcare team will explain what your result means for you and your family.

A genetic cause is found
A gene change is found that helps explain the heart or aortic condition. Relatives may be offered genetic testing.

No genetic cause is found
No gene change that explains the condition is found. This does not always mean there is no genetic cause. Relatives usually have cardiac or aortic screening.

An uncertain result
Sometimes a gene change is found, but we do not yet know whether it causes the condition. Relatives usually have cardiac or aortic screening.

• What could a result mean for my family?

If a genetic cause is found, it may help us understand whether other people in the family could have the same condition.

A genetic result only tells us about the person who was tested. It does not tell us whether the gene change was inherited from a parent or started for the first time in that person.

Testing other relatives can sometimes help us understand where the gene change came from and who else in the family may have it.

• Deciding whether to have genetic testing

Having genetic testing is your choice.

Some people know quite quickly whether they would like genetic testing. Others have questions or want more time to think about it.

You can talk through any questions or concerns with your healthcare team before you decide.

• Sharing the genetic result with your family

One of the main reasons for genetic testing is to provide useful information for your family.

For this reason, we offer genetic testing on the understanding that a result that is important for your relatives can be shared with them. Most people are happy to share their result letter with relatives themselves.

If a medical team looking after one of your relatives asks us for a copy of the genetic result, we will share this with them. **If you have any concerns about this, please discuss them with us before agreeing to genetic testing.**

• Is there anything else I should know?

There are a few other things to be aware of before deciding whether to have genetic testing.

Unexpected findings
Occasionally, genetic testing may find information that was not expected.

Testing is not always perfect
Very rarely, errors can happen during genetic testing or a result may not be clear.

Sample quality
Sometimes there may not be enough DNA, or the quality of the sample may mean that testing cannot be completed.

What about insurance?

Having genetic testing may sometimes have implications for insurance.

Diagnostic genetic test results can form part of a person's medical information. Relevant medical information may need to be provided when applying for some types of insurance.

If you have any questions or concerns about insurance, you can discuss these with your healthcare team before deciding whether to have genetic testing.

What happens if I decide to have genetic testing?

Your healthcare team will guide you through each step.

1. **Talk it through**
   Ask questions and discuss genetic testing.

2. **Sign the consent form**
   If you decide to go ahead.

3. **Blood sample**
   A blood sample is taken if DNA has not already been stored.

4. **DNA is prepared**
   If needed, DNA is extracted and prepared for testing.

5. **Genetic testing**
   Your DNA is tested in a specialist laboratory.

6. **Your result**
   Your healthcare team will explain your result and what happens next.

Things I don't want to forget

You can use this page to write down questions, things you would like to talk about, or anything you want to remember.

Support and resources

You may find these sources of information and support helpful.

NHS – Genetic and genomic testing
Information about genetic testing, results and what they may mean for you and your family.
NHS genetic and genomic testing

Amber's story
A parent's experience of loss, cardiac screening and genetic testing. NT GMS website: 'Highlighting our Sudden Cardiac Death project'.

The Ripple

A family member once described the impact an inherited condition can have on a family as being like a stone dropped into a pond. The ripples spread outwards, touching every member of the family in different ways.

Those words have stayed with me ever since.



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This guide has been developed to support people and families considering diagnostic genetic testing for an inherited heart or aortic condition.

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Developed by Claire Kirkby,
North Thames ICC Coordinator