Consent and confidentiality

You will need to consent to any genetic or genomic testing. This means you agree to the test after understanding its purpose, benefits, possible results and risks.

Your information will be kept confidential.

If there is a need to share your results for example, with another healthcare professional, they will only be shared with your consent and approval.

Questions to think about and ask your healthcare professional

We encourage you to ask questions of your GP (family healthcare professional) or nurse. Here are some questions other patients asked.

1. Why have you recommended this test for me?
2. What are the risks of doing the test?
3. What if I choose not to get tested?
4. What if I have a hereditary condition?
5. Will I have to pay for the test?
6. Where will my test be done?
7. How long will it take to get my results back?
8. How will I get my results?
9. What is the chance of the test finding an answer?
10. Could the test show a finding unrelated to the purpose of original test?
11. What support is available during testing and after I receive the results?
12. Will the test change the way I or my child receive healthcare?

Support and further information

There is a network of patient support groups that can help you through your experience.

You can access these groups at any time in the process. Please see some support groups listed below.

Rare Ireland	www.rareireland.ie
Irish Cancer Society	www.cancer.ie
Irish Maternity Support Network	www.maternitysupport.ie

Plain English
Approved by NALA

Genetic and genomic testing

This Patient Information Leaflet is for people who are considering having a **genetic** test, a **genomic** test or both types of tests. These terms are explained later in this leaflet.

HSE

What are genes and genetics?

Genes are the instructions that tell your body how to grow and develop. Your genes carry characteristics that you inherit from your parents. For example, the colour of your eyes or your blood type. Your genes are made up of sequences of DNA. DNA is the genetic information inside the cells of your body. Your genome is made up of all your genetic information including your genes.

Each person has many differences **(variants)** in their genome. Most variants do not impact how a gene works, but some people have variants that do. This can cause or increase the chance of developing a genetic condition. **Variants** can be passed down from parents or can appear for the first time in a person.

What is the difference between a genetic and genomic test?

A genetic test looks at a single gene or a small number of genes at a time. The test may look for a single variant or several variants of genes.

A genomic test looks at many genes, sometimes 20,000 to 25,000 genes at once.

Genetics is the study of genes and how characteristics are passed from one generation to the next. Your healthcare professional might suggest a **genetic test** or **genomic test** to you. The reason is to look for variants that change how a gene works. This can be for many reasons including:

- to help find a reason for your features
- to help guide treatment of your condition
- to identify and understand a risk to you and your family, or
- to confirm a diagnosis.

How is the test done?

Blood tests or mouth swabs (saliva) are the most common samples taken to provide a DNA sample for testing.

What is the purpose of the test?

Genetic and genomic tests are used to:

- diagnose a genetic condition **(diagnostic)**
- find out if you have a variant (a difference in a gene) that has caused a genetic condition in your family. This could impact your health in the future **(predictive)**
- see if you carry a variant that could be passed onto your children, but which is unlikely to affect your own health **(carrier)**
- diagnose a genetic condition in your pregnancy **(prenatal)**
- confirm a clinical diagnosis you have previously received **(confirmation)**, or
- understand how a variant that has been found previously has been passed down in your family **(segregation analysis)**.

Discuss the information in this leaflet with healthcare professionals, family and friends to help you decide whether to have testing. It can help you think about any other questions you might have. It is important for you to give your consent before the testing starts. Testing is your choice.

What are the potential benefits of tests?

- To find out the cause of you or your child's condition.
- To support or guide medical care for you or your child.
- To improve understanding of a condition.
- To access support.
- To help you know the chance of developing a condition.
- To provide information about the chance of having a family member with the same condition.

What are the possible results?

The result of your test may show:

- **no variants** found that identify the cause of your or your child's condition
- a **variant** found that is likely to cause your or your child's condition
- a **variant of uncertain significance** found. This means that, at this time, it is not known whether the variant found is the cause of your or your child's condition
- an unexpected or additional finding. Your healthcare professional will discuss this with you.

Are there any risks to the tests?

A finding from a genetic or genomic test may be additional or unexpected and this can be upsetting. Before you consent to taking a test, the person taking your consent will discuss possible test findings with you and any related risks.