Understanding Genetic Testing

This leaflet explains why genetic testing may be offered, what happens and what results can mean. Use it to prepare for a conversation with your healthcare team and decide what you want to ask.

What is a genetic test

Genetic tests look for changes in genes, chromosomes or related proteins. They may help explain symptoms, identify an inherited condition or assess the chance of developing or passing on a condition. The terms **genetic testing** and **genomic testing** are both used; genomic testing often examines a wider amount of genetic information. [1,2]

DNA carries biological instructions. **Genes** are sections of DNA, and **chromosomes** are structures containing DNA. A **variant** is a difference in DNA. Many variants are harmless; finding a variant does not automatically explain an illness. [1,3]

A genetic condition can occur without a known family history. A new variant, sometimes called **de novo**, may be present in a child but not found in either parent. Genetic does not always mean inherited. [3]

Why might testing be offered

- **Diagnosis:** to investigate symptoms or a suspected condition. [4]
- **Future risk:** to assess the chance of developing a condition before symptoms appear. [4]
- **Carrier testing:** to find a variant that could be passed to a child, even when the person tested has no symptoms. [4]
- **Care or family planning:** to help guide some treatment decisions or explore reproductive options. Availability varies. [2,4]

A test may provide an explanation, but it cannot promise an answer or a treatment. Ask what useful information this particular test could give you. [2,5]

Before you decide

Different tests answer different questions

A **single-gene test** examines one gene. A **gene panel** examines a selected group. **Exome sequencing** focuses on the parts of genes that provide instructions for making proteins. **Genome sequencing** examines DNA much more widely. Other tests look for larger chromosome changes. A broader test still cannot detect or explain every possible genetic cause. [1,6]

What happens during testing

A sample may be taken from blood, saliva or the inside of your cheek; some tests need other tissue. The laboratory analyses the sample and sends a report. The sample required and the way results are returned depend on the test and service. [7]

Most blood or cheek-swab tests have small physical risks. Some procedures used for testing during pregnancy carry additional risks, including miscarriage. Ask about the risks of the exact procedure being offered. Results can also bring worry, relief or difficult family conversations. [5]

Consent means understanding your choices

Before agreeing, ask about the purpose, limits, possible results and implications for relatives. You can ask for time, an interpreter or accessible information. For children or people unable to give consent, ask how local consent rules apply and how the person will be involved. [8]

Some tests may reveal findings unrelated to the original question. Ask which additional findings might be reported and what choices you have about receiving them. Discuss sample storage, data sharing and whether any research participation is optional. [8,9]

Questions to take to your appointment

- Why this test, and how could each possible result affect my care?
- What does it miss, and might relatives also need testing?
- What will it cost, when should results arrive and who will explain them?
- Can I decline or change my mind, and what are the limits once testing or data sharing has begun?

Making sense of results

The meaning depends on the test, your symptoms and family history. Ask for an explanation of your own report rather than interpreting the words “positive” or “negative” on their own. [9,10]

A relevant genetic change is found

A **pathogenic** variant is a change known to cause disease; **likely pathogenic** means the evidence strongly suggests this. The team must still consider whether it fits your symptoms and how the condition is inherited. A finding may support a diagnosis, show carrier status or indicate future risk. It does not always predict whether symptoms will occur, when they will start or how severe they will be. [9,10]

No relevant change is found

A negative or uninformative result means the test did not identify a change that answers the question. It may miss some kinds of change, or current knowledge may be insufficient. It does not necessarily rule out a genetic cause or invalidate your symptoms. A negative test for a specific, known family variant can be more informative; ask what your result rules out. [9,10]

A variant of uncertain significance is found

A **variant of uncertain significance (VUS)** is a change for which there is not enough evidence to know whether it causes disease. It is not a confirmed diagnosis. A VUS alone should not guide treatment changes or predictive decisions for relatives. Your team may suggest further investigation, including selected family testing to help clarify it. [9,10]

Uncertain does not mean harmful. Interpretation can change as evidence grows. Ask whether reanalysis is possible, who would arrange it and how you would hear about a change. Do not assume your result is being continuously checked. [9]

Can a result be wrong or incomplete

Tests have technical and interpretive limits. False positive and false negative results can occur. Sometimes another sample, a different test or later reassessment is needed. Keep a copy of the report and ask what follow-up is appropriate. [10]

After the test

Agree a follow-up plan

Ask who will explain the result and what happens next, including if it gives no answer. A genetic counsellor or clinical genetics specialist can help with the medical and family implications. Continue care based on your symptoms and agreed plan; do not change treatment from a report alone. [2,9]

If the result matters for biological relatives, ask which relatives may benefit from advice and whether the service can provide a family letter. You do not have to explain complex genetics on your own. Results can raise unexpected questions about family relationships, so support may be useful. [2,9]

Tests bought online and research results

A consumer DNA test is not a substitute for a clinical assessment. A health-related finding may need confirmation through a healthcare service before it is used for diagnosis or care. Do not treat an ancestry or wellness report as a medical diagnosis. [11]

If testing is offered through research, ask whether individual results will be returned and whether clinical confirmation would be needed before acting on them. [9]

Privacy, costs and local rules

Ask who can access your sample, report and genetic data, where they are stored, whether they may be shared and what withdrawal or deletion is possible. Keep reports secure. You do not need to send genetic results or medical information to Rare Disease Watch. [8,9]

Referral routes, eligibility, costs and waiting times differ between healthcare systems. Consent, data protection and insurance rules also vary by country and policy. Confirm the rules that apply to you with the testing service or an appropriate local adviser before deciding; protection against genetic discrimination is not the same everywhere. [2,8,12]

Key points to remember

- Know what question the test is intended to answer.
- A negative result may leave the cause unexplained.
- A VUS is not a confirmed diagnosis.
- Agree who will explain results and arrange follow-up.

One useful question: “What will this result change for me now, and what is still unknown?”

General information for patients and carers; individual decisions depend on your circumstances and the test being considered.

Sources and further reading

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Numbers in the leaflet refer to the sources below. Links were checked on the date above. Sources from the UK and USA explain the science and testing process; their descriptions of services and legal protections apply to their own settings.

1. MedlinePlus Genetics. What is genetic testing?
https://medlineplus.gov/genetics/understanding/testing/genetictesting/

2. NHS. Genetic and genomic testing.
https://www.nhs.uk/tests-and-treatments/genetic-and-genomic-testing/

3. MedlinePlus Genetics. What is a gene variant and how do variants occur?
https://medlineplus.gov/genetics/understanding/mutationsanddisorders/genemutation/

4. MedlinePlus Genetics. What are the uses of genetic testing?
https://medlineplus.gov/genetics/understanding/testing/uses/

5. MedlinePlus Genetics. What are the risks and limitations of genetic testing?
https://medlineplus.gov/genetics/understanding/testing/riskslimitations/

6. MedlinePlus Genetics. What are whole exome sequencing and whole genome sequencing?
https://medlineplus.gov/genetics/understanding/testing/sequencing/

7. MedlinePlus Genetics. How is genetic testing done?
https://medlineplus.gov/genetics/understanding/testing/procedure/

8. MedlinePlus Genetics. What is informed consent?
https://medlineplus.gov/genetics/understanding/testing/informedconsent/

9. NHS England, National Genomics Education Programme. Consent conversation for genomic testing.
https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/the-consent-conversation-for-genomic-testing/

10. MedlinePlus Genetics. What do the results of genetic tests mean?
https://medlineplus.gov/genetics/understanding/testing/interpretingresults/

11. MedlinePlus Genetics. What is direct-to-consumer genetic testing?
https://medlineplus.gov/genetics/understanding/dtcgenetictesting/directtoconsumer/

12. MedlinePlus Genetics. What is genetic discrimination?
https://medlineplus.gov/genetics/understanding/testing/discrimination/

https://rarediseasewatch.com/ Rare Disease Watch©. Highstone House, 165 High Street, Barnet, Herts. EN5 5SU, UK.