

Genetic Testing and Results:

A 6-step Guide for Healthcare Professionals



Latin-SEQ+

This poster guides healthcare professionals through a 6-step process to support informed consent for genetic testing. Use it with the patient toolkit to explain what testing involves, its possible outcomes and implications.

1. Explain the test to your patient

- Why are you offering the test?
- What sample is required?
- Where will you send the sample?
- When do you expect the results?

Informed consent is an ongoing conversation, not a single event.

2. Speak of potential benefits

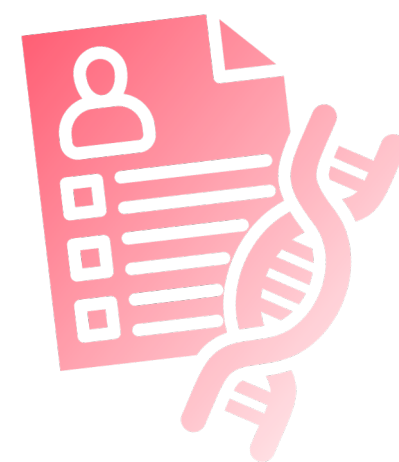
For your patient will the test:

- Identify or confirm a genetic cause.
- Inform treatment, monitoring and future care.
- Provide prognostic information.
- Support reproductive decisions.
- Identify risks for relatives.
- Help explain the condition?

3. Be clear about its limitations

Explain that:

- A cause may not be found.
- A negative result may not exclude a genetic cause.
- Results may not change care.
- Unexpected findings may arise which can cause worry and uncertainty.
- Some results may remain uncertain.



4. Mention potential emotional and family implications

Testing and results may:

- Cause anxiety, uncertainty, guilt, shame or worry.
- Generate tensions within the family
- Raise questions about how and whether to share results.
- Reveal information relevant to relatives
- Lead to differing family views.



5. Explain possible results



Pathogenic or likely pathogenic variant:

A genetic change explains or contributes to the condition.



No significant variant found:

No genetic explanation identified, although a genetic cause may still exist.



Variant of Uncertain Significance (VUS)*:

this means that a genetic change was found but there is not enough evidence to know whether it is related to disease.

***Clinicians and patients should understand that:**

- A VUS does not confirm a diagnosis.
- A VUS does not rule out a diagnosis.
- Interpretation of a VUS may change over time as scientific knowledge develops.



6. Discuss wider implications

- Whether and how results can affect relatives.
- Reproductive options and family planning.
- Possible implications for insurance, employment or travel.
- Future re-analysis or reinterpretation (particularly if there is a VUS).

Before consent, confirm your patient understands:

- Why testing is offered.
- Its benefits and limitations.
- The possibility of uncertain or unexpected results.
- The potential impact of results on relatives.
- How and when results will be communicated.
- Their right to ask questions or decline testing.