

A simple way to guide patients through consent

You can use this framework to walk patients through exome and genome testing.



Why is this test being offered?

This test looks at genetic information to see if we can find a reason for your patient's symptoms and help guide care, next steps, or future planning.

What results might come back?

There may be an answer from this test, or there may not be, but it can move you a step closer to figuring things out.

- + Positive:** A genetic change is identified that explains the patient's symptoms. This can help guide care and management.
- Negative:** No genetic cause is found based on current knowledge. This does not rule out a genetic condition, and additional testing or follow-up may be considered.
- ? Variant of Uncertain Significance (VUS):** A genetic change is found, but it is unclear whether it is related to the patient's symptoms. These results may require follow-up or future clarification.

In some cases, the test may find additional information that isn't related to why you ordered the test but it is on the report. These can be either additional findings or ACMG secondary findings.

What are ACMG secondary findings?

If a patient chooses to receive them, genetic testing may identify what are called ACMG secondary findings. These are genetic changes in specific genes that increase the chance to develop certain health conditions like hereditary cancer risk or hereditary cardiovascular conditions. ACMG secondary findings are identified in a small number of patients, about 2–3%. If the patient chooses to learn about these findings, it will be included in a separate section of the report, if detected.

What are additional findings?

Additional findings are variants in genes beyond the ACMG secondary findings list that are associated with serious health problems in children may also be identified and reported. These are not commonly reported and are not optional to receive.

Should family members be tested as well?

Testing two biological relatives, preferably parents, along with the patient, can help to provide more informative results. This is called trio testing. Comparing family member DNA can increase the chance of finding an answer and help reduce the likelihood of reporting uncertain findings.

Could anything unexpected come up when trio testing is ordered?

In rare cases, testing may reveal unexpected information, such as biological relationships that are not what were reported, like non-paternity.

How is genetic information protected?

There are protections in place to help keep your genetic information safe. Laws like GINA help protect against discrimination in health insurance and employment. There are some limitations, such as life, disability or long-term care insurance that can request access, and there are groups that are not covered by GINA. Additional information about GINA can be found at <https://ginahelp.org>.

What choices do patients have when it comes to their data?

Patients have control over many aspects of their genetic testing experience and can choose whether or not to receive ACMG secondary findings, be contacted for research opportunities, and participate in Health Information Exchange (HIE).

A few more key concepts and definitions

What genetic testing looks for

Genetic testing looks at DNA to identify changes that may explain a patient's symptoms or condition. The amount of DNA, and way that it is looked at, varies based on the test chosen. Results can help guide diagnosis, care decisions, and future planning.

Exome sequencing

Exome sequencing is a comprehensive genetic test that looks at a person's genes (genetic instructions that are found within the DNA) to identify genetic changes that may explain developmental differences, health conditions, or medical concerns.

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GINA

The Genetic Information Nondiscrimination Act (GINA) protects against discrimination in health insurance and employment. However, it does not apply to life, disability, or long-term care insurance, and some groups may not be covered. Additional information about GINA can be found at <https://ginahelp.org>.

Health information exchange

Health Information Exchange (HIE) allows healthcare providers and organizations to securely share a patient's medical information to support coordinated care.

- Participation depends on the patient's healthcare institution
- Information is shared securely to help improve the speed, quality, and safety of care



We're here to support you

If questions come up at any point, your GeneDx team is here to help:

Support@genedx.com | 888-729-1206



You can share our patient-friendly consent education video to walk patients through genetic testing, including results, secondary findings, and what to expect.

genedx.co/consent