

Genetic Testing for Hereditary Cancer



Our bodies are made of small structures called **cells**.



Each cell contains instructions that tell the cell what to do. These instructions are called **genes**, and are made of **DNA**.



Genes tell the cell how to make structures called **proteins**, which each have a specific job to do in the body.



We all have differences in our DNA. A **mutation** is a difference in the DNA that can harm our health. Some people are born with mutations that cause an increased risk of cancer. This is called a **hereditary cancer predisposition**.

To test for a hereditary predisposition to cancer, either a blood or saliva sample is sent to a special lab for **genetic testing**, which looks at genes related to hereditary cancer.



For each gene tested, the results can be **positive**, **negative**, or **uncertain**.



• A **positive** result means that the test has found a mutation that is known to increase cancer risk and sometimes other health problems. Positive results may explain why some people in the family developed cancer.



• A **negative** result means that nothing was found that would increase cancer risk. Negative results are reassuring, but they do not explain any of the cancer in the family, and they do not answer every question.



• An **uncertain** result happens when a difference in the DNA is found, but there is not enough evidence to know whether the difference is related to cancer or health problems, or whether it is normal variation. In time, uncertain results may be reclassified as positive or negative.



Signs of Hereditary Cancer in a Family



A family member with a known genetic mutation



Multiple family members diagnosed with cancer



Cancer below age 50



High-risk heritage, such as Ashkenazi Jewish ancestry



Bilateral cancers, such as cancer in both breasts or both kidneys



A person with two or more separate cancers



Rare cancers, such as:

- Pancreatic cancer
- Ovarian cancer
- Male breast cancer
- Pheochromocytoma
- Neuroendocrine tumors

Family history is not a perfect tool for predicting who will test positive, but it can help to identify people who may benefit from genetic testing.