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What is germline genetic testing in cancer and what might it mean for you?

Being diagnosed with cancer can be overwhelming, and some of the tests can be complex. But genomic testing and understanding your genes doesn't have to be confusing.

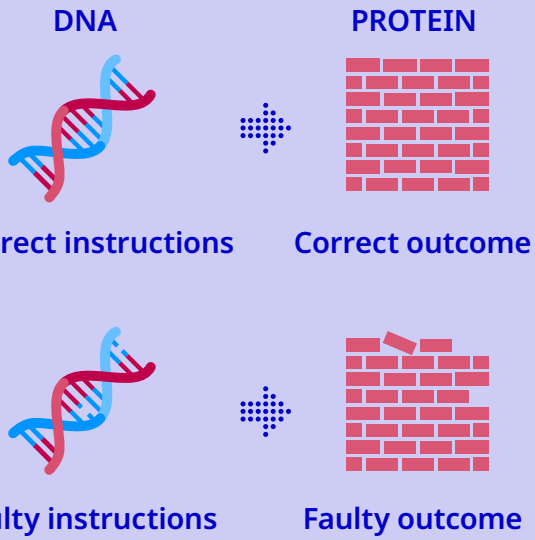
If your doctor thinks it is appropriate to proceed with genetic testing, you can have a better understanding of your risk of developing cancer and potential treatment options.

The information provided on these pages aims to make genomic testing easier to understand.

What is a gene?

A gene is like an instruction book full of words (DNA), that tell your cells how to build proteins.

Proteins are the building blocks of your cells and have many roles. Our bodies are made up of many cells which are small units that work together for specific jobs within the body.[1,2]



What is a mutation?

A mutation is a change in your DNA. If the words in the instruction book change it may mean that the building blocks (proteins) form differently.[2]

Mutations can be caused by many things[1], including:

- **Environmental factors** (eg, smoking or UV light)
- **Infections** (eg viruses such as human papillomavirus)
- **The ageing process**

Some gene alterations can be inherited (passed down) from your parents. This is called a 'germline mutation'.[1,2]

Scientists have found that some genetic changes can play a role in cancer development, growth and spread.[1]

Cancer genomic testing

Jargon buster what does it mean?

Doctors and healthcare experts sometimes use complex scientific words and technical language (called jargon) when they talk to patients. Understanding what these words mean may help you feel more confident about the choices and decisions you have to make.

Germline

Germline mutations are gene alterations found in all the cells of the body as they have been inherited from a parent and can be passed down through the generations.[1,2]

Cell

Cells are the building blocks of your body that contain a copy of your genes. The genes within each cell are like instructions that tell the cell how to how to make proteins.[1]

Chromosome

DNA is packaged into structures called chromosomes, found in the cells of living things.[2]

Somatic

Somatic mutations occur randomly during our lifetime in response to environmental factors. They are gene alterations only found within cancer cells and don't affect healthy cells in the body.[3]

Genetic vs Genomic

Genetics looks at the function of a single gene on its own, whereas genomics looks at all the genes and surrounding structures together, and how they influence how we grow and develop.[4]

Biomarker

A biological molecule that is a sign of a normal or abnormal process, or of a condition or disease.[5]

Sequencing

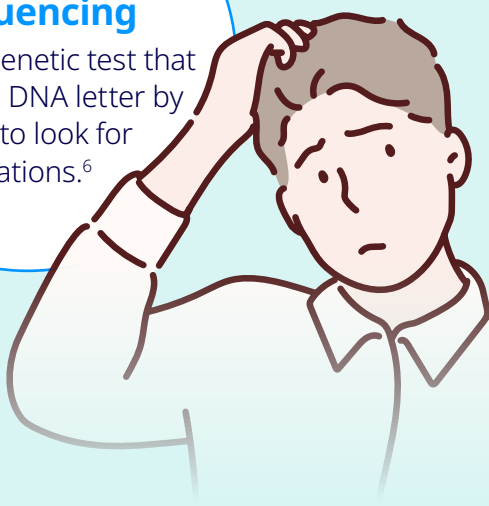
A type of genetic test that reads the DNA letter by letter to look for mutations.[6]

ctDNA

Scientists have discovered that dying cancer cells may release small pieces of their DNA into the bloodstream. These pieces are called cell-free circulating tumor DNA (ctDNA).[7]

Tumour biopsy

A sample of the tumour or cancer is taken to characterise the tumour which might include looking at genetic alterations[6]



What is germline genetic and genomic testing?

Inheriting certain mutations raises your risk of developing cancer. Mutations in some genes might mean they are unable to repair damaged cells, which can then lead to uncontrolled growth and formation of a tumour.[8]

Genetic testing can tell you if you have inherited one of these 'cancer risk genes' or a specific mutation.[8]

Genomic testing may be used to look at all of your genes to see if there is a risk of developing cancer.[4,6]

Why is it important?

Germline genetic testing can estimate your risk of developing cancer if you have inherited a mutation in a cancer risk gene.[9]

It does not mean you have cancer or are going to develop it.[8]

It just means it may help identify those with a higher risk of cancer due to family history.[10]

It may help you and your doctor plan risk reducing interventions and certain treatments.[8]

Your doctor may recommend germline genetic testing if you:

Have extensive family history of cancer

Show sign(s) of an inherited cancer

Are in a high-risk population group

What does germline genetic testing involve?

1. **Your doctor will discuss if genetic testing could be useful for you**
2. **You may have a blood test which will be sent to a laboratory to look for mutations**
3. **A healthcare professional will explain what your cancer risk is, and potential treatment options based on your results[1]**

Blood test

Blood tests are used for **germline testing**. A blood test can look for inherited changes in the family.[10,12]

Germline genetic and genomic testing is a personal decision and should only be made after you've spoken to your doctor

Tumour test

Somatic testing is different. To look at somatic mutations, (those not passed down from your parents) a tumour biopsy may be taken to look at a particular tissue or organ.[12]

Questions you may want to think about

What will the results mean?

Germline genetic tests can tell you if you have a **mutation in a known cancer-related gene** which may increase your risk of cancer.[1,2]

With this information, you and your doctor may be able to plan risk reducing interventions and certain treatment options.[1]

What about my family?

If you have a mutation in a cancer-related gene, **there is a chance it can be inherited and passed on to your children** if it is present in sperm cells or egg cells.[3]

You can't inherit somatic mutations because they occur in cells that are not sperm or egg cells.[3]

Your family members may want to explore the possibility of genetic counselling and testing too.

What are the advantages of germline testing?

Knowing your genes means you can have a better understanding of your risk.[11]

You can make lifestyle changes to lower your risk of developing cancer. [11]

You can discuss treatment options with your doctor, and have regular screening to increase the chance of finding any abnormalities early.[8]

Knowing your result may reduce any anxiety or stress you have that comes from not knowing your risk.[10]

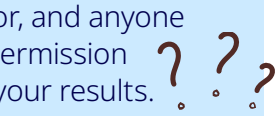
Are there any disavantages?

Sometimes doctors may find an inherited mutation in a gene but not know what the consequence will be.[10]

Identifying an increased risk of developing cancer might also cause some anxiety within the family, and this is why receiving genetic counselling is important.[11]

What happens to my results?

Your genetic testing results, like all of your health information, is strictly confidential. They can only be discussed between you, your doctor, and anyone you give permission to access your results.



When will I get my results?

Initial germline blood test results for the individual affected may be available at approximately 6 weeks.[10]

More information & references

Where can I find more information?

Visit **https://www.nhs.uk/conditions/genetic-and-genomic-testing/** for more information on genetic testing.

References

Links to external websites are provided as a resource to the viewer. Pfizer Ltd. accepts no responsibility for the content or services of the linked site other than the information or other materials relating to Pfizer medicines or business which it has provided or reviewed.

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