Pathway

Olivia.
Your Ovarian Cancer Support

Gene Testing

Each one of us is unique, as a result of small differences in our DNA - the biological code that gives instructions to every cell in our bodies. DNA consists of two strands, running in opposite directions, that form a shape like a twisted ladder.[1]

Your DNA determines the colour of your eyes, your hair and skin, but it also controls the smaller-scale activities, such as sending chemical messages between individual cells. Sections of DNA that share a purpose are called **genes**.

The same gene, serving the same purpose, can be different between two people – it all comes down to what gets passed on from your parents. You get half your genes from each parent.[1]

Our genes can change ("mutate") either randomly on their own, or due to things they are exposed to, e.g., cigarette smoke, that can damage our DNA. [1]

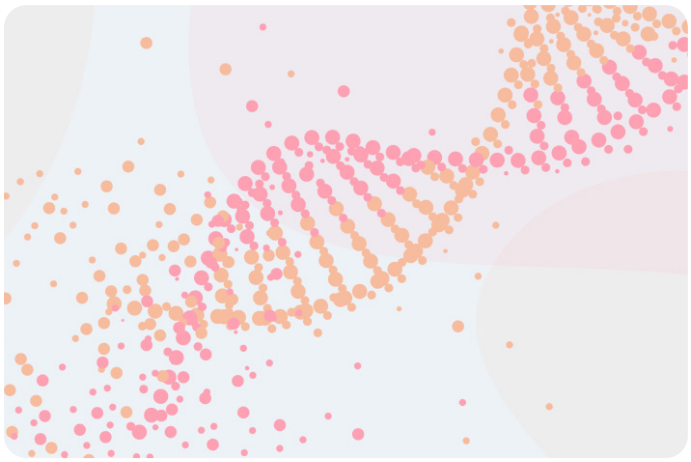
Recent advances in gene mapping technology allow researchers to identify genes that are associated with an increased risk of cancer. This has also helped in the development of targeted treatments that work on tumours with specific mutations.[2]

BRCA

One such mutation is known as BRCA. BRCA stands for BReast CAncer. This gene was first recognised in connection with breast cancer, and that is where it gets its name. However, it is now recognised that BRCA is also associated with ovarian, prostate and pancreatic cancers.

The BRCA 1 and BRCA 2 genes contain instructions to create chemicals that help repair DNA damage in the cell and thus ensure the normal functioning of cells. Sometimes one of the BRCA genes mutates, causing DNA repair to stop working, which can lead to BRCA-positive tumour formation.[3]

If the mutation occurs in germ cells (eggs or sperm), the mutant gene can be passed on to children and increase their risk of developing ovarian, breast, prostate and pancreatic cancers. In that case, the mutant BRCA would be present in every cell in their body rather than just the tumour cells.[3]



Significance of BRCA:

- If you have a family history of breast, ovarian, pancreatic, or prostate cancer, on either side of your family and especially at a young age, you may wish to speak to your doctor about having genetic testing for the BRCA mutation because it can be inherited. Genetic testing will include genetic counselling to help you decide on the best course of action for you, if it is confirmed that you have the mutation.
- If you have already been diagnosed with cancer, it is important to know whether you have the BRCA mutation because it may open up different treatment choices. Results of BRCA testing can be used to personalise treatment options for people with ovarian cancer.[4,5]

Homologous recombination deficiency

DNA repair is an ongoing process in cells that is essential for survival. DNA damage can be caused by the chemicals produced naturally by breathing or by external environmental factors such as UV rays, other radiation, synthetic chemicals, toxins, etc., resulting in DNA damage throughout your body

every day. Fortunately, there are several automatic repair systems within our cells depending on the type of damage.

These repair systems (such as the ones managed by the BRCA genes) scan the strands of DNA for errors and try to fix any that they find.

When both strands are damaged, the chemical process to repair them involves gluing the two ends of the broken strands back together. If this is done incorrectly, it can actually result in new mistakes and cause more problems. If your cells can't perform double-strand repair, it's known as homologous recombination deficiency or HRD.[6]

The repair systems are particularly important when a gene that controls cell growth has been damaged. If it isn't fixed, that gene can be switched on permanently and lead to cancer.

As with BRCA, knowing your HRD status can help your healthcare team personalise treatment options for you.

References

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Ovarian Cancer Commitment

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