Genomics and Cancer

10 top tips

Useful Resources:

- Family history, genes and cancer risk
- Pre-cancerous and genetic conditions
- Other inherited conditions that can increase cancer risk
- Genetic testing for cancer
- Genetic counselling

This edition: January 2026
Next planned review: January 2029

We make every effort to ensure the information in these pages is accurate and correct at the date of publication, but it is of necessity of a brief and general nature, and this should not replace your own good clinical judgement, or be regarded as a substitute for taking professional advice in appropriate circumstances. In particular check any drug doses, side-effects and interactions. Save insofar as any such liability cannot be excluded at law, we do not accept any liability in relation to the use of or reliance on any information contained in these pages, or third-party information or websites referred to in them.

Macmillan Cancer Support, registered charity in England and Wales (261017), Scotland (SC039907) and the Isle of Man (604). Also operating in Northern Ireland.
MAC19358_Managing_Sexual_Dysfunction_tips

1 As the availability of genomic testing increases within the NHS and private sectors, part of a GP's role will incorporate a clinical understanding of genomics. Primary Care are likely seeing an increasing number of patients presenting to them with questions about their personal risk or test results, those with a family history of cancer who may benefit from genetic testing, and those undergoing treatment who are seeking advice about targeted therapies.
GPs need to have the resources and skills to deliver genomic conversations, to identify those at risk of a genetic condition, and to clinically manage genetic condition.
The **NHS England's Genomic notes for clinicians website** is a useful resource that contains clinical case studies of genomics in **primary care**.

2 Cancer is a disease of the genome. Now that next-generation sequencing technologies are available, genomics has rapidly become an integral part of cancer care and a priority for **Government strategy**.
Its clinical significance will only increase in time as we use genomic information to develop a better understanding of the causes of cancer, how to treat it and how to prevent it.
Related pages: Demystifying genomics in cancer care

3 A copy of the genome (all of someone's DNA) is found in nearly every cell in the body. Genomic changes in a person's DNA, also called variants, are either acquired over time or inherited/hereditary (germline).

Acquired variants that build up over time are the most common cause of cancer. These are usually caused by "wear and tear" damage on genes over time like: aging, exposure to hormones, smoking, environmental toxins and certain viruses.

Germline variants: these changes are present in every nucleated cell in the body and have been since birth. These changes can be passed on to the next generation, so may have an impact on family members.

4 Germline variants can lead to around 5% to 10% (1 in 10) of cancers.

Commonly known examples are:

- Breast or ovarian cancer associated variants in the BRCA1, BRCA2, and PALB2 genes
- Or familial cancer syndromes, such as Lynch Syndrome.

This has an impact on the treatment of the patient with cancer, but also on their relatives. It is important to initiate cascade genetic testing to family members who are at risk of inheriting a genetic variant that has been identified. This will enable family members to access preventative interventions, such as increased monitoring, screening, or treatment.
Related pages: Genetic testing for cancer

5 Most changes within the genome are harmless, and do not lead to disease. However, there are a small number of inherited variants in the genome that can increase an individual's risk of cancer over their lifetime. These result in a higher than usual chance of recurrence after primary cancer. For example, people who have a CDK4 variant have a high lifetime risk of **Melanoma** , which is up to 80% compared to a population risk of 2.5%. **Related pages:** Family history, genes and cancer risk

6 Each germline variant is associated with a different risk of developing cancer in a person's lifetime, even within the same tumour grouping.
For example, within the breast cancer predisposition genes, those with **a BRCA1 or BRCA2** variant carry a 69% to 72% lifetime risk of breast cancer. However, those with a PALB2 variant only have a 44% to 63% lifetime risk

7 Somatic variants only occur in the non-germline cells of the body. Therefore, they cannot be passed from parent to child.

Changes in these cells can be caused by exposure to environmental factors over a person's lifetime. For example, damaging UV rays from the sun cause changes in the base pairs of DNA within the cell. These changes can accumulate over time which can lead to uncontrolled cell growth and tumour formation.

8 Analysing the genome of a tumour is used to make a precise diagnosis. This enables a more accurate prognosis and better tailored treatment.

Increasingly, drugs are available that are targeted to the genetic features of a cancer, requiring genetic testing of the cancer cells to determine potential response. **Related pages:** Targeted therapy

9 It is important for GPs to understand the concept of germline testing for hereditary predisposition to cancer and somatic testing and its role within targeted therapies by identifying a tumour's 'genetic signature'.

This is because language is important. When patients hear the term "genetics linked to their cancer diagnosis" this can generate considerable anxiety. It may lead to an inappropriate referral for testing of family members when the reference is to somatic testing.

Our Hallmarks of Heredity Risk: Our Hallmarks of Heredity Risk: When to Test poster is a helpful resource for GPs to understand when a patient may need to be referred for further testing.

10 Macmillan has information which can be shared with patients to support discussions about family history and genomics. Below we have included links to pages and information booklets that may be useful. See the links on the green ribbon.

MACMILLAN CANCER SUPPORT