

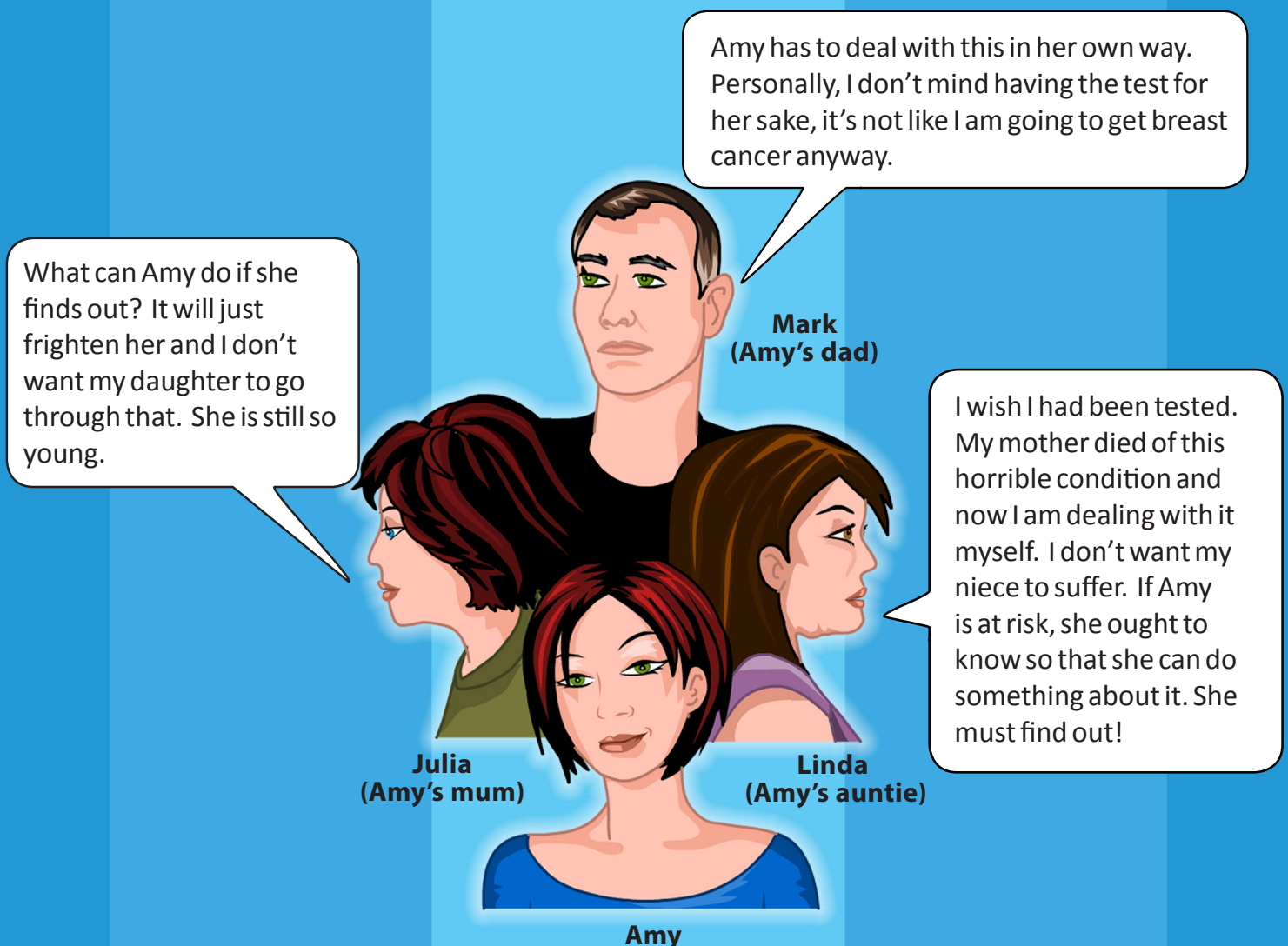


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Amy is just 15. Her family has been rocked by the recent news that Amy's Auntie Linda has breast cancer. Amy's father Mark is especially worried about his sister Linda. Mark and Linda lost their mother to breast cancer shortly before Amy was born. A genetic test has revealed that Auntie Linda has a mutation of her *BRCA1* gene, which increases a person's risk of developing breast cancer.

Amy wants to know if she also has the *BRCA1* mutation, but the standard procedure would be to test her father Mark first to see if he has the *BRCA1* mutation. If this is confirmed Amy could then have the test, as there is evidence that a direct relative carries the mutation. Mark is willing to be tested but Amy's mother, Julia, is against it. Julia thinks Amy is too young and is concerned about how Amy would react if she found out she was at risk of having the *BRCA1* mutation. Mark will only do the test if Julia and Amy can come to some agreement. Amy leads a healthy lifestyle – eating healthily and exercising regularly – but she is willing to do anything to avoid getting breast cancer.

**Should Amy insist on her father Mark being tested for the *BRCA1* mutation?**





Is it better to find out as early as possible or  
is Amy too young to cope with this type of  
information?

If Mark takes the test, who should have access  
to his results?



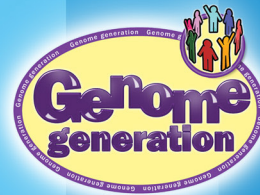
If Mark discovers that he does not have the *BRCA1* mutation, how will that impact Amy and the rest of the family? What does Amy gain from knowing that information?

If Amy discovers she may be at risk, what should she do next? For example, should she have the test for the *BRCA1* mutation herself?



If Mark discovers that he does have the *BRCA1* mutation what does that mean for him?

Should this test be freely available to everyone through national health services?



## The genetics of breast cancer

Mutations in the *BRCA1* and *BRCA2* genes are strongly linked to breast cancer risk. They are also linked to an increased risk of ovarian cancer.

- Women with a *BRCA1* mutation have a 75-85% lifetime risk of developing breast cancer.
- Women with a *BRCA2* mutation have a 50-80% lifetime risk of developing breast cancer.
- Fewer than 1 in 500 women carry a high-risk mutation for breast cancer.

If a person carries a mutated copy of the *BRCA1* or *BRCA2* gene, it does not mean they will definitely develop cancer.



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## Our genes

Most of the DNA in human cells is packaged into 46 chromosomes, in 23 pairs. Our genes are arranged along these chromosomes and carry the instructions to make molecules, such as proteins. To function properly, each cell depends on thousands of proteins doing the right job, in the right place, at the right time. Our complete set of DNA, containing all the instructions required to make us, is called a genome.



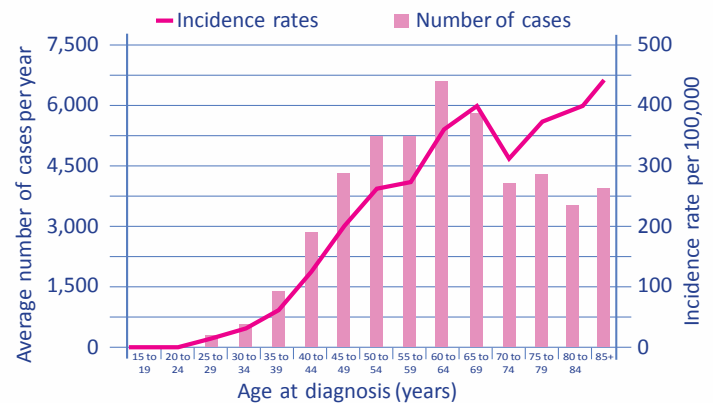
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## How common is breast cancer?

1 in 8 women will get breast cancer by the time they are 80. Because breast cancer is so common, having more than one relative with the disease does not necessarily mean that the family carries a higher genetic risk. The highest risk factor (after being female) is age. 80% of breast cancers occur in women over 50.

Number of new cases and incidence rates of breast cancer in the UK 2007-2009

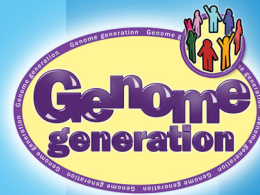


Source: Cancer Research UK



## Gene mutations

A mutation is a permanent change in the DNA sequence. Mutations come in a number of different forms and can change the functioning of our genes. Some mutations may lead to a difference in the amount, or structure, of a protein produced by specific genes. Some mutations may switch genes on at the wrong time, preventing a protein from doing its job, or changing the way it does its job.



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## Health care advice for breast cancer

All women, regardless of their family history or genetic risk, are advised to:

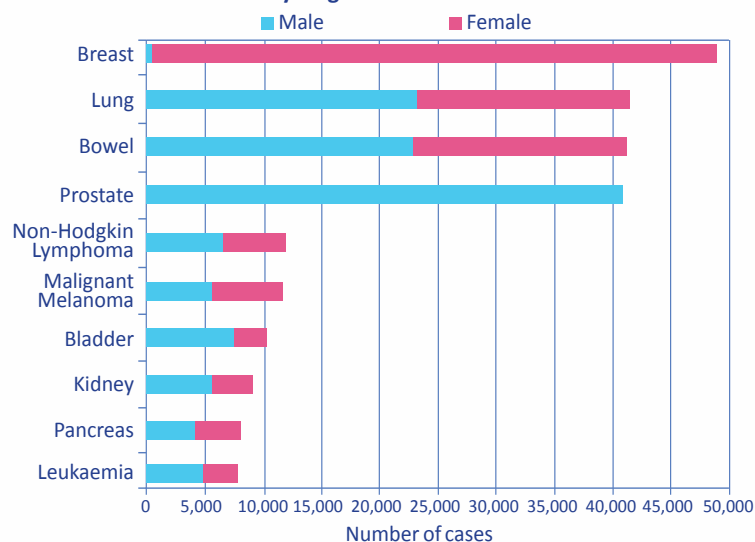
- examine their own breasts and report any changes immediately
- eat a healthy diet and keep fit
- maintain healthy weight, especially after menopause
- moderate alcohol consumption.



## Breast cancer statistics

Breast cancer is the most common cancer in the UK and represents 16% of all diagnosed cancers. In 2009, 48,788 people in the UK were diagnosed with breast cancer. There were 11,633 deaths from the disease in the UK in 2010.

Ten most commonly diagnosed cancers in the UK in 2009



Source: Cancer Research UK



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## Tumour suppressor genes

*BRCA1* and *BRCA2* are examples of tumour suppressor genes, which play an important role in preventing cancer. Normally, if the cell's DNA is damaged, these genes halt cell division or promote cell death. We have two copies of every gene – one inherited from each parent. As long as you have one functioning tumour suppressor gene it does not matter if the other is mutated. However if the functioning copy also becomes mutated then cancer may develop.



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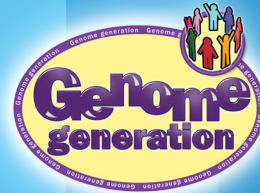


## Breast cancer in men

Breast cancer is not only a threat to women. Men can get breast cancer, although it is much rarer, accounting for less than 1% of cases (around 300 per year in the UK). Lower awareness of male breast cancer means it tends to be detected at a later stage in men than in women, which means that the outcomes tend to be worse.



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## Breast cancer screening in at-risk families

In genetic screening, a living relative with breast cancer has to be tested first. If they have one of the mutations associated with the cancer, other family members can be tested for the same mutation.

Genetic testing of under-16s for diseases that occur in adulthood is not normally advised. In this scenario, it would be clinical practice to test the father (who has a relative with the gene mutation) first.



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## Monitoring women at high risk

Women with a family history of breast cancer are categorised as either 'raised risk' or 'high risk'. Less than 1% of women are at high risk. Women at high risk aged between 20 and 49 may receive yearly MRI screening and will be offered genetic counselling. Some women choose to remove all of their breast tissue as a preventive measure, a procedure known as a pre-emptive mastectomy.



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## Our DNA

Our bodies are made up of around 50 trillion cells. Most of these cells contain a person's genetic information within a long molecule called DNA (deoxyribonucleic acid). This DNA contains the genetic information that we inherit from our parents.

DNA is made up of specific sequences of letters or bases that encode instructions on how to make proteins. These bases [Adenine (A), Guanine (G), Cytosine (C) and, Thymine (T)] are lined up along two strands of DNA. The DNA is coiled into a double helix structure that resembles a twisted ladder.



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## What is cancer?

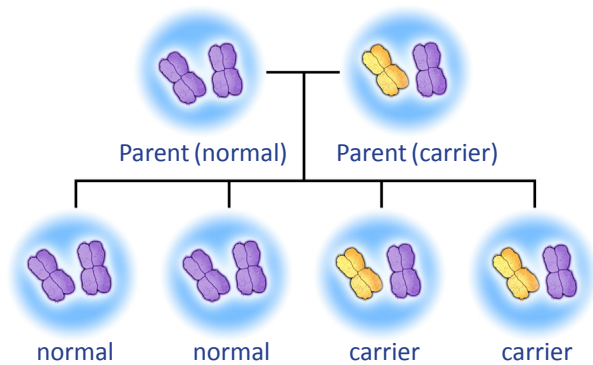
All cancers are genetic diseases. Cancer develops from single cells that continually divide in an uncontrolled manner. Cancer cells behave in this abnormal way because of changes (mutations) in the DNA sequence of key genes, which are known as cancer genes.

The changes in the DNA provide a growth advantage to the cell. This means that the cancer cell and its offspring divide at a faster rate than that of their neighbours. This results in tumour formation, invasion of surrounding tissue and, if left untreated, eventually 'metastasis' where the cancer spreads to other parts of the body.



## What is the risk of inheriting a *BRCA* mutations?

The mutated *BRCA1* and *BRCA2* genes are inherited in an autosomal dominant pattern. This means that a child only needs to inherit one copy of the mutated gene to have an increased risk of developing cancer. If one of the parents is a carrier of a mutated *BRCA1* or *BRCA2* gene, each of their children has a 1 in 2 (50%) chance of inheriting the mutated gene from the carrier parent.



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