

## About hereditary breast and ovarian cancer :

- Hereditary breast cancer and ovarian cancer are rare forms of these diseases, accounting for around 5-10 per cent of all cases. Men in affected families have a slightly increased risk of prostate cancer, and an increased risk of breast cancer.
- Hereditary breast and ovarian cancer is caused by an inherited change (a mutation) in any one of several different genes. The two major genes identified so far are called BRCA1 and BRCA2. People with a BRCA1 or BRCA2 gene mutation have a 50 per cent chance of passing it on to each of their children.
- BRCA1 and BRCA2 gene mutations increase a woman's lifetime risk of developing breast cancer from around 12 per cent to between 50-85 per cent. The risk of developing ovarian cancer rises from 1.5 per cent to 40-60 per cent in women with BRCA1 mutations, and 10-15 per cent in women with BRCA2 mutations.
- Early diagnosis is important for detecting cancer as it increases the chances of successful treatment. People who are known to be at risk of breast cancer are advised to carry out regular self-examination, and have an annual mammogram. For women with a very high risk of developing breast or ovarian cancer, preventative surgery can be offered.
- Once a BRCA1 or BRCA2 gene mutation has been identified in an affected person, a genetic test may be offered to other family members who might have inherited the condition. However, some of the genetic changes that cause hereditary breast and ovarian cancer have not yet been discovered, and so not all affected families can be offered this type of test.

## Find out more

To find out more about hereditary breast and ovarian cancer, and for information and support, contact:

Breast Cancer Care helpline free on  
0808 800 6000 (textphone 0808 800 6001)  
or visit [www.breastcancercare.org.uk](http://www.breastcancercare.org.uk).

BREAST CANCER CARE

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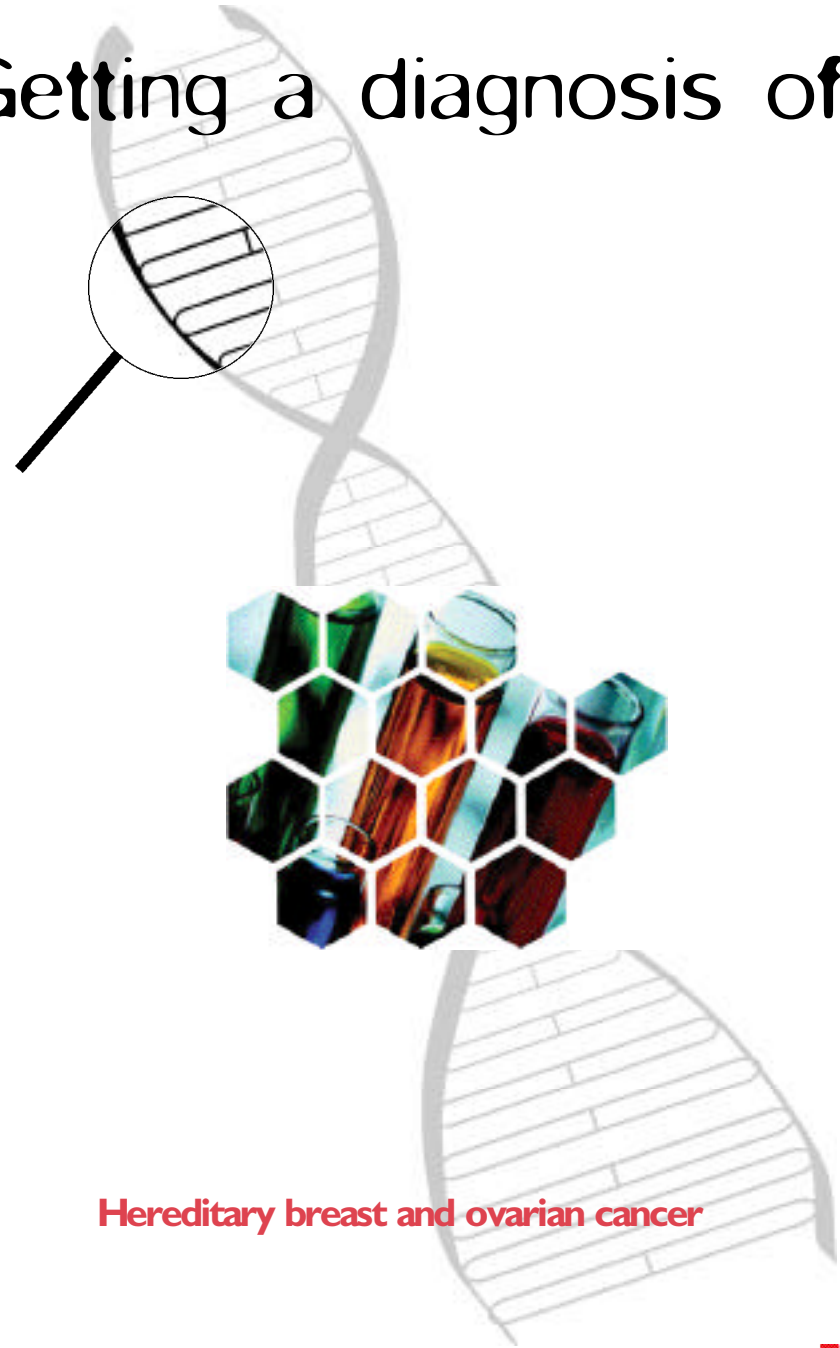


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# Getting a diagnosis of...



## Hereditary breast and ovarian cancer



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## BRCA

### Who should have a Breast Cancer (BRCA) test?

Most cases of breast and ovarian cancer are not inherited. However, two genes have been identified that confer susceptibility to breast and ovarian cancer in a small minority of cases - BRCA1 and BRCA2 – and they are found on chromosomes 17 and 13 respectively. These genes code for proteins that can repair errors occurring during the process of copying DNA as cells are dividing to form new cells.

People have two copies of nearly every gene, one inherited from their father and one from their mother. Women with inherited forms of breast cancer caused by BRCA1 and BRCA2 inherit one faulty copy of the gene; if the second gene copy then becomes damaged, any errors occurring in the DNA in that cell cannot be corrected. An accumulation of errors within the DNA leads to the development of a cancerous cell and eventually a tumour.

Less than 10% of all breast cancers are caused by mutations in the BRCA-1 or BRCA-2 gene, and the current opinion is that only selected groups of women should have the BRCA test. The likelihood of developing cancer when a BRCA1 or a BRCA2 mutation is found is variable. Therefore, the BRCA test is currently not recommended as a screening test for all women. However, the test can be particularly useful for women that already have or have had breast cancer or where breast or ovarian cancer has occurred in a woman's family.

In this case, it can be useful to check whether she is carrying a mutation of one of the two genes. If so, this has implications for the frequency and intensity of further preventive medical checkups, or even for preventive surgery. It would also be useful to test relatives. The risk of developing breast cancer when a mutation is present by the age of 70 is significantly higher than in the rest of the population.



Annette lives in Essex with her husband and two children. She was treated for breast cancer when she was thirty, and nine years later

she has inherited a tendency to develop both breast and ovarian cancer.

*'We had a family history of breast cancer and ovarian cancer – my grandmother had ovarian cancer and my mum had breast cancer, followed by ovarian cancer...they both died when they were 49. A year after mum died, I discovered that I had breast cancer so there seemed to be a family link..., I consequently had the standard treatment that you would have for breast cancer.'*

Several years later, Annette's sister asked her if she would be prepared to give blood for a genetic test, to find out if an inherited genetic change was responsible for all the cases of cancer in the family:

*'They need the blood of someone who's had the cancer for them to identify the gene.... I assumed that if there was a gene I'd be carrying it, but I thought no more about it. When I went up there to help my sister, they told me there are two genes that they've identified so far, and one was linked to breast cancer and in a small percentage of cases ovarian cancer – that was BRCA2 – and BRCA1, was linked to breast and ovarian cancer. So then the pieces started to fit together.'*

After receiving genetic counselling about the implications of taking a test for hereditary breast and ovarian cancer, Annette decided to find out if she had inherited a BRCA1 mutation:

*'After 4 months I heard from them...the counsellor was there with the consultant, and they told me that I was carrying the gene, BRCA1. And so suddenly I was faced with...a big decision...I'd already had breast cancer, and I'd had a lumpectomy, not everything removed but quite a bit, and gone through all this...and suddenly I'm facing something else which I wasn't expecting'.*

Annette was advised to have a hysterectomy, to remove the risk of ovarian cancer, which is very difficult to detect in its early stages. She continues to receive regular check-ups, and has a mammogram (breast examination) every year. Following the identification of the BRCA1 mutation responsible for Annette's cancer, it was possible to find out if her sister was also at risk:

*'It turned out that the day after my hysterectomy, when I was just waking up, my sister received the result of her test. It was much easier to find the faulty gene after they'd identified it in one person in the family, so they picked it up very quickly. The day after my operation, she got her result, and she was free of it, she hasn't inherited it. So we were both 'clear' on the same day really'.*

Despite being initially 'devastated' by the BRCA1 test result, Annette says she is pleased that she was then able to have preventative surgery, which has cut her lifetime risk in half, to around 32 per cent:

*'Because I've had as much as I could have had done, I probably feel more comfortable, knowing that I won't die the same way as my mum did – the ovarian bit is out the way. I know I'm at risk of breast cancer, but I always was...I developed it when I was very young and I've learnt to live with that more'.*