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Case studies: gene testing for ovarian cancer

A BRCA testing pathway developed by the Mainstreaming Cancer Genetics Programme could save the NHS £2.6 million a year.

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[The Royal Marsden NHS Foundation Trust](#) [6] offers [the gene test](#) [7] as standard at patients' routine cancer clinic, rather than having to be referred to a separate genetic testing clinic.

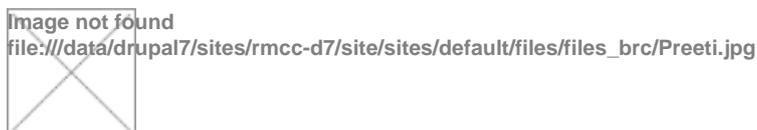
This can improve and personalise the treatment of women diagnosed with ovarian or breast cancer and also provides information useful for their relatives about their own risk of developing these cancers.

Five women share their experiences of genetic cancer testing:

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- [Simplifying gene testing access for women with ovarian cancer improves treatment choices and could save lives](#) [7]

Preeti Dudakia, 49



Preeti had suffered from abdominal pain since 2007, and underwent operations to remove cysts at her local hospital. But by the end of 2013 she was in unbearable pain and had to go

back to the local hospital.

In 2014, she went to The Royal Marsden and was diagnosed with stage three ovarian cancer and had a full hysterectomy the following week, followed by chemotherapy.

Preeti was offered BRCA testing that summer and was found to have BRCA1 mutation, and has now been on the SOLO1 clinical trial for two years.

Her mother died from ovarian cancer. Her three brothers were tested but don't have the mutation. Preeti said:

When I was offered the BRCA testing it was an easy decision for me to make. I already had cancer but didn't want my brothers, their children, or anyone else in the family to go through the same thing. If they did have the mutation they could make a decision about what to do.

When I found out that they didn't have the mutation it was such a relief because I now know their children won't go through this.

I already had cancer but didn't want my brothers, their children, or anyone else in the family to go through the same thing?

Elaine Wolk, 55

207

ovarian cancer patients on the study accepted the offer of BRCA testing

Elaine was diagnosed with ovarian cancer at the age of 45, and had a hysterectomy, chemotherapy and took part in a drug trial.

She had no family history of breast or ovarian cancer at that time so was not offered testing, but was offered the test through the Mainstreaming Cancer Genetics programme eight years later.

It was at this point she found out she carried BRCA2 mutation, which has subsequently been identified in her father.

Her mother doesn't want to know if she is carrying the gene and her brother lives in New Zealand, so neither of them have been tested.

Elaine is currently considering having a bilateral risk-reducing mastectomy to manage her breast cancer risk in the future. Elaine said:

I'm really glad I had it done and think it should be mainstream for everyone. I know a lot of people are hesitant but isn't it better to know so you can do something about it?

When it comes to your health it's the most important thing, without your health everything else is meaningless.

The next big decision for me is to decide whether to have a mastectomy or to carry on taking

drugs for the next five years. I think there's only one option but I'm in no rush at the moment.

I have a clear vision of what I'm going to do going forward and I'm lucky enough to have an amazing network of friends. Having the test was the best thing I have ever done. I know there's that chance of getting breast cancer, so I have the opportunity to make some decisions.

The new genetic testing pathway has been a huge success in the clinic. It all runs very smoothly and I know that patients and their families are really most appreciative.

Professor Martin Gore, Consultant Medical Oncologist at The Royal Marsden NHS Foundation Trust

I'm really glad I had it done and think it should be mainstream for everyone?

Carolyn Farrar, 53

Carolyn was diagnosed with breast cancer in 1999. She had a bilateral mastectomy at The Royal Marsden but BRCA genetic testing wasn't available at the time. However, there were suspicions that it was related to genetics, as both her mum and great grandmother died from breast cancer.

Carolyn was diagnosed with advanced ovarian cancer in April 2013 - she had absolutely no symptoms at all. She was offered the BRCA testing and said it was a 'no brainer' to be tested. She had chemotherapy and surgery and went into remission. She found out that she has the BRCA1 mutation and has been on the SOLO1 clinical trial.

Both of Carolyn's daughters have the BRCA1 gene. One daughter had a bilateral mastectomy aged 26, and her 21-year-old daughter, Georgia-May, will have the same surgery early next year once she graduates. Carolyn said:

"I was one of the first patients to have the BRCA gene test done at The Royal Marsden outpatients clinic. It was an absolute no brainer to do the genetic test because I was thinking about my two daughters, and they both decided they wanted it too after seeing me go through cancer treatment.

It's enabled us to make decisions instead of having to wait - if you get cancer the decision is taken out of your hands.

Being pre-warned about the risk of getting cancer means my daughters could decide for themselves on whether to have a preventive mastectomy.

7,000

women a year are diagnosed with epithelial ovarian cancer in the UK

?It?s enabled us to make decisions instead of having to wait - if you get cancer the decision is taken out of your hands?

Georgia-May Farrar, 21

£2.6m

could be saved if this testing pathway was implemented nationally

Georgia-May, Carolyn Farrar's youngest daughter, had the BRCA gene test aged 19, where it revealed she had the BRCA1 gene.

She has decided to go down the preventative route and is due to have a double mastectomy early next year after she has graduated from university Georgia-May said:

?Having the test done was an easy decision because I knew I had a high risk of getting cancer. Initially, I thought that it would be either me or my sister with the mutation, because I was told there was a 50% risk, so it was quite a shock when the results showed that we both had it.

?For me it?s been quite a hard decision on whether to have the surgery, but it?s better than the alternative ? because my mum had cancer it was like looking into the future.

?Having the test is scary and is something that?s hard to deal with, but by having all the available information you gain back control and can plan ahead.?

?Having the test done was an easy decision because I knew I had a high risk of getting cancer?

Lesley Leahy, 51

Lesley was diagnosed with advanced ovarian cancer aged 47. She was given nine weeks of chemotherapy, surgery and more chemotherapy, but after six months her cancer returned. It was following this that she was offered BRCA testing which revealed she has the BRCA2 mutation.

She was not known to have a family history of breast or ovarian cancer prior to testing, so would not have been offered testing previously.

She is currently receiving olaparib after responding well to more chemotherapy, but it is early days as this is only going into her second month on the drug.

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