ovarian cancer action

A policy report by
Ovarian Cancer Action

Acting on BRCA: Breaking down barriers to save lives.

Acting on BRCA Contents

Foreword.

Around 15% of cases of ovarian cancer are linked to BRCA - this equates to over 1,000 women a year in the UK whose lives could potentially be saved.

Ovarian cancer claims the life of a woman every two hours. Treatment lags behind other, better known, cancers and survival rates remain low. BRCA testing is one of our strongest weapons in the fight against this disease. Since 2013, Ovarian Cancer Action has been campaigning for all women with non-mucinous epithelial ovarian cancer to be offered testing for the BRCA gene mutation at the point of diagnosis.

In July 2015 our campaign had success with the introduction of a new NHS England Clinical Commissioning Policy recommending women with ovarian cancer be offered testing at the point of diagnosis. That same year we published our 'Acting on BRCA' policy paper. We set out what we wanted the NHS to provide for those diagnosed with a BRCA gene mutation and their families and what we as a charity would do.

Two years on from our last paper, there's been positive steps forward in BRCA testing around the country. The landscape is shifting with more families becoming aware of hereditary cancer, in part thanks to the 'Angelina Jolie effect', and the next generation acting early to prevent cancers that they have watched the women in their families suffer from.

As well as a tool for cancer prevention, BRCA testing has become a crucial part of the ovarian cancer treatment pathway. New drugs have been made available across the UK for women with the BRCA gene mutation who have relapsed ovarian cancer, with discoveries that those with the gene mutation respond better to certain treatments.

When the Independent Cancer Taskforce was setting out its bold strategy for the next five years, we took action to make sure BRCA gene testing was included. It was an important step forward when "Achieving world class cancer outcomes: a strategy for England 2015-2020" included a recommendation that 'all women with non-mucinous epithelial ovarian cancer are offered testing for BRCA1/BRCA2 at the point of diagnosis'.[1]

Despite all this, we were hearing from many of our supporters that they were still facing barriers in accessing BRCA testing. From misinformation in primary care to regional policies restricting testing, these barriers are still standing in the way. The consequences of these barriers can be devastating, not just for the individual, but for whole families, as we know from Rosie's story (p17).

With so many stories like Rosie's, we wanted to discover what the picture of BRCA testing really looked like around the UK. We spoke to hundreds of people up and down the country about their experiences and found that despite the Clinical Commissioning policy in place, there are still many eligible women not being offered a BRCA test when they are diagnosed with ovarian cancer. Individuals with significant family history are also struggling to be referred for testing.

The NHS is still missing the opportunity to use BRCA testing as an effective cancer prevention strategy, and women are dying as a result. With the Clinical Commissioning Policy in place, no one eligible should slip through the net or be denied testing. The Government must ensure that the Policy is being uniformly implemented across the country.

Our report highlights seven areas where the Government must take action, from making sure those who are eligible are offered testing, to providing every patient with good quality information to help them make life-changing decisions. We've made recommendations in each of these areas to improve services and fulfil BRCA testing's potential as a cancer prevention tool.

Over the years we've heard wonderful success stories where BRCA testing has allowed young women to make informed decisions about managing their risk, and prevent cancers from occurring in the first place. These women had the knowledge that everyone with a family history should have access to, giving them the power to change their future. Their stories inspire us to keep campaigning until every woman and family are given the same opportunity.

The landscape around BRCA testing is always moving. New drugs are coming onto the market and ground-breaking research findings are constantly changing what it means to have a BRCA gene mutation. The future in managing hereditary cancer is exciting and ever-changing. The Government needs to be responsive to these changes and always ensure women have access to the best quality treatments and up to date information.

At Ovarian Cancer Action we're on a mission to stop women dying before their time. We'll continue to take action on BRCA testing until it fulfils its potential to save thousands of lives.

Katherine Taylor,
Chief Executive

"As well as a tool for cancer prevention, BRCA testing has become a crucial part of the ovarian cancer treatment pathway."

Executive summary.

From our research, we have made a series of recommendations for the Government to go further to act on BRCA and save lives.

At Ovarian Cancer Action, we've long campaigned for BRCA gene mutation testing to be used by the Government as a cancer prevention tool. 2015 saw the introduction of the Clinical Commissioning Policy recommending testing at ovarian cancer diagnosis. This was a great start - but our campaign hasn't stopped there.

Since then we've carried out research, collected stories, visited clinics and more to find out what the picture of BRCA testing really looks like in the UK today. This report is the outcome of our research.

There's been improvements in provision of BRCA testing around the country. However our key findings show that much more needs to be done. People up and down the country are still facing barriers to access testing and reduce their risk of cancer. As a result women are still developing ovarian cancer that could, and should, have been prevented.

From our research, we've identified seven priorities for the Government and the NHS. Throughout the report we've made recommendations in each of these priorities to break down these barriers and take BRCA testing forward. We are calling on the Government to take action on preventable cancers in the future and stop women dying before their time. ■

KEY FINDINGS

Priority 1 - Testing
29% of women diagnosed with ovarian cancer are not being offered BRCA testing.

Priority 2 - Counselling
33% received no counselling before deciding whether to go ahead with BRCA testing.

Priority 3 – Standardised information
34% received no information about choosing the right cancer risk reducing procedure for them.

31% were not fully informed of their HRT choices.

28% received no information about fertility options.

36% received no resources to help them speak to their families.

Priority 4 – Waiting times
Waiting times for risk-reducing surgery ranged from 2-104 weeks.

Priority 5 – Follow up
The NHS is not keeping in touch with those with a BRCA gene mutation after testing.

Priority 6 – Men
83% of men have never heard of a BRCA gene mutation.

Priority 7 – Devolved nations
There is a lack of data from Wales, Scotland and Northern Ireland.

What are BRCA gene mutations?

Genes are the instruction manual that defines how our bodies work. A mutation in our genes is like a spelling error in that manual, which alters the function of the gene.

Every individual has BRCA1 and BRCA2 genes and we inherit two copies, one from each of our parents. They are called tumour suppressor genes and their job is to repair damage in cells and prevent them from growing and dividing too rapidly. Mutations in these genes can cause cells to become abnormal and grow in an uncontrolled way and increase a risk of both ovarian and breast cancer, as well as other cancers. Around 17% of cases of ovarian cancer are linked to BRCA gene mutations.

It is thought that, in the general population, around 1 in every 400 to 1 in every 800 people carry a BRCA1/2 gene mutation. This figure is significantly higher in certain backgrounds, including Ashkenazi Jewish and those from Dutch, Icelandic, Norwegian, Polish and Swedish ancestry.

These gene mutations can be inherited and passed on from a mother or father. When a mutation is present in either of the BRCA1/2 genes, there is a 50% chance of passing it on to any children. Men have an equal chance of inheriting and passing on BRCA gene mutations as women. ■

ESTIMATED LIFETIME CANCER RISK (UP TO AGE 70)

Type of cancer	General population	BRCA1	BRCA2
Ovarian cancer	2%	40-60%	10-30%
Breast cancer in women	11%	60-90%	45-85%
Breast cancer in men	0.1%	0.1-1%	5-10%
Prostate cancer	12%	~10% similar to normal population	20-25%
Pancreatic cancer	1.40%	~3%[2]	~5%[3]

Source: The Royal Marsden NHS Foundation Trust[4]

BRCA as a cancer prevention opportunity.

When a family member has an identified BRCA gene mutation, other family members can also be tested and access screening and cancer risk-reducing options.

Reducing ovarian cancer risk

- Risk-reducing bilateral salpingo-oophorectomy: surgical removal of the ovaries and fallopian tubes. Surgery will reduce the risk of developing ovarian cancer to less than 5%. This surgery triggers immediate menopause.
- Screening: there is currently no cancer screening programme for ovarian cancer. CA-125 blood tests and transvaginal ultrasounds are currently offered for monitoring. Vital research is ongoing into an ovarian cancer screening tool.
- Lifestyle: Long-term use of the combined contraceptive pill can offer some risk reduction for ovarian cancer in all women. However, it is thought that this also comes with a slight increase in breast cancer risk.

Reducing breast cancer risk

- Risk-reducing bilateral mastectomy: surgical removal of breast tissue, with reconstruction if desired. Surgery will reduce the risk of developing breast cancer to 5-10%.
- Earlier/additional screening: MRIs are available for BRCA+ women from the age of 30, and mammograms from the age of 40. These options do not prevent cancer, but aim to detect any cancer that may develop at the earliest possible stage.
- Chemoprevention: tamoxifen or raloxifen can be prescribed to reduce the risks of breast cancer developing. However, there are side effects which must be considered and chemoprevention is not suitable for all women with a BRCA gene mutation.

The next generation

- Preimplantation genetic diagnosis (PGD) is available on the NHS for those with a BRCA gene mutation who are planning a family and have not yet had any children. Through IVF, embryos are screened for the gene mutation and non-mutated embryos are implanted. This prevents any risk of the BRCA gene mutation from being passed to the next generation. ■

MORE INFORMATION

More information about risk reducing options and family planning can be found at **www.ovarian.org.uk/brca**

CASE STUDY
ALISON AND GABY

Knowledge is power.

"Before my diagnosis, I did not know that you can inherit a BRCA mutation from your father."

Alison: "I always presumed (incorrectly) that breast and ovarian cancer risk was passed down the maternal side.

If I had known about my BRCA mutation, my story would be one of inspiration, of a "Previvor", someone who has had preventative surgery and stopped cancer before it starts.

But sadly, we discovered all too late that I am a BRCA1 gene mutation carrier after I was diagnosed with stage 4 ovarian and breast cancer in July 2014- a shocking diagnosis. However, what I found most upsetting, was that my cancer could have been prevented. I could have opted for surgery, and although this surgery is extreme, it would have ultimately saved my life (at far lower a cost than the gruelling years of harsh chemotherapy). I did not have the luxury of making this choice, of having control over my future. My only option was to go ahead with the chemo. If I stop it, I will die.

Not long after my diagnosis, we discovered I had also passed this mutation down onto my wonderful daughter, Gaby. But every cloud has a silver lining. We learnt everything there was to know about the mutation, searching far and wide and seeing countless specialists.

With this knowledge came great power: the power to take control over my daughter's destiny. She is such an inspiration to other young girls who will be facing these same decisions and blows me away with her courage and great strength. I am so very proud of her.

"Taking control of my future is invigorating and frightening - but I can do it"

Gaby: "I remember the day so clearly when Mum was diagnosed with breast cancer. I was on holiday when Dad called and I felt as if I had been punched in the stomach. Mum needed surgery followed by chemotherapy.

It was after Mum's first surgery that the doctors discovered things were more serious: they discovered that she also had inoperable ovarian cancer. We were devastated, how could she have breast and ovarian cancer? But things became clearer when they my mum tested positive for the BRCA1 gene mutation. I had never heard of it but I quickly learnt. She had inherited it through my healthy, cancer-free 90 year old grandfather.

We found out that my brother and I had a 50/50 chance of inheriting it too. I wanted to get tested as soon as possible so I decided to have counselling to try and get my head around everything. I was in the park with my friend when I got the phone call from the geneticist telling me that I had the BRCA1 gene mutation too. Even though I was expecting it, it was still such a shock to have my fear confirmed.

I had a double mastectomy with reconstruction last year at the age of 26: I saw my breasts as ticking time bombs waiting to explode. When my family is complete, I will have surgery to remove both my ovaries and fallopian tubes by the time I am 35. Together these surgeries will bring the risk of developing breast and or ovarian cancer down to below the national average.

It's scary knowing what I have to do to my body but watching my mother suffer going through chemotherapy and seeing how hard she is finding battling this horrible disease, preventative surgery is the right option for me. Taking control of my future is invigorating and frightening - but I can do it." ■

What we said we'd do.

In our 2015 Acting on BRCA policy paper, we pledged to do our bit alongside the NHS to increase awareness of BRCA gene mutations, and to continue to campaign around BRCA testing. We said:

"We will develop and provide patient guide materials to help family members access existing genetic services and cancer prevention options."

We've developed our online BRCA hub to deliver extensive, high quality information for patients and family members about genetic services and cancer prevention options. We regularly review the hub both internally and externally to ensure the information is kept up to date. For those who are not online we have a printed guide to hereditary cancers which is also under regular review.

"We will lobby the government, and the devolved assemblies and parliaments to deliver a life-saving cancer prevention strategy across the UK as a whole."

We've lobbied the Government, Parliamentarians and Regional Assembly Members to implement a cancer-prevention strategy around BRCA but we believe more needs to be done by Governments across the nations in the UK to halt these preventable cancers.

"We will monitor international advice and protocols through our extensive global connections from supporting international forums."

We've worked with BRCA organisations across Europe, the USA, Australia, Israel and others, keeping up to date with international advice and protocols to make sure women in the UK are receiving the same standard of care as the rest of the world.

"We will monitor the roll out of the current NHS England Clinical Commissioning Policy to ensure consistent and high quality services are available to all BRCA families in England."

We've lobbied the Government for access to relevant data to monitor the Clinical Commissioning Policy and continue to hold NHS England to account in rolling out the policy to all corners of the country.

"We will help women understand their BRCA risk through our online risk tool."

We have developed our BRCA risk tool to help people explore whether their family history puts them at risk of ovarian cancer. The tool is regularly reviewed in line with new developments. ■

TAMPON TAX

In 2016 we were the recipients of HM Treasury's Tampon Tax grant. This allowed us to fund:

- Scientific research focussed on hereditary risk at the Ovarian Cancer Action Research Centre.
- The UK's first Ovarian Cancer Prevention Officer to focus on hereditary ovarian cancer.

Methodology.

We carried out several pieces of research across 2016-2017 to find out what the picture of BRCA testing really looks like in the UK today.

BRCA testing survey
The BRCA testing survey was used to measure variation in BRCA testing and related services around the country. The survey was open to anyone in the UK, male or female, who had any experience of trying to get a genetic test, had a cancer diagnosis or already knew they had a genetic mutation.

The online survey ran from February - April 2017. The survey was advertised on BRCA social media groups as well as Ovarian Cancer Action's own media channels. Advertisements for the survey were also placed in hospitals and oncology departments in various locations across the UK. Participants were asked to share the survey with relevant family members, with a particular focus on men to combat the expected gender bias in the sample. A prize draw was used as an incentive for participation in the survey. In total 531 participants completed the survey.

The questionnaire was designed in-house with guidance from experts Dr Jonathan Krell, Imperial College and Dr Angela George, Royal Marsden as well as patient advocate Carla Atherton. In total there were 110 possible questions, including a mix of qualitative and quantitative research to capture the sample's experiences as widely as possible.

Case studies
We contacted respondents from the BRCA testing survey and used our pool of supporters for further information about their experiences of the BRCA testing pathway. These case studies can be read throughout the report.

Family contact survey
The family contact survey was used to measure attitudes to NHS responsibility in reaching out to family members at risk of having a BRCA gene mutation.

In Autumn 2016 we commissioned research agency Populus to design and carry out two online surveys. Populus interviewed 104 Ovarian Cancer Action supporters online through a self-selecting survey. The survey was advertised through our social media channels and digital outreach. In parallel Populus also interviewed 1,093 members of the British general public online, selected through a random sample and data was weighted to be nationally representative.

Freedom of Information requests
In August 2017 we sent a Freedom of Information request to NHS England to establish their activity for monitoring uptake of the Clinical Commissioning Policy E01/pb.

Observation
In September 2017 we carried out observational research at Guy's Hospital in London at their BRCA family services BRCA clinic.

Literature review
Throughout 2016-2017 we have conducted research into the literature around BRCA testing. This has included information from studies, journal articles, international guidelines and NHS publications. When data from these sources have been used, these have been referenced and a full list of the literature reviewed can be found at the back of this report.

Parliamentary questions
With thanks to Peter Bone MP, we asked the Government for information about genetic testing activity held by the UK Genetic Testing Network. ■

Priority 1 Testing

Ovarian cancer patients.

CURRENT NHS POLICY

NHS England Clinical Commissioning Policy E01/pb recommends that women with ovarian cancer be offered testing at the point of diagnosis.[5]

The Independent Cancer Taskforce's report (Recommendation 36) states that "NHS Commissioners should ensure that all women with non-mucinous epithelial ovarian cancer are offered testing for BRCA1/BRCA2 at the point of diagnosis."

The recent UK Genetic Testing Network report on genetic testing activity has shown an increase in the number of genetic tests for BRCA in 2015/16 since the guidelines were introduced: 11% higher in England and 14% higher in Scotland compared to the rates in 2014/15.[6] The proportion of these tests that relate specifically to ovarian cancer patients is not known, as disease-specific data is not captured.[7]

Relevant data from Wales and Northern Ireland was not included in the report. We support the report's recommendation to ensure this data is included in the future.

In our BRCA testing survey, we determined BRCA testing eligibility in our respondents using the criteria set out in the Clinical Commissioning Policy (ovarian cancer diagnosis, cancer subtype, date of diagnosis). Of those who met the criteria, 71% had been offered BRCA testing.

"[I was referred] by oncologist. Had appointment with genetic counselor and offered testing following this. Very straightforward."

"Mum was tested when she was first admitted to hospital following her ovarian cancer diagnosis."

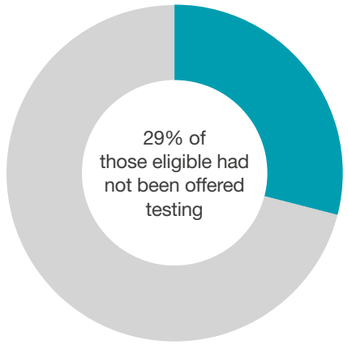
This is encouraging and we welcome the fact that this commissioning policy is largely being implemented. However, 29% of those eligible had not been offered testing.

Responses from across the UK showed a regional variation in access to testing, with some regions significantly better at offering testing to eligible patients. A postcode lottery in access to testing is unacceptable.

Through a Freedom of Information request, NHS England confirmed that they do not not carry out any evaluation of the implementation of the commissioning policy across the patient population:
"There is no national mechanism in place to monitor and evaluate implementation; individual commissioning teams may monitor this on an ad hoc basis through their discussions with individual providers."[9]

Without national monitoring to capture this information, the true picture of access to testing and regional disparity will remain unknown.

"Mum was tested when she was first admitted to hospital following her ovarian cancer diagnosis"



AROUND THE WORLD

Monitoring genetic testing activity
In France, the genetic counselling units and laboratories that are funded through the National Cancer Institute are obliged to deliver annual reports on their activities to the Institute. As a consequence, exemplary statistics regarding hereditary breast and ovarian cancer and BRCA1/2 testing are easily available.[8]

Barriers
There are still several barriers that stand in the way of the commissioning policy being fully implemented:

- **Time-frame –** The commissioning policy recommends testing is offered at diagnosis. A four-week timeframe from the point of diagnosis is recommended for breast cancer patients, however a timeframe within which to offer this testing to ovarian cancer patients has not been set.

- **Pathway –** BRCA testing does not currently have a set place in NICE Clinical Guideline 122.[10] These guidelines set out the pathway for treating women with ovarian cancer.

- **Responsible Team –** Of those in the survey who were offered genetic testing, there was variation on where in the care pathway this testing was offered, with 58% of testing offered by the oncology team, 32% by the genetics team and 5% by the surgical team. This variation can be accounted for with different NHS Trusts using differing methods for BRCA testing, some offering testing as part of their patients' cancer care and some referring patients to genetic centres. However, unless each NHS trust has a defined policy detailing their team responsible for offering testing, there is a lack of accountability in ensuring testing is taking place.

Next steps
A group of respondents in our survey met the criteria for eligibility for testing, but had been diagnosed before 2015. 68% of these women had not been offered testing. The NHS must reach out to women diagnosed with ovarian cancer before 2015 with information about their eligibility for BRCA gene mutation testing.

Cancer Alliances, alongside the National Cancer Vanguards, are responsible for the delivery of the Independent Cancer Taskforce's cancer strategy locally. Each Alliance has been tasked with creating a delivery plan to achieve the recommendations before 2020. These plans will be instrumental in ensuring all women diagnosed with ovarian cancer are offered testing as standard across the country by 2020. ■

AROUND THE WORLD

TRACEBACK
In Australia, a new project TRACEBACK provides a framework that aims to proactively identify women who were diagnosed with ovarian cancer but were not offered genetic testing at the time. This would allow these otherwise lost high risk families to access genetic testing, which in turn provides the opportunity to take action and reduce their risks.

Professor David Bowtell: "We are working through different approaches to finding patients and next of kin. We expect to start the first level of testing of Australian patients in the next few months."

RECOMMENDATIONS

1A The UK Genetic Testing Network must capture disease specific data when monitoring activity of genetic tests.

1B The NHS must find a mechanism to collect data to monitor implementation of Clinical Commissioning Policy E01/pb at CCG level.

1C Clinical Commissioning Policy E01/pb to include a timeframe for offering BRCA testing within four weeks of ovarian cancer diagnosis.

1D BRCA testing must be embedded into NICE CG122 at the point of diagnosis.

1E Each NHS Trust must have an explicit team responsible and accountable for offering BRCA testing.

1F The NHS must reach out to women diagnosed with ovarian cancer before 2015 to inform them of their eligibility for BRCA testing.

Ovarian Cancer Action
We will work with CCGs in England and health bodies in Scotland Wales and Northern Ireland to raise awareness of BRCA testing and the relevant guidelines to clinicians across the UK.

We will monitor the Cancer Alliances' delivery plans to ensure effective delivery of Recommendation 36 of the Cancer Strategy by 2020.

Priority 1 Testing

Family members at risk.

CURRENT NHS POLICY

NICE Clinical Guideline 164: Familial breast cancer: classification, care and managing breast cancer and related risks in people with a family history of breast cancer' states that a person should be offered genetic testing if there is at least a 10% (1 in 10) combined BRCA1 and BRCA2 mutation carrier probability.[11]

"I was told I did not meet the criteria. I was offered testing after my sister's cancer diagnosis. If I had been tested earlier it may have prevented development of cancer in myself and three relatives"

In our BRCA testing survey, respondents who had not been diagnosed with ovarian cancer were asked for their family history of cancer and whether they had tried to access testing through the NHS.

Of those that tried to access genetic testing through the NHS, 91% were able to access the testing they wanted. There was positive feedback from some of these patients who accessed testing quickly and easily:

"My GP was very supportive and I got into the system easily."

"The GP made a referral and I had a genetic counsellor appointment fairly quickly."

"Really easy - referred from doctors to breast clinic at the hospital - referred for testing with amazing support."

However, for some who eventually accessed testing, it was not such an easy process:

"I saw a GP who was also my mother's, so knew the history. Would not refer for genetic testing, would not entertain the idea there could be a link. Walked out of the appointment, asked to see another GP who I saw minutes later & referred straight away. Found to have BRCA2 [mutation]."

"My sister had to fight really hard to get tested. Her GP was most dismissive despite family history."

33 respondents attempted to access testing, but for various reasons were prevented from doing so.

Barriers

Some of the barriers individuals are facing in accessing testing when they have not been diagnosed with cancer are:

Individuals are judged not to be at high enough risk

A respondent in Scotland:
"They said [I was] low risk but my cousin in England got it straight away."

"I mentioned to my GP that my mother died of breast cancer in her mid-40s and he suggested I look into my family history of cancer. My maternal aunt died of ovarian cancer in her 60s, but this family history wasn't significant enough to suggest I have genetic testing at that time."

Contrary to guidelines some regional policies prevent testing without a living relative with cancer to test first

"[I was] told that because no one is alive with cancer at present in the family then they cannot test us."

"I could only get gene test on NHS if I had a living blood relative who had had cancer. Unfortunately I didn't. I only got gene test after being diagnosed with breast cancer."

"I was absolutely distraught to find that after a year of waiting there was no tumour sample for my mum - my assumption is that it wasn't saved or had been lost/destroyed!"

AROUND THE WORLD

Relaxing the referral criteria

BRCA recommendations in France do not specify a degree of relatedness among women affected by cancer (only that they must be on the same side of the family), or between the patient and their affected relatives in a family risk assessment. This may be done to help solve the problem of an intervening healthy male in an analysis of cancer in female relatives.

In France and Germany, an individual has direct access to genetic counselling and does not rely on a specialist referral.

Like many other European countries, BRCA referral guidelines - coordinated by the French National Cancer Institute - rely on a family history and/or ovarian cancer. France has a notably inclusive genetic testing criteria. It was broadened so fewer BRCA gene mutations went undetected. For this reason, their recommendations now included a single instance of ovarian cancer diagnosed before the age of 70 within the family as a criterion for genetic counselling. These guidelines are much less strict than the UK's in a number of ways:

- With the familial criteria that focuses upon multiple female relatives affected by breast cancer, there is no precise number of affected women or age at diagnosis required for the individual to access genetic testing.
- Their indication criteria do not state a mandatory degree of relatedness between the patient and their affected female relatives. This may be to help overcome the fact a gene mutation may go undetected if passed down through a healthy, paternal line when assessing female cancer incidence.

An isolated individual case of early onset breast cancer before the age of 40; bilateral breast cancer in one woman without age limit; and an isolated case of ovarian cancer before 70 years are considered grounds for genetic counselling.[12]

Some healthcare professionals are not fully informed about BRCA and eligibility for genetic testing for those with family history, so are denying testing through misinformation with possible life-threatening consequences

"I was told that my mother's and daughter's ovarian cancer was not linked."

"My GP told me there was no point in being tested as I already had cancer."

"[I] was told only people with cancer can be tested."

Next Steps

Every eligible woman who has been denied testing and goes on to develop ovarian cancer represents a cancer prevention failure:

"I was told I did not meet the criteria. I was offered testing after my sister's cancer diagnosis. If I had been tested earlier it may have prevented development of cancer in myself and three relatives."

In our survey, 8 of the 33 who could not access NHS testing sought it out privately. 100% of these 8 were found to have a mutation. These individuals had the resources to pay for these tests themselves, however many others will not be in the same position.

Many of these families were denied the right to take steps to prevent cancers from developing. The NHS must take responsibility for the Clinical Commissioning Policy being accurately implemented around the country to ensure that eligible family members at risk are not being denied testing.

The current 10% eligibility threshold is a matter of debate among scientists and clinicians, with research demonstrating "that the widespread use of 10% is not appropriate for all models, clinics or purposes".[13] NHS England's own economic analysis has shown that a 5% carrier probability threshold would be cost-effective.[14] Using the 10% threshold "will result in substantial numbers of those with a BRCA mutation being missed."[15] It limits access to testing for those who have smaller families, or limited contact with relatives, making it not fit for purpose.

Priority 1 Testing

Family members at risk.

AROUND THE WORLD

Tackling incomplete or inconspicuous family histories of cancer

Medical commentary and guidelines in both Europe and North America address the reasons why many BRCA gene mutations go undetected when access to genetic testing is based upon strict criteria of family history. As a solution, multiple national recommendations suggest a more flexible approach.

In Sweden, Regional Cancer Centres (RCC) coordinate and develop cancer care. Their recommendations on hereditary breast and ovarian cancer recognise that a complete reliance upon a family's medical history can be problematic. They reference a validation study that showed this model underestimates the risk of High Grade Serous Ovarian Cancer, especially in younger patients.[16 17] Furthermore, the RCC guidance states a family history of hereditary cancer is lacking in over 40% of women with BRCA germline mutations. The RCCs therefore advise that genetic testing should be offered to all women with ovarian, tubular, or primary peritoneal cancer, regardless of family history.[18]

The Cancer Society of Finland also acknowledges risk-prediction models are not always valid or effective. The Society recommends that if a doctor feels an individual's concerns about increased genetic risk of cancer are justified, the patient should be re-directed to a genetic counselling policlinic for further investigation and possible genetic testing.[19]

In the USA, the National Comprehensive Cancer Network guidelines offer a similarly common-sense approach, acknowledging family size and structure will affect the probability of detecting a mutation.[20] Individuals with a limited or unknown family history may have an underestimated probability of a familial gene mutation detection, whereas the likelihood of mutation detection may be very low in families with a large number of unaffected female relatives.

Of the 33 in the survey who could not access testing, even the limited family history taken suggested a likelihood that 22 of these would qualify for BRCA testing if the eligibility threshold was lowered to 5%. This would open the door to many more families to find out their BRCA status and have the opportunity to take risk reducing action.

Of the women diagnosed with ovarian cancer who are found to have a BRCA mutation, up to half report no family history:

"I feel there is too much emphasis on family cancer history with BRCA. Although I have extensive family on both sides there was no trace of this gene because it came down through males."

"I was told that I didn't have enough close relatives with cancer. I have no idea about my father's side of the family and only had 2 female relatives on mother's side. This was prior to my own cancer diagnosis."

"We have no idea of family history on my father's side, as he died young and we have no contact with any surviving relatives." ■

RECOMMENDATIONS

1G Tumour samples from women with ovarian cancer should be retained as standard so that families are not prevented from accessing testing if patients have died.

1H The testing eligibility threshold should be lowered from 10% to 5% combined BRCA 1 and BRCA 2 mutation carrier probability

Ovarian Cancer Action
We will continue to raise awareness with GPs about eligibility for BRCA testing for those with family history.

CASE STUDY ROSIE

The consequences of missed opportunities.

If only I had known about BRCA...If only I had known about testing.

"My maternal aunt died of ovarian cancer in her 60s, but this family history wasn't significant enough to suggest I have genetic testing at that time"

Then my family and I would have been saved so much pain and heartache and the NHS would have been saved a small fortune in treatment costs.

I was born in 1950, the fourth of five children (four girls and a boy). Our mother died of breast cancer in 1960 and all four girls grew up with a fear of breast cancer but no idea that our mother had a BRCA gene mutation that sealed her fate.

In 2014, I mentioned to my GP that my mother died of breast cancer in her mid-40s and he suggested I look into my family history of cancer. This revealed that my maternal aunt died of ovarian cancer in her 60s, but this family history wasn't significant enough to suggest I have genetic testing at that time.

In January 2016 my younger sister was diagnosed with Stage 3-4 fallopian tube cancer and was advised to have genetic testing. This confirmed that she had a BRCA1 mutation and so we as her siblings were offered testing. I was tested in July 2016 and was also found to have the mutation. By this time one other sister had also been found to have fallopian tube cancer. So now we had two sisters who had extensive surgery followed by chemotherapy. Our fourth sister also has the mutation but thankfully no ovarian cancer. Our brother does not have the mutation.

In July 2016 I was offered a bilateral salpingo oophorectomy which was undertaken in October 2016 and I had an initial appointment to discuss my options for prophylactic double mastectomy. In spite of two normal pelvic ultrasounds and two normal CA125s in May 2016 and October 2016 the histology on my ovaries showed that I had a stage 1c aggressive ovarian cancer. This resulted in a hysterectomy, node removal and omentectomy in November 2016 followed by six cycles of chemotherapy. Thankfully, although my treatment was not pleasant I have recovered well. My two other sisters have had significant health issues as a result of chemotherapy.

Sadly this was not the end of the story. My children were offered genetic testing. Thankfully my son who has two young daughters did not have the mutation but my daughter did. She immediately arranged for a prophylactic double mastectomy. This was planned for January 2017. As part of the work up for this she had an MRI. She was not quite 39 at the time and had not, therefore, entered the national breast screening programme. An MRI revealed a small but aggressive breast cancer. She had chemotherapy from December 2016 until April 2017 followed by double mastectomy. The treatment nearly cost her her life. Her body did not tolerate chemotherapy and finally rejected her implants. She is still planning to have reconstruction and bilateral salpingo oophorectomy in the near future.

Thankfully the next generation will all have the advantage of genetic testing and advice. ■

Priority 2 Counselling

Pre-testing.

CURRENT NHS POLICY

NICE Clinical Guideline 164

Pre-test
Two sessions of genetic counselling should be given before genetic testing. The patient should be fully informed of the counselling process before their first session.

During genetic counselling, a genetic specialist should discuss the risks of carrying a gene mutation and give a written summary of this discussion afterwards. The patient should be told about the process of testing, what a positive, inconclusive or negative result will mean for them and their family, and a likely timescale for receiving the results.

Post-test
Women with no personal history of cancer considering risk reducing surgery should have genetic counselling in a specialist cancer genetic clinic before making a decision. Pre-operative counselling about psychosocial and sexual consequences of the surgery should be undertaken.

Women with a personal history of cancer considering risk-reducing breast or ovarian surgery should be referred for appropriate genetic and psychological counselling before surgery. Counsellors should discuss the risks and benefits of risk-reducing surgery, which would cover such issues as body image, sexuality, and anxiety.

Risk and psychological counselling should be offered to women undergoing surveillance.[21]

Genetic counselling plays a necessary role in the BRCA testing pathway, both pre- and post-testing. While some genetic counsellors give patients support, their primary role is to provide factual information around testing and risk-reducing options. The role is not what many would understand as "counsellor":

"Stop calling it genetic counselling for starters, completely misleading. It's testing not counselling - you don't get any "counselling" at all which is poor."

We believe that patients need better access to psychological support both pre- and post-testing.

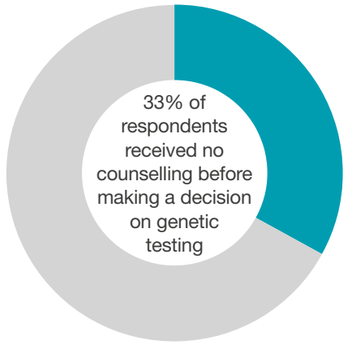
In our survey we asked respondents whether they received any psychological counselling or support before and after their genetic test, and whether this support was adequate.

Pre-test counselling & support
The survey revealed 33% of respondents received no counselling before making a decision on whether to go ahead with genetic testing:

"I requested counselling and was told it was not available. I REALLY could have used it."

"Just me and Google – it was a lonely time."

There were a number of cases where the level and detail of information given to counselees failed to meet the standard set in NICE Guidelines.



Patients were not always informed about the process of genetic counselling, and some genetic counsellors neglected to cover fundamental topics, such as what a positive, negative, or inconclusive result would mean for the patient and their family:

"The genetics counsellor called me before the appointment to tell me what to bring but that was about it."

"Didn't realise implications if test positive and how my family could be affected."

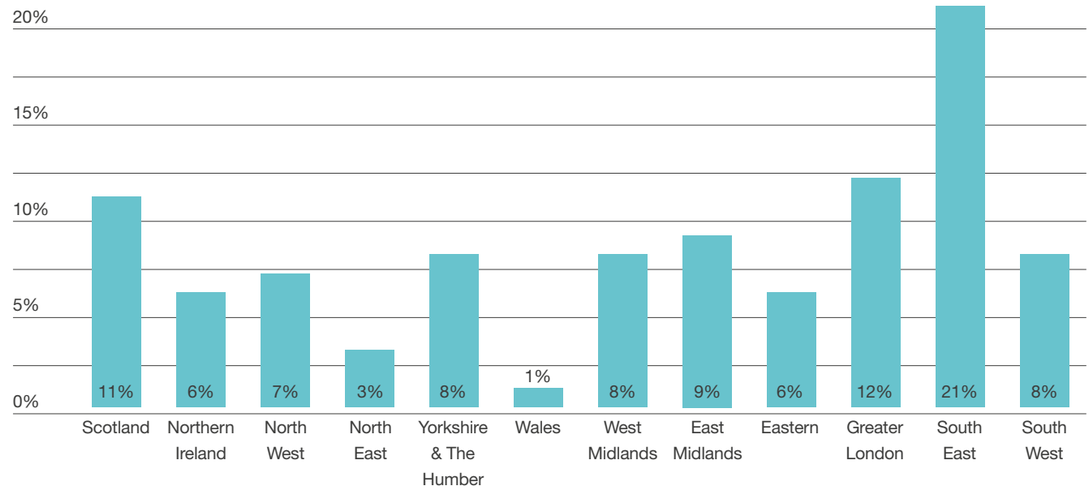
These inadequacies forced some patients to look elsewhere for support, such as online forums or a different NHS Trust.

"Minimal information was provided before the test, I did my own research."

One respondent joined her mother's genetic counselling service in Wales after a disappointing counselling session in Hampshire:

"The two could not have been more different, with [the counselling] offered in Wales being far more thorough."

REGIONAL BREAKDOWN OF 145 RESPONDENTS WHO DID NOT RECEIVE PRE-TEST COUNSELLING OR SUPPORT



Our survey revealed regional variation in the level and quality of support given to patients. Genetic counselling should not be a postcode lottery.

Of the 33% who received no counselling, 84% were found to have a genetic mutation. It is imperative that patients are informed through counselling of their right "not-to-know" as well as their right "to-know" their genetic status and discuss all the implications.

Where counselling is taking place, our survey showed a system working well. Of those who received pre-test counselling, 82% said it was adequate. The survey gave examples of genetic counselling sessions that offered patients not only adequate, but "fantastic" levels of pre-test support:

"The genetic counsellor discussed all the possible options and outcomes with me in great detail. She also involved my husband in our chats to help gauge if I was 'ready' for testing as it was quite soon after my mother passed away."

"I feel that I was very well informed of what would happen and what my options were if the results come back positive."

"Geneticist was very thorough and followed up with a letter explaining everything."

It was encouraging to see exemplary cases of genetic counselling, where patients received detailed, high-quality and comprehensive information and support, as per NICE Guidelines.

AROUND THE WORLD

Informed consent
Across national guidelines, the principle of informed consent is recognised as fundamental to pre-test genetic counselling.[22] The German Consortium for Hereditary Breast and Ovarian Cancer (GB-HBOC) places great emphasis upon it, giving equal weight to the right "not-to-know" as the right "to-know". GB-HBOC guidelines state the counsellee must be informed about the limitations of the test and the consequences of all possible results beforehand and must agree to the the test in written form - a point also advised by the Association of Gynaecologic Oncology Austria.[23]

Priority 2 Counselling

Post-testing.

Post-test counselling & support
We asked respondents whether they received any counselling immediately after their genetic test and whether they felt this was adequate.

Some respondents said they were made to feel incredibly supported in the light of their test results:

"The Royal Marsden has always given lots of info including education days and support groups."

"It was and is still helpful."

"The counsellor was great she was able to really help me to think hard about what 'I' wanted to do and focus on me rather than on me trying not to upset others."

"There was just the right amount for me. I could have had more if I had felt it was needed."

Again, it is encouraging to see some NHS Trusts giving high quality support to the patient. However, it is evident that post-test support is not consistent in its provision or quality. Of our respondents who accessed genetic testing through the NHS, and were found to have a mutation, 42% received no counselling or support after their genetic test results. In absolute terms, that is 165 people who were shown to have a genetic mutation and were given no support to deal with the news.

"I felt alone."

"No support. Just told my surgical options."

"I felt overwhelmed by the sea of paper I received and like I was simply left to deal with the outcome with no offer of a follow up appointment to discuss my options and feeling."

"Since finding out 3 months ago that I have BRCA2 gene mutation, I am still waiting on more information. I feel I have been left to find out more on my own."

Of those who did receive some support after their results, 21% said it was not adequate:

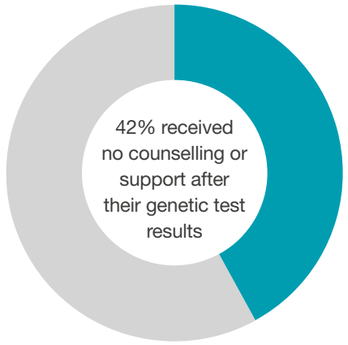
"The information was adequate in so much as I understood what the results could mean. However, I didn't feel I received any support with where I go next once I received the results showing my BRCA1 mutation."

"I wish there was more support after the test. I found out I had BRCA2 and after initial advice and support from my genetic counsellor I have been left to deal with it alone. Ironically the one person I wish I could have talked about this with is my Mum who had already passed away without even knowing she had the mutation. Thank goodness I found BRCA Umbrella."

"Afterwards I had some queries so I wrote to the counsellor and never got any reply."

"She knew nothing about options and offered a hug?!"

After testing, only 27% of our respondents were given information about where to find additional support if they needed it. There are a number of support groups and charities that can help those that need this support.



"I have been left to deal with it alone. Ironically the one person I wish I could have talked about this with is my Mum who had already passed away without even knowing she had the mutation"

Long term support
Once test results have been received and understood, genetic counselling sessions end. The period after this was identified in our survey as the time when patients need the most psychological support.

"It only really hit me a few months later when all the other medical intervention began. Would advocate this is a time when a patient needs as much or more support."

Depending on the age of the patient, they may move immediately into being referred to specialists for screening or risk-reducing surgery or it may be years before this is appropriate. If a woman is tested in her mid-twenties, it may be five or ten years before the appropriate time to discuss breast screening or surgery, and another five or ten years before discussing the removal of her ovaries. During this time those with a gene mutation are left to deal with the emotional consequences of their diagnosis themselves.

AROUND THE WORLD

Post-test counselling
The Association of Gynaecologic Oncology Austria: "The [BRCA] test results must be explained to the patient in a second personal genetic counselling session by a Medical Geneticist or a medical specialist for the particular indication as defined by the Austrian Gentechnikgesetz (GTG, Genetic Engineering Act). Counselling must be concluded with a counselling letter that contains all relevant points of the discussion, including the relevance of the findings for the patient's family."[24]

AROUND THE WORLD

Tailored counselling
The USA's National Comprehensive Cancer Network (NCNN) guidelines on genetic counselling are notable in their detail and their consideration of the patient as an individual with particular needs.

The emphasis upon context, delivering tailored and practical information, and empowerment is clear:
"Counselling for hereditary breast and/or ovarian cancer uses a broad approach to place genetic risk in the context of other related risk factors, thereby customizing counselling to the experiences of the individual. The purpose of cancer genetic counselling is to educate individuals about the genetic, biological, and environmental factors related to the individual's cancer diagnosis and/or risk factors related to the individual's cancer diagnosis and/or risk for disease to help them derive personal meaning from cancer genetic information, and to empower them to make educated, informed decisions about genetic testing, cancer screening, and cancer prevention."

Pre-test counselling should include a discussion of why the test is being offered, the significance and impact of the results, and practical issues such as confidentiality, economic considerations and risks of genetic discrimination.

NCCN Guidelines' detailed directions for post-test counselling makes a point of highlighting the need for emotional support and for health care providers to signpost accordingly:
"Post-test counselling must also be performed and includes disclosure of results, a discussion of the significance of the results on the emotional state of the individual, a discussion of the impact of the results on the medical management of the individual, and how and where the patient will be followed."

"Counselling should also include making the individual aware of any available resources, such as disease-specific support groups, advocacy groups, and research studies. Individuals who have tested positive for a mutation may have greater distress than anticipated, so provisions for supportive interventions should be provided." [25]

"I've got a 6-year old daughter and I feel guilty a lot of the time that this could be passed on."

Despite NICE guidelines in place, many of our respondents expressed the wish for better emotional and psychological support when accessing risk-reducing surgery, specifically on the surgery's impact:

"There could definitely have been more emotional support about the impact of the preventative surgery I was advised to have."

"I did not know quite how much a radical hysterectomy would impact my life emotionally or physically - no counselling was offered."

Priority 2 Counselling

Pre and post test.

In particular, women in our survey struggled with a lack of support around:
Risk-reducing surgery meaning an end to fertility;

"I have found the fact I can no longer have children quite hard to deal with even though it was totally my choice, it's been mentally harder to deal with than I thought."

"I think the fact that my child bearing days are over has had an impact."

Changes to their sexual relationships;

"No sex drive which impacts on relationship with husband."

The impact of early menopause;

"I think that doctors currently underestimate the significance of the menopause on a woman's quality of life. For example, before breast surgery, it was compulsory to see the psychologist but no similar appointment or counselling has been offered to me in respect of the ovarian surgery... I would like doctors to understand that this is a huge decision for pre-menopausal women (even those who have 'completed' their families)."

Barriers
The barriers preventing patients from receiving adequate psychological support throughout the BRCA testing pathway are:
Psychological counselling – psychological support is seen as secondary to information provision in genetic counselling and is not a standardised part of the testing pathway;

"I didn't really have much chance to explore my feelings – it was more about getting all the facts."

"The information was good but emotional/psychological support was non-existent."

Mental health services – There is no clear pathway for patients diagnosed with a genetic mutation to access psychological counselling from mental health services;

"I've approached the doctors and they have no resources to help other than mental health and they're only interested in you if you have tried or thought about suicide."

Support groups – Patients are not receiving information about relevant support groups;

Time after diagnosis – Patients can go for years between receiving test results and undergoing risk-reducing surgery without any contact with the NHS.

Next steps
Patients who are not fully supported emotionally through the BRCA testing pathway might struggle with making life-changing decisions around having risk-reducing surgery and telling family members that they might also be at risk. Feelings of guilt and anxiety were found to be common among our survey respondents who had been diagnosed with the BRCA gene mutation.

It is imperative that whilst reducing the risk of patients developing cancer, the NHS is not creating a new population of men and women struggling with their mental health. The NHS must take responsibility for the mental health as well as the physical health consequences of a diagnosis. Mental health services must be properly resourced to support those with a genetic mutation.

Genetic test results may have far-reaching emotional implications for the individual and their family; therefore psychological counsellors should be involved and emotional support built into the genetic testing pathway to prevent more serious issues later.

"Generally people are having the test because they have lost someone close to them, or have been diagnosed themselves. They are already going through emotional trauma so need support to have the strength to deal with the test's outcome." ■

RECOMMENDATIONS

2A Patients should receive tailored emotional and psychological counselling as part of the genetic counselling process in the genetic testing pathway.

2B Patients diagnosed with a BRCA gene mutation should have access to relevant mental health services adequately trained to support those with a genetic mutation at any point in their BRCA journey, whether pre or post testing, at the time of considering or undergoing risk-reducing surgery or at any point beyond this.

Ovarian Cancer Action
We will continue to support women and men diagnosed with a BRCA gene mutation through good quality information and peer-to-peer support.

CASE STUDY
JO

Emotional support.

"I don't want to take up the time of a stretched mental health service but can't afford the expense of private counselling."

"I was 22 when I lost my mum to ovarian cancer and found out her genetic test was positive for a BRCA1 mutation. I attended two genetic counselling sessions soon after but decided to wait to be tested for a few years. At the age of 27 I decided to go ahead with the test, so made contact with the genetics team to request this. Once I had the test results over the phone, I had one additional face-to-face session with the genetic counsellor to discuss the results, but I wasn't at the stage to start with either additional screenings or be referred for risk-reducing surgery so I went home with no contact with anyone for several years.

I next made contact when I was nearly 30, after I had had my second child and decided I was ready to discuss risk-reducing mastectomy. There was no additional support at this point to discuss the psychological or emotional impact of having a double mastectomy and no further sessions with the genetic counsellor. Although there was a breast care nurse available, she seemed busy and didn't contact me when she said she would so I didn't bother her. I was not required to have a session with a psychologist before being put on the list for the surgery. The breast care nurse was on maternity leave when I had questions soon after surgery, I had complications and was very stressed about who to call, and the other breast care nurses in her office did not return my calls.

I found my own support through online channels, to discuss experiences with other BRCA women and cry on their shoulder (from afar). It isn't the same as having counselling as you don't want to overload a friend. The whole experience was incredibly stressful. Although I haven't ever regretted having the surgery, it was made a lot harder due to lack of communication and a lack of understanding of the mental health implications.

The next stage for me is to have a BSO after the age of 35, and I have started the process of referrals to discuss this. I haven't had any contact with my genetic counsellor for some time, so I don't really know who to talk to about my options. No one has mentioned emotional support for going through the early menopause and I'm really worried about where I should go to if I struggle. I don't really have a relationship with my genetic counsellor where I could ask for support in that way. I don't want to take up the time of a stretched mental health service but can't afford the expense of private counselling." ■

"The breast care nurse was on maternity leave when I had questions soon after surgery, I had complications and was very stressed about who to call, and the other breast care nurses in her office did not return my calls"

Priority 3 Standardised Information

CURRENT NHS POLICY

NICE Clinical Guideline 164 states that "People should be provided with standardised written information about risk, including information on HRT, hormonal contraceptives, breast feeding, alcohol, smoking, weight and physical activity"

"Standard information should be evidence based wherever possible, and agreed at national level." [26]

The BRCA testing survey revealed frustration in patients at the inconsistencies in information provided at each stage of the genetic testing process, and through their risk-reducing decisions.

"The one thing I'm passionate about is everyone getting the same information. For example, some surgeons recommend a full hysterectomy and some only recommend tubes and ovaries."

"I would like to see greater consistency of information between the medical professionals involved in the process."

"[We need] cohesive information from medical profession, every single doctor and genetics person has given different and sometimes conflicting information."

Often patients discover the differences in information due to increased use of social media as a means of support. Some of our respondents had family members in different areas across the UK who were told different things.

It's vital that information provided to patients be accurate and evidence based, as the concern over which information to trust is a cause for anxiety.

Risk-reducing procedures

NICE CG164 states that "Patients should have the opportunity to make informed choices about any treatment and care and to share in decision making".

The opportunity to reduce the elevated risk of ovarian cancer through surgery is significant and life-changing. The impact can be hugely positive and provide relief from years of worry:

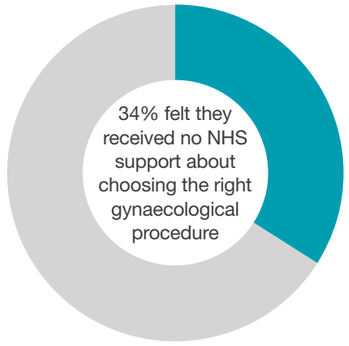
"Having had my children already this was an amazing option which has changed my life for the better. Not just my life has changed for the better but all those around me."

"Great sense of relief reducing my risk of ovarian cancer."

However, 34% of our respondents felt they received no support from the NHS about choosing the right gynaecological risk-reducing procedure for them.

"I found little guidance on whether to go for uterus removal or not with BRCA1."

"I feel I seriously lacked information regarding preventive surgery... I've recently been finding out about increased risks of dementia. Knowledge is power and I feel I'd rather have known all info to make a balanced informed decision."



"[People] need information on these surgeries - REAL information about side effects and complications, not just one chart in a PR booklet."

This is of great concern as women with a BRCA gene mutation are forced to try and research their options themselves, either attempting to decode journal articles or consulting non-qualified peers on social media. This is a recipe for misinformation and frequent anxiety.

The implications of making a life changing choice without the full evidence base can be long lasting. The risk of ovarian and breast cancer starts to increase at certain ages. There are risks and benefits to weigh up when choosing the type of risk reducing surgery to have, as well as the right age to have it.

"I would like to see greater consistency of information between the medical professionals involved in the process"

Menopause and Hormone Replacement Therapy (HRT)

Respondents in our survey highlighted early menopause and HRT as an area of particular concern where information was lacking or variable.

Following the removal of a woman's ovaries at an early age, premature menopause can have a significant impact on quality of life and long term health. This earlier-than usual loss of natural hormones in the body can have long lasting effects causing osteoporosis, cardiovascular disease and dementia as well as decreasing quality of life and impact sexual health." [27 28 29 30]

"The menopause has been brutal and I feel like a different person."

Many of these can be ameliorated with the use of HRT and the standard of care is to continue providing this until natural menopause age. There is not a "one-size-fits-all" approach and women must be able to work with their medical team to find the best options for them.

Most women who carry the BRCA gene mutation are medically able to have HRT, yet information and support in this area is inconsistent.[31 32]

AROUND THE WORLD

Surgical menopause follow-up

Clinical Practice Recommendations from Cancer Australia: state that an ongoing assessment of the effects of surgical menopause is required after surgery.[33]

In our survey, 31% of those who had gone ahead with risk-reducing surgery felt they were not fully informed of their HRT choices. This is not only a cause for concern due to anxiety and day to day struggles that are often related to menopause, but also the major risks this poses for these women's future health and the costs this will bring at a later date. This lack of clarity and consistency relating to HRT choices is highlighted by our respondents' comments:

"This was the worst part of my cancer journey. Absolute complacency and indifference about consequences of a surgical menopause for a 40yr old woman."

"Junior doctor wouldn't prescribe HRT. Said it wasn't urgent."

"Hit the menopause like a brick wall... no info given before or after ...hellish experience."

"No one mentioned it!"

"Nightmare! Was given minimal info on options 40 mins before going into surgery. Agreed with consultant post-anaesthetic I'd opt for gel, which was out of stock in hospital pharmacy so discharged without HRT."

"I faced a mix of opinions from medics about whether HRT patches were safe for me and there was not clear guidance on whether it would affect osteoporosis."

"I haven't gone for HRT as so confused by it & don't really know who to talk to.....don't want to make the wrong decision & do any harm & put myself at any risk."

"I have been told HRT is not recommended due to BRCA1 mutation."

"I had my op three years ago and I am still attempting to get the HRT balance right."

Women must be provided with accurate, up to date and standardised information about their HRT options as part of their pre-surgery support and after-care.

"Nightmare! Was given minimal info on options 40 mins before going into surgery. Agreed with consultant post-anaesthetic I'd opt for gel, which was out of stock in hospital pharmacy so discharged without HRT"

Priority 3 Standardised information

Family planning options
Family planning is an important and sensitive topic for those carrying a genetic mutation. It's vital that information is provided to both men and women with BRCA mutations so that they can make informed decisions about planning their families. This information is key for preventing the BRCA gene mutation from being passed to the next generation.

There are fertility options available for those who want to have a family. In England and Wales, couples that meet certain criteria are eligible for 3 cycles of IVF using Preimplantation Genetic Diagnosis (PGD).[34 35] Couples in Scotland are eligible for two cycles.[36]

In the BRCA testing survey we asked relevant respondents if their genetics team gave them any information about options relating to having children when you carry a genetic mutation. 28% of these respondents received none.

For those patients who did not receive information regarding fertility options, the consequences can be life altering.

"If I'd known about egg storage options whilst I was still age eligible, I may have looked into it further. By the time I thought about it, I was too old."

Contacting family members
A commonly reported concern in those with a BRCA gene mutation is the issue of when and how to contact family members to inform them that they are at risk. In order for BRCA testing to truly fulfil its potential as a cancer prevention tool, it is essential for those tested to pass on the information to their close relatives, who will have 50% chance of also carrying the mutation if first degree (parents, siblings, children). However, this is life-changing news, and combined with family politics, ongoing grief and changing roles within the family, it can be a difficult topic to navigate.

In the BRCA testing survey, we asked respondents to rate on a scale of 1 to 5 how much stress or anxiety they felt relating to contacting their family members. 65% of respondents felt some anxiety, with 10% experiencing "extreme anxiety".

"My daughters are in their 20s-they had just gone through the stress of my illness only to find that they are also implicated."

AROUND THE WORLD

Reproductive options
USA - NCCN Guidelines: "The outcomes of genetic testing can have a profound impact on family planning decisions for individuals of reproductive age who are found to be carriers of BRCA1/2 mutations. Counselling for reproductive options such as prenatal diagnosis, PGD, and assisted reproduction may therefore be warranted for couples expressing concern over the BRCA1/2 mutation carrier status of their future offspring.

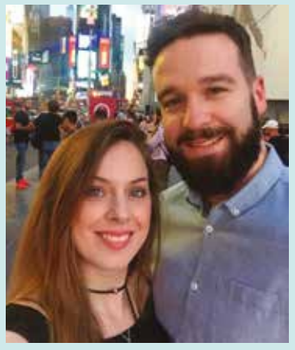
Such counselling should include a comprehensive discussion of the potential risks, benefits, and limitations of reproductive options."[37]

CASE STUDY CARLA

"I want the next chapter of our lives to be BRCA free."

"The fact that PGD is available to us is life-changing. We have chosen this route as we want this gene risk to stop with me. We do not want to have to go through life worrying about our child's health, worrying if they too carry the faulty gene, having to think about when the best time is to tell your child of their potential increased risk of cancer. If we had a girl I just couldn't image her having to go through the pain and suffering I have had to to avoid getting cancer. It would break our hearts.

My biggest fears would be if I was to pass away that my other half would be left with having to support our children going through preventative surgeries. With him supporting me through mine and then having to support our children on his own, I can't think how that would feel. I can't even begin to think of how our child would handle that, to have to go through these surgeries because of something their mum gave to them. The thought of passing the gene down makes me feel sick and stricken with guilt, I just don't want to take that chance."



"I felt like I was to blame for them maybe carrying a genetic mutation too."

"Some family hadn't been spoken too for a long time. It felt a bit like I was delivering a death sentence!"

"I found it more difficult telling my children than actually having the test myself."

In the Family Contact survey, 81% of our supporters, and 69% of the general public agreed that they would want the help of the NHS in telling relatives who could have also been affected.

"I wasn't in contact with my brother. I was only given a letter for him. I was left to contact and explain it to the rest of the family. Could have done with more support in explaining this to my children."

The most common resource given to respondents to help contact relatives about their potential risk was a letter - still only 42% of those found to have a genetic mutation were given this. Of our respondents, 36% were given no resources at all to help them explain the complicated details of genetic mutations and their implications to their families.

Barriers
Regional variation in information – Each NHS Trust has its own information resources that it provides to its patients. These resources differ in depth of information and topics covered. Some resources are a couple of pages, and others go into much further detail in almost 50 pages. Quality of information about BRCA gene mutations should not be based on where patients are being treated.

Language – Complex medical information for patients can be difficult to disseminate when English is not their first language.

Next steps
In a digital age, each NHS Trust using and updating its own information resources is unnecessarily costly and time consuming. A centralised digital resource for all NHS Trusts to provide information for patients would ensure all patients receive the same essential information at minimum to support life-changing decisions and help patients prevent cancer in themselves, their families and the next generation.

New developments in medical research, including possible future screening methods for ovarian cancer will be extremely important for those with a BRCA mutation to be informed about. Relevant updated information must be communicated to those with a BRCA gene mutation, especially those who have not had risk-reducing surgery. This is already taking place in some NHS Trusts, but must be standard practice throughout the country.

RECOMMENDATIONS

3A A standardised, evidenced-based digital resource to be created by the NHS and made available to all NHS Trusts and their medical professionals for patients covering (at minimum) the following:

- Screening options for both breast and ovarian cancer including explanations of why certain options are not available
- Evidence-based reviews of each risk-reducing surgery option including recommended age
- HRT options including risks, benefits, and contraindications
- Advice for contacting family members at risk
- Specific advice for men who carry the BRCA gene
- Rights regarding financial concerns such as insurance/ mortgage/ sick leave from work
- Family planning options (PGD)
- Where to get support

This resource must be regularly updated in line with developments and available in a wide range of languages for hard to reach groups.

3B The NHS must find a mechanism to contact those with a BRCA gene mutation with new relevant medical breakthroughs that could affect their decisions to reduce their cancer risk.

Ovarian Cancer Action
We will work with the NHS to develop this digital resource for NHS Trusts around the country.

We will continue to provide high quality, far reaching information for those seeking information on BRCA gene mutations through our BRCA hub and our Cancer Prevention Officer.

Priority 4 Waiting times

At each stage of the genetic testing process, from initial appointments and referral to surgery, respondents in the BRCA testing survey showed waiting times differing greatly from patient to patient. Some patients were able to go through the process quickly:

"I asked my doctor and was referred, counselled and given test all in a matter of months."

But many others had a long wait:

"The process was a very slow one. I had to personally keep chasing up and requesting the testing. I had it done eventually, but the whole process from start to finish was extremely prolonged."

"The process was easy but took 1.5 years from first referral and submittal of my family history questionnaire to the genetics clinic, to actually seeing a genetic counsellor."

"It took a while - getting a GP appointment to get a referral to a breast clinic, assessment by the breast clinic and waiting for referral from them to Guy's for the test, waiting for the results. There was no issue at each stage, but it took about 9 months from start to results."

"[We need] more consistent referral times across NHS trusts. Mine was 18 weeks, my sibling's was 3 weeks"

Initial appointments and referral
Many respondents described long waits and variations across their family members also going through the process:

"[We need] quicker waiting times for initial appointments, I have been on a waiting list for 8 months currently to have an initial consultation with gynae about moving forward with my preventative surgery."

"[We need] more consistent referral times across NHS trusts. Mine was 18 weeks, my sibling's was 3 weeks."

In some cases, long waiting times are driving people to undergo private testing rather than wait:

"My father found out he had BRCA1 but NHS insisted I needed to complete genetic family history for them to assess whether I should be tested. This would have meant a 4-6 month process best case scenario which was too long a wait so I went privately for a £200 cost and received results within 10 days as they were looking for a known variant."

"I could not wait for their process, so went private. They got back to me by the time I had already found out I had BRCA1 and had surgery which found pre-cancer cells so I was right not to wait."

Testing results
Worryingly, some respondents faced long waits to find out the results, causing stress and anxiety:

"The process is very slow and rigid, it needs to be more responsive. In total I waited an agonising 7 months for a [genetic testing] result. This is distressing and stressful when all you want is your result I feel you are dealt with as a process rather than a person who has feelings and anxieties, this needs to change."

If genetic laboratories do not have the resources to carry out testing in a timely way, more resources must be provided or testing must be outsourced to other laboratories so that patients do not suffer prolonged anxiety.

Surgery
Risk-reducing surgery is included under the 18 weeks from referral target for non-urgent surgical procedures. Although risk-reducing surgery is not regarded as elective, these patients sit lower on the priority list than other patients. The result is that waiting times are unpredictable, with surgical dates frequently cancelled due to emergencies.

Patients referred to hospitals with a waiting list longer than 18 weeks have the legal right to chose alternate hospitals to have their surgery, including private hospitals.[38]

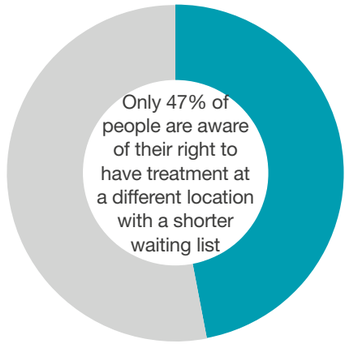
In the BRCA Testing survey, the results showed a wide range of waiting times for ovarian cancer risk-reducing surgery:

- Waiting time ranged from 2 weeks to 104 weeks.
- The average waiting time for surgery was 15 weeks
- 7% of respondents waited over a year.
- 77% of respondents waited less than 18 weeks.

"BRCA1 runs widely across my family who [live in] different areas. My [breast surgery] was completely done in 3-4 months, [my sister] eventually finished after 3 years."

Research suggests that during the waiting period for surgery patients experience decreased health and an affected psychological and social life. Patients experience anxiety during the wait for surgery, with longer waiting times correlating with higher anxiety. Emotional reactions to the waiting were found to be less negative when patients received information on the date of surgery when they were placed on the waiting list.[39]

While resources are stretched in the NHS, it is important to have a target in place for surgeries to take place, so that risk-reducing surgeries are not jettisoned to the bottom of the pile. 77% of respondents having surgery within the 18 week target is promising, and the NHS must ensure this does not decline. Funding risk-reducing surgery in a timely way is of cost-benefit to the NHS: it is cheaper to fund risk-reducing surgery than to fund the high likelihood of a later cancer diagnosis and expensive treatments subsequently required, on top of the same surgical procedure.



Barriers
Lack of information about rights and options – Many patients waiting for surgery under the 18-week waiting time are not aware of their right to have treatment at a different location with a shorter waiting list. NHS Research shows only 47% of people are aware of these rights.

In the BRCA survey 73% of patients who had undergone risk-reducing surgery were not aware that their treatment came under this 18-week wait time, so were not aware of their rights.

When on a waiting list, patients are not made aware of alternative shorter options. Patients also have to weigh up the risk of moving to a different waiting list and being placed at the back of another queue.

NHS resources – An increase in the number of genetic tests and risk reducing surgeries will increase costs in the NHS in the short term (saving money on costly cancer treatment in the future).

Next steps
The varying waiting times across each stage of the genetic testing process is concerning. While we campaign for more people to be offered testing to reduce the number of deaths from ovarian cancer, this means there will be greater numbers of patients and their families accessing genetic testing and genetic counselling. This will put more strain on resources and waiting lists, causing greater anxiety to those going through testing and risk-reducing options who have to wait even longer.

Greater investment in cutting down waiting times along the genetic testing pathway and for risk-reducing surgery is necessary for long term NHS cost savings in the future. ■

RECOMMENDATIONS

4A The NHS must instate a target for receipt of results of genetic testing.

4B The NHS must ensure genetic testing laboratories have sufficient resources and funding to carry out testing quickly.

4C The NHS must ensure that patients are informed of their right to have risk-reducing surgery in another location if their wait is more than 18 weeks.

Priority 5 Follow up after testing

After patients receive their genetic testing results, there are inconsistencies in their experiences of referrals for screening and surgery. This is not just in psychological support, but also contact about screening and risk-reducing surgeries:

"I feel that I have been very much left to my own devices with no follow up from the medical profession unless I initiate it."

"In my experience, mutation is diagnosed and no further follow up from anyone. I have to go to my GP annually and ask to be referred for an ovarian ultrasound."

"[We need] a joined up approach between providers and an understanding that whilst some of us don't have cancer, we're still freaking out massively at the mere possibility of it."

This post-testing support is just as vital when results do not show a mutated BRCA gene. Research is evolving into new inherited gene mutations that still increase cancer risk. Patients found not to have a BRCA gene mutation are having to turn to other services so they do not lose out:

"There needs to be support after receiving results revealing that you don't have a BRCA gene mutation. No further advice was forthcoming for me and I was merely told that I had a moderate risk of breast cancer. I have had to research the best way to deal with this myself as I cannot be given regular mammograms on the NHS. Luckily, Action Cancer in Northern Ireland offer a free breast screening service which I will avail of."

OBSERVATION

Guy's & St Thomas' BRCA family services clinic

A monthly BRCA clinic is held at Guy's & St Thomas' hospital for patients who carry a BRCA gene mutation.

The clinic is attended by the whole multi-disciplinary team -the breast team (breast surgeon/plastic surgeon/breast care nurse); Gynae-oncologist; Oncologist (if patient has a current cancer diagnosis); Clinical Psychologist; Research team; Clinical genetics team. The patient can request which clinicians they would like to see on the day of the clinic and highlight any concerns or questions they may have.

Patients are invited to attend face-to-face but can have telephone consultations if they prefer. Patients are encouraged to bring a support person to the clinic days at Guy's hospital where they will spend approximately 2-3 hours. They are allocated their own consultation room and are visited in turn by the staff they have requested time with. Patients are encouraged to discuss any worries or concerns they may have. They can also review information about topics such as screening options, risk-reducing options, talking to their family members and prenatal options. Individually tailoring each patient's appointments allows the clinic to offer an individual plan for risk management to each patient.

Patients are seen in a morning or afternoon session, with a multi-disciplinary team meeting in the middle of the day to discuss each patient attending the clinic. This meeting ensures that the best plan of management for each patient is made with input from each speciality.

Once a patient is part of the BRCA service they can request to come back to the clinic when needed. The BRCA Family Service encourages an "open door policy" to allow patients to have access to professional support at relevant stages of a patient's life. Genetics clinicians are also available on the phone on weekdays from 9-5 to answer patient queries and provide support. Issues raised during the patient's visit to clinic are followed up by the relevant clinician. If patients want to proceed with risk-reducing surgeries then their GP will refer them to the relevant specialist to take this forward. Standard follow-up invites are sent at specified ages to discuss relevant management options.

The BRCA clinic is part of the BRCA family service, which involves update days, newsletters and a BRCA database so patients can be recalled at appropriate times. The monthly clinic consists of approximately 10 patients and has a waiting list for patients of approximately 2-3 months.

It was initially set up as a pilot in 2006 with funding from the Guy's & St Thomas' charity, but subsequent funding has been taken over by the Trust. Service use has been steadily increasing, resulting in additional clinics being added. The clinic has high satisfaction rates and non-attendance is low at less than 5%. The clinic adapts to changes in needs of patients and works to provide the best service to its patients. The BRCA family service is held in high esteem both within the trust and internationally.

AROUND THE WORLD

Regular follow-up action

A Finnish working group - set up by the Finnish Medical Society Duodecim and the Finnish Gynaecological Association - advised women with BRCA1/2 mutations should have annual gynaecological visits that involve a gynaecological examination, transvaginal echography and a CA-125 test. Measures should be put in place to reduce the cancer-risk.[40 41]

Sweden's Regional Cancer Centre ovarian cancer care programme recommends that healthy women with a BRCA1/2 gene mutation have annual follow-ups. Psychosocial care should be included in these appointments alongside the standard monitoring tests. Furthermore, women with a BRCA gene mutation and potential mutation carriers should have a regular gynaecological contact.[42]

Barriers

NHS resources – In a time of stretched NHS budgets, resources for BRCA services can be harder to justify. Individuals championing better BRCA services within NHS Trusts are necessary to drive change.

Next steps

Patients falling off the radar after receiving their genetic testing results is unacceptable and can cause negative consequences in both their mental and physical health.

The NHS must explore the most effective means of keeping in touch with patients with a BRCA gene mutation on an annual basis, whether this is through face-to face appointments, by telephone or by invitations. This could be done in a similar way to screening invitations for cervical cancer.

Once the patient reaches the age for relevant screening and risk-reducing surgery, the responsibility should lie with the NHS to reach out to them to inform them of this.

Some patients will be found not have a BRCA mutation, but are still deemed to have a higher risk of cancer or a variant of unknown significance, a mutation where the risk is still unknown. These patients should be kept informed of research developments that could affect their treatment in the future. ■

RECOMMENDATIONS

5A The NHS must find a mechanism to keep in touch with all patients found to have a BRCA gene mutation on an annual basis.

5B When a patient reaches the relevant age, the NHS must send reminders for eligibility for breast screening, (and ovarian cancer screening once a method becomes available) and for risk-reducing surgery.

5C Patients who are found not to have a BRCA mutation, but are at a higher risk of ovarian cancer, should be kept informed of research development that could affect their treatment in the future.

5D All NHS Trusts should introduce a BRCA family service, and BRCA clinic, run with the multi-disciplinary team.

Ovarian Cancer Action

We will continue to keep our website up to date with research developments that could affect those without a BRCA mutation but still found to be at high-risk of cancer."

"I feel that I have been very much left to my own devices with no follow up from the medical profession unless I initiate it"

Priority 6 Men and BRCA

Despite common misconceptions, men are just as likely to carry a BRCA gene mutation as women and have the same probability of passing it onto their children.

Men are less likely than women to have even heard of the BRCA gene mutation. Our Family contact survey of the general public showed that 83% of men have never heard of BRCA compared to 59% of women.

This is worrying, as men need to have awareness for their own increased risk of breast, prostate and pancreatic cancer.

Beyond their own health risks are the implications for the man's family. A man becoming aware of his BRCA status may well be the bridge to his daughters, sisters and aunts finding out their status and being able to take risk-reducing action themselves. For every man who is unaware of the importance of his BRCA status, there could potentially be an entire family at risk.

Unfortunately it is not just members of the public that are unaware that men can carry the BRCA gene mutation. Our survey found that GPs and healthcare professionals were also not fully informed about the importance of family history in males and cancer history on the men's side of the family:

"[there was] misinformation / lack of information in healthcare professionals about whether men can be BRCA carriers."

AROUND THE WORLD

Understanding risk in BRCA positive males

USA: NCCN Guidelines: "Men testing positive for a BRCA1/2 mutation should have an annual clinical breast examination, and undergo training in breast self-examination with regular monthly practice starting at age 35 years. Regularly scheduled mammography is not recommended by the panel, as there are only limited data to support breast imaging in men, since male breast cancer is rare. Screening for prostate cancer starting at age 40 years should be recommended for BRCA2 carriers and considered for BRCA1 carriers."[43]

"[there was] misinformation/ lack of information in healthcare professionals about whether men can be BRCA carriers"

Barriers

Framing the discussion – BRCA is often talked about in relation to breast and ovarian cancer risk, so it is not surprising that the general population are not fully aware that men can have BRCA gene mutations. When BRCA is seen as a women's issue, half of the target population for cancer prevention will be missed. The broader problem is that men aren't engaged in BRCA groups- of the respondents in our BRCA survey, only 8 out of the 531 respondents were men, despite active outreach for male participants.

Overcoming myths – There are several myths surrounding BRCA gene mutations that need to be overcome. Our survey revealed that these myths, including only women being carriers, and BRCA gene mutations only being passed down the maternal line, are dangerous and important to quell – especially when they are believed by healthcare professionals.

"[We need] more publicity about genes being passed down by male family members. I thought I didn't have to worry about BRCA genes as there was no breast cancer and lots of males in my family."

Next Steps

Men are just as likely to have a BRCA gene mutation as women, therefore 50% of the BRCA patient population should be men. The Government should commission research into patients undergoing genetic testing to ascertain the gender balance of patients and identify the true picture of whether men are aware of their BRCA status.

CASE STUDY ALAN BLASSBERG

Pink and blue.

"The majority of people do not realise men can be BRCA+."

"I received quite an education testing positive for the BRCA mutation after my BRCA2 positive sister passed away from triple negative breast cancer. Sitting with 20 women in an oncology waiting room filling in paperwork about vaginal dryness or pregnancy certainly makes you aware of the pink walls around you. Certain steps have to be made to make men feel more comfortable and help dispel the social stigma associated with a so called female disease.

Everyone knows the pink side of things in the cancer realm, but the majority of people do not realize men can be BRCA+ and get breast cancer too. In fact, a higher percentage of men die from breast cancer than women because they are unaware and don't undergo genetic testing – so male breast cancer is often found at a later stage.

Carrying the mutation also increases a man's risks for other cancers, and a parent that carries the mutation has a 50 percent chance of passing the gene on to either their son or daughter.

This is not about one colour or the other, but rather how we can work together, because greater awareness of these facts is paramount in saving lives." ■

Alan Blassberg is the Director/Producer of the documentary 'Pink and Blue: Colors of Hereditary Cancer'

A common complaint from men who go through the genetic testing process is that it is geared towards women: questions about pregnancy and vaginal dryness and resource booklets with only a few sentences about male BRCA carriers. Tailored resources and support for men is essential going forward

"I really feel there is no support from a male's point of view at all." ■

RECOMMENDATIONS

6A A national public awareness campaign about BRCA mutations in men and the importance of considering family history on both male and female sides.

6B A national awareness campaign for medical professionals to raise awareness of BRCA mutations in men and the importance of considering family history on both male and female sides.

6C The NHS should commission research into patients undergoing genetic testing, to identify the true picture of men aware of their BRCA status.

6D The NHS must have separate forms, resources and tailored psychological support for men going through the process of BRCA testing.

Ovarian Cancer Action

We will continue to raise awareness of BRCA mutations in men, and explore new methods to engage men in conversations around BRCA.

Priority 7 Devolved nations

The devolved nations across the UK each run their own health services independently and the picture of BRCA testing and the barriers are different in each of the four countries.

Our previous research into BRCA testing in the four nations was undertaken in 2014.[44] Since this time there have been changes, not least Wales' lowering of the eligibility threshold from 20% to 10%.

Our BRCA survey included respondents from England, Scotland, Wales and Northern Ireland. However, it did not include sufficient respondents from the devolved nations for us to draw conclusions about each country's specific picture and challenges. Coupled with missing data from the UK Genetic Testing Network from Wales and Northern Ireland, there is not enough quantitative data to make country specific recommendations at this time.

There is real need for an updated piece of in-depth research to analyse the full picture of strengths and barriers areas for development in the devolved nations and to understand each country's unique challenges.

While this report primarily focusses on the barriers in the NHS in England, many of them were echoed across the nations.

Respondents from Scotland, Wales and Northern Ireland gave their priorities for where their country's guidelines needed to be built on:

Scotland
The Scottish Intercollegiate Guidelines Network (SIGN) has recommended BRCA testing for all women diagnosed with non-mucinous ovarian cancer since 2013.

SIGN guideline 135: Management of epithelial ovarian cancer states:

- All women with non-mucinous ovarian or fallopian tube cancer should be offered BRCA1 and BRCA2 mutation testing.
- BRCA1 and BRCA2 mutation analysis should be considered in a family where there is a 10% or greater risk of a mutation being present.[45]

"Try to make wait times fair. Differs so much between health boards."

"More information pre-operatively and support post op... I had nothing and it really has affected me."

"If results are positive have more mental health support afterwards as feel this really let the whole thing down."

CASE STUDY
CHRISTINA, EDINBURGH

"I feel I have been lucky with my BRCA testing experience in the sense that it all happened relatively quickly and smoothly.

However this isn't the case for all women going through this process. Having spoken with other BRCA positive women across the UK, it would appear that there are inherent disparities in guidance and support given based on geographical region.

I would like to see a universal approach to genetics testing and support across the UK where there are clear milestones and guidelines covering the initial appointment with your GP, through to surgical and screening options.

Also, information on counselling services that are available locally, as this is an invaluable part of this journey. I would also like to see GP awareness raised to ensure patients are given the correct information from the onset of their genetics process."

"All women that are diagnosed with ovarian cancer should be tested and more support groups should be set up"

Wales
As of July 2015, Wales has been following NICE Clinical Guidelines 164. Prior to this, Wales was only testing individuals with over 20% chance of having a mutation, despite the threshold being lowered in the rest of the UK.

"Regular way of being updated with risks and new treatments after surgery. I would also like to be made aware of additional risks e.g pancreatic cancer for BRCA carriers."

"More counselling after the surgery has taken place. I feel like I had plenty of people to talk to before my surgery but nobody to talk to afterwards."

"In terms of ovarian cancer I feel I seriously lacked information regarding preventive surgery."

"Some form of-post surgery follow up even every 5 years to check on how things are, and to give information. Equal access to same surgery and reconstruction, and better HRT advice please."

Northern Ireland
Since 2013, the Health and Social Care endorsed NICE Clinical Guidelines 164.

"Private healthcare should cover preventative surgery. More information on HRT and more information on the types of surgery."

"All women that are diagnosed with ovarian cancer should be tested and more support groups should be set up."

CASE STUDY
EMMA, NORTH WALES

"My initial meeting with the genetics team was positive as they covered aspects I hadn't considered if a BRCA mutation was present.

I was comforted in knowing there were annual follow ups, however this is no longer the case. Personally I found there are inconsistencies with the age a risk-reducing surgery is offered and whether a hysterectomy is also advised. Ideally the genetic testing should have consistent advice and through every stage of testing, surgery and follow up."

"My older sister was offered counselling, nobody has mentioned counselling to me. More counseling should be offered."

CASE STUDY
GWYNETH, BELFAST

"There are inadequate resources within the Regional Genetic Service for genetic testing.

There is a waiting time of about six months for patients to be seen for genetic testing. Women diagnosed with triple negative breast cancer are often making decisions regarding treatment options without the knowledge of their genetic status.

There is also very limited testing of non-affected people with a family history of breast and ovarian cancer.Often women are given the results of their genetic test over the phone with little or no follow up from the genetic counsellors."

Gwyneth Hinds, Associate Specialist, Breast Family History Clinic

RECOMMENDATIONS

7A The UK Genetic Testing Network must ensure data is captured across England, Scotland, Wales and Northern Ireland when monitoring activity of genetic tests.

Ovarian Cancer Action
We will carry out an update to our 'BRCA1/2 gene testing and Ovarian Cancer: The UK picture' 2014 report.

Conclusion

Ovarian cancer claims the life of a woman every two hours.

While treatment options are limited and survival rates remain low, cancer prevention is one of our strongest weapons in the fight against this disease.

Around 15% of cases of ovarian cancer are linked to BRCA gene mutations. BRCA testing gives women who have a high risk of ovarian and breast cancer the power to change their future. The Government has made important progress in improving access to testing in the last two years. However, our research has found that there are still barriers in a number of areas that are preventing BRCA testing from fulfilling its potential as a cancer prevention tool. We've made a series of recommendations to break down these barriers and save lives.

We're calling on the Government to implement these recommendations, and act on BRCA to stop women dying before their time.

Priority 1: Testing
Recommendation 1A: The UK Genetic Testing Network must capture disease specific data when monitoring activity of genetic tests.

Recommendation 1B: The NHS must find a mechanism to collect data to monitor implementation of clinical commissioning policy E01/pb at CCG level.

Recommendation 1C: Clinical Commissioning Policy E01/pb to include a timeframe for offering BRCA testing within four weeks of ovarian cancer diagnosis.

Recommendation 1D: BRCA testing must be embedded into NICE CG122 at the point of diagnosis.

Recommendation 1E: Each NHS Trust must have an explicit team responsible and accountable for offering BRCA testing.

Recommendation 1F: The NHS must reach out to women diagnosed with ovarian cancer before 2015 to inform them of their eligibility for BRCA testing.

Recommendation 1G: Tumour samples from women with ovarian cancer should be retained as standard so that families are not prevented from accessing testing if patients have died.

Recommendation 1H: Lower the testing eligibility threshold from 10% to 5% probability.

Priority 2: Counselling
Recommendation 2A: Patients should receive tailored emotional and psychological counselling as part of the genetic counselling process in the genetic testing pathway.

Recommendation 2B: Patients diagnosed with a BRCA gene mutation should have access to relevant mental health services adequately trained to support those with a genetic mutation at any point in their BRCA journey, whether pre or post testing, at the time of considering or undergoing risk-reducing surgery or at any point beyond this.

Priority 3: Standardised information
Recommendation 3A: A standardised, evidenced-based digital resource to be created by the NHS and made available to all NHS Trusts and their medical professionals for patients covering (at minimum) the following:

- Screening options for both breast and ovarian cancer including explanations of why certain options are not available
- Evidence-based reviews of each risk-reducing surgery option including recommended age
- HRT options including risks, benefits, and contraindications
- Advice for contacting family members at risk
- Specific advice for men who carry the BRCA gene
- Rights regarding financial concerns such as insurance/ mortgage/ sick leave from work
- Family planning options (PGD)
- Where to get support

This resource must be regularly updated in line with developments and available in a wide range of languages for hard to reach groups.

Recommendation 3B: The NHS must find a mechanism to contact those with a BRCA gene mutation with new relevant medical breakthroughs that could affect their decisions to reduce their cancer risk.

Priority 4: Waiting times
Recommendation 4A: The NHS must instate a target for receipt of results of genetic testing.

Recommendation 4B: The NHS must ensure genetic testing laboratories have sufficient resources and funding to carry out testing quickly.

Recommendation 4C: The NHS must ensure that patients are informed of their right to have risk-reducing surgery in another location if their wait is more than 18 weeks.

Priority 5: Follow up after testing
Recommendation 5A: the NHS must find a mechanism to keep in touch with all patients found to have a BRCA gene mutation on an annual basis.

Recommendation 5B: When a patient reaches the relevant age, the NHS must send reminders for eligibility for breast screening, (and ovarian cancer screening once a method become available) and for risk-reducing surgery.

Recommendation 5C: Patients who are found not to have a BRCA mutation, but are at a higher risk of ovarian cancer should be kept informed of research development that could affect their treatment in the future.

Recommendation 5D: All NHS Trusts should introduce a BRCA family service, and BRCA clinic, run with the multi-disciplinary team.

Priority 6: BRCA and Men
Recommendation 6A: A national public awareness campaign about BRCA mutations in men and the importance of considering family history on both male and female sides.

Recommendation 6B: A national awareness campaign for medical professionals to raise awareness of BRCA mutations in men and the importance of considering family

Recommendation 6C: The NHS should commission research into patients undergoing genetic testing, to identify the true picture of men aware of their BRCA status.

Recommendation 6D: The NHS must have separate forms, resources and tailored psychological support for men going through the process of BRCA testing.

Priority 7: Devolved Nations
Recommendation 7A: The UK Genetic Testing Network must ensure data is captured across England, Scotland, Wales and Northern Ireland when monitoring activity of genetic tests.

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