The Health Policy Partnership

Genetic testing for *BRCA* mutations:

country profile for United Kingdom

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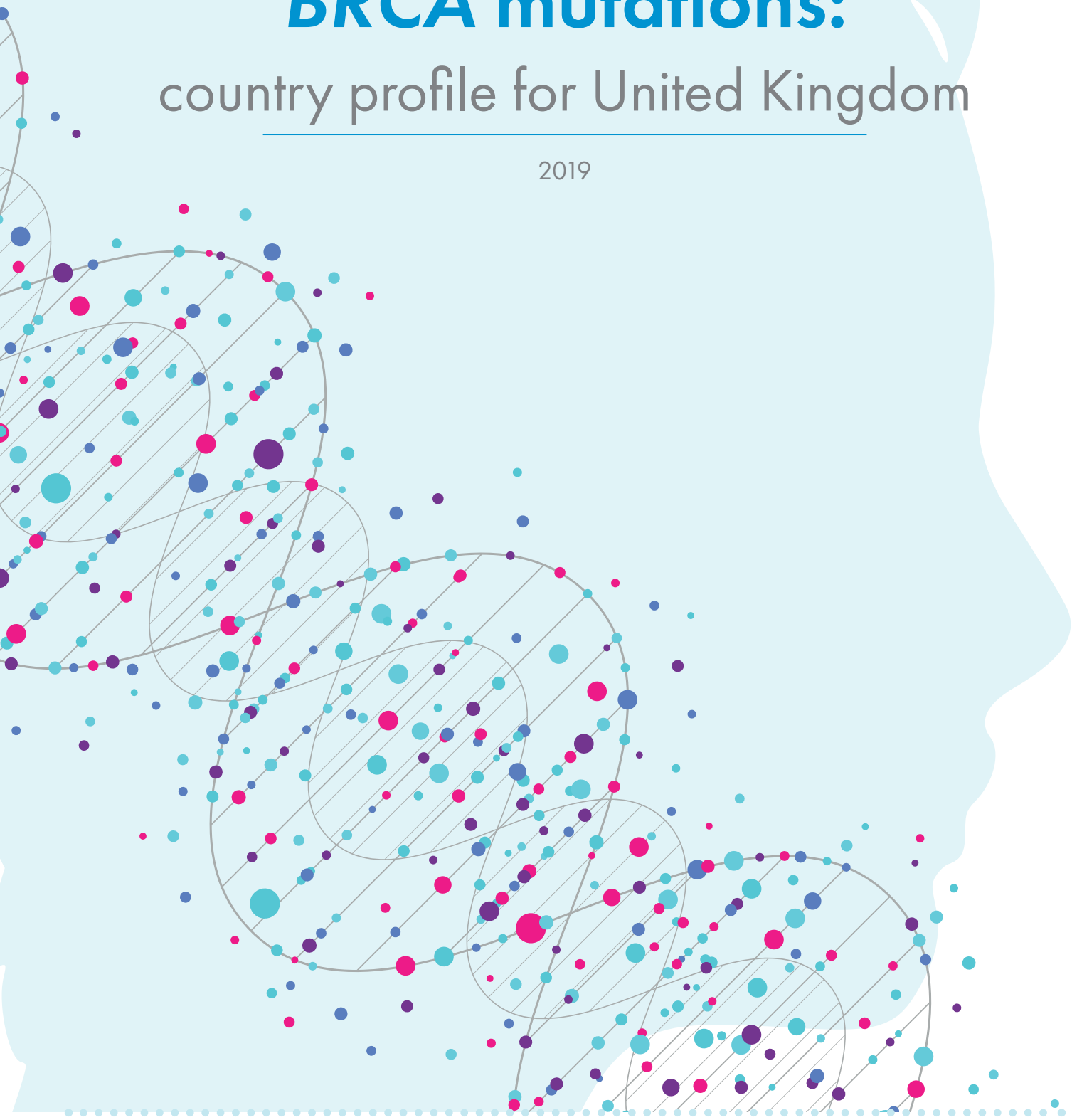

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The experts who provided interviews and/or feedback for this country profile are:

- **Gareth Evans**
  Professor in Medical Genetics and Cancer Epidemiology,
  University of Manchester

- **Nick Meade**
  Director of Policy, Genetic Alliance UK

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Key facts and figures on breast cancer and *BRCA* mutations in the United Kingdom

- The UK has the 5th highest incidence rate and the 15th highest mortality rate of female breast cancer in Europe.[1]
- Between 1.5% and 2% of all breast cancer cases in the UK are due to *BRCA* mutations,[2 3 4] although this rises to around 3–7% among those under 35.[2 3]
- It is thought that 16% of all hereditary breast cancer cases in the UK are associated with *BRCA1* or *BRCA2* mutations.[5]
- 60% of women with a *BRCA1* mutation and 55% of those with a *BRCA2* mutation will develop breast cancer by age 70.[6] This compares to a risk of around 9% among all women by age 74.[7]

Introduction

Mutations in the *BRCA1* and *BRCA2* genes put women at significant risk of developing breast cancer. Based on international data, it is thought that most women have a 12.5% risk of developing breast cancer in their lifetime. This risk increases to 60–90% among women with a *BRCA1* mutation and 45–85% among women with a *BRCA2* mutation.[8-10] Breast cancers related to *BRCA1* mutations are more likely to be triple-negative breast cancer, which is difficult to treat.[11]

It is important for women to know whether they carry a *BRCA* mutation. *BRCA*-mutation carriers who are asymptomatic can be supported to make an informed choice to reduce their risk of developing breast cancer through, for example, preventive surgery and, in some countries, chemoprevention. They can also reduce their chance of breast cancer mortality through regular monitoring which can pick up earlier cases of the disease. Knowing whether a woman with breast cancer carries a *BRCA* mutation can also help to inform decisions around treatment and surgery. In addition, identifying *BRCA*-mutation carriers allows for family members to be tested.[12]

Men can also benefit from knowing they carry a *BRCA* mutation. *BRCA* mutations can increase the risk of prostate and other cancers, so men with identified mutations can be closely monitored for early signs of the disease.[13] In addition, men who carry a *BRCA* mutation can pass it on to their daughters.

However, many people at high risk of *BRCA*-related breast cancer cannot access a genetic test, while many who have been found to carry a *BRCA* mutation face gaps in accessing ongoing care and support.

This country profile presents an overview of the *BRCA* policy landscape in the UK with a view to guiding policy development. It considers gaps, challenges and advances in four areas policymakers should prioritise when looking to improve access to genetic testing and care for people with *BRCA* mutations:

1. Comprehensive, evidence-based policies for *BRCA* testing and the management of *BRCA*-related breast cancer
2. Greater public and patient understanding
3. Comprehensive care pathways
4. Improved awareness and knowledge among healthcare professionals.

Comprehensive, evidence-based policies for *BRCA* testing and the management of *BRCA*-related breast cancer

The UK has a clear strategic vision that recognises the importance of improving the prevention of hereditary cancers, including those related to *BRCA*.

Inclusion of *BRCA* genetic testing in national plans for cancer and genetics

The UK has committed to improving early diagnosis and care for patients with rare diseases, including hereditary cancers. In 2013, the Department of Health published a strategy for rare diseases, with input from all four UK countries. The strategy contains 51 commitments which aim to ensure that all people living with rare conditions, including hereditary cancers, have access to the high-quality care and treatment they need. These commitments include supporting and empowering patients, improving awareness and supporting effective coordination among healthcare professionals.[14] Scotland published its implementation plan for this strategy in 2014,[15] Northern Ireland in 2015[16] and Wales in 2017.[17] Scotland recently reported on progress in implementing its plan, highlighting progress in the availability of testing for somatic (acquired) *BRCA1* and *BRCA2* mutations.[18]

Unlike the UK's other countries, England only recently published its implementation plan, following an inquiry from the All-Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions.[19] The inquiry found that neither National Health Service (NHS) England nor the UK's Department of Health and Social Care was taking responsibility for implementing the strategy, leaving services poorly coordinated and patients unable to access the care they need. The plans for England have now been published[20] but patient support organisations argue that, with only two years until the strategy is due to end, it is time for it to be refreshed.[21]

Despite the lack of a rare diseases plan for England until recently, NHS England has invested heavily in advancing diagnosis for people with genetic disorders. It has, for example, been working in partnership with the private sector to provide quicker and more accurate diagnoses, including for people with hereditary breast cancer. The 100,000 Genomes Project established by Genomics England in 2012 forms part of this ambition. The project has sequenced 100,000 genomes from people with rare diseases and certain cancers, along with their families, in order to help with faster diagnosis and identify the most effective treatments.[22] Northern Ireland, Scotland and Wales have agreements in place so that their citizens can also take part in the project.[23-25]

NHS England is building on this investment by establishing the NHS Genomic Medicine Service, which aims to make genetic testing accessible to all in England. The service is establishing a national genomic laboratory network,[26] a National Genomic Test Directory, 13 Genomic Medicine Centres which provide a clinical service,[27] and a Genomics Unit within NHS England.[28]

National policies support cost-free access to *BRCA* genetic testing and personalised follow-up care

Genetic testing, care and follow-up for those eligible is fully funded by the NHS.[28] Cost is therefore unlikely to be a barrier for people considered at high risk.

National registries to collect data on *BRCA* genetic testing

There appear to be very few recent, national data on *BRCA* mutations, which may be hampering effective policymaking and planning. There seem to be no comprehensive registries collecting data on *BRCA* mutations. The Cancer Variant Interpretation Group (C-VIG), however, is an initiative delivered through Genomics England which draws on *BRCA*-mutation data from laboratories across England to discuss and classify specific variants.[29] In addition, there are plans to roll out the National Hereditary Cancer Registry in England, which would focus initially on *BRCA* mutations.[30]

Legal protection of *BRCA*-mutation carriers

Anti-discrimination legislation in the UK does not cover discrimination based on genetic predisposition for disease. In 2002, the statutory body charged with monitoring the Disability Discrimination Act (now absorbed into the Equality Act 2010) recommended the Act be extended to cover people who have a genetic predisposition. This would have made it illegal for an employer to require an individual to undergo a genetic test; however, the extension has not yet been adopted.[31] [32] The issue of genetic discrimination has continued to been debated, including during a seminar organised by the Human Genetics Commission, but a gap in legislation remains.[32]

Insurance companies in the UK have, however, signed up to a voluntary code of practice to protect people from genetic discrimination. The UK Government and Association of British Insurers have an agreement in place which features a number of commitments, including not treating an applicant differently based on test results or requiring an applicant to share results of a predictive test, except in certain cases, for example where life insurance cover exceeds £500,000. It also prevents insurance companies from imposing conditions, exclusions or disproportionate terms based on genetic test results.[33] The code does not, however, prevent insurance companies from requesting information regarding an individual's family history, or acting on this information.[34] Despite these gaps in legislation, an expert interviewed for this country profile noted that few, if any, cases of discrimination have occurred in the UK.[34]

Greater public and patient understanding of *BRCA* mutations and their association with breast cancer

Awareness of *BRCA* mutations has been raised through the efforts of patient organisations and media reporting.

A number of breast cancer advocacy groups have focused efforts on raising awareness about *BRCA* gene mutations, and support people who may be considering testing. *BRCA* Umbrella, for example, works to raise awareness of *BRCA* mutations and supports those who may be considering or undergoing testing. It provides an online community of *BRCA*-mutation carriers by encouraging conversation, support and information-sharing.[35] Other organisations that are active in promoting awareness include the National Hereditary Breast Cancer Helpline and Breast Cancer Now.

The media also have a powerful role in improving awareness – as was demonstrated when actress Angelina Jolie spoke openly about her *BRCA* test results. One study estimated that related media coverage led to a tenfold increase in calls to the UK's National Hereditary Breast Cancer Helpline. This, in combination with reporting on changes to eligibility criteria for testing, is thought to have contributed to a 2.5-fold increase in referral rates for *BRCA* testing compared with the same period in the previous year.[36]

Comprehensive care pathways

The UK has clear eligibility criteria and referral pathways for genetic testing, but inequalities in access persist.

Evidence-based national eligibility criteria for *BRCA* testing

The UK has comprehensive guidelines to support the diagnosis, care and management of women who carry a *BRCA* mutation. These guidelines, published by the National Institute for Health and Care Excellence (NICE) in 2013,[37] are used across the UK, are well regarded internationally and are referred to by the European Society for Medical Oncology (ESMO) 2016 guidelines.[38] The NICE guidelines include information on eligibility for genetic testing, referral pathways, taking a family history and providing information and support to those undergoing testing.[37]

Recommendations regarding eligibility for *BRCA* genetic testing have recently been expanded. Guidelines for the management of patients with early or locally advanced breast cancer were published by NICE in 2018. These recommend that all women in England with triple-negative breast cancer under the age of 50 are tested for *BRCA* mutations, regardless of family history.[39] In addition, in Scotland, all women with high-grade ovarian cancer, regardless of their age, have access to *BRCA* genetic testing.[40]

Equitable and timely access to genetic testing

Despite clear eligibility criteria, *BRCA* genetic testing is not being offered to all those in the UK who are eligible. Estimates suggest that there is a gap of over 3,000 women every year who undergo a risk assessment and are eligible for a *BRCA* test but are not offered it.[41] It is likely that, with advances in technology and the ability to identify personalised treatment based on genetic-testing results, the number of people accessing genetic testing will increase.[42]

These gaps in testing are compounded by lower referral rates for women from minority ethnic groups.[43] There are many reasons for this (see *Box 1*). Among South Asian communities, for example, some female patients have reported that discussing breast cancer with male practitioners was difficult and embarrassing. These women and their families may associate cancer with stigma and death, which may make them reluctant to raise the subject with other family members.[44 45]

Box 1 | What are the reasons for lower genetic testing rates among black and minority ethnic women?[43-45]

- Lack of awareness of testing
- Language difficulties and challenges around cross-cultural communication
- Limited awareness of support services available, including genetic counselling
- Culturally held beliefs about breast cancer and stigma related to the condition, which also influence communication with family members

There is also geographic variation in *BRCA* genetic testing throughout the UK. The number of *BRCA* tests performed is lower in England than in Scotland (see *Table 1*). There is also wide variation within England, with some regions such as Yorkshire and the Humber having particularly low rates of *BRCA* testing.[46] Understanding the reasons behind these differences will be critical in ensuring that access to genetic testing services is more evenly spread across the UK.

Table 1 | *BRCA* genetic testing provision across the UK

Country	Number of BRCA genetic tests per 100,000 women in 2016/17[46]	Number of laboratories offering genetic tests for familial breast/ovarian cancer[47]
England	54.2	16
Scotland	69.8	4
Northern Ireland	Data not available	1
Wales	Data not available	1

Two laboratories in the UK, both of which are in England, test samples against a panel of genes.[47] Panel tests for hereditary breast cancer test for mutations in 13 genes associated with an elevated risk of breast and ovarian cancer, using whole-gene sequencing. These laboratories provide results within 42–112 days depending on urgency.[48 49] Panel tests for known mutations in family members, however, take 14 days.[49]

Testing samples against a larger number of genes, however, runs the risk of leading to more uncertain results and greater anxiety. Genetic tests that identify mutations which are new or have an unknown association with cancer risk are referred to as variants of unknown significance (VUS). The VUS rate for *BRCA1/BRCA2* testing is less than 1%, but this increases to 14% when panel testing is used. This has implications for those being tested, who are likely to experience more stress and anxiety than with a more definite result.[50 51] In the longer term, however, more mutations will be identified and the proportion classified as unknown should reduce.

Role of private genetic-testing providers

Although genetic testing, care and follow-up for those who are eligible is fully funded by the NHS, private testing is also available, including for those who are not eligible for a test through the NHS or who do not want to wait. Although data are not available on the number of *BRCA* tests being delivered by private laboratories, experts suggest uptake of private *BRCA* genetic testing is low.[42] Private testing providers include those providing clinical genetic testing services and those that offer direct-to-consumer (DTC) testing. DTC testing is not regulated in the same way as genetic testing in clinical settings.[52] This can mean DTC providers test different, or fewer, mutations than the NHS and may not provide genetic counselling along with their results, potentially leaving people unsupported.[53]

Access to genetic counselling

Appropriate genetic counselling is an essential component of care for those eligible for *BRCA* testing. NICE guidelines recommend that all who are eligible for *BRCA* genetic testing are referred to genetic counselling teams for (preferably) two sessions of pre-test counselling.[37] Genetic counsellors provide support to those being tested as well as their families, to help them understand their breast cancer risk and options available to them.[37]

Psychosocial support following test results is critical. Despite this, psychological support may not always be provided as part of genetic counselling for those undergoing *BRCA* testing. In one survey undertaken by Ovarian Cancer Action, 42% of women who were tested and found to have a *BRCA* mutation received no counselling or support after they received their results.[9]

The UK has one of the highest numbers of genetic counsellors per capita globally. Currently there are around 310 genetic counsellors for the UK population of 60 million.[54] To support genetic counsellor development, the UK has three accredited master's programmes which train around 40 genetic counsellors per year and now include genomics.[55] This number includes 10–20 who are trained in England as part of the Scientist Training Programme (STP). Inclusion in the STP means they are included in NHS workforce planning, and will be awarded statutory regulation as clinical scientists when this is established in 2019.[54 56 57]

Unlike in many other countries in Europe, genetic counsellors in the UK are qualified to work independently or as part of a multidisciplinary team. Registration for genetic counsellors in the UK is voluntary and most work in regional genetics centres or associated outreach clinics.[54]

Despite the higher numbers of genetic counsellors compared with other countries, there are still not enough to meet demand and waiting times can be long in some areas.[9 42] This creates a significant bottleneck, slowing access to *BRCA* genetic testing.[58]

Follow-up care and support for *BRCA*-mutation carriers

Those who receive a positive result from a *BRCA* genetic test and do not have breast cancer have three main options available to them for reducing their risk of developing the disease: surgery, hormonal therapy, and changes to lifestyle coupled with surveillance. Although national data are not available on the extent to which different options are selected, studies have found that 34–40% of asymptomatic women who carry a *BRCA* mutation undertake preventive double mastectomies.[59 60] Uptake of chemoprevention in England is low, with a recent study suggesting that around 15% of women with a *BRCA* mutation initiate this therapy.[61]

Improved awareness and knowledge among healthcare professionals

The UK is investing in improving the genetics knowledge of healthcare professionals who are not genetics specialists.

Gaps in knowledge about *BRCA* testing among non-genetic-specialist healthcare professionals may play a role in lower-than-optimal referral rates and uptake of genetic testing. In a primary-care-driven health system such as the UK, GPs play an important role in addressing patients' initial enquiries and referring them to appropriate specialist genetic services to gather further information on family history, or for testing.[62] Yet a 2006 survey of GPs found that, while they felt genetics was an important topic for practice, they did not believe they had adequate knowledge to best guide their patients.[63]

Other factors may also impact on low rates of referral from primary care to specialist genetics services. These include time constraints and lack of experience in making a referral to a genetics specialist.[64] Poor communication between non-genetic-specialist healthcare professionals and people at risk of *BRCA* mutations may also create a barrier for people in seeking genetic testing, which is exacerbated in those who do not speak English as a first language.[44]

There is evidence that breast cancer specialists may feel uncertain about how to interpret *BRCA* test results or know what guidance to give their patients. For example, in a 2015 survey of 155 breast cancer specialists, 12% reported not having received any genetics training. In the same study, although 95% of the breast cancer specialists surveyed had referred patients for *BRCA* genetic tests, 71% felt unsure about the clinical implications of the test reports presented.[65] Clinicians seemed to particularly struggle to understand and communicate the clinical implications of results when there was no family history of breast cancer.[65]

Health Education England is aiming to address these knowledge gaps by developing the £20 million Genomics Education Programme (GEP) to build knowledge of genetics among healthcare professionals. The GEP is funding 500 master's places and additional healthcare science training places, and provides access to a range of genetics educational resources and tools for healthcare professionals, including for those working in primary care.[66]

Conclusions and recommendations

People with *BRCA* mutations in the UK are supported by comprehensive clinical guidelines, strong patient support organisations and well-developed services. Furthermore, existing policy initiatives seek to improve early detection for people with rare and genetic cancers.[14] Despite this, there are several gaps which must be addressed to ensure that all *BRCA*-mutation carriers have access to the comprehensive care and support they need.

Genetic testing services and information must be tailored to ensure they are accessible to all, regardless of location or ethnic background. Geographic and ethnic inequalities in awareness of and access to genetic testing[43-47] are a serious concern in the UK and must be addressed.

Plans are needed to manage the increasing demand for *BRCA* testing, to ensure that all *BRCA*-mutation carriers can access a timely genetic test and be supported as they manage their results. As treatments for *BRCA*-related breast cancer become available, demand for genetic testing is likely to increase, putting strain on existing infrastructure and workforce. Scrutiny is needed to ensure that services can keep up with this rising demand.

Greater efforts are required to ensure that everyone with a *BRCA* mutation is able to access the full range of care they need, including psychosocial care both before they are tested and following their results. The UK has one of the highest numbers of genetic counsellors per capita,[54] but waiting times vary and not all women receive the psychological support they need as part of their genetic counselling.[9]

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