



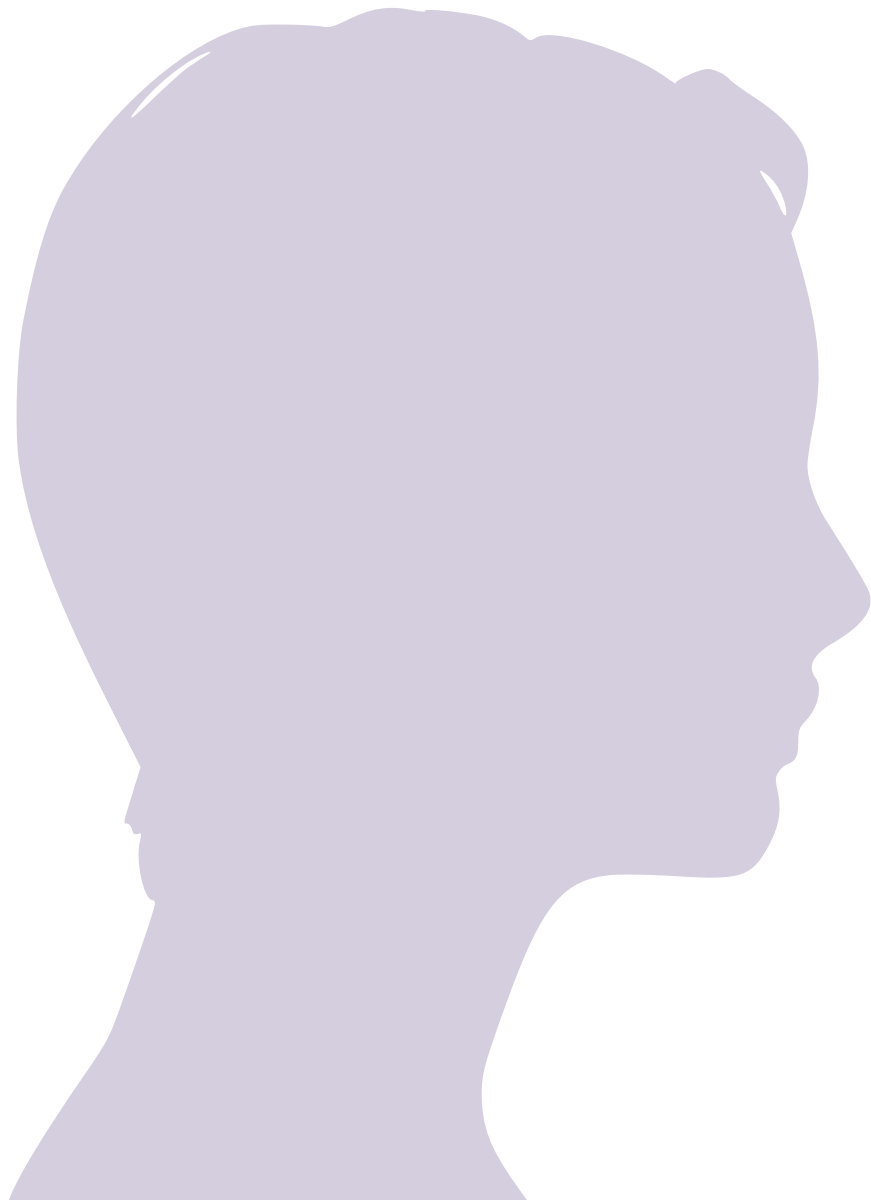
Genetic testing for *BRCA* mutations: country profile for Ireland

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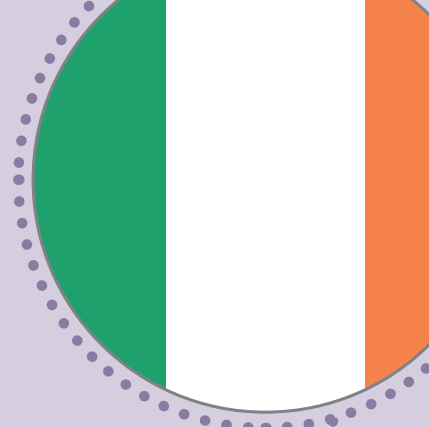
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Genetic testing for *BRCA* mutations: country profile for Ireland



Key facts and figures on breast cancer and *BRCA* mutations in Ireland

- Ireland has the 11th highest incidence rate and the 3rd highest mortality rate of female breast cancer in Europe.¹
- It is estimated that there were 3,334 new cases and 791 deaths from breast cancer in 2018.¹
- There are no national data on the proportion of breast cancer cases which are due to *BRCA* mutations.



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.²⁻⁴ Breast cancers related to *BRCA1* mutations are more likely to be triple-negative breast cancer, which is difficult to treat.⁵

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.⁶

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.⁷ 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 Ireland 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

Significant gaps in hereditary cancer services have been identified by the Irish government, which has developed plans to boost access to comprehensive care for people with *BRCA* mutations.

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

There is no comprehensive national policy on genetics and genetic testing in Ireland, but there is a clear intention to scale-up access to these services.⁸ In 2012, the National Cancer Control Programme (NCCP) established the Hereditary Cancer Programme to scale-up advanced diagnostics in genetics and meet increasing demand from healthcare professionals and patients for these services.⁹ The programme is implemented in partnership with the Department of Clinical Genetics at Our Lady's Children's Hospital, Crumlin (OLCHC), with St James's Hospital acting as the National Clinical Lead for cancer genetics.⁹

The government has identified significant gaps in the Hereditary Cancer Programme and outlined a range of recommendations to improve access to genetic services as part of its National Cancer Strategy 2017–2026. The strategy recognises that the Hereditary Cancer Programme is underfunded and underdeveloped.⁹ It plans to boost staffing by specialist healthcare professionals and training on cancer genetics among oncology graduates. It also calls for the Health Service Executive (HSE) to combine existing services under a coordinated National Cancer Genetics Service.⁹

National registries to collect data on *BRCA* genetic testing

There is a lack of data on *BRCA*-mutation carriers in Ireland, hampering the government's ability to make evidence-based policy decisions. There are no national registries which collect data on *BRCA* mutations and *BRCA*-related breast cancer.

Legal protection of *BRCA*-mutation carriers

Under Irish law, genetic data cannot be used for insurance, pension, mortgage or employment purposes.¹⁰ However, there are no further anti-discrimination regulations with regard to genetic data.¹¹ In addition, Ireland has not signed the European Convention on Human Rights and Biomedicine, which protects individuals in other European countries from discrimination based on genetic test results and mandates the individual's consent and right to be informed about genetic test results.¹²

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

People in Ireland have access to a limited range of information on *BRCA* mutations and *BRCA*-related breast cancer, although national patient organisations are working to address this and raise awareness among the population.

Public awareness about hereditary breast cancer and genetic testing has increased, contributing to a significant rise in referrals for genetic testing.⁹ Referrals to cancer genetics increased by 60% between 2000 and 2013, and testing for *BRCA* mutations is the most common reason for a request.⁸ A large proportion of these referrals are healthy women worried about their genetic predisposition.⁹

A limited range of online and printed information from Ireland is available for people who are concerned about their risk of hereditary breast cancer.¹³ Action Breast Cancer, a programme of the Irish Cancer Society, provides information about the likelihood of a *BRCA* mutation and the process of genetic testing.¹⁴ Comprehensive information is also available from patient support organisations such as the Marie Keating Foundation.

There appear to be gaps in additional support available for those with *BRCA* mutations in Ireland.

Until recently there were no peer-support groups for people with *BRCA* mutations, nor *BRCA*-specific patient organisations – but the Marie Keating Foundation has now developed a peer programme through which volunteers have been trained on topics including a patient's journey from genetic testing to surgery and strengthening self-care and resilience. The programme, which was launched in April 2019, allows individuals to submit questions through the organisation's website, which are then answered in confidence by a trained volunteer or a *BRCA* expert.¹⁵ The Foundation also offers workshops and seminars for those at high risk of breast cancer.¹⁶ Furthermore, in 2016, Cancer Trials Ireland organised a *BRCA* conference for people with a family history of breast cancer.¹⁷



Comprehensive care pathways

Restrictive eligibility criteria for genetic testing and limited capacity have an impact on access to and uptake of *BRCA* genetic testing and follow-up care.

Evidence-based national eligibility criteria for *BRCA* testing

Ireland has established criteria for genetic testing for hereditary breast cancer, but these are more restrictive than in many other European countries.^{13 18} The criteria, developed by the National Centre for Genetic Medicine (NCMG), define those who may be considered at high risk of carrying a *BRCA* mutation and are therefore eligible for referral by a specialist or their GP for a consultation. This may lead to either diagnostic genetic testing in patients with existing breast cancer, or predictive genetic testing in those without symptoms, to confirm whether they carry the *BRCA* mutation.

In contrast with many other European countries, Ireland does not make genetic testing available to all asymptomatic women at high risk of *BRCA*-related breast cancer.¹⁸ The NCCP noted that, with the currently available resources, the introduction of testing for all women at high risk regardless of whether they are affected by cancer – as is practised in the UK – is not feasible.⁸

Equitable and timely access to genetic testing

Three public hospitals offer genetics services to people considering *BRCA* testing in Ireland. However, it has been noted that with current resourcing, these hospitals are not equipped to provide comprehensive genetics services on a national level.⁹

A lack of investment in cancer genetics has led to the low availability of oncologists with specialist training in cancer genetics. Such healthcare professionals are needed to lead on cancer genetics in their local cancer centres and to coordinate with NCMG.⁹

Consequently, waiting times for an initial consultation, genetic testing and counselling are high, placing a burden on people waiting for tests and potentially delaying initiation of necessary treatment.⁹ People may have to wait up to 12 months to be seen in a cancer genetics clinic after the initial referral.¹⁸ Only half of those referred have an initial appointment within the first six months of referral.⁸

Role of private genetic testing providers

Long waiting times and strict eligibility criteria have led some people to choose private BRCA genetic testing at high cost.¹³ Private clinic-based and direct-to-consumer (DTC) testing for BRCA mutations are becoming increasingly popular.^{11 13 19} The test costs between €1,400 and €1,800²⁰ and, although it is now covered by one private health insurance company,²¹ cost is likely to be a barrier for some people.¹³

Furthermore, limited regulation and oversight have led to concerns about the quality of genetic testing services provided in the private sector in Ireland.

There is a lack of standards to ensure the clinical and analytical validity of genetic test results, leading to the potential for misinterpretation. In addition, advertising of DTC genetic testing is not regulated under Irish law, and genetic counselling is often not provided. This may contribute to increased distress and anxiety among those being tested.¹¹

Access to genetic counselling

Genetic counselling is not required by law but it is demanded by the Irish Medical Council and recognised as critical in the National Cancer Strategy.¹¹ The National Cancer Strategy also recognises the need for significant investment in the genetic counsellor and genetic nursing workforce.^{8 9}

In proportion to its population, Ireland has one of the lowest numbers of genetic counsellors and genetic consultants in Europe. In 2011, Ireland had only six geneticists and six genetic counsellors; the recommended numbers, given the population size, would be 14 and 46, respectively.²²

Novel forms of genetic counselling are being explored and could improve access and quality of cancer genetics services. The National Cancer Strategy proposes telemedicine services, which are already provided by some centres as a possible option for genetics counselling.⁹ A recent study showed that the majority of people surveyed value having the choice between different disclosure methods, and about 45% preferred to receive their results via post or telephone.²³

Follow-up care and support for BRCA-mutation carriers

Recommendations for surveillance of BRCA-mutation carriers are in place but are not implemented on a nationwide basis. The Health Information and Quality Authority (HIQA) developed recommendations for surveillance of BRCA-mutation carriers as part of a comprehensive health technology assessment. Recommendations include breast cancer surveillance for women with an identified genetic breast cancer mutation and annual digital mammography for women aged 40–49 who have high familial risk but no identified genetic mutation. To ensure equity of access, the HIQA suggests establishing an organised surveillance programme at a national level.²⁴ Despite this, free annual breast checks are currently offered only to women aged 50–69 on a routine basis, regardless of their risk status.²⁵

There is limited guidance available for the management of women with BRCA mutations.

The National Clinical Guideline for breast cancer provides recommendations for women with BRCA-related breast cancer,²⁶ but guidance for reducing breast cancer risk among asymptomatic BRCA-mutation carriers appears to be lacking. This may be contributing to low uptake of preventive double mastectomy. One study found that only 12% of asymptomatic BRCA-mutation carriers in Ireland decided to take up this surgery.²⁷

Improved awareness and knowledge among healthcare professionals

Knowledge gaps among GPs in Ireland limit their ability to facilitate access to genetic testing.

Non-genetic-specialist healthcare professionals, such as GPs, can request BRCA-mutation testing for their patients. NCMG has published guidelines for genetic testing and risk assessment for primary care practitioners.²⁸ These explain the process of genetic testing in Ireland. The NCCP has also developed a guidance document for non-genetics healthcare professionals.²⁹

However, GPs may not be equipped to adequately respond to increased interest in testing for genetic cancer predispositions or to pre-assess their patients' risk. Patients may seek help from GPs to discuss potential genetic risks or query their test results from DTC genetic testing services.⁹ In a national survey in 2016, around 90% of GPs who responded said they did not have sufficient information to discuss genetic testing with their cancer patients.³⁰ In response to the role of GPs as a gatekeeper for referrals to cancer genetics centres, the NCCP has called for an assessment service for GP referrals.⁹

Conclusions and recommendations

Ireland faces some significant challenges in meeting the needs of people who carry *BRCA* mutations. Eligibility criteria for *BRCA* genetic testing are more restrictive than in many countries in Europe, meaning that many people who may be carrying a *BRCA* mutation are not being identified.^{13 18} In addition, the availability of specialist staff, including genetic counsellors, is too low to meet demand – contributing to long waiting times for genetic tests.⁸

Information and support for those with *BRCA* mutations must be improved. There are no *BRCA*-specific patient organisations in Ireland, although there are general cancer organisations providing information and limited support services for *BRCA*-mutation carriers.^{14 16 17} Many non-genetic-specialist healthcare professionals, such as GPs, seem to lack the knowledge required to meet the needs of their patients.³⁰

National guidelines which support access to comprehensive care for *BRCA*-mutation carriers are urgently needed. Access to genetic testing services for those at high risk of *BRCA*-related breast cancer is severely hampered by restrictive eligibility criteria. Recommendations on surveillance are not being adopted and there are no national guidelines on risk-reducing options for women with *BRCA* mutations who do not yet have breast cancer.

Ireland must follow through on its strategic commitments and urgently invest in the genetic testing infrastructure and workforce needed to improve access to timely *BRCA* genetic testing. This includes building staff numbers and streamlining current cancer genetic services.⁹ Progress in relation to these commitments must be monitored to ensure that all those with *BRCA* mutations in Ireland have access to the care they need.

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