The Health Policy Partnership

Genetic testing for *BRCA* mutations:

country profile for Germany

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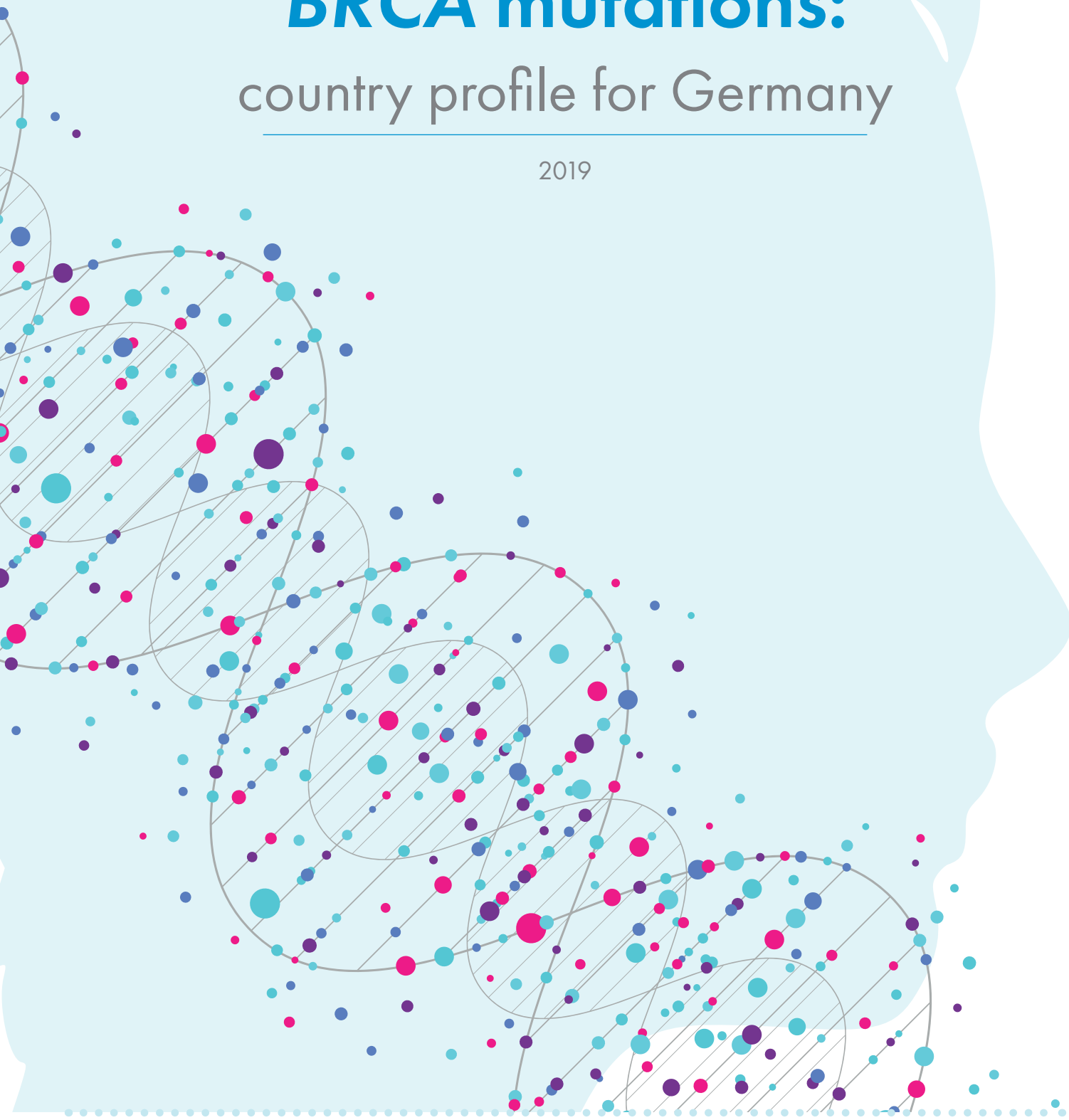

Pfizer Oncology

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

- Germany has the 15th highest incidence rate and the 9th highest mortality rate of female breast cancer in Europe.[1]
- 16% of women with triple-negative breast cancer in Germany carry a *BRCA* mutation.[2]

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.[3-5] Breast cancers related to *BRCA1* mutations are more likely to be triple-negative breast cancer, which is difficult to treat.[6]

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.[7]

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.[8] 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 Germany 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

Germany's enabling policy environment has supported the development of a network of familial breast and ovarian cancer centres.

Inclusion of *BRCA* genetic testing in the national plan for cancer

Germany's national cancer plan, launched in 2008, aimed to advance early detection and provision of care for cancer patients in addition to the expansion of data registries, including for breast cancer.[9] The plan fostered the growth of a network of specialist familial breast and ovarian cancer centres in Germany and encouraged their certification.[9] Furthermore, data on treatment for breast cancer patients were incorporated into the national tumour registry, marking an important step towards comprehensive data collection and monitoring.[10]

The German Consortium for Hereditary Breast and Ovarian Cancer is active in implementing individualised, risk-adjusted prevention for hereditary breast cancer. It aims to ensure women with high genetic risk of breast cancer are legally entitled to intensified screening and risk-reducing options.[11] Consortium centres provide those women with access to care through multidisciplinary teams, working closely with the patient organisation *BRCA*-Network. They also collect and store data in a common database which allows them to share information for improved patient care. As part of this, the German Consortium is leading the European expert group RiskAP, which is developing a position paper on hereditary breast cancer prevention that accounts for genetic risk factors.[12]

To further improve care for people with rare diseases, including *BRCA*-related breast cancer, the Germany Ministry of Health initiated the National Action League for People with Rare Diseases (NAMSE), a multi-stakeholder coalition.[13] Partners include the patient advocacy group Allianz Chronischer Seltener Erkrankung (ACHSE), of which the *BRCA*-Network is a member.[14] In 2013, the league devised a National Action Plan for people with rare diseases, which aims to establish a network of referral centres, specialist centres and collaborative centres for patients with rare diseases.[14] Furthermore, several German genetic centres are part of the European Reference Network for Genetic Tumour Risk Syndromes (ERN GENTURIS), including the Munich Medical Genetics Centre, the Centre for Hereditary Tumour Syndromes (CHT) at the University Hospital in Bonn, and the Hereditary Cancer Syndrome Centre Dresden at the Faculty of Medicine of TU Dresden.[15]

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

There are differences between statutory and private health insurance coverage, which could create difficulties in accessing *BRCA* genetic testing. While genetic testing is fully reimbursed by statutory health insurance, people with private insurance need to request the *BRCA* test from their insurer and are not guaranteed approval. Equally, prophylactic treatment may not be automatically covered.[16] Some private health insurers do not have contracts with the specialist centres, which may complicate or prohibit reimbursement for treatment in those centres.[17]

National registries to collect data on *BRCA* genetic testing

Germany benefits from a number of registries, although greater efforts are needed to aggregate data for policymakers. Cancer and tumour registries collect data on *BRCA*-mutation carriers, but an expert consulted for this research has highlighted that these data should be aggregated in order to validate clinical evidence and generate robust data for policymakers.[16]

Legal protection of *BRCA*-mutation carriers

German legislation is in place to protect those with *BRCA* mutations from employment and insurance discrimination. It guarantees every citizen the authority over their genetic information and appears to successfully prevent people from experiencing discrimination as a result of a positive *BRCA* test result.[18] An expert interviewed for this country profile, however, notes that some people do not request a test due to fear that it may affect their private insurance coverage.[19]

However, this legislation may present a barrier to testing family members of *BRCA*-mutation carriers. Unlike in some other countries, such as France, healthcare professionals in Germany are not allowed to contact family members of a mutation carrier regarding their potential genetic risk.[17]

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

BRCA-specific patient organisations and increased reporting in the media have contributed to growing awareness of *BRCA* mutations among the population.

Awareness of *BRCA* mutations has increased notably in recent years, largely due to advocacy from patient organisations and prominent cases in the media. Yet experts have highlighted that more needs to be done to increase knowledge about *BRCA* mutations and the benefit of genetic testing in the general population and among policymakers.[16 17]

The organisation Deutsche Krebshilfe funds specialist centres where people with familial breast cancer can seek information and receive adequate diagnosis and treatment. It also played a major role in funding and establishing the patient organisation *BRCA*-Network, which educates and supports those who may carry a *BRCA* mutation.[20 21]

The magazine *Mamma Mia!* – available online and in all breast cancer centres – also provides a range of information on *BRCA*. In 2013, it published a special edition with detailed information for mutation carriers about their options relating to *BRCA*. It also offered information on possible financial issues and available support programmes.[20] Updates are published regularly to improve knowledge among people with *BRCA* mutations of all aspects of medical care and living with cancer.[19]

Despite efforts to improve understanding and awareness, gaps remain. An expert interviewed for this country profile notes that some women continue to have low levels of knowledge about *BRCA* and the importance of multidisciplinary care in supporting women's physical and psychosocial needs.[19]

Comprehensive care pathways

Comprehensive guidelines provide detailed recommendations on care for *BRCA*-mutation carriers, while a network of specialist centres provides the infrastructure for delivery.

Evidence-based national eligibility criteria for *BRCA* testing

Updated comprehensive guidelines on breast cancer (S3 guidelines) were published in 2018 and provide recommendations for *BRCA*-related breast cancer. This includes eligibility criteria for genetic testing and recommendations regarding genetic counselling, long-term monitoring and risk-reducing strategies for mutation carriers.[22] Recommendations for *BRCA* genetic testing are also included in the guidelines from the working group for Arbeitsgemeinschaft Gynäkologische Onkologie (AGO).[23] To facilitate and promote guideline use in clinical practice, a checklist has been developed by the Consortium for Hereditary Breast and Ovarian Cancer to help healthcare professionals assess family history. The checklist is recommended for use by practitioners.[24 25]

Equitable and timely access to genetic testing

People can seek genetic testing for *BRCA* mutations either through one of the 18 certified specialist centres of the Consortium for Hereditary Breast and Ovarian Cancer, through Departments of Human Genetics at Universities, or through private genetic testing providers. It is mandatory for physicians to assess the family history of all people requesting a genetic test and to inform them about all aspects of testing for genetic mutations associated with breast cancer. Genetic testing is offered to all who meet the eligibility criteria specified in the S3 guidelines.[22]

While specialist centres are located across Germany, there are fewer centres in some areas which may lead to geographic inequalities in access to *BRCA* genetic testing. People living in north-east Germany, for example, may face long travelling times to specialist centres.[17 26] The consortium of specialist centres is, however, building networks with other breast and gynaecological cancer centres in an attempt to ensure nationwide access.[27]

Waiting times for genetic test results from the specialist centres have reduced in recent years. In 2013, waiting times for genetic test results from one of these centres were between one and eight months, depending on medical urgency.[28] One expert interviewed for this country profile, however, indicated that this has now reduced to an average of one month.[27]

Role of other genetic testing providers

In addition to the specialist centres, genetic testing and counselling are also offered by human geneticists working in private practice. They may not provide the comprehensive, multidisciplinary care that is available through Consortium centres, but they may refer patients to these services.[20 28] Genetic counselling and *BRCA* genetic testing are also provided by all university-based Departments of Human Genetics.[19]

Access to genetic counselling

Genetic counselling is required following the results of diagnostic genetic testing. In the case of predictive genetic testing, genetic counselling is mandatory both before testing and once results are available.[29] People receive information on what it means to live with a *BRCA* mutation and about implications for other family members.[28]

As in several other European countries, genetic counselling in Germany is performed only by physicians. This includes those certified in human genetics and, in the case of diagnostic genetic testing, physicians with other specialties who have received the appropriate training.[29]

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

As part of the care pathway for *BRCA*-mutation carriers, national breast cancer guidelines give guidance for long-term monitoring, prophylactic surgery and treatment options. Recommendations are frequently updated to incorporate emerging evidence and are implemented on a case-by-case basis and depending on the *BRCA*-mutation carrier's preferences.[22 30] However, risk-reducing surgery for asymptomatic women is not routinely reimbursed by all German health insurance, limiting access for some women.[31]

Improved awareness and knowledge among healthcare professionals

There are recognised gaps in *BRCA* knowledge among healthcare professionals who are not genetic specialists.

Non-genetic-specialist healthcare professionals, such as gynaecologists, breast surgeons and GPs, are often not adequately equipped to initiate referrals for *BRCA* genetic testing, make the right referral decisions or recommend appropriate risk-reducing interventions.[16 17 32] A survey from 2013 found that only 27% of GPs and 66% of breast surgeons in Germany were in favour of prophylactic mastectomy as a treatment option.[32]

The recent projects iKNOW and EDCP-*BRCA* aim to address this gap by supporting physicians to interpret and communicate *BRCA* testing results and provide accurate advice to *BRCA*-mutation carriers. The projects implemented by Charité – Universitätsmedizin, the Technische Universität in Berlin and the University of Cologne involve the development and roll-out of online tools, which will support doctors to discuss test results and breast cancer risk, and offer evidence-based prevention and treatment options in line with current guidelines.[33 34]

Conclusions and recommendations

Multi-stakeholder efforts at policy level in recent decades have significantly advanced the provision of care for women who have or are at risk of hereditary breast cancer.[9 20] The establishment of the German Consortium for Hereditary Breast and Ovarian Cancer and the foundation of its now 18 specialist centres has been an important step towards offering high-quality care in hereditary breast cancer.[23] Furthermore, the current national cancer plan aims to advance the risk-adjusted early detection of people at high risk of cancer, which includes women who may carry a *BRCA* mutation.[9 11] There are, however, a number of challenges and gaps which policymakers should address when considering ways to improve care for those with *BRCA* mutations.

Greater attention is needed to ensure that *BRCA*-mutation carriers and those with a high chance of carrying a mutation can access comprehensive, affordable care. This should cover the full range of services that people would be expected to access, including support and information before they are tested, and clinical and psychosocial support both during the testing process and, if they are found to have a mutation, following the test. This would address the geographical barriers and differences in insurance coverage which have led to inequalities in access to comprehensive *BRCA*-related care.[17 26]

GPs and gynaecologists must be supported with evidence-based guidance and up-to-date knowledge to enable them to make appropriate referrals of those at high risk of *BRCA*-related breast cancer. GPs and gynaecologists play a critical role in supporting women who may be carrying a *BRCA* mutation, but knowledge gaps among these professionals present a barrier to accessing high-quality care.[32]

Data from different registries, including hereditary tumour and cancer registries, should be aggregated for the validation of clinical evidence and the generation of robust data for policymakers. While Germany benefits from several registries which collect information on *BRCA* carriers and breast cancer patients, the data are not aggregated,[16] limiting the potential to support policymakers, researchers and clinicians.

Awareness of *BRCA* mutations among the general population must be improved. There is also a need to improve understanding of genetic testing in general and the importance of interdisciplinary care that supports the full range of physical and psychosocial needs of *BRCA*-mutation carriers.

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