Barriers to and opportunities for genetic testing of BRCA mutations in breast cancer across Europe

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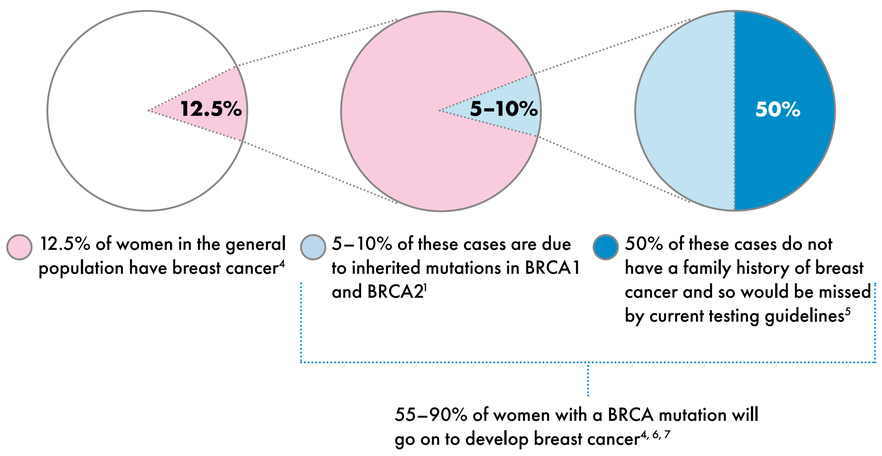
Background

Inherited mutations in BRCA1 and BRCA2 genes account for 5–10% of breast cancer cases[1] (*see Figure 1*). Breast cancer associated with BRCA mutations can develop at an early age,[2] has a higher chance of recurrence and, with fewer treatment options, may be more difficult to treat than other breast cancers.[3]

With the growing prevalence of breast cancer, priority must be given to the development of comprehensive cancer control plans that provide high-quality prevention, treatment and care for all cases of breast cancer. Such plans must address the unmet needs of more difficult-to-treat cancers, such as those associated with BRCA gene mutations or triple-negative breast cancer, and rapidly adapt to the evolving diagnostic and treatment landscape.

Figure 1. Epidemiology of breast cancer associated with BRCA gene mutations



As genetic testing for BRCA mutations becomes increasingly available, appropriate information and counselling, along with treatment and care for those who develop breast cancer, must be made more accessible. Safeguards must also be put into place to protect women with BRCA mutations against any form of genetic discrimination. Patients, healthcare professionals and governments must work together to implement programmes and policies to effectively meet the needs of all women with breast cancer, women with BRCA mutations and their families.

Objective of the research

Our research aims to understand the European policy landscape for women with BRCA mutations, and the barriers they may face when trying to access genetic testing. This will provide a solid basis to evaluate the gaps in policies and programmes, allowing us to present concrete recommendations to improve access to information, testing and care.

Material and methods

In this first phase of our research, we undertook a pragmatic review of the published and grey literature. This was followed by national-level literature searches to inform six individual country profiles. The desk research will be complemented by semi-structured, qualitative telephone interviews with thought leaders, a sub-group of whom will form a steering committee to guide our research and help to validate findings.

Results

Our literature review found that specific policy frameworks and reliable data on BRCA testing are often lacking, while awareness and access are highly variable across Europe.

Access and uptake of genetic testing may vary considerably depending on race, age, location (rural vs urban) or socioeconomic status.[2, 3, 8-12]

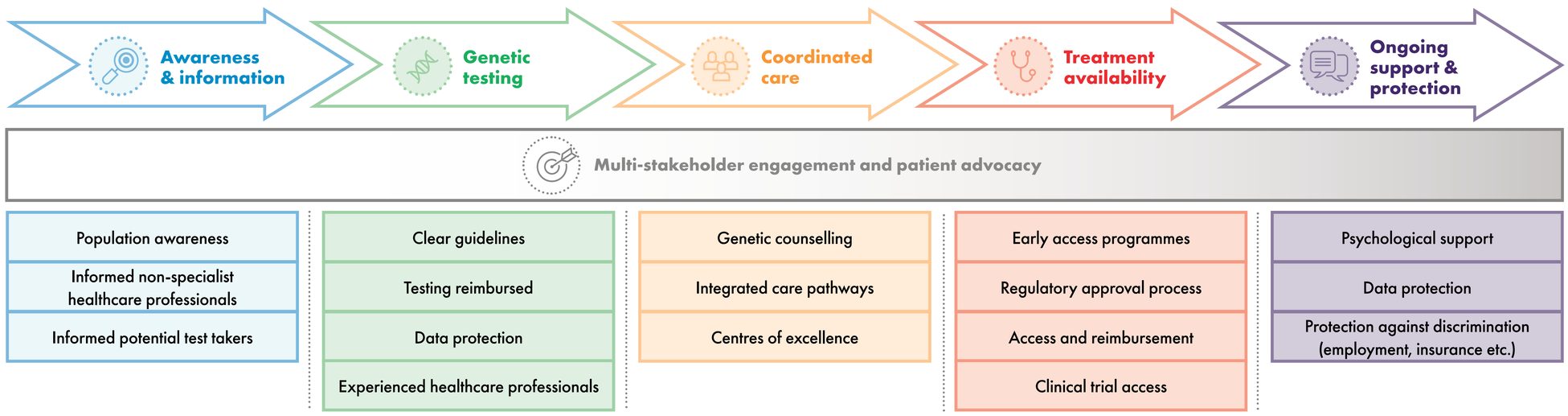
Additional barriers include the following:

- **Limited awareness among healthcare professionals** can lead to low referral rates. There is a clear lack of knowledge when it comes to interpreting the 10% of cases where results of a genetic test are uncertain or inconclusive. This can lead to an overestimation of cancer risk and significant distress for the woman concerned.[13, 14]
- **Limited access to genetic counselling and inadequate information.** Access to appropriately qualified genetic counsellors is uneven across Europe. This can lead to increased levels of patient anxiety and may cause delays and bottleneck early treatment access.[15, 16]
- **Unclear and restrictive testing guidelines.** Family history tends to be the basis for BRCA testing criteria. However, around half of BRCA mutation carriers do not have a family history of breast or ovarian cancer before diagnosis, and would be missed by these criteria.[17]
- **Concern about the impact a positive result may have on family members** and relationships. Some individuals will not tell their loved ones for fear of misinterpreting information and causing distress to a family member unnecessarily. Some women also deal with feelings of guilt about passing on the mutation to their children.[18]
- **Fear of discrimination** from insurance providers or employers when genetic test results are shared. This may act as a deterrent for women considering testing and therefore delay diagnosis.[19]

Conclusions from initial research

Our research informed the creation of a policy framework, developed to identify key opportunities to improve access to and utilisation of appropriate testing, diagnosis, treatment and care for women with BRCA mutations (*see Figure 2*). From this framework, concrete recommendations can be made for policymakers to improve national cancer control plans and public health policies. Moreover, breast cancer policy frameworks can be developed to benefit women with BRCA mutations within the breast cancer community. This framework will also be used to guide subsequent phases of our research.

Figure 2. Key issues of relevance to BRCA testing



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