



# Genetic testing for *BRCA* mutations: country profile for Sweden

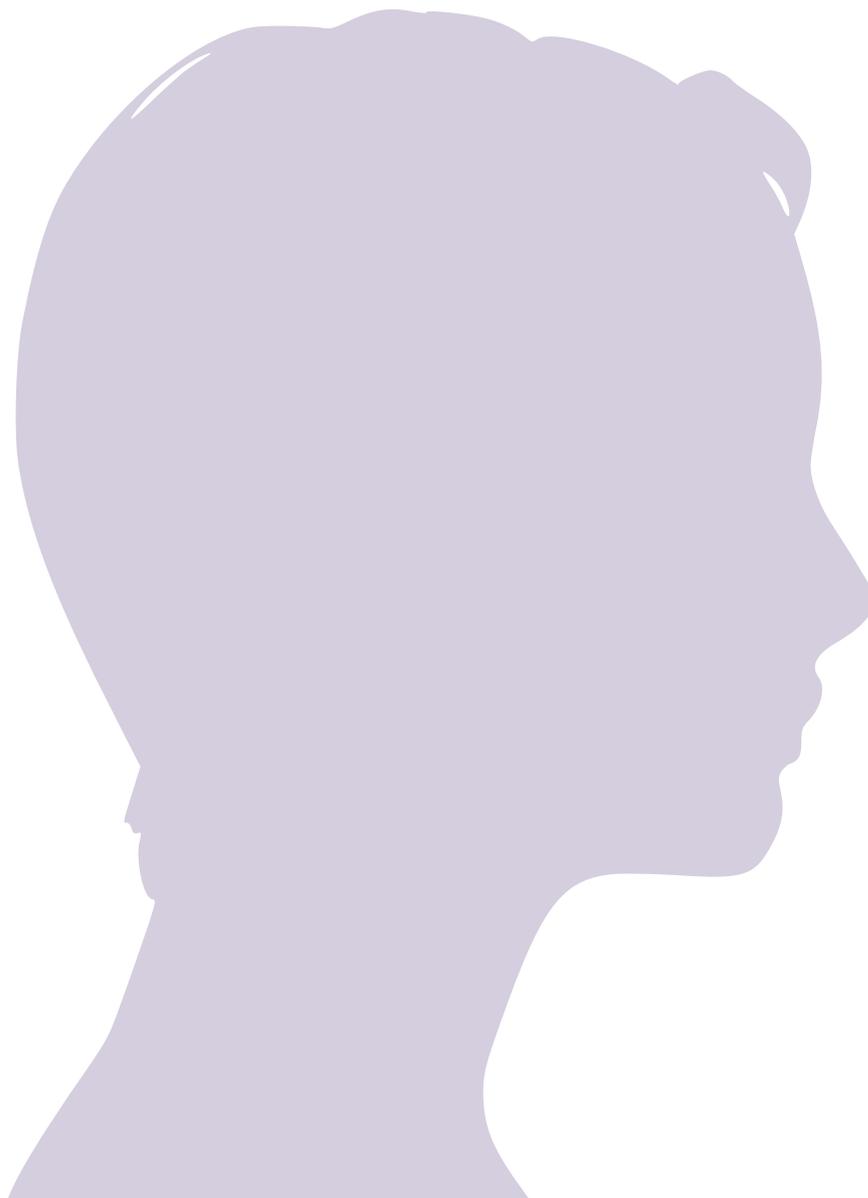
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# Genetic testing for *BRCA* mutations:

## country profile for Sweden



### Key facts and figures on breast cancer and *BRCA* mutations in Sweden

- Sweden has the 6th highest incidence rate and the 26th highest mortality rate of female breast cancer in Europe.<sup>1</sup>
- Approximately 2% of breast cancer cases in Sweden are estimated to be associated with *BRCA* mutations.<sup>2</sup>
- Between 50–80% of Swedish women with a *BRCA* gene mutation will develop breast cancer.<sup>3 4</sup>



# 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.<sup>5-7</sup> Breast cancers related to *BRCA1* mutations are more likely to be triple-negative breast cancer, which is difficult to treat.<sup>8</sup>

**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.<sup>9</sup>

**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.<sup>10</sup> 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 Sweden 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

Sweden's national cancer strategy is being applied at the regional level with the aim of improving cancer prevention, while investments are being made on advancing genomic research.

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

**In 2009, the Swedish central government launched a national cancer strategy prioritising improved primary prevention and quality of cancer care and equal access to care across the regions.**<sup>11</sup>

This led to the creation of six regional cancer centres with the aim of better responding to local care needs and encouraging care delivered by multidisciplinary teams.<sup>11 12</sup> These centres have developed activities in collaboration with healthcare providers and the regional health administrations, including breast-cancer-related activities and plans.<sup>12</sup>

**Unlike many other European countries, Sweden has not yet implemented a national strategy for rare genetic diseases.**<sup>13</sup> In 2011, Nationella Funktionen för Sällsynta Diagnoser (NFSD), the Swedish National Agency for Rare Diseases,<sup>14</sup> was founded. In conjunction with the National Board of Health and Welfare, it developed a national strategy for rare diseases – but this has not yet been officially adopted.<sup>13</sup>

**Sweden has four designated centres of excellence which deal with rare and genetic disease, including hereditary breast cancer.** Three of these centres are at the Karolinska University Hospital in Stockholm. They include the Centre for Rare Diseases, which is a member of the European Reference Network on Genetic Tumour Risk Syndromes (GENTRIUS), and an Expert Team for Rare Gynaecological Diseases, which is a member of the European Reference Network on Rare Adult Cancers (EURACAN).<sup>15</sup>

**There are efforts to advance research and implementation of genomic medicine in Sweden. The multi-stakeholder initiative Genomic Medicine Sweden launched in 2017, with the aim of building new infrastructure to enable the use of novel diagnostics and precision medicine across Sweden.** It focuses on people with rare inherited diseases and cancers.<sup>16</sup> In 2011, the large population-based cohort study LifeGene was launched, which aims to analyse biosamples from more than 500,000 participants and follow them up for more than 20 years.<sup>17</sup> Collected data will be used by various research groups, including a cancer working group.<sup>18</sup>



## National registries to collect data on BRCA genetic testing

Although there are no *BRCA* or rare disease registries at national or regional level, Sweden has a range of disease-specific registries, including for cancer.<sup>13</sup> The National Breast Cancer Quality Register (NKBC) collates data from the regional cancer centres on quality and outcomes of breast cancer care. This includes information on preventive surgery but not on people with *BRCA* mutations.<sup>19</sup>

## Legal protection of BRCA-mutation carriers

Legislation is in place to ensure data privacy and prevent discrimination based on genetic test results. Doctors are not allowed to talk to anyone about test results or pass them on without the permission of the person being tested. In addition, in most cases, insurance companies are not allowed to request information about pre-symptomatic testing from prospective customers. Genetic counsellors can advise those being tested on the kind of information on genetic testing that insurance companies may request or access. Furthermore, the Association of Swedish Insurers released a statement in 1998 clarifying that insurers would not enquire about genetic test results or use information from results when assessing risks below SEK 250,000 (€24,050 [05/19]).<sup>20</sup>

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

There are no national *BRCA*-specific patient organisations in Sweden and only limited information for people considering a *BRCA* genetic test.

Some information on *BRCA*-related breast cancer is available to those considering testing, but availability varies by region.<sup>21</sup> The limited information available includes a leaflet for people considering *BRCA* genetic testing, published by the Swedish Society of Medical Genetics and Genomics (SFMG) in collaboration with EuroGentest and Genetic Alliance. The leaflet advises on choosing the right time to get a genetic test, privacy rights and available support.<sup>20</sup>

A number of rare disease organisations engage in advocacy, patient information and support. These include Orphanet Sweden,<sup>22</sup> the national competence centre Ågrenska<sup>23</sup> and the patient organisation Riksförbundet Sällsynta diagnoser (Rare Disease Sweden).<sup>24</sup> However, there is no national patient organisation for hereditary breast cancer.<sup>25</sup>

## Comprehensive care pathways

Sweden's national breast cancer programme provides comprehensive information for people with *BRCA* mutations at all stages of care. But geographic barriers may limit access to *BRCA* genetic testing, while a severe lack of genetic counsellors may leave some unsupported.

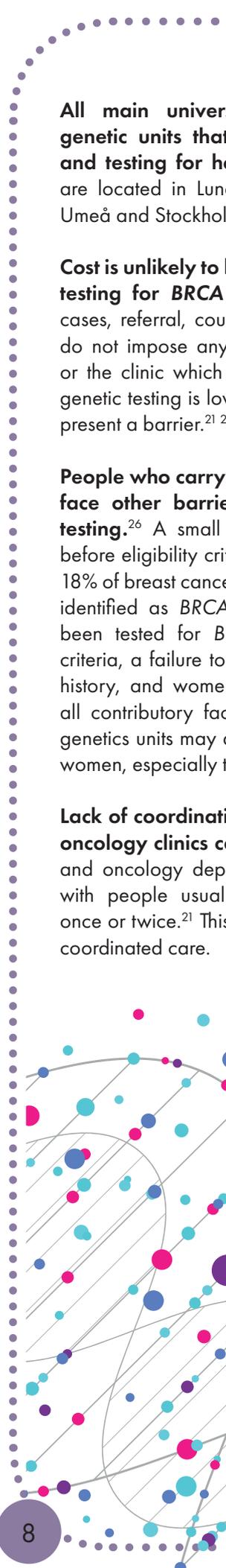
### Evidence-based national eligibility criteria for *BRCA* testing

**Comprehensive care pathways for those who carry *BRCA* mutations are well integrated in the national breast cancer care programme.** This includes recommended criteria for genetic testing.<sup>4</sup> Eligibility criteria for *BRCA* genetic testing are mainly based on age at diagnosis and family history of cancer.<sup>26</sup> The criteria are more inclusive than they were in the past; for example, they now include women with breast cancer below the age of 40, rather than the previous cut-off age of 35, which means more women are now eligible. In addition, all women with ovarian cancer are also eligible for a *BRCA* genetic test.<sup>27</sup> While these recommendations are not mandatory, there seems to be national agreement and most clinicians and oncologists are familiar with them.<sup>26</sup> There may, however, be some regional variation in their implementation.<sup>21</sup>

### Equitable and timely access to genetic testing

**The Swedish government is investing in improving access to more consistent and timely care.** In response to long waiting times for cancer diagnosis and treatment, in 2015 the Swedish government launched Standardised Care Pathways to achieve consistent, timely, high-quality diagnosis and care for cancer patients in all regions. This includes a care pathway for breast cancer which highlights the importance of understanding possible hereditary risk at the point of diagnosis.<sup>28</sup>

**People considering *BRCA* testing are usually referred to genetic counselling by their GP, but self-referral is also common.**<sup>26 29</sup> One study on referrals to a hospital in Stockholm found that 44% of people self-referred.<sup>30</sup> In these cases, people submit a personal assessment of their cancer-related family history.<sup>30</sup>



**All main university hospitals have specialist genetic units that offer assessment, counselling and testing for hereditary cancer.** These hospitals are located in Lund, Göteborg, Linköping, Uppsala, Umeå and Stockholm.<sup>31</sup>

**Cost is unlikely to be a barrier to accessing genetic testing for BRCA mutations in Sweden.** In most cases, referral, counselling and BRCA genetic testing do not impose any cost on the woman being tested or the clinic which referred her. Utilisation of private genetic testing is low, so the costs of testing should not present a barrier.<sup>21 26</sup>

**People who carry BRCA mutations may, however, face other barriers in accessing BRCA genetic testing.**<sup>26</sup> A small study in one region (undertaken before eligibility criteria were relaxed) found that only 18% of breast cancer patients who were retrospectively identified as BRCA-mutation carriers had previously been tested for BRCA mutations. Restrictive testing criteria, a failure to accurately report or assess family history, and women deciding not to be tested were all contributory factors.<sup>26</sup> The geographic spread of genetics units may also be an access barrier for some women, especially those in remote areas.<sup>21</sup>

**Lack of coordination between some genetics and oncology clinics can be a further barrier.** Genetics and oncology departments tend to work separately, with people usually visiting a genetics clinic only once or twice.<sup>21</sup> This may limit access to consistent and coordinated care.

## Access to genetic counselling

**Cancer genetics clinics have integrated genetic counselling into the management of hereditary cancer patients.** Pre-test and post-test counselling is mandatory and provided during in-person sessions for individuals or families.<sup>3 32</sup> Post-test counselling includes psychological support by genetic counsellors and psychologists.<sup>20 33</sup> Those who are found to carry BRCA mutations are further encouraged to seek help from family members and patient groups.

**There is variation between genetics centres in how genetic counselling is practised due to a lack of national standards, registration, licensing, or undergraduate or postgraduate training courses.**<sup>21</sup>

Genetic counsellors can, however, voluntarily register to be certified by the European Board of Medical Genetics (EBMG) and the SFMG.<sup>32 34 35</sup> The Swedish Professional Association for Genetic Counsellors (SFGV) aims to improve access to standardised training and accreditation,<sup>34</sup> and is developing national standards and a voluntary registration system together with SFMG. However, uptake of this registration scheme to date has been low.<sup>21</sup>

**The limited number of genetic counsellors and high administrative workload can limit their ability to directly engage with people wishing to be tested, and cause bottlenecks.**<sup>21</sup> The number of genetic counsellors per capita in Sweden is lower than in many other countries such as France, Ireland and the UK.<sup>35</sup>

**Modifying the process of genetic counselling in Sweden, however, could help to meet the increasing demand of testing while maintaining quality of the service.** Recent studies in Sweden show that new methods, such as telephone counselling or written pre-test information, reach similar satisfaction compared with the standard method of face-to-face counselling sessions before the test, and have the potential to reach more people.<sup>30 36</sup> Currently, however, telephone-based genetic counselling is not believed to be commonly practised in Sweden.<sup>21</sup>

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

The national breast cancer care programme provides detailed guidance on supporting women with BRCA mutations in line with current international best practice. It is, for example, current practice in Sweden to recommend risk-reducing salpingo-oophorectomy (RRSO) at the age of 35–40, depending on parenthood planning.<sup>3</sup> The uptake of

RRSO among BRCA-mutation carriers has increased steadily, as has the uptake of prophylactic mastectomy, which is estimated to be around 50% in Sweden.<sup>3</sup> In addition to the population-wide mammography screening offered to all Swedish women over 40, carriers of a BRCA mutation are offered magnetic resonance imaging (MRI)-based screening from the age of 25 onwards.<sup>3</sup> Chemoprevention is not commonly used as a risk-reducing strategy for BRCA-mutation carriers in Sweden.<sup>3</sup>

## Improved awareness and knowledge among healthcare professionals

The multidisciplinary SFMG and its cancer genetics working group play a role in training of healthcare professionals.

**Hereditary cancers are a priority of the multidisciplinary SFMG, which supports training of healthcare professionals.** It established a cancer genetics working group due to an increased demand for genetic tests and a rise in referrals. This group provides training for healthcare professionals and shapes the agenda of the SFMG.<sup>37</sup>

**The role of regional patient process leaders (RPPLs) for hereditary cancer has also been established in some regions to improve genetic testing, diagnosis and treatment.**<sup>38</sup> In the southern Swedish region, for example, a healthcare professional who is appointed as RPPL is responsible for advocating cancer-genetic health research, facilitating training in hereditary cancer for healthcare professionals and the general public, and reducing inequalities in access to cancer-genetic testing.<sup>39</sup>



## Conclusions and recommendations

**Sweden's national breast cancer care programme contains detailed guidance on supporting BRCA-mutation carriers.** This includes clear recommendations on eligibility for *BRCA* genetic testing and for ongoing management and support of those who carry mutations.<sup>4</sup> While these recommendations are not mandatory, they seem to be well-accepted nationally. There are, however, some areas of development that policymakers in Sweden could take into account as they consider how to improve access to high-quality care for *BRCA*-mutation carriers.

**Greater recognition and investment in genetic counselling is needed.** There are no university-level training courses for genetic counsellors in Sweden, and no requirement for licensing or registration. Furthermore, there are not enough genetic counsellors practising to meet demand, leading to bottlenecks in access to *BRCA* genetic testing.<sup>32</sup> Greater emphasis on strengthening this profession is needed to ensure everyone who needs it has access to high-quality, standardised care. Widening access to innovative approaches such as the use of telemedicine could also play a role.

**Robust and up-to-date national and regional data are needed on BRCA-mutation carriers to support effective planning.** There is no dedicated registry for *BRCA*-mutation carriers, and *BRCA* is not included in the breast cancer registry. A comprehensive *BRCA* registry could be one way of collecting these data and making them available to policymakers as they work to understand the level of care needed for those who carry a *BRCA* mutation, and where gaps exist.

**More comprehensive, locally relevant information would support BRCA mutation carriers and those considering BRCA genetic testing.** There are no *BRCA*-specific patient organisations working nationally in Sweden. Although there are organisations providing support to *BRCA*-mutation carriers, this does not seem to be as well developed as in some other European countries. This is limiting the availability of tailored information and support for the general public and *BRCA*-mutation carriers in Sweden.

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