



Inequities in genetic testing for hereditary breast cancer: implications for public health practice

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Abstract

The Ontario Breast Screening Program for women with a genetic predisposition to breast cancer is one of the first international models of a government-funded public health service that offers systematic genetic screening to women at a high risk of breast cancer. However, since the implementation of the program in 2011, enrolment rates have been lower than anticipated. Whilst there may be several reasons for this to happen, it does call into consideration the ‘inverse equity law’, whereby the more advantaged in society are the first to participate and benefit from universal health services. An outcome of this phenomenon is an increase in the health divide between those that are at a social advantage versus those that are not. Using an intersectionality lens, this paper explores the role of the social determinants of health and social identity in creating possible barriers in the access to genetic screening for hereditary breast cancer, and the implications for public health practice in recognising and ameliorating these differences.

Keywords Genetic testing · Social determinants of health · Hereditary breast cancer · Breast cancer · BRCA · Equity · Disparity · Social justice · Intersectionality · Social location · Public health

Introduction

In Canada, one out of every eight women will face a diagnosis of breast cancer over their lifetime (Canadian Cancer Society’s Advisory Committee on Cancer and Statistics 2017). Five to 10% of these cancers are hereditary in nature and can be attributed to the inheritance of specific gene mutations, most commonly in the tumour suppressor genes BRCA1 and BRCA2 (Edlich et al. 2005). Breast cancers associated with BRCA gene mutations are typically aggressive, resistant to treatment, and occur overwhelmingly in women (van der Groep et al. 2011). Since the introduction of genetic testing for hereditary breast cancer (HBC), it has been possible to categorise women as high risk and offer them methods of risk reduction through strategies such as bilateral mastectomy with oophorectomy, chemotherapy with tamoxifen, or intensive

monitoring with mammography and MRI (Berliner et al. 2013; Burke et al. 1997). Accurately identifying women with inherited predispositions to breast cancer and offering them genetic testing with subsequent risk reduction therapeutic options has shown to reduce the incidence of breast cancer-related mortality by 90% (Domchek et al. 2010; Rebbeck et al. 2004), and is therefore an important way to improve survival for this group of women.

In 2011, the Ontario Ministry of Health and Long-Term Care (MOHLTC) introduced a publicly funded system of genetic testing for hereditary breast cancer through its accountable body Cancer Care Ontario (CCO). The Ontario Breast Screening Program (OBSP) represents the first government-organised genetic screening program available to all women of Ontario who are deemed medically eligible. Enrolment into the program is based on a referral from a physician for individuals with no history of breast symptoms with either (i) a known BRCA1 or BRCA2 mutation; (ii) a first-degree relative known to have a BRCA mutation, and having declined genetic testing after counselling; (iii) a lifetime risk of breast cancer $\geq 25\%$ based on the IBIS or BOADICEA tools; or (iv) a history of radiation to the chest (Cancer Care Ontario 2011). This program is novel in many ways: for starters, it is a pioneering effort to introduce a population-wide, publicly

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funded genetic test to high-risk individuals; secondly, it is expected that the knowledge bank created through the enrolment of all provincial high-risk individuals will pave the way for innovations in how HBCs are managed (Chiarelli et al. 2014); and more pragmatically, contribute to the development of international standards and quality benchmarks with regard to genetic testing for HBC (Auditor General of Ontario 2012).

The Ontario Breast Screening Program for High Risk Women is funded by the government; therefore, financial barriers in terms of access to care do not exist for individuals seeking screening. Despite this, the program has had low levels of enrolment based on their population targets (Cancer Care Ontario 2011). Whilst access to health care services is an important social determinant of health (Whitehead 1992), the structure and design of health services can render them structurally unavailable and socially unacceptable to certain population groups (Gilson et al. 2007). Indeed, recent studies clearly demonstrate how socially disadvantaged individuals, such as those with lower levels of education, and those from ethnic minority groups consistently under-utilise health services despite the lack of a financial barrier to care (Maddison 2011).

Although it remains too early to understand the exact cause for under-enrolment in the OBSP high-risk screening program, this paper serves to illuminate how screening programs that are used as targeted interventions to improve health outcomes must take into consideration the complexities associated with utilisation and need across the entire population. A failure to do so may further disenfranchise socially disadvantaged individuals and widen the health equity gap that currently exists between population groups based on social location.

Theoretical underpinnings

McGibbon and McPherson (2011) describe the ‘synergies of oppression framework’, which is built on the premise of complexity theory, feminist political economy, and feminist intersectionality. This analytical lens allows us to conceptualise how the social determinants of health, social identity and social geography intersect to create ‘structures of constraint’ (Young 2002) that hinder access and utilisation of health services. In addition, Hankivsky et al. (2010) call us to refrain from categorising women as a single group, and to recognise instead the intrinsic heterogeneity that exists within women as a result of their social location; whilst Young (2002) and Weldon (2006) urge us to move beyond intrinsic components of gender identity such as race and class, and to consider instead the intersections of social structures that create an unequal distribution of power, resources and opportunities that lead to gendered social inequality.

Whilst each of these theories guides us on how to understand gender, inequality and social structural hierarchies, the OBSP high-risk screening program represents a ‘real-world’ (Hankivsky et al. 2010) problem. It is in the context of the

design of this intervention that we can see how structures of constraint can create gendered opportunities for care, conferring a potential survival advantage to some women with a high risk of breast cancer over others. Given that population-wide health inequities that currently exist are projected to further increase after the introduction of genetic testing (Hall and Olopade 2005), it is important that genetically based screening programs that target high-risk population groups take proactive measures when designing the delivery and funding of such interventions to mitigate these effects.

Social location and genetic testing for heredity breast cancers

Studies have demonstrated that women from ethnic minority groups (Mai et al. 2014) with lower levels of education and income (MacNew et al. 2010) participate less frequently in genetic testing because of the cultural inappropriateness of genetic tests (Charles et al. 2006), financial limitations (Kieran et al. 2007), fear of discrimination (Peterson et al. 2002), and an overall lack of trust in the healthcare system (Ramirez et al. 2006). Indeed, self-identified, ethnic minority groups represented fewer than 10% of the initial cohort that utilised BRCA gene testing in the USA (Frank et al. 2002), and subsequent studies have continued to demonstrate under-utilisation (Armstrong et al. 2003) as well as a lower likelihood of receiving genetic counselling (Armstrong et al. 2005) in non-white population groups.

Given that genetic testing is a relatively new concept in healthcare, it is not surprising that the literature linking the social determinants of health and utilisation of genetic testing for HBC is sparse. Furthermore, studies have not assessed the impact of either education or income in direct correlation to the utilisation of genetic testing by women, but rather education and income have been some of the many variables included in study design. As a result, findings are more incidental rather than designed. Nonetheless, what is clearly apparent is that the way genetic testing is both accessed and utilised follow similar trends, such that higher levels of both income and education correlate with an increased awareness of genetic testing, a greater likelihood of receiving referrals for genetic testing, appropriateness of genetic counselling and the final decision to proceed with genetic testing.

Overall, white Caucasian women have the highest levels of knowledge and awareness about genetic testing (MacNew et al. 2010; Mai et al. 2014), whereas, ethnic minority groups including Asians, African Americans and Hispanics do not (Mai et al. 2014). Furthermore, the levels of awareness correlate with degree of education and income status such that women with higher levels of education and greater personal affordability are more knowledgeable and have positive attitudes towards genetic testing (MacNew et al. 2010). Ironically, low-income women from ethnic minority groups

may have high levels of interest in the concept of genetic testing, but this attitude may not be backed by any significant understanding of the risks and responsibilities associated with personal genetic information (Chalela et al. 2012).

A study conducted in the Brazilian public health system demonstrated how educated women are able to drive the process of genetic testing even if they are clinically at low risk and not considered eligible candidates for screening (Palmero et al. 2007). In stark contrast, a study on African-American women illuminated how up to 50% of women with young age highly invasive breast cancer were never referred for genetic testing prior to the onset of their cancer despite clear clinical indications (Cragun et al. 2015). Not surprisingly, women in this study were more likely to have undergone timely genetic testing if they had a college education and an income greater than USD 35,000 (Cragun et al. 2015). In a universal public health system, such as the one that exists in Canada, it has been shown that educated individuals are more likely to utilise health services (Allin 2008), visit health specialists (Allin 2008) and negotiate better terms of care (Maddison 2011). Intersecting identities and social class distinctly shape opportunities to seek pre-emptive genetic care, validating the need to further explore utilisation of genetic testing across sociodemographic variables in the Canadian context.

According to the National Society of Genetic Counsellors (2016), 87% of genetic counsellors are white Caucasian and speak only in English. As expected, this can create a significant structural barrier for women who speak other languages and have different religious and cultural beliefs. Studies have shown that genetic counselling sessions that are culturally adapted are received with greater patient satisfaction (Joseph and Guerra 2015). Unfortunately, language barriers are most pressing, and despite the use of translators during genetic counselling, it has been shown that important issues such as the risks and liabilities associated with genetic testing are difficult to convey and may never be adequately communicated to the patient (Joseph and Guerra 2015). This is critical as genetic information does not come without baggage (Surbone 2011) and may place an increased burden of ethical and moral responsibility on women to do whatever they can to remain healthy for their family and to share personal genetic information for the sake of the wellbeing of the extended kinship (d'Agincourt-Canning 2001).

Financial limitations have traditionally played a direct role in the utilisation of genetic testing in the USA where women may lack health insurance all together or not have the economic means to afford the test (Kieran et al. 2007; Peterson et al. 2002). Although this situation is irrelevant in Ontario due to the publicly funded nature of the tests, it is important to note that lower levels of income are associated with other structural deterrents to health care access such as precarious working conditions, expensive childcare, or unaffordable transportation (Douglas et al. 2015).

Implications

This paper explores equity in the utilisation of genetic testing for HBC using an intersectionality lens. It is through this process that we can appreciate how a woman's social location and her degree of social advantage or disadvantage can be pivotal in her ability to access and benefit from health resources. Social structures of inequality, as represented by the social determinants of health (SDH) and social identity, can create gendered opportunities for care. Historically, ethnic minorities have been underrepresented in genomic research and participation from clinical trials, and current gene testing standards for BRCA1 and BRCA2 are based predominantly on gene sequence analysis obtained from Ashkenazi Jews and other white populations (Hall and Olopade 2006). Furthermore, socially disadvantaged women including low-income women and recent immigrants are known to refrain from utilising preventative cancer services despite publicly funded care (Kerner et al. 2015). Whether these factors are responsible for the low OBSP uptake rates is unclear such that it is important that demographics of the program participants be considered.

Targeted interventions such as genetic testing have the potential to increase health inequities (Hall and Olopade 2005). This necessitates consideration of how research efforts and scarce resources are allocated. Key questions requiring attention include (1) to what extent do social determinants of health such as income and education intersect with social identities and biological risk to produce health inequities? (2) do treatment choices vary based on the economic feasibility of risk reduction options? and (3) most crucially, is there a significant cancer-free survival difference between socially advantaged and disadvantaged women with these gene mutations?

A robust cancer care system should ensure that all women have equal opportunity and access to preventative genetic care. Suggestions have been made for a population-wide genetic screening program versus one that is a targeted intervention for high-risk groups as a means to improve access to testing across all population cohorts (King et al. 2014). There are however several arguments against this, such as the low prevalence of the BRCA gene mutation across the general population as well as the economic efficiency of rolling out such a program (D'Andrea et al. 2016; Lippi et al. 2017) versus investments in other tools that would lead to overall uplift of women and marginalised groups as a social class (Raphael 2016).

It is vital that all women regardless of their social location be able to avail themselves of these new programs and achieve positive health outcomes. Attention must be directed—at least—towards reducing inequities in access to these programs. A failure to integrate this recognition into a high-risk screening program runs the risk of increasing the divide between advantaged and disadvantaged women with a genetic predisposition to breast cancer.

In the long-term however, attention must be given to reducing the profound differences in advantage and disadvantage that exist among women in Ontario. Such efforts will not only reduce inequities in access to the genetic testing programs which are the focus of this paper but also to a wide range of other health outcomes in which difference in advantage and disadvantage are implicated (Raphael 2016).

Compliance with ethical standards

Conflict of interest The author declares that she has no conflict of interest.

Ethical approval This article does not contain any studies with human participants or animals performed by any of the authors.

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