A POLICY PROPOSAL FOR A DEDICATED HEREDITARY BREAST AND OVARIAN CANCER SYNDROME FOLLOW-UP AND NAVIGATION PROGRAM

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A report submitted to the School of Graduate Studies in partial fulfillment of

the requirements for the degree of

Master of Nursing

Faculty of Nursing

Memorial University of Newfoundland

May 2021

St. John’s Newfoundland and Labrador

Abstract

Background and Purpose: Individuals who carry certain inherited pathogenic genetic variants (PVs) have an increased lifetime risk of developing hereditary breast and ovarian cancer (HBOC). Despite the effectiveness of risk reduction modalities, many PV carriers in Newfoundland and Labrador (NL) are unidentified. Moreover, for known PV carriers in the province, there is no systemic support available to them in their long-term risk management. Therefore, these high-risk individuals and families are not receiving the information and support needed to live a long, healthy life vis-à-vis their carrier status. The purpose of this practicum is to provide a rationale and recommendations for a prospective nurse navigation and follow-up program for HBOC PV carriers in NL.

Methods: A literature review, key informant consultations, and an environmental scan were conducted and used to inform a policy proposal for a novel program.

Results: Significant systemic barriers exist for PV carriers; many HBOC PV carriers and families have unmet information and psychosocial needs in the current primary care provider-dependent follow-up processes. A need exists for a novel follow-up and navigation and program. The literature review, consultations, and environmental scan were used to inform the five sub recommendations for the follow-up program including: a 1) carrier registry, 2) a nurse-led navigation program for PV carriers, 3) a multidisciplinary approach, 4) a person and family-centered approach, and 5) virtual and electronic delivery methods.

Conclusion: There is an unrivaled, cost-effective opportunity to improve outcomes in NL HBOC PV carriers through the proposed follow-up and navigation program.

Keywords: hereditary breast and ovarian cancer, genomics, patient navigation, nurse roles

Acknowledgements

I would like to give my thanks to the stellar individuals who made this project possible

To my practicum supervisor, Dr. Joy Maddigan, for her unwavering guidance, encouragement, belief in me, and commitment to helping me achieve my goals. I was deeply fortunate to go through this experience with you as my navigator, Joy.

To Dr. Lesa Dawson whose vision for the future of HBOC prevention inspired my interest in this topic. Moreover, for her expertise, encouragement, and willingness to collaborate during this project.

To Dr. Kerri Smith for her participation in early meetings that were pivotal to getting this practicum off the ground and for her willingness to share her expertise and resource materials that have been invaluable to this project.

To all the informants who so willingly agreed to participate in the consultations. I am in debt to you all for the rich contributions you made to this project.

To ALL of my beloved friends for everything you bring to my life. Special thanks to Chels for proofreading, Johanna for your endless support, Leanna for embarking on the MN journey with me, and Mike and Dan for my second home.

To my parents for a foundation that instilled the value of an education. For the drives to the library and for always rooting for me. Also, to my brother, Walter for our rapport and your help when I need it. Finally, to my feline family, Ace, Abner, and Annie for your unconditional love. A special shout out to the feline editor-in-chief, Sheila who is my biggest fan and loyal compadre.

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A Policy Proposal for a Dedicated Hereditary Breast and Ovarian Cancer Syndrome Follow-Up and Navigation Program

The burden of breast and ovarian cancer in Canada is significant. Breast cancer accounts for 4,992 deaths annually in Canada (12.9% of cancer-related deaths in females) (Canadian Cancer Statistics Advisory Committee, 2019). Ovarian cancer, while less prevalent than breast cancer (3,000 new cases annually in Canada), is the most lethal gynecologic malignancy and accounts for approximately 1,896 deaths annually (Canadian Cancer Statistics Advisory Committee, 2019). Various pathogenic variants (PVs), such as genetic variants in BRCA1, BRCA2, MLH1, and MLH2 genes, are associated with increased breast and ovarian cancer risk. For example, BRCA 1 & 2 PV carriers have a 51-72% lifetime risk of breast cancer and an 11-44% lifetime risk of ovarian cancer (Kuchenbaecker et al., 2017; Rebbeck et al, 2015). Several recommended strategies are proven to mitigate HBOC risk in known, asymptomatic PV carriers. Risk-reducing salpingectomy oophorectomy (RRSO), has been associated with 80% reduction in ovarian/fallopian tube cancer risk and a 50% reduction in breast cancer risk in asymptomatic women who carry PVs in the BRCA1 &2 genes (Rebbeck et al., 2009). Furthermore, annual breast MRI, alternating every six months with annual breast mammography has a combined sensitivity of > 90% for detecting early stage breast cancer and is recommended for high risk women (Warner, 2018). Risk-reducing mastectomy is also discussed as a potential risk-reduction option which has been shown to virtually eliminate the risk of breast cancer in female BRCA 1 & 2 PV carriers (Li et al., 2016).

HBOC syndrome is most attributed to PVs in the BRCA 1 & 2 tumour suppressor genes but has also been associated with up to 24 various inherited PVs (Nielsen et al., 2016). An estimated 20% of cases of ovarian cancer are associated with a hereditary predisposition

syndrome (Susyznska et al., 2020). There is no proven method of primary ovarian cancer screening effective in reducing ovarian cancer morbidity and mortality in the general, average risk population (Buys et al., 2011; Jacobs et al., 2016). Therefore, the identification of an inherited ovarian cancer predisposition syndrome, and adherence to risk reduction options, offers an unprecedented opportunity to prevent ovarian cancer.

Despite the survival advantage and cost-effectiveness offered by these HBOC risk reduction strategies, in many Canadian jurisdictions, including Newfoundland and Labrador (NL), there are no systemic follow-up programs that address the unique needs of HBOC PV carriers. Once the initial genetic counselling appointment and disclosure of an individual's PV genetic testing results have occurred, individuals and/or their primary care providers are solely responsible for facilitating screening and risk-reduction recommendations. In an unpublished manuscript by Roebothan et al. (n.d.) the study authors characterized the population of female BRCA1 & 2 carriers in NL and evaluated factors that influence their HBOC screening and prevention modality adherence. They noted that only 41.6% of eligible BRCA PV carriers in NL had undergone the recommended annual MRI screening in an 18-month period ($p < 0.001$) (Roebothan et al., n.d.). They found that female BRCA PV carriers who were not seen by gynecologic and medical oncology specialists were less likely to follow care guidelines, "which may be explained by lack of patient knowledge of cancer risks and recommendations, lack of primary care physician comfort and/or expertise in BRCA1 & 2 care, or patient-specific factors related to anxiety or avoidance" (Roebothan et al., n.d, p.20). In so doing, the authors shone a spotlight on the need for a systemic follow-up program, such as a navigation program, for individuals with hereditary breast and ovarian cancer syndrome in NL.

Background to Practicum Project

In response to this identified need, my MN practicum project was a policy proposal for a HBOC PV carrier navigation and follow-up program in NL. My personal interest in the topic stemmed from the beginning of my nursing career when I was working on an acute gynecology surgery unit. Frequently, I was providing care to women living with ovarian cancer. I quickly realized that ovarian cancer outcomes are poor and that by the time many of these women came to us for surgery, they were in the advanced stages of the disease. I came to appreciate the urgent need for strategies to improve ovarian cancer outcomes. What I did not understand at the time was that furthering our understanding of inherited breast and ovarian cancer risk offers an opportunity to prevent those diseases. It was not until I attended an ovarian cancer education educational symposium, offered through my affiliate university, that this link became fully clear to me. Genomic content was virtually absent from my undergraduate nursing education. The more I learned, I began to see this omission of genomics for what it is: an enormous, missed opportunity. This practicum project was therefore an opportunity to propose recommendations to mitigate the current gaps in PV carrier follow-up care and to advocate for an increased role of nurses in the provision of hereditary cancer and genomic follow-up care.

Goal and Objectives

The overall goal of the practicum was to provide a rationale and recommendations for a prospective Navigation and Follow-Up Program for HBOC PV carriers in NL.

The key objectives of the practicum were the following:

1. Describe the current process of follow-up and supportive care for HBOC PV carriers in NL in comparison to other regions outside of NL
2. Identify priority health care needs for the HBOC PV carrier population in NL and how a

program would address these needs

3. Develop recommendations for a HBOC PV carrier navigation program based on a literature review, consultation from provincial stakeholders, and an overview of programs in regions outside of NL
4. Explore how integration of the nursing profession in genetic care could improve outcomes for HBOC carriers and families
5. Demonstrate advanced nursing practice competencies.

Overview of Methods

To meet these key objectives, an integrated literature review, key stakeholder consultations, and an environmental scan were conducted. In the integrative review, barriers and facilitators of HBOC PV carrier follow-up care were examined, as were various inherited cancer prevention (ICP) follow-up interventions reported in the literature. (N=8) key stakeholder consultations were conducted with health care professionals and researchers in NL who had perspectives in either genetics, hereditary cancer, or primary care. These stakeholders provided further insight into the barriers and facilitators of PV carrier follow-up care in NL and provided their priority recommendations for a prospective follow-up program. An environmental scan was also conducted by exploring the related hereditary cancer/ genetic follow-up and surveillance services offered in other jurisdictions. Informed by the literature search, stakeholder consult, and environmental scan, a program policy proposal document for a HBOC PV carrier navigation program model in NL was proposed. These project methodologies are summarized in further detail in the sections below.

It should also be noted that the conceptual framework used to guide the development of these documents was Hartrick-Doane and Varcoe's (2015) relational inquiry approach to

nursing. Using a relational inquiry approach to HBOC care, health care providers acknowledge the interpersonal, intrapersonal, and contextual factors (Hartrick-Doane & Varcoe, 2015), influencing PV carriers' conceptualization of risk, and their adherence to risk-management strategies. By using a relational inquiry approach, attention is also given to denounce the harmful effects of medical paternalism and to adopt an emancipatory approach to PV carrier follow-up care. The influence of this conceptual framework is evident throughout all development stages of this project.

Summary of the Literature Review

To develop an effective policy proposal, it was crucial that the proposal was sufficiently supported by evidence in the literature. A literature review was conducted in two parts, reflecting my two questions of interest. These questions were: 1) what are the identified barriers/issues in the care of individuals who carry PVs for HBOC in the current follow-up processes? and 2) What strategies/models have been examined in the literature for the supportive and follow-up care of individuals with HBOC PVs? A search was conducted in the Cumulative Index of Nursing and Allied Health Literature (CINAHL), Memorial University Library, and Google Scholar electronic databases. The full version of the integrated literature review is found in Appendix I of this practicum report. In this section of the practicum report, I will overview key findings of the literature review that were directly pertinent to recommendations in the policy proposal.

Part One Study Findings

In response to question one, common themes emerged from the studies of the PV carriers' self-reported barriers and experiences in their HBOC navigation journey. These themes served to elucidate priority needs for a follow-up and navigation program. These themes

included: health care provider-centered barriers, personalized considerations in risk management decision-making, unmet information needs, and the need for a coordinated approach to follow-up care.

Health Care Provider-Centered Barriers

Literature review findings were suggestive that PV carriers' experience significant health care provider-centered barriers in the current follow-up processes. Some PV carriers felt like they were the ones guiding their primary care provider in their HBOC journey, as they were not always provided accurate nor reliable information from their primary care provider (Cherry et al., 2013; Leonarcyzk & Mawn, 2015; Watkins et al., 2011). PV carriers reported inconsistencies in medical advice and surveillance recommendations from the various members of their health care team (Caita-Zufferey et al., 2015; Cherry et al., 2013; Watkins et al., 2011). A focus group of health care providers also noted that HBOC PV carriers were being missed in the disintegrated lines of communication involving multiple health care providers (Komatsu & Yagasaki, 2014). Watkins et al. (2011) noted that breakdowns in lines of communication about Lynch Syndrome management occurred most frequently between medical specialists and primary care providers. It was also reported that scarcity of health care providers, especially in rural areas posed challenges to adherence to recommended screening modalities (Leonarcyzk & Mawn, 2015). Moreover, with limited resources, breast care providers noted that they had limited time to focus on preventative measures when they were dealing with active of cases breast cancer and hereditary breast cancer prevention was placed lower on their list of priorities (Komatsu & Yagasaki, 2014).

Personalized Considerations in Risk-Management Decision-Making

The literature review findings were indicative that there are considerable nuances in PV carriers' risk management preferences. Yet, PV carriers reported that interactions with health care providers left them feeling as though they were "not being seen as a whole person" (Leonarcyzk & Mawn, 2015, p.77). Family planning preferences were reported as being influential in the uptake and timing of RRSO for many women (Cherry et al., 2013; Etchegary et al., 2015). HBOC PV carriers also noted that both their family history of cancer and family dynamics had tremendous impacts on their value appraisal and adherence to risk management modalities (Caita-Zuffery et al., 2015; Etchegary et al., 2015). For some women who lost family members to HBOC, making HBOC risk-management decisions was triggering to those feelings of loss (Caita-Zuffery et al., 2015). Others indicated that having experienced a breast/ovarian cancer diagnosis of someone close to them was influential in their adherence to recommended risk reduction modalities (Etchegary et al, 2015). Some asymptomatic PV carriers reported they felt a strong sense of moral obligation to both their ancestors and their dependents to make use of the genetic and medical information at their disposal and to stay healthy for their loved ones (Caiata-Zufferey et al. 2015).

HBOC PV carriers verbalized different levels of comfort about discussing PV carriership in their families. It was noted there was potential for guilt and/or resentment among families when one family member carried a PV and another did not (Hughes & Phelps, 2010). HBOC PV carriers also reported varied levels of comfort with openly discussing their carrier status and while some voiced that seeking support from other carriers was beneficial, others felt that there was a stigma associated with seeking professional and peer support (Hughes & Phelps, 2010). For many women, adequate, informed HBOC decision-making involved a combination of both

professional and peer support (Cherry et al., 2013; Hughes & Phelps, 2010; Rauscher & Dean, 2017).

Many PV carriers reported that they needed time to process information prior to making decisions about risk-management (Dean et al., 2017; Etchegary et al., 2015). Yet some women reported that they felt "pushed" (Caita-Zufferey, 2015, p.730) by their healthcare provider to adhere to risk-management guidelines. It was clear from the literature that there is no one-sized-fits-all approach to PV carrier care following the disclosure of genetic testing results. It was also evident that HBOC PV carriers seek more than just medical information, they need personalized, on-going support as they navigate the peaks and valleys in their PV carrier journey.

Unmet Information Needs

Many HBOC PV carriers indicated their information needs were not being met by the current PV carrier follow-up processes. This was evidenced during data collection in two studies when HBOC PV carriers made erroneous statements about risk-management (Cherry et al., 2013; Hughes & Phelps, 2010). While this finding was not universal among all the studies, it highlights that many women are not given the clear information to make a truly informed decision about HBOC risk-management. In a study by Etchegary et al. (2015), premenopausal women who underwent RRSO reported that prior to surgery, they did not have an adequate understanding of the full extent of surgical menopause and thus felt unprepared when these distressing symptoms occurred. In a study by Pezario et al. (2012), 73% (n=104) of women stated they received no on-going follow up with their Gynecology surgeon following the initial post-operative check up.

There were calls made by PV carriers in the literature for centralized and up-to date resources where PV carriers could retrieve reliable medical and research updates about HBOC such as an e-mail subscription, or other type of online resource (Hughes & Phelps, 2010). Other women indicated that they wanted decisional aid tools and/or prescriptive plans of action for their risk-management (Dean et al., 2017; Leonarcyzk & Mawn, 2015). Health care providers reported that the information and support needs of HBOC PV carriers could not be met by the current routine breast care follow-up and recommended separate outpatient follow-up clinics where due attention could be given to the information and supportive care needs of HBOC PV carriers (Komatsu & Yagasaki, 2014).

The Need for a Coordinated Approach

Several study authors concluded that there was a need for an overhaul in many current HBOC risk-management and follow-up care policies (Caita-Zufferey et al., 2015; Cherry et al., 2013; Komatsu & Yagasaki, 2014; Pezario et al., 2012; Watkins et al., 2011). Caita-Zufferey et al. (2015) and Komatsu and Yagasaki (2014) both concluded that there was a need to establish specialized, multidisciplinary hereditary cancer clinics to meet the current navigational needs of HBOC PV carriers. Similarly, there was a call made by Watkins et al. (2011) for an overhaul of the current fragmented, physician dependent screening paradigm for Lynch Syndrome PV carriers. Other authors highlighted that there was capacity to expand the role of nurses in the HBOC follow-up paradigm. Cherry et al. (2013) purported that a nurse ICP navigation model could be a promising alternative to the current processes and could provide BRCA PV carriers with support, access to other resources, and assistance with referrals and appointment scheduling. Komatsu and Yagasaki (2014) also noted that nurses have an opportunity to act as a

communication bridge among multidisciplinary HBOC team members and to improve the coordination of care.

Part Two Study Findings

In part two of the integrative review, quantitative study findings were examined wherein authors examined HBOC PV follow-up care models and interventions (models/interventions not currently in use in NL). These interventions included psychoeducational groups and workshops, cognitive behavioral interventions, dedicated HBOC follow-up clinics and technology platforms, In this section, there is a focus on interventions in the literature that were directly used as prototypes to inform the key policy proposal recommendations.

An iPhone Application for Screening Adherence

Cohen et al. (2018) provided pilot data on an iPhone application intervention designed to assist BRCA PV carriers in their adherence to recommended BRCA screening modalities. Cohen et al. (2018) provided preliminary data, suggestive that their iPhone app meets a practical need and is highly acceptable for BRCA PV carriers. While 94.3% of study participants reported their intention to engage in a BRCA surveillance plan, only 72.6% reported perceived health care system support for surveillance (Cohen et al., 2018). By the same vein, 50% of respondents reported they have difficulty keeping track of when to schedule their next BRCA screening appointment, and 20% reported that they rely on their primary care provider to do so. At baseline, the majority of the (n=69) participants who were provided a download code for the BRCA iPhone app agreed or strongly agreed that iPhone applications had a positive impact in their lives (Technology Acceptance Model Scores ranging from 3.4±1.1 to 4.1±0.7), and the majority of respondents also reported comfort with completing iPhone tasks (Comfort with

Technology Scores ranging from 3.5±0.92 to 3.8±0.82) (Cohen et al., 2018). Electronic smartphone applications are likely to become integrated into routine health care. These apps are a potentially valuable tool for PV carriers, as part of a dedicated follow-up program, when navigating the recommended ICP screening appointments.

Psychoeducational Group Sessions

In the studies examining group interventions for HBOC PV carriers, group interventions where psychosocial and educational content was offered were met with positive outcomes. ≥ 96% of participants in an annual Lynch Syndrome educational workshop reported overall satisfaction with the workshop (Corines et al., 2017); 91% reported that they found the content at the workshop useful and >87% reported that they were satisfied with the technical medical information components of the workshop. In other studies of outcomes of psychoeducational groups, there has been improvement noted in BRCA PV carriers' psychometric measures of anxiety, worry, and distress following participation in these groups (Esplen et al., 2004; Kwiatkowski et al., 2019; Listøl et al., 2017). Therefore, periodic group sessions where family members can attend and connect with other carriers may be a valuable component of a dedicated PV carrier follow-up program.

Dedicated HBOC Follow-Up Clinics

During the initial literature search, there were surprisingly few peer-reviewed studies of outcomes in multidisciplinary follow-up clinics for HBOC PV carriers (n=1). However, this number grew to (n= 5) later in the project development when I expanded my search strategies. (Bancroft et al., 2010 ; Engel et al., 2012; Firth et al., 2011; Pichert et al., 2010; Yerushalmi et al., 2016). Yerushalmi et al. (2016) reported on a specialized, multidisciplinary BRCA follow-up

clinic that PV carriers attended for bi-annual screening and follow-up clinic visits, with additional psychosocial support available to clinic attendees if needed. While the overall quality of the evidence was somewhat low, the data on patient outcomes in the clinic were promising. Only 7.2% of clinic attendees to date developed cancer. Of those 7.2% cases of cancer, 17 were breast cancers, one ovarian cancer, and three were additional cancers. Of the 17 cases of breast cancer, 94.1% of those cancers were detected at stage I disease when treatment outcomes are generally far more encouraging. Of those breast cancer cases, 70.6% were detected by MRI and 17.6% were detected by mammography (Yerushalmi et al., 2016). It is impossible to conclude with certainty that the low incidence of malignancy occurred exclusively as a result of the dedicated follow-up clinic, as clinic outcomes were not compared with outcomes from a matched control of a family physician based BRCA follow-up model. Still, the rate of RRSO uptake at age 40+ at the clinic in Yerushalmi et al. (2016) was high at 87.3% and the median and mean ages at time of RRSO were 46.5 and 48 years, ranging from 33-68. This high rate of RRSO uptake before natural menopause in clinic attendees was higher than in most other reported registries and in the literature (Yerushalmi et al., 2016). The median age at the time of RRSO in the multidisciplinary clinic was also lower than the median age at time of RRSO of 49.6±9.7 in NL BRCA PV carriers (Roebothan et al., n.d.) Further studies are needed to compare outcomes in dedicated follow-up clinics with family physician-based follow-up, still, Yerushalmi et al. (2016) provided a glimpse of outcomes in a successful multidisciplinary follow-up clinic. In other study findings, PV carriers reported high levels of satisfaction with multidisciplinary BRCA follow-up clinics (Firth et al., 2010). Pichert et al. (2010) found that BRCA PV carrier participation in a dedicated follow-up clinic was associated with significantly greater participation in related clinical trials ($p < 0.001$).

Summary of Consultations

Following the literature review, the next step to inform the policy proposal was to conduct a series of interviews with key informants in NL. There were four objectives for conducting the consultations. First, was to confirm with stakeholders that a systematic approach to HBOC PV carrier follow-up is relevant and acceptable to them. Second, was to identify issues in the current process of HBOC PV carrier follow-up care in NL. A third objective was to explore how the role of the nursing profession could be optimized in the proposed policy, as well as in genetic/genomic care. And the final objective was to provide stakeholders with an opportunity to recommend priority features for a HBOC navigation program policy, from their vantage point. Individuals who were identified as potential stakeholders were sent a letter explaining the nature of the consultations. Participants who agreed to participate in the consultations were: (n=2) genetic counsellors, (n=1) oncologist who provides high-risk PV carrier follow-up, (n=2) primary care physicians, (n=2) researchers involved in patient-centered research pertinent to these high-risk populations in NL, and an (n=1) individual involved in the development of a cancer prevention registry in NL. Consultations took place via telephone while some participants chose to respond to the questions through email. Different semi-structured question guides were developed, customized to the vantage point of the informant. Content analysis was used was used to analyze the raw data generated from the interviews. A full summary of the consultation methodologies, findings, and the full complement of semi-structured question guides is included in Appendix II of this report. Here, I highlight themes from the key informant interviews that directly informed the policy document. These themes were: the need for a centralized registry and follow-up program, and the potential nursing role.

A Centralized Registry and Follow-Up Program

The consensus among key informants was that PV carriers' needs are not being adequately met by the current primary care provider-dependent follow-up processes in NL. Some PV carriers in NL have reported frustrated with the uncoordinated, multiple appointments, lack of reliable information sources, and lack of psychosocial support. Some PV carriers even stopped attending their prevention and screening appointments because it all became too overwhelming. Another commonly reported theme in the consultations was the health care provider-centered barriers in PV carrier primary care follow-up. An informant who practices as an oncologist and works with PV carriers, was quick to shift the blame for this away from primary care physicians. She noted that inherited cancer genomics are complex and risk reduction recommendations can change rapidly, beyond a reasonable expectation of primary care providers' level of awareness. Primary care providers also have extremely busy family practices. Informants noted that because of this, there is a lack of quality assurance in the current primary care system for ensuring these tests are ordered and that specialist follow-up is arranged. There is no electronic or systemic recall to ensure PV carriers are getting the recommended screening tests at the appropriate intervals. One primary care provider found in his experiences, patients tend to be over-reliant on their GPs to remember and coordinate all screening appointments. He added that his 'greatest fear was that [he] will miss a screening and early detection will be missed'.

Informants agreed that a dedicated inherited cancer prevention registry would be ideal where data in the registry was connected to a carrier program that would assume responsibility for coordinating high risk follow-up, arranging screening, and providing assistance with the psychosocial impacts of carriership. An informant noted that there is an ethical responsibility to address the emotional needs of carriers and to support them in the genetic results disclosure with

their families. Therefore, it was unanimous among key informants that a prospective program should not have a sole focus on screening but also on helping PV carriers navigate the psychosocial and family implications of carriership.

Potential Nursing Role

Several informants expressed that there is unmet potential for nurses to become involved, and to ultimately improve the delivery of genetic health care. There are nurses who work as genetic counsellors in Canada who were 'grandfathered in' prior to the advent of the Canadian Association of Genetic Counsellors. Since then, the entry level requirement for certification as a genetic counsellor is a master's degree in genetic counselling. One informant agreed that the Masters of Genetic Counselling should be the entry level requirement but maintained that there are many other ways that nurses may contribute to genomic care. These included, but are not limited to collecting adequate family histories, conducting, and assisting in genetic research, engaging in genetic follow-up and supportive care. The successful implementation of nurse-led navigation programs in other populations, such as patients diagnosed with cancer, was also referenced to give credence to the argument that a nurse navigator could provide a similar service to PV carriers in this program. One informant also suggested that a nurse practitioner associated with a hereditary cancer screening program could be the one to order the recommended screening tests. Informants stated that there are clear opportunities to mobilize genetic nursing in nursing practice, research, education, and in professional governance.

Summary of the Environmental Scan

Also included in Appendix II of this practicum report is the full environmental scan summary. To complement the key informant interviews, it was also important to determine what works well for PV carrier follow-up programs in other health jurisdictions. Therefore, an

environmental scan was conducted, and several objectives of the environmental scan were used to inform the policy proposal. These objectives were: 1) to gain an idea of what services are common features of familial/hereditary cancer follow-up programs in areas of Canada outside of NL and globally. 2) To examine features of a high-risk breast screening registry.

Familial Cancer Clinics

In other areas of Canada and around the world, familial cancer clinics are available as part of routine health care to individuals with inherited cancer predisposition syndromes after they receive their genetic testing results. Many of these centers offer multidisciplinary care from professionals such as geneticists, genetic counsellors, nurses, dieticians, gynecologists, oncologists, social workers, and psychologists. By having a broad multidisciplinary lens in these programs, the multiple facets of PV carriership are addressed. Also, a few of these clinics/programs offer periodic carrier support groups and sessions where PV carriers can liaise with other PV carriers and families and attend support sessions with guest speakers and genetic/hereditary cancer experts. Clinic attendees may be given the option to participate in research that may be of benefit to them. For example, clinic attendees who are followed long-term may be given the option to participate in trials of new prevention modalities, as the field of genetic medicine continues to rapidly evolve. PV carriers are scheduled to visit the clinics annually or bi-annually for surveillance, follow up and supportive care. In the province of Ontario, many of these clinics work with the High-Risk Ontario Breast Screening Program (OBSP) to coordinate breast surveillance of eligible high-risk individuals.

High-Risk Ontario Breast Screening Program

Definitions of 'high risk' of breast cancer across all Canadian provinces, generally include: known carriers of breast cancer predisposition PVs, and first-degree relatives of a known PV carrier who did not opt for genetic testing, among other risk factors (Canadian Partnership Against Cancer, 2018). Most provinces, including NL, have guidelines for recommended high-risk screening intervals, consisting of annual mammography, MRI and/or ultrasound beginning at age 30, 40 or 50, to stop at age 69-74 (CPAC, 2018). Despite these policies, women at high risk of breast cancer in NL are referred back to their primary care provider for further management (CPAC, 2018).

In the province of Ontario, there is a dedicated follow-up program for women at high-risk of breast cancer. To be enrolled in the High Risk OBSP, a referring physician must submit a requisition form to a designated High Risk OBSP site (Cancer Care Ontario, n.d.) It is implicit in the program requisition form that the ordering physician is requesting future MRI testing and in some cases, image guided biopsies, which under current Ontario regulations requires a physician's signature (Cancer Care Ontario, n.d.) Women are either directly enrolled in the program because they carry a known PV associated with increased breast cancer risk, or are a first-degree relative of someone with a known PV and underwent genetic counselling but opted not to have genetic testing (Cancer Care Ontario, n.d.) Women who are a first degree relative of someone with a known PV must be assessed by a genetic counsellor prior to enrollment in the high risk OBSP, even if they do not opt for genetic testing. The high risk OBSP program is operated by high risk OBSP navigators responsible for screening and breast assessment appointments, following up on abnormal results, arranging annual recalls, and communicating all imaging results to women and the referring physician (Cancer Care Ontario, n.d.)

Summary of the Policy Developed

The primary resource output of this practicum project was the 65+ page policy proposal document. The influence of the literature review findings, key informant interviews and environmental scan are evident in the proposal. The primary recommendation in the policy proposal was the establishment of a novel, dedicated HBOC PV carrier navigation and follow-up program in NL. There were five key sub recommendations outlined in the policy proposal:

1) **A central HBOC PV carrier registry in NL**
2) **The establishment of a nurse navigator position to coordinate PV carrier surveillance and follow-up.**
3) **Coordinated involvement of a multidisciplinary HBOC team**
4) **A person and family centred approach to care**
5) **Virtual and electronic infrastructure to support delivery of the program**

While the full version of the policy proposal document is located in Appendix III of this report, a brief overview of the key policy recommendations is summarized in this section. Under the prospective program, individuals carrying pathogenic variants, likely pathogenic variants, and variants of uncertain significance would be entered into a PV carrier registry, following the disclosure of their genetic testing results. It was recommended that information in the carrier registry should include demographic information, information on the pathogenic variant, information on the testing panel ordered, and information on the recommended clinical actions for the variant. Moreover, the registry data should be updated as PV carriers undergo the recommended screening and risk-reduction modalities. The registry data should be accessible to

all approved multidisciplinary team members in the PV carrier follow-up program. The participation of PV carriers in the follow-up program would be voluntary.

The second recommendation, and central feature of the follow-up and navigation program, is the nurse PV carrier navigator role. The nurse navigator would have several responsibilities in the delivery of the program. (S)he will be responsible for maintaining ongoing follow-up with PV carriers, coordinating and scheduling screening appointments, connecting them with other multidisciplinary team members, and assisting with discussing genetic testing results with family members. The nurse navigator will rely on the carrier registry data to facilitate booking and screening reminders and to develop a personalized plan of care. Moreover, under the prospective program, PV carriers will be connected to expert multidisciplinary care providers as needed including, but not limited to, gynecologists, oncologists, breast surgeons, social workers, psychologists, dieticians, and genetic counsellors as needed, following ongoing assessment with the nurse navigator.

A person-centered approach is recommended for this program when working with PV carriers to deliver a follow-up care plan that is psychologically suited to their needs, preferences, and individual life circumstances. Given that these individuals are generally asymptomatic and lead busy, active lives, this program should have flexible delivery options. This may include evening and weekend appointment offerings and different communication options, such as in person appointments and/or video and telephone conferencing. The framework recommended to guide the delivery of this program is Hartrick Doane and Varcoe's (2015) relational inquiry. The principles of relational inquiry are relevant when delivering person-centered follow-up care that acknowledges all the systemic, familial, and individualized factors influencing adherence and

appraisal of risk mitigation in PV carriers. The use of this framework is a strategy to ensure a person and family centred approach.

A family centered approach is also a key feature recommended for this proposed program. Both affected and unaffected family members of PV carriers may experience psychologic distress emanating from the awareness of their own risk and/or the worry of their relatives' increased risk of cancer. In the current approach to PV carrier follow-up, family considerations are unaccounted for. This oversight would be addressed in the prospective program by encouraging the involvement of family members in a follow-up program. PV carrier families would also be supported by the follow-up program in family genetic results disclosure sessions. There, HBOC follow-up team members would be present to help explain the implications of PV carriership to PV carriers and to their potentially at-risk family members. The program would also support at-risk relatives who wish to pursue genetic testing by streamlining them with Provincial Medical Genetics.

The final recommendation in the proposal is that virtual and electronic delivery methods should be available for this program. The use of telehealth and virtual appointment delivery will ensure that PV carriers provincewide have equitable access to quality follow-up services, regardless of their geographic location. PV carriers should also be able to opt for electronic text appointment reminders and online information/updates and to connect with other PV carriers should they desire. It is recommended that the delivery of the proposed program is compatible with the province-wide electronic medical record and electronic health record software so that relevant information pertaining to their PV carriership can be clearly communicated and shared to all relevant providers in the circle of care, ensuring continuity and consistency of care.

Discussion of Advanced Nursing Practice (ANP) Competencies

In addition to the aforementioned documents, a key objective of this practicum project was to demonstrate advanced nursing practice (ANP) competencies. In an ANP Framework, the Canadian Nurses Association (CNA) (2019) outlined six categories of ANP competencies: direct comprehensive care, health system optimization, education, research, leadership, and consultation and collaboration. The ANP competencies of research, health system optimization, and consultation and collaboration were demonstrated in the completion of this practicum project.

Research

This ANP competency refers to "generating, synthesizing, critiquing and applying research evidence" (CNA, 2019, p. 32). This ANP competency was demonstrated through the utilization and application of research findings in each stage of policy development. In the integrative literature review, findings in peer-reviewed articles were appraised and used as a basis to inform the consultations, environmental scan and ultimately, the policy proposal. Although this was not a formalized research project, in the key informant consultations and environmental scan, I engaged in data collection, data analysis, and undertook the appropriate steps to safeguard the data and to ensure ethical conduct in data collection. This data was used as evidence to inform the policy proposal.

Health System Optimization

Advanced practice nurses demonstrate this competency by contributing "to the effective functioning of health systems through advocacy, promoting innovative client care and facilitating equitable, client-centered health care" (CNA, 2019, p. 30). This program policy is largely an

advocacy paper. I presented the argument that PV carriers have an unequivocally increased of cancer when compared to the general population and that it therefore unjust to treat them the same as people of average risk. In the policy paper, I made recommendations to mitigate the systemic barriers that PV carriers experience in their access to follow-up care and to improve clinical and satisfaction outcomes in PV carrier populations. I also demonstrated how a prospective program could improve clinical efficiency, as I presented in the policy document that several of the proposed program features are validated in the literature as being cost-effective.

Consultation and Collaboration

This ANP competency is hallmarked by "effective collaboration and communication with clients, others health-care team members and stakeholders whose services impact the determinants of heath" (CNA, 2019, p. 33). This practicum project would never have gotten off the ground without the interdisciplinary consultation with my contact persons for the project. These two individuals are researchers in the area of hereditary cancer genetics, and they endorsed the need for this project and shared resources and input that were invaluable to this project. Furthermore, the key informant consultations were conducted with individuals with important clinical and research vantage points in the NL health care system. Their perspectives provided rich, pragmatic data, which informed the program policy proposal. The interdisciplinary collaboration as part of this project was an entry point for potential partnerships and future professional collaborations in research and policy development.

Next Steps

The policy proposal is an in-depth document with detailed recommendations for the proposed program. However, the proposal is only an important starting point in what is

ultimately required to bring the proposed program to fruition. It is important to continue to disseminate findings, seek feedback, amend the proposal as necessary, and to gain the support of stakeholders and health system funders. I have several strategies to accomplish this; firstly, is through continued collaboration and communication with key stakeholders and contact persons for the project. The completed program policy proposal was shared with my identified contact person for the project who is actively involved in genetic high-risk care in NL and is a founding member of the NL Ovarian Cancer Research and Education Fund. I informed her that she has permission to share findings from the program proposal to inform future studies in this area and/or to share the proposal with the administrators of her organization(s) for approval and the resources needed to implement. The work in this practicum project led me to become involved in a research team that she is leading, who are endeavoring to development a framework for a Lynch Syndrome hereditary cancer registry. I am hopeful that my work on this practicum project was a starting base for future multidisciplinary research collaborations, both in NL and in other regions, on the topic of ICP and genomic nursing. Future research may include patient-orientated research with carriers who can provide input and feedback on a proposed follow-up program model, as well as an in-depth cost-analysis of a prospective program when compared with the NL health system costs of non-surveyed HBOC. Ultimately, my desired goal is to inform a pilot project of a novel follow-up program for HBOC PV carriers.

Secondly, it will be important to keep an eye out for professional opportunities to promote the visibility of my project and other related work. I recently wrote a related article for submission in a Canadian nursing journal. The purpose of submitting an article for publication is to generate conversation about the potential nursing role in the delivery of genomic follow-up health care.. In the spring of 2021, I will be presenting this practicum project as part of the

Tuesday teleconference sessions offered by the College of Registered Nurses of Newfoundland and Labrador.

Finally, I will also keep an eye out for educational opportunities to increase my own genomic literacy and competencies. In the long-term, I hope to pursue both further graduate and professional development education that is aligned with my interest in hereditary cancer prevention and genomics. This will enhance my credibility to disseminate research findings, as well as to make recommendations for improved inherited cancer prevention delivery models. In short, the buck does not stop here in this report with the work that I have completed to date as part of this project.

Conclusion

The project objectives were met over the course of this practicum project. An overview of the current HBOC syndrome follow-up processes in NL were presented with a rationale for how this current approach is inadequately meeting the complex medical, informational, and psychosocial needs of PV carriers and families. To understand and present possible solutions to these gaps, a literature review, key informant consultations, and an environmental scan were conducted. These documents were used to inform five key recommendations in a policy proposal for prospective registry and follow-up program for HBOC PV carriers in NL. Through completion of this project, I was able to demonstrate advanced practice nursing competencies of research, health system optimization, and consultation and collaboration. A compelling case was made in the proposal document for both the need for, and the feasibility of prospective follow-up and navigation program for NL HBOC PV carriers. The policy proposal document is poised to help shape healthy public policy for PV carriers and to inform future exploratory and pilot studies of a dedicated, novel follow-up and navigation program.

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Appendix I Literature Review

The Case for Navigation Programs for Individuals and Families with Hereditary Breast and Ovarian Cancer Syndrome

An Integrative Literature Review

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The Case for Navigation Programs for Individuals and Families with Hereditary Breast and Ovarian Cancer Syndrome: An Integrative Literature Review

Various pathogenic variants (PV) such as mutations in the BRCA1, BRCA2 genes are associated with an inherited breast and ovarian cancer predisposition (HBOC) syndrome. For example, individuals who carry PV (deleterious changes) in the BRCA1/2 genes have a 51-72% lifetime risk of breast cancer and an 11-44% lifetime risk of ovarian cancer (Kuchenbaecker et al., 2017; Rebbeck et al, 2015). Despite the high lifetime risk, breast and ovarian cancer associated with germline mutations can be prevented with the uptake of recommended risk-reduction modalities proven to be effective in known PV carriers.

Despite clear evidence that targeted HBOC prevention and screening modalities are effective, there is a paucity of follow-up programs in Canada that address the health considerations specific to this high-risk population. Once the genetic results disclosure has occurred, PV carriers and their primary care providers are often solely responsible for the coordination and compliance with screening and prevention recommendations (Roebothan et al., n.d.). This is concerning, given that in a study of participants with a personal or family history of genetic disease, 64% of study participants reported receiving no genetics education from their primary provider (Harvey et al., 2007). In an unpublished manuscript by Roebothan et al. (n.d.) the study authors characterized the population of female BRCA1 & 2 PV carriers in Newfoundland and Labrador (NL), and evaluated which factors influence their HBOC screening and prevention modality adherence. They found that “women who did not access care from [cancer and genetic] specialists were less likely to follow care guidelines, which may be explained by lack of patient knowledge of cancer risks and recommendations, lack of primary

care physician comfort and/or expertise in *BRCA1/2* care, or patient-specific factors related to anxiety or avoidance" (Roeboethan et al., n.d, p.20). In so doing, the authors highlighted the need for a psychosocial and educational follow-up model, such as a navigation program, for individuals with HBOC predisposition syndrome.

To situate this review, I will provide a background of the prevalence and implications of HBOC syndrome in Canada, and specifically in the province of NL. From there, I have two specific objectives in conducting this integrative review; first is to examine the barriers to quality follow-up care for individuals with HBOC predisposition syndrome. The second is to determine to what extant HBOC PV carrier follow-up program models/interventions have been implemented. The end goal of this literature review is to present the available evidence stemming from these two questions so it may be used to inform policy recommendations for a HBOC patient navigation program in the province of NL. For this literature review, the bulk of the information retrieved will be generalizable to all individuals and families with germline PVs known to be predisposing to breast and/or ovarian cancer.

Background

Hereditary Breast and Ovarian Cancer Syndrome (HBOC) is an adult-onset cancer predisposition syndrome associated with significantly increased lifetime risks of developing breast and ovarian cancer (Jackson Laboratory, 2020). It should be noted that while HBOC syndrome is most often associated with BRCA1 &2 PVs, it has also been associated with up to 24 various inherited PVs (Nielsen et al., 2016). This includes germline mutations in the mismatch repair genes associated with Hereditary Non-Polyposis Colorectal Cancer or Lynch Syndrome (LS) which is also associated with an increased lifetime risk for ovarian cancer.

Prevalence of HBOC in Canada and NL

The incidence of BRCA PV carriers in the general population is estimated at approximately 1 to 300 to 1 in 500 (Nelson et al., 2014). 354,965 Canadians have been or will be diagnosed with a hereditary breast or ovarian cancer and therefore as many as 709, 930 (1.9%) of Canadians carry a known PV that predisposes them to hereditary breast and/or ovarian cancer (Hereditary Breast and Ovarian Cancer Society, 2018). Roebothan et al. (n.d.) conducted a retrospective study to characterize the entire dataset of female BRCA1/2 mutation carriers in NL and the factors influencing their adherence to recommended risk-reduction modalities. They identified a total of 276 BRCA mutation carriers since the introduction of genetic testing in NL. While the BRCA carrier prevalence rate of 0.05% in the general NL population was lower than expected, they cited the current opportunistic genetic testing paradigm, resulting in the under-identification of BRCA carriers in the province, as a possible cause for their findings. Using population risk estimates of 1 in 300, Roebothan et al. (n.d.) purported that they estimate the prevalence of BRCA 1/2 PV carriers in NL to be upwards of 1,700. The low prevalence of BRCA PV carriers in NL could also be attributable to the fact that NL is considered a founder population, primarily of Irish and English descent (Zhai et al., 2016). Despite the low prevalence of BRCA PV carriers, NL has the highest rate of breast cancer mortality and the fourth highest rate of ovarian cancer mortality in the country (Canadian Cancer Statistics Advisory Committee, 2019).

Dawson et al. (2020) recently published their case/control study on the molecular genetics of HBOC in NL where they performed multigene paneling on five female probands with a personal history of breast and/or ovarian cancer who tested negative for known high and

moderate risk HBOC variants, but all shared a variant of uncertain significance (VUS) in the RAD51C gene. Interestingly, when Dawson et al. (2020) performed control haplotype analysis, there was a 52-fold increase of the RAD51C VUS in the NL population versus in general Caucasian population control (0.165% vs 0.0032%). From this, they concluded that the RAD51C(NM_058216.3: c.571 + 4A > G) variant is pathogenic; this and "other yet undiscovered variants may explain the increase incidence and perhaps mortality associated with HBOC in NL" (Dawson et al., 2020, p.11). The unique genetic composition of NL is characterized by the historical isolated nature of the island, an increased inbreeding coefficient, and reduced heterozygosity (Zhai et al., 2016). As new evidence emerges surrounding the molecular genetic makeup of HBOC in both NL and global populations, and as additional VUS in HBOC are reclassified as pathogenic, this will further the need for programmatic follow-up for these high-risk women in NL.

Recommended HBOC Risk-Reduction Modalities

There is strong evidence of the favorable impact of BRCA risk-reduction modalities on morbidity and mortality in asymptomatic BRCA mutation carriers. Risk-reducing salpingectomy oophorectomy (RRSO) has been associated with 80% reduction in ovarian/fallopian tube cancer risk and a 50% reduction in breast cancer risk in asymptomatic women who carry mutations in the BRCA1/2 genes (Rebbeck et al., 2009). Risk-reducing mastectomy (RRM) is also discussed as a potential risk-reduction intervention which has been shown to essentially eliminate the risk of breast cancer in asymptomatic female BRCA 1/2 carriers (Li et al., 2016).

Likewise, annual breast magnetic resonance imaging (MRI), alternating every 6 months with annual breast mammography has been shown to have a combined sensitivity of > 90% for detecting early stage breast cancer and is therefore recommended in this population (Warner,

2018). The Breast Disease Site Group (2017) of the Eastern Health (EH) Regional Health Authority (and NL tertiary care provider) established a policy recommending alternating annual MRI and mammography for women with an increased risk of breast cancer, starting at age 30. The Breast Disease Site Group (2012) also established a policy stating that premenopausal women ≥ 35 and postmenopausal women with a high risk of hereditary breast cancer should be offered oral Tamoxifen (a selective estrogen receptor modulator) once daily for five consecutive years. Use of oral contraceptive medication for six or more years has been associated with decreased risk of developing ovarian cancer in BRCA 1/2 carriers (OR 0.62, 95% CI 0.35-1.09) (Whittemore et al., 2004).

Complexity of Decisions Surrounding HBOC Syndrome Carriership

While the evidence surrounding risk-reduction modalities is extremely encouraging and while increased breast surveillance and prophylactic surgery are widely available in most Western countries, decisions surrounding risk-reduction remain complicated for both patients and healthcare providers. For example, RRSO and RRM offer the most significant protective factors for HBOC but the decision to undergo prophylactic surgery must take into consideration other factors in a woman's life. Women who underwent RRSO reported distressing vasomotor and urogenital symptoms associated with surgical menopause, reduced sexual pleasure, and for women of childbearing age, an RRSO meant that they were not able to have any further children (D'Alonzo et al., 2018). In the same study, women who underwent RRM reported the negative effects of the surgery on their body image and sexuality (D'Alonzo, 2018). HBOC PV carriers also face complex decisions related to the disclosure of this information to family members, and/or the decision to have children given the chance of passing on the mutation (White et al., 2014). There may be disagreement among family members about whether the knowledge of

one's carrier status is beneficial or if this knowledge may cause iatrogenic psychological harm (Speice et al., 2002).

Problem with the Current Follow-up Processes in NL

Once an individual is referred to the NL provincial genetics program, genetic testing is exclusively delivered by medical genetics specialists after in-person counselling (Adams & Etchegary, 2015). Following the disclosure of genetic results, navigation of annual recommended follow-up modalities, complicated treatment decisions and family considerations are left entirely in the hands of the individual and their primary care provider (Roebothan et al., n.d.). Dr. Dawson, a gynecologic oncologist and Associate Professor of clinical genetics at Memorial University of Newfoundland, described BRCA carriers in the province as being "orphaned by the healthcare system" (Mercer, 2018, para 2). Other than the work done through Dr. Dawson's gynecologic oncology inherited cancer prevention clinic at Memorial University, there is no programmatic follow-up for these women in the province, resulting in a significant missed opportunity. Roebothan et al. (n.d.) noted that only 41.6% of BRCA PV carriers in NL had undergone the recommended MRI screening in the past 18 months ($p < 0.001$). While proven effective to prevent disease, the recommended modalities are still largely underutilized by HBOC PV carrier populations who stand to benefit from them.

The current follow-up processes in NL, and in many other jurisdictions, is not only ineffective, but this approach is in contradiction with policy statements in fundamental Canadian health promotion framework documents such as the Canadian Lalonde (1974) report, *A New Perspective on the Health of Canadians* and *Achieving Health for all: A Framework for Health Promotion* (Epp,1986). Furthermore, with the high costs of cancer treatments and an aging

Canadian population, “a cancer care system that focuses on treatment over prevention is not sustainable” (Roebothan et al., n.d., p.4).

It is clear the need exists for improved HBOC previvor support and follow-up care. Therefore, there are two questions that must be posed in order develop an effective healthy public policy for a HBOC previvor care model. 1) What are identified barriers/issues in the care of individuals who carry PVs for HBOC in the current paradigm? and 2) What strategies/models have been examined in the literature for the supportive and follow-up care of individuals with PVs predisposing them to HBOC? A literature search was conducted to respond to these questions.

Conceptual Framework

Two conceptual frameworks were selected to guide this literature review. HBOC is truly a family affair and targeted strategies should be approached accordingly. Therefore, I selected Street and Soldan’s (1998) conceptual framework of psychosocial issues in families with genetic conditions. The authors purported that individuals require routine psychosocial care during the pre-illness phase of genetic disease and not just in the incidence of poor coping. They postulated that when caring for PV carriers, health care providers should move away from “disease-specific framework with its limited acknowledgment of psychosocial issues, to one informed by the family systems life cycle and the therapeutic practices that emanate from it” (Street & Soldan, 1998, p.231).

The second conceptual framework used to inform the literature review is Relational Inquiry which was developed as an approach to nursing practice. Hartrick-Doane and Varcoe (2015) enlisted an approach consisting of two main elements, “relational consciousness and

inquiry as a form of action" (p.3). To engage in relational consciousness, health care providers must be attentive to what is going on interpersonally (among and between people), intrapersonally (within people), and contextually (within factors and structures), in all health situations (Hartrick-Doane & Varcoe, 2015). They described inquiry as the "how-to" (Hartrick-Doane & Varcoe, 2015, p.6) of relational inquiry. In other words, inquiry involves using the insight gained through relational consciousness when implementing health interventions. Using a relational inquiry approach to the care of HBOC PV carriers acknowledges the complexities of the interpersonal, intrapersonal, and contextual factors at play in their journey as HBOC previvors. Relational inquiry was influenced by critical theory, a philosophical movement where disparities in sociopolitical structures are highlighted and there are calls for action to mitigate the lasting effects of disadvantageous socioeconomic, political, and historical ideologies (Polit & Beck, 2017). By using a relational inquiry approach, attention is also given to denounce the harmful effects of medical paternalism and adopt an emancipatory approach to HBOC care.

Search Strategy

The literature search was conducted in two parts, reflecting my two separate questions of interest. I searched the Cumulative Index of Nursing and Allied Health Literature (CINAHL), Memorial University Library, and Google Scholar electronic databases for both literature review questions. Additionally, both the ancestry and descendancy approaches as described by Polit and Beck (2017) were used as search strategies. Using the ancestry approach, I located relevant studies that were listed in the reference pages of retrieved studies and included them in the review. Using the descendancy approach, I used the 'cited by' option in the MUN Library database to find more recent, relevant articles wherein the authors had cited my retrieved studies. Adhering to the guidelines of Polit and Beck (2017), studies included in this review were all

written in English language within the past 15 years. Retrieved studies were peer-reviewed and featured either a qualitative or quantitative design. Inclusion criteria for studies in both question one and two were: studies pertinent to individuals and families carrying pathogenic variants known to predispose to HBOC. The strength and quality of the evidence presented in the quantitative studies were evaluated using guidelines from the Public Health Agency of Canada (PHAC) (2014) critical appraisal tool, while the Joanna Briggs Institute (2017) critical appraisal checklist for qualitative research was used to appraise the qualitative studies.

In part one, I sought to answer my question, "What are reported barriers to adequate follow-up care of individuals at high risk for HBOC in the current paradigm?" To this end, combinations of the descriptors 'HBOC', 'carriers', 'Familial Cancer', 'Hereditary Cancer', 'Lynch Syndrome' were used in combination with the search terms 'AND' 'follow-up adherence', 'barriers', 'screening', "psychosocial", 'educational needs'. A total of (n=10) studies were retrieved that were deemed applicable to part one of this review. Most of these studies (n=9) had a qualitative design. HBOC PV carriers provided rich, first-person testimony of their experiences as PV carriers and their perceived barriers with HBOC follow-up and risk management. In one study, health care providers provided first person accounts of their perceived health care barriers to HBOC management (Komatsu & Yagasaki, 2014). A summary of these studies can be found in Appendix A of this integrative review.

For the second question, I sought to answer, "What strategies/models have been examined for the supportive and follow-up care of individuals and families with PVs predisposing them to HBOC?" To do so, I used combinations of the search terms 'HBOC', 'BRCA carriers', 'Familial Cancer', 'Hereditary Cancer', 'Lynch Syndrome' in combination with 'AND' 'patient navigation', 'follow-up care', 'patient support programs', 'supportive care'

and ‘psychoeducational interventions.’ Inclusion criteria specific to question two was that the interventions in the studies had to occur in the phase post-genetic counselling. While the interventional studies retrieved in question two were of varied time lengths and modes of delivery, they were all of sufficient relevance to one of two the key aims of this literature review. A total of (n=9) studies were deemed applicable to the focus of question two. Strength of the study designs ranged from strong to weak, and the quality of the evidence in the studies ranged from high to low as per the PHAC (2014) critical appraisal criteria. A summary of these studies can be found in Appendix B.

Part One Study Findings

In response to question one, five common themes emerged from the studies of the participants’ reported barriers and experiences in their HBOC navigation journey. These themes were: health care provider-centered barriers, personalized considerations in risk management decision-making, unmet information needs, the need for peer support, and the need for a coordinated approach to follow-up care.

Health Care Provider-Centered Barriers

Reiteratively, in many current Canadian HBOC paradigms, once disclosure of the individual’s PV carrier status has occurred, the navigation of health considerations specific to their carrier status becomes the responsibility of the individual and/or their primary care provider (Roebothan et al., n.d.). Taking this into consideration, it is concerning that study participants commonly reported receiving insufficient guidance from their primary care provider. Several participants voiced that they felt as though they were the ones guiding their primary care provider in their HBOC journey, as the information they were provided was not always accurate

nor reliable (Cherry et al., 2013; Leonarcyzk & Mawn, 2015; Watkins et al., 2011). Participants reported challenges in obtaining relevant HBOC risk-management information from their primary care provider, especially about subjects considered taboo such as the potential adverse sexual implications of HBOC risk management (Cherry et al., 2013). Some participants reported that if PV carriers were younger in age, their primary care provider discredited the importance of adherence to recommended screening and essentially 'kicked the can further down the road' (Watkins et al., 2011).

There was also considerable confusion reported by PV carriers about inconsistencies in medical advice and surveillance recommendations from various members of their health care team (Caita-Zufferey et al., 2015; Cherry et al., 2013; Watkins et al., 2011). This caused the women to feel overwhelmed and frustrated by the sometimes-conflicting advice they received (Caita-Zufferey et al., 2015). A similar concern was echoed in a focus group of health care providers who noted that there was a high probability of HBOC PV carriers being missed in the disintegrated lines of communication involving multiple health care providers (Komatsu & Yagasaki, 2014). Watkins et al. (2011) noted that breakdowns in lines of communication about Lynch Syndrome management appeared to be most salient between medical specialists and primary care providers. It was also reported that scarcity of health care providers, particularly in rural regions posed challenges to adherence to recommended screening modalities (Leonarcyzk & Mawn, 2015). Moreover, with limited resources, breast care providers noted that they had limited time to focus on preventative measures when they were dealing with active of cases breast cancer and thus hereditary breast cancer prevention was placed lower on their list of priorities (Komatsu & Yagasaki, 2014).

Personalized Considerations in Risk-Management Decision-Making

Another theme that emerged from the data was the imperative of personalized considerations when assisting individuals to navigate their HBOC journey. Some participants noted that interactions with health care providers left them feeling as though they were "not being seen as a whole person" (Leonarcyzk & Mawn, 2015, p.77). In one example, family planning vis-a-vis HBOC PV carriership is a highly personalized decision. Individuals and families making these decisions require sensitivity and support from health care providers to make the most suited, informed choice for their life circumstances. This is perhaps best summarized by one HBOC PV carrier that, "[family planning for PV carriers] is not a statistic, it's what they feel in their heart is the right thing to do" (Rauscher & Dean, 2017, p. 491). Some participants stated it was important that family planning and counselling involve a two-way dialogue about both previvor and their spouse's feelings about family planning, yet it was common for the spouse to be overlooked in these discussions (Rauscher & Dean, 2017). Family planning was also influential in the uptake and timing of RRSO for many women (Cherry et al., 2013; Etchegary et al., 2015).

HBOC PV carriers also noted that both their family history of cancer and family dynamics had tremendous impacts on their value appraisal and adherence to risk management modalities. For some women who had lost family members to HBOC, making HBOC risk-management decisions was triggering to those feelings of loss (Caita-Zuffery, 2015). Others indicated that having experienced a breast/ovarian cancer diagnosis of someone close to them was influential in their stringent adherence to recommended risk reduction modalities (Etchegary et al, 2015). Other women reported that the risk-management decision experiences of family members who were also PV carriers were influential factors in their own risk-reduction decision

making (Cherry et al., 2015). Some asymptomatic PV carriers reported they felt a strong sense of moral obligation to both their ancestors and their dependents to make use of the genetic and medical information at their disposal and to stay healthy for their loved ones (Caiata-Zufferey et al. 2015).

HBOC PV carriers verbalized different levels of comfort about discussing PV carriership in their families. It was noted there was potential for guilt and/or resentment among families when one family member carried the gene and another did not, and opinions often greatly varied between family members about whether or not PV carriership should be disclosed (Hughes & Phelps, 2010). HBOC PV carriers also reported varied levels of comfort with openly discussing their carrier status and while some voiced that seeking support from other carriers was beneficial, others felt that there was a stigma associated with seeking professional and peer support (Hughes & Phelps, 2010). Many women reported that they needed time to process information prior to making decisions about risk-management (Dean et al., 2017; Etchegary et al., 2015). Yet some women also reported that they felt "pushed" (Caita-Zufferey, 2015, p.730) by their healthcare provider to adhere to risk-management guidelines. It was clear from the literature that there is no one-sized-fits-all approach to HBOC previvor care following the disclosure of genetic testing results. It was also evident that mere provision of medical information is insufficient, HBOC PV carriers need personalized, on-going support as they navigate the peaks and valleys in their journey as a HBOC previvor.

Unmet Information Needs

There is a need for information that many HBOC PV carriers reported is not being met in the current HBOC care paradigm. This was evidenced during data collection in two studies when HBOC PV carriers made erroneous statements about risk-management (Cherry et al., 2013;

Hughes & Phelps, 2010). One woman stated she believed an RRSO would increase the risk of breast cancer when in fact, RRSO decreases the risk of breast cancer (Cherry et al., 2013). While this finding was not universal among all the studies, it highlights that many women are not given the clear information to make a truly informed decision about HBOC risk-management. In the study by Etchegary et al. (2015), premenopausal women who underwent RRSO reported that prior to surgery, they did not have an adequate understanding of the full extent of surgical menopause and thus felt unprepared when these distressing symptoms occurred. These symptoms appear to be more pronounced in women who were premenopausal at the time of RRSO; in a study by Pezario et al. (2012), the distribution of women who reported "persistent severe" menopausal symptoms showed a linear correlation with younger age at the time of RRSO ($p=0.002$). Moreover, in the same study, 73% ($n=104$) of women stated they received no on-going follow up with their Gynecology surgeon following the initial post-operative check up. As these symptoms can be distressing and interfere with a woman's quality of life, it is important that pre-menopausal women who opt for RRSO are informed and adequately supported if these symptoms occur.

There were calls made by study participants for additional resources that could be useful to them in their HBOC journey. Some women reported that a directory including a list of relevant care providers (i.e. oncologists, gynecologists) would be a beneficial resource for use when navigating the health care system (Dean et al., 2017). Women also indicated the need for a centralized and up-to date resource where PV carriers could retrieve reliable medical and research updates about HBOC such as a newsletter, e-mail subscription, or other type of online resource (Hughes & Phelps, 2010). Other women indicated that they wanted decisional aid tools and/or prescriptive plans of action for the next steps in their risk-management (Dean et al., 2017;

Leonarcyzk & Mawn, 2015). Health care providers reported that the information and support needs of high risk HBOC women could not be met by the current routine breast care paradigm and recommended the establishment of separate outpatient follow-up clinics so that due attention could be given to the information and supportive care needs of HBOC previvors (Komatsu & Yagasaki, 2014).

The Need for Peer Support

A prevailing theme among the studies was the participants' reported desire and/or acclaim for formalized peer support in their HBOC navigation journey. HBOC PV carriers reported feeling different from the rest of the population and desired the opportunity to liaise with someone who could relate to their experiences as a HBOC previvor (Hughes & Phelps, 2010). Some HBOC previvors participated in online support groups such as Facing Our Risk of Cancer Empowered (FORCE) and Bright Pink (Dean et al., 2017; Leonarcyzk & Mawn, 2015). Feedback from participants who engaged in these online platforms was quite positive. For risk-reduction decision making, participants emphasized that they wanted to hear both the positive and negative risk-management experiences of other PV+ carriers, to make an informed decision in their own journey (Cherry et al., 2013; Dean et al., 2017). For many women, adequate informed HBOC decision-making involved a combination of both professional and peer support (Cherry et al., 2013; Phelps & Hughes, 2010; Rauscher & Dean, 2017). It became clear from study findings that when developing follow-up programs for HBOC PV carriers, the need for formalized peer support is an important consideration.

The Need for a Coordinated Approach

Several study authors concluded that there was a need for an overhaul in many current HBOC risk-management and follow-up care paradigms (Caita-Zufferey et al., 2015; Cherry et al., 2013; Komatsu & Yagasaki, 2014; Pezario et al., 2012; Watkins et al., 2011). Caita-Zufferey et al. (2015) and Komatsu and Yagasaki (2014) both concluded that there was a need to establish specialized, multidisciplinary hereditary cancer clinics to meet the current navigational needs of HBOC PV carriers. Similarly, there was a call made by Watkins et al. (2011) for an overhaul of the current fragmented, physician dependent screening paradigm for Lynch Syndrome PV carriers. Other authors highlighted that there was capacity to expand the role of nurses in the HBOC paradigm. Cherry et al. (2013) purported that a nurse navigation model for could be a useful alternative to the current paradigm and could provide BRCA PV carriers with support, access to other resources, and assistance with referrals and appointment scheduling. Komatsu and Yagasaki (2014) also noted that nurses have an opportunity to act as a communication bridge among multidisciplinary HBOC team members and to improve the coordination of care. To this effect, expanding the role of nurses in genetic and hereditary cancer previvor care could be a beneficial strategy for improving the follow-up care of this high-risk population.

Quality of the Evidence

As most of the evidence retrieved for question one came from qualitative studies with relatively small sample sizes, there is limited generalizability of the study findings. Though generalizability is not an expectation of qualitative research, qualitative research findings can be transferrable; this was the case with the qualitative studies in question one. Similarly, there was limited sociodemographic diversity among study participants. While this should be taken into consideration when weighing the evidence, there is an unassailable value in the rich, first-person

accounts of barriers and experiences as a HBOC previvor. As they are the ones who stand to benefit from targeted strategies, it is important that their priority needs are seen from their vantage point. Moreover, there was considerable overlap among participant and health care provider reported themes in the studies, which were conducted in a variety of countries and settings. This suggests that these are common issues and experiences shared by many HBOC PV carriers. While further studies are needed to quantify the findings discussed here, these study findings provide useful direction on priority needs and preferences of HBOC PV carriers when designing follow-up care models. It is also of note that all studies in question one were appraised using the Joanna Briggs (2017) Qualitative checklist and found to be of sufficiently high quality for inclusion in this integrative review.

Part Two Study Findings

In this section of this review, I will present quantitative study findings (n=9) wherein authors examined alternative HBOC PV follow-up care models and interventions. These interventions included psychoeducational groups and workshops, peer-support interventions, cognitive behavioral interventions, and dedicated HBOC follow-up clinics and technology. The effects of these follow-up strategies/interventions on psychosocial functioning, adherence to risk-reduction modalities, and unmet information need outcomes in HBOC PV carriers were explored.

Psychoeducational Support Groups

Group settings (facilitated by both peers and/or medical professionals) have been shown to be useful for facilitating psychological adjustment in cancer patients (Goodwin et al., 2001). Such groups have been linked with establishing feelings of mutual support and a sense of normal

through shared experiences, the opportunity to learn through the experiences of others, enhanced coping, as well as the acceptance of one's reality (Landsbergen et al., 2010). The purpose of psychoeducational support groups for HBOC PV carriers is to "assist women in making an informed choice, respecting and taking into account their private lives and circumstances" (Landsbergen et al., 2009, p. 214). This intervention model was examined by Corines et al. (2017), Kwiatkowski et al. (2019), Landsbergen et al. (2009), Listøl et al. (2017), and McKinnon et al. (2007). Three of the study interventions (Kwaitkowsi et al., 2019; Listøl et al., 2017; McKinnon et al., 2007) were one-time psychoeducational intervention group retreats for HBOC PV carriers, the psychoeducational support groups in Corines et al. (2017) and Landsbergen et al. (2009) occurred over the course of multiple months, or as quarterly and annual sessions. All the group interventions featured both psychological and medical content pertinent to HBOC PV carriers, such as risk-reducing surgery, considerations about genetic insurance discrimination, and family communication about genetic testing. Three major impact outcomes were identified of the psychoeducational group interventions including psychosocial impact outcomes, risk management adherence outcomes, and unmet information needs outcomes.

Psychosocial Impact Outcomes

There is evidence that psychoeducational support groups are acceptable to HBOC PV carriers; ≥96% of responding participants reported overall satisfaction with an annual Lynch Syndrome Educational workshop (Corines et al., 2017). Young female BRCA PV carriers who participated in a two-day psychoeducational retreat showed increases in psychometric measures of hope, self-esteem, and quality of life, one year following their participation in the intervention ($p= 0.00032$) (Kwiatkowski et al., 2019). Using the Hospital Anxiety and Depression Scale (HADS) metric, Listøl et al. (2017) reported that participant HADS-anxiety sub scores decreased

significantly from 6.2 to 5.2 (p=0.003), following a one-day psychosocial intervention for BRCA PV carriers. There were significant improvements noted in psychosocial functioning in a study of a multisite supportive-expressive group intervention for BRCA PV carriers, though this study did not meet inclusion criteria for this review as it was written > 15 years ago (Esplen et al., 2004). However, it is still worth noting that Esplen et al. (2004) observed significant improvements in participants' psychosocial functioning including cancer worries (p= 0.005), anxiety (p=0.033), and depression (p=0.015) following the intervention. While no differences were seen in participant Impact of Events (IES) scores following a one day BRCA psychoeducational intervention in the study by McKinnon et al. (2007), the authors noted this was potentially attributable to the high baseline IES scores in some of the participants. Given that the participant feedback on the intervention in McKinnon et al. (2007) was overwhelmingly positive, and the overall strength of the study design and quality of the evidence were relatively low, it is likely that a different research design would have yielded more favorable results.

Risk-Management Adherence Outcomes

Participation of HBOC PV carriers in psychoeducational support groups appeared to have a positive effect on their adherence to recommended risk-reduction modalities. In Landsbergen et al. (2009), individuals who participated in at least six out of eight BRCA psychoeducational group sessions were more likely to proceed with their initial preference for RRM within two years, when compared to a control group of BRCA PV carriers who did not partake in psychoeducational group sessions, 89% vs 63% respectively, (OR 4.8, p = 0.04). In a subsequent study by Landsbergen et al. (2010), for BRCA PV carriers who participated in an educational support group, their intention to undergo RRM increased from 37% to 44% following the intervention (p=0.7), and their intention to undergo RRSO increased from 71% to 81% (p=0.6).

It should be noted however that statistical significance was not achieved and thus these findings must be interpreted with considerable caution.

Unmet Information Needs Outcomes

91% of responding participants at a LS educational workshop (LSEW) over years 2-5 of the program reported that they found the information presented at the workshop to be useful and clear, and ≥87% reported they were satisfied with technical medical information provided to them through LSEW (Corines et al., 2017). In Landsbergen et al. (2010), there was a 19% increase in percentage of participants' information needs met following participation in a BRCA psychoeducational group, this was close to achieving statistical significance ($p=0.06$). In Esplen et al. (2004), 75% of participants noted improvements in their BRCA decision-making, and 60% reported an increase in assertiveness and knowledge regarding medical professional/patient communication following participation in a supportive-expressive therapy group. The evidence that participation in HBOC psychoeducational groups improves the unmet information needs of HBOC previvors must be cautiously interpreted and compared with other modalities of previvor information provision. Further robust studies to confirm this assertion are required.

Targeted Follow-Up Interventions

There were three interventional studies retrieved where the authors implemented an intervention to target a specific facet of HBOC PV carriership. This included a mindfulness-based stressed reduction training (MBSR) for menopausal symptoms in women who underwent RRSO (van Driel et al., 2019), a BRCA peer-support telephone intervention (White et al., 2014), and pilot data on an iPhone application to assist BRCA carriers in their adherence to recommended screening modalities (Cohen et al., 2018).

MBSR Intervention for Women who Underwent RRSO

In an RCT, (n=34) women participated in an 8-week intervention consisting of weekly two and a half hour sessions, a four-hour silent retreat evening, and 30-45 minutes of home exercises six times a week. (van Driel et al., 2019). When compared with a control group, these women had significant improvements in psychometric measures of menopausal related quality of life at 12-months post initiation of intervention, with menopause-specific quality of life: (MENQOL) scores of 3.6 (95% CI 3.1-4.0) versus 3.9 (95% CI, 3.5-4.4) respectively (p = 0.04). van Driel et al. (2019) noted that MBSR may be an acceptable complementary therapy to hormone replacement therapy (HRT) for women who report distressing RRSO menopausal symptoms, and may be particularly useful for women who have contraindications to HRT such as increased risk of breast cancer. There is strong evidence from van Driel et al. (2019) that the inclusion of MBSR could be an important component of an effective HBOC follow-up program.

Peer Telephone-Based Intervention

White et al. (2014) conducted an RCT where an intervention group of BRCA PV carriers received phone calls from trained peer volunteers over a 4-month period (an average of 3.7 calls). There were both short-term and long-term benefits observed from this intervention. The intervention was shown to have a positive effect on participants' psychological stress as reflected in their Impact of Events (IES) scale Breast cancer distress scores. Participants in the intervention group had a significantly greater reduction in breast cancer related distress when compared to a control group both immediately following the intervention -5.96 (95% CI, -9.80-2.13; p=.002), and two months later, -5.96(95% CI, -9.80-2.13; p=.002). There was also a greater reduction in unmet information needs in the intervention versus control group immediately following the intervention, -5.17 (95% CI, -7.96 to -2.37; p= < .00). However, this effect did not

reach statistical significance two months later, -.67 (95% CI -4.28 to - 0.93; p =0.21). Still, this was a well-designed and adequately powered study and White et al. (2014) gave considerable credence to trained, peer-based telephone support as an effective intervention as part of a holistic follow-up model for BRCA previvors.

An iPhone Application for Screening Adherence

Cohen et al. (2018) provided pilot data on an iPhone application intervention that they developed for BRCA PV carriers to assist them in their adherence to recommended BRCA screening modalities. While the 18-month follow-up data of this pilot project is still yet to be published, the baseline and preliminary data from Cohen et al. (2018) is suggestive that this iPhone app will be well-received and meet a practical need for BRCA PV carriers. While 94.3% of study participants reported their intention to engage in a BRCA surveillance plan, only 72.6% reported perceived health care system support for surveillance (Cohen et al., 2018). By the same vein, 50% of respondents reported they have difficulty keeping track of when to schedule their next BRCA screening appointment, and 20% reported that they rely on their primary care provider to do so. At baseline, the majority of the (n=69) participants who were provided a download code for the BRCA iPhone app agreed or strongly agreed that iPhone applications had a positive impact in their lives (Technology Acceptance Model Scores ranging from 3.4±1.1 to 4.1±0.7), and the majority of respondents also reported comfort with completing iPhone tasks (Comfort with Technology Scores ranging from 3.5±0.92 to 3.8±0.82) (Cohen et al., 2018). While it is still too premature to make sound conclusions on the utility of a BRCA iPhone app, Cohen et al. (2018) noted that in the first 21-months, 68 out of 69 participant provided codes were successfully downloaded and participants accessed the app an average of 6.28 times, ranging from 2-57 times. When the follow-up data is available and as health applications become

more routinely integrated into routine care, this intervention could be an extremely valuable tool for HBOC risk-reduction in a technology-dependent era.

Dedicated HBOC Follow-Up Clinics

There were surprisingly few peer-reviewed studies of the outcomes in multidisciplinary follow-up clinics for HBOC PV carriers (n=1). This was unexpected given in many countries, including some urban locations in Canada, familial cancer follow-up clinics are routinely integrated into regional genetics programs. Yerushalmi et al. (2016) reported on a specialized, multidisciplinary BRCA follow-up clinic that PV carriers attended for bi-annual screening and follow-up clinic visits, with additional psychosocial support available to clinic attendees if needed. While the overall quality of the evidence was somewhat low, the data on patient outcomes in the clinic were promising. Only 7.2% of clinic attendees to date developed cancer. Of those 7.2% cases of cancer, 17 were breast cancers, one ovarian cancer, and three were additional cancers. Of the 17 cases of breast cancer, 94.1% of those cancers were detected at stage I disease when treatment outcomes are generally far more encouraging. Of those breast cancer cases, 70.6% were detected by MRI and 17.6% were detected by mammography (Yerushalmi et al., 2016). It is impossible to say with certainty if the low incidence of malignancy occurred exclusively as a result of the dedicated follow-up clinic, as clinic outcomes were not compared with outcomes from a matched control of a family physician based BRCA follow-up model. Still, the rate of RRSO uptake at age 40+ at the clinic in Yerushalmi et al. (2016) was high at 87.3% and the median and mean ages at time of RRSO were 46.5 and 48 years, ranging from 33-68. This high rate of RRSO uptake before natural menopause in clinic attendees was higher than in most other reported registries and in the literature (Yerushalmi et al., 2016). The median age at the time of RRSO in the multidisciplinary clinic was also lower

than the median age at time of RRSO of 49.6±9.7 in NL BRCA PV carriers (Roebothan et al., n.d.) Further studies are needed to compare outcomes in dedicated follow-up clinics with family physician-based paradigms, still, Yerushalmi et al. (2016) provided a glimpse into how a successful specialized follow-up clinic can be implemented to meet the needs of the HBOC PV carrier population.

Quality of the Evidence

The quality of the evidence examined in question two was varied. Some studies featured a strong RCT design and statistically significance evidence was produced that these interventions had a positive effect on participants' stress and quality of life (White et al., 2014; van Driel et al., 2019). The inclusion of these studies made a strong case that these types of peer and MBSR interventions could be an effective part of follow-up programs for this population. Moderate quality evidence of the effect of psychoeducational groups on outcomes in HBOC populations is indicative that a similar pilot for an annual HBOC psychoeducational group session may be part of an effective follow-up program model. Furthermore, while I found limited evidence about the effect of multidisciplinary specialized follow-up clinics on HBOC outcomes, a program model similar to the one used by Yerushalmi et al. (2016) could be piloted in another setting to compare pre and post effects of the program implementation. It may be useful to complete a Controlled/Uncontrolled-Before-After study about the effects of a pilot program dedicated to HBOC on screening adherence, disease outcomes, as well as psychosocial adjustment. For the latter metric, a psychometric scale was developed by Watkins et al. (2013) that is designed to measure psychosocial adjustment challenges in cancer predisposition syndrome populations (the PAHDS scale). Further research is warranted to determine if the implementation of a multimodal

HBOC previvor follow-up program would have significant positive effects on PAHDS scores, other quality of life measures, and unmet information needs in HBOC PV carrier populations.

Interpretation

It is clear from the literature summarized in Appendix A that there are gaps in the current paradigm of care for HBOC PV carriers, leaving this high-risk population at an equally high-risk of falling between the cracks of the healthcare system. HBOC PV carriers interviewed in the studies reported that the current primary care provider-dependent model of HBOC follow-up did not meet their unique needs as high-risk individuals. Participants indicated that they had unmet information needs about several ongoing aspects of PV carriership including family planning, family communication, symptoms of surgical menopause, and the need for an articulated plan of risk-management. Participants verbalized that the current health system often left them feeling like a statistic rather than a human being and that they felt as though little consideration was given to the highly personal implications that their carriership status had in their lives. Some study participants felt a sense of isolation and a sense of feeling different from the rest of the population because of their PV carrier status. Many participants desired a formalized channel where they could seek peer support from women with HBOC syndrome who previously underwent testing and could relate to what they were going through. Both health care providers and HBOC PV carriers reported that the current approach is uncoordinated and ineffective.

These findings are unacceptable for several reasons. Firstly, the current approach is contradictory to the positions in several key Canadian Health policies. ‘Health’ is determined by the interplay of several complex factors such as income and social status, social support networks, education and literacy, physical environments, gender, and biology and genetic endowment, among others (Federal Provincial and Territorial Advisory Committee on

Population Health, 1999). In an effective population health policy, there is an active effort to reduce the health disparities experienced by certain populations due to the interplay of health determinants in their life circumstances (Health Canada, 2001). Due to their genetic predisposition, female BRCA PV carriers are almost guaranteed to have breast cancer in their lifetime, while the rest of the general population has a 1 in 8 lifetime risk of breast cancer (Canadian Cancer Statistics Advisory Committee, 2019). Yet, the current opportunistic model of genetic testing and lack of coordinated follow-up in many Canadian jurisdictions does little to mitigate the health inequities experienced by individuals with inherited cancer predisposition syndromes. A systemic, upstream approach to health promotion and disease prevention for individuals with hereditary cancer predisposition syndromes, frankly, is long overdue. Health is "seen as a resource for everyday life, not the objective of living" (World Health Organization, 1986, p.1). Yet, there are limited programs in Canada that offer any psychosocial and navigational resources to women in their everyday life as HBOC previvors. As stated by Thiruchlevam et al.(2018), "genetic testing can be life-changing and indeed life-saving, but it is crucial that it comes with all of the facts and appropriate professional support to enable individuals to live and plan for a healthy life" (p. 2091).

Not only is the current paradigm of care for high-risk individuals in Canada unacceptable, but it is also "the most expensive and least effective" (Roebothan et al., n.d, p.18). The costs of genetic testing have significantly decreased over time and there is irrefutable evidence that HBOC risk-reduction modalities are effective. Still, in an 18-month period, only 41.6% of eligible BRCA PV carriers in NL were compliant with all the annual breast screening modalities recommended for high risk individuals ($p < 0.001$) (Roebothan et al, n.d.). Simply put, the current paradigm does not make sense given the plethora of evidence in favor of a coordinated, outcome

driven hereditary cancer care policy (Roebothan et al., n.d.). Another area for future research will be a cost analysis of the current system and burden of HBOC-associated disease compared with the costs of genetic testing and risk-reduction modalities. Recently, Manchanda et al. (2018) determined that general population-based testing for PVs in the BRCA1/BRCA2/RAD51C/RAD51D/BRIP1/PALB2 genes is more cost-effective than the current opportunistic model of genetic testing based on clinical criteria and family history.

The findings in part two and Appendix B of this literature review provide some insight into what an effective HBOC policy and systemic follow-up program may look like in NL. In an examination of outcomes in a dedicated BRCA follow-up clinic in Israel, only 5.8% of BRCA PV carriers who attended the clinic developed breast cancer in the 5-year follow-up after genetic testing (Yerushalmi et al., 2016). This is compared to the 10.2% of breast cancer cases detected in NL BRCA PV carriers in the period following genetic testing (Roebothan et al., n.d.). Moreover, in Yerushalmi et al. (2016), 94.1% of the cases of breast cancer detected post-genetic testing were stage I disease versus only 40% of cases of breast cancer detected in NL post-genetic testing that were stage I disease (Roebothan et al., n.d.). A pilot study of a systemic navigation clinic in NL, modelled similarly to the clinic in Yerushulmi et al. (2016), may be useful in determining the appropriateness of this model as part of an effective Canadian public health strategy.

An effective HBOC care program will involve multiple strategies to meet the needs of the HBOC previvor population. There is moderate quality evidence that ongoing or annual psychoeducational group workshops, retreats, and interventions are an effective way to meet information needs of this population and to reduce their feelings of burden experienced as PV carriers (Corines et al., 2017; Kwiatkowski et al., 2019; Landsbergen et al., 2009, Listøl et al.,

2017; McKinnon et al., 2007). Ongoing psychoeducational group interventions may be highly acceptable for some HBOC PV carriers as a component of a systemic follow-up program. Targeted, effective interventions such as mindfulness-based stress reduction and peer-telephone support may also be acceptable interventions to include as part of a follow-up model for HBOC PV carriers who are experiencing distressing symptoms associated with RRSO and for those who seek formalized support from other HBOC PV carriers (van Driel et al., 2019; White et al., 2014). Technology will likely play an effective role in disseminating key information and screening reminders to participants in a HBOC follow-up program. The pending results of the pilot study by Cohen et al. (2018) may have great implications for how iPhone technology is used in a variety of hereditary cancer risk-management settings to improve outcomes.

It should also be noted that in two studies (Cherry et al., 2013; Komatsu & Yagasaki, 2014), the authors purported that the nursing profession has a potential role to play in improving the delivery of services to HBOC PV carriers. Nurses are ideally situated to provide of genetic health care, with the discipline's "focus on health promotion, caring, and the understanding of individuals, including their relationships with families, the community, and society" (Calzone et al., 2013, p.1). As one of the largest groups of health care providers, nurses frequently engage in multidisciplinary collaboration, and are situated to provide a psychosocial and ethical perspective to genetic care (Bortoff et al., 2004). The expansion of genetic nursing practice in Canada is trailing significantly behind that of other developed countries. Despite several key recommendations identified in a 2004 Canadian forum on genetic nursing practice, progress on genetic nursing practice in Canada, for the most part, has been at a stand-still for the last 16 years. This is a significant missed opportunity as it has been demonstrated in the US and the UK how "nurses' awareness of genetics enabled them to skilfully address patient and family concerns

related to specific hereditary conditions, and lead to the development of specialist nursing roles in genetics" (Bortoff et al., 2004, p.25). It is overdue to launch genetic nursing practice in Canada and to implement strategies shown to be acceptable and feasible in other countries. There are several barriers to mobilizing genetic nursing in Canada; Canadian professional nursing organizations have not outlined genetic competencies for nursing practice, there is lack of nursing faculty in Canada with adequate experience in genetics, and due to the lack of genetic content on RN licensure exams, there is little onus on nursing schools to include genetic content in their curriculum (Bortoff et al, 2004). While there are barriers to genetic nursing in Canada, they are not insurmountable. The influence of nursing's "holitstic, humanistic complement to the biomedical approach" (Bortoff et al., 2004, p. 24) should be incorporated in the development of a tailored, navigation program for HBOC PV carriers.

Conclusion

With the rapid advancement of genetic technology, the clinical identification and management of HBOC is an emanating priority health issue, with several proven treatment options available to affected PV carriers to reduce their risk of developing HBOC. In the current fragmented paradigm, individuals not only face barriers to obtaining genetic testing but face considerable challenges in navigating the implications of PV carriership. As a result, individuals with HBOC PVs are under identified, under-screened, and under-supported. Ultimately, they are getting ill, and in many cases dying from a disease that we know how to prevent in this population. From this literature review, I conclude that the current HBOC paradigm in NL has a narrow focus on treating a disease rather than treating a person and enabling them to live a healthy life. However, extant models and interventions examined in the literature provide a primer of what a basket of primary care services for HBOC PV carriers in NL could look like.

This may include but is not limited to a dedicated follow-up clinic, navigational assistance with screening and surgical decisions, individual and health care provider education sessions, assistance with the family implications of PV carriership, and targeted supportive interventions. to manage specific facets of carriership, such as surgical menopause. Coupled with stakeholder consultation, and an environmental scan, the findings of this literature review can be used in the development of an effective healthy public policy for HBOC PV carriers in the province of NL.

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Appendix A

Study/Design	Methods	Key Results	Comments
Authors: Caita-Zuffery et al. (2015) Design: Qualitative, Grounded theory Purpose: Explore challenges unaffected HBOC PV carriers face when managing risk, psychosocial processes behind the challenges	N: (n=32) French-Italian speaking females, unaffected BRCA 1/2 carriers, age 26-30 yrs, diagnosed atleast 3 yrs prior (58% agreed to participate) Country/setting: Switzerland, participants recruited from genetic counselling centers, Data collection: Interviewed 1-2 times, each interview 3 hrs on average, Data Analysis: Use of constant comparative method, inductive coding (grounded theory methodology), ATLAS qualitative software program used to confirm robustness, quotations	Key Participant Themes: 1)Sense of moral duty to both off-spring and ancestors to make use of risk-reducing modalities at their disposal 2)Risk-reduction decision brought back memories of family members affected by hereditary cancer 3) Opposing views of medical management among providers, contradictory attitudes reinforced women's feelings of disorientation 4) Women sometimes felt "pushed" (p.730) by providers to adhere to risk management guidelines	JBI Checklist (2017) Overall Appraisal (Include) Strengths: Good adherence to grounded theory methodology Data collection/analysis continued until data saturation Narrative verified with participants; supplemental medical information confirmed Limitations: May be selection bias of women who were willing to participate Recommendations: Multidisciplinary hereditary cancer clinics would be appropriate for follow-up of BRCA1/2 PV carriers

Study/Design	Methods	Key Results	Comments
Authors: Cherry et al. (2013) Design: Qualitative Purpose: To explore BRCA previvor's perceptions of their ovarian cancer risk; understanding of their ovarian cancer risk-reduction options, and decision-making needs related to their risk reduction options	N: (n=12) females w/ confirmed BRCA 1/2 PV (n=3 w/ a hx of breast cancer) who were ≥21 years old, no history of ovarian cancer, atleast 1 intact ovary Country/setting: USA, participants recruited from high-risk cancer clinics Data collection: Standardized set of open-ended questions (interview approx. 1 hour) Interviews audio-recorded and transcribed Data Analysis: Grounded inductive method of data analysis Coding in 2 steps, 1st step: 2 co-authors coded separately 2nd step: whole team coded together	Key Participant Themes: 1) Carrier decision making needs & resources (reported challenges getting information from primary care provider, especially about sexual adverse events considered taboo, info from providers was variable and not always reliable) 2) Valued the opportunity to liaise with other women who have faced BRCA risk decisions (wanted to hear both the positives and negatives to make an informed decision) 3) Confusions about risk-assessment (erroneously believed RRSO increases breast cancer risk, knew very little details about procedures) 4) Timing of surgery important vis-à-vis family planning 5) Experiences of family who were BRCA+ influenced decision	JBI Checklist (2017) Overall Appraisal (Include) Strengths Rigorous adherence to qualitative methodology Script developed based on counselling with 500+ women who received BRCA 1/2 test results Limitations: Purposive sampling, not generalizable Sample lacked socioeconomic diversity Implications Suggested nurse navigator model so nurses can provide BRCA previvors with support/access to other resources, referrals, appointment scheduling Suggested BRCA support groups led by medical professionals for women considering RRSO

Study/Design	Methods	Key Results	Comments
Authors: Dean et al. (2017) Design: Qualitative, grounded theory Purpose: To investigate the experiences of BRCA previvors' information needs following a positive BRCA test result	N: (n=25) females w/ BRCA 1/2 PV, no person history of cancer, ≥18 years old, English Speaking Country/setting: USA, participants recruited from social media Data collection: Semi-structured interviews (22-90 mins) conducted via telephone Interviews audio-recorded and transcribed by professional transcription company Data Analysis: Constant comparative method of analysis 3 stages of coding Open coding, axial coding, selective coding (consistent with grounded theory)	Key Participant Themes: Post-testing information needs: 1) Previvors desired information including a list of relevant care providers (i.e. oncologists, gynecologists) 2) Previvors desired an action plan for what the next steps were 3) Previvors reported needing time to come to terms with decisions 4) Availability of support groups such as FORCE and Bright Pink were beneficial to previvors Pre-management needs 1) Previvors placed emphasis on the input of how other previvors made their decisions 2) Some risk management decisions could not begin until health insurance policies had changed/been updated	JBI Checklist Overall Appraisal (Include) Strengths Rigorous adherence to grounded theory methodology Use of Theory of Motivated Information Management as theoretical framework Krippendorff's alpha values calculated at 2 points in coding for intercoder reliability T1, $\alpha = 0.926$; T2, $\alpha = 0.843$) both within limits of reliability ($\alpha = 0.7$) Limitations: Small sample size Homogenous, affluent white women Only small number of young age women Retrospective, relied on participant recall Practice Implications The mere provision of medical information to BRCA previvors is inefficient Need to explore further educational interventions for this population

Study/Design	Methods	Key Results	Comments
Authors: Etchegary et al. (2015) Design: Qualitative, description Purpose: To explore the experiences of women affected by LS related to their RRSO risk management decisions	N: (n=10 females w/ confirmed LS predisposition PV who was considering or underwent RRSO) 74% contacted agreed to participate Country/setting: Canada, NL provincial medical genetics registry Data collection: telephone interviews (30 min-1hr) use of question guide, audio recorded, transcribed verbatim Data Analysis: Qualitative description, Naturalistic inquiry (no *a priori* assumption) Data presented in language of participants	Key Participant Themes: Factors associated with prophylactic RRSO decisions 1) Motivation to reduce personal risk of cancer, affected by seeing the cancer experiences of others 2)Needing time to process information before surgery 3)Recommendation of physician was influential to surgery decision 3) Demographics/ child-bearing considerations were influential to decision to undergo RRSO 4) Information needs prior to surgery, some unaware full extent of surgical menopause/sexual adjustment	JBI Checklist (2017) Overall Appraisal (Include) Strengths Voices of participants accurately represented Congruence between stated methodology and representation and interpretation of data Limitations: Small sample size Potential recall bias Lack of philosophical/theoretical assumption Cannot be generalized to women who declined RRSO Recommendations: Continued efforts to educate primary care providers and oncologists about screening recommendations for LS

Study/Design	Methods	Key Results	Comments
Authors: Komatsu & Yagasaki (2014) Design: Qualitative, Grounded theory Purpose: To explore implementation & challenges of HBOC risk-assessment from the perspective of HCPs providing breast care; to explore provider readiness for personalized cancer risk management	N: (n=17) health care providers (n=7 breast specialists, n=5 staff physicians, n=4 nurses, n= 1 genetic counsellor) Country/setting: Japan, 2 institutions providing Rx to HBOC patients in Japan Data collection: semi-structured focus groups lead by nurse researcher (60-70 min), audiotaped & transcribed verbatim Data Analysis: Use of constant comparative method, open coding Interviews deconstructed sentence by sentence	Key Participant Themes: 1)Providers were too busy treating disease; little time left for risk management (lower priority with limited resources) 2)Recommended the development of outpatient follow-up clinics separate from routine breast care 3) Individuals at risk likely to be missed by the fragmented communication among multiple care providers 4) Oncology nurses have a role to play in bridging communication among multidisciplinary HBOC team	JBI Checklist (2017) Overall Appraisal (Include) Strengths: Good adherence to grounded theory methodology Adherence to symbolic interactionism as theoretical basis Limitations: With nature of focus group, results possibly influenced by group dynamics Recommendations: Specialized hereditary cancer clinics should be established for multidisciplinary collaboration/partnership

Study/Design	Methods	Key Results	Comments
Authors: Leonarcyzk & Mawn (2015) Design: Qualitative, Phenomenological Purpose: Explore the experiences of cancer risk management decision-making in unaffected BRCA1/2 Carriers	N: (n=15) females, age ≥ 18, self-reported, asymptomatic BRCA 1/2 carriers Country/setting: USA Data collection: purposive sampling, interviews 25 min to 1 hr, telephone or in person, semi-structured interview guide Data Analysis: Use of appropriate phenomenological techniques; Epoche & Bracketing NVivo9 qualitative software used to organize/identify data Interviews audiotaped	Key Participant Themes: 1) Lack of knowledge of HCPs, scarcity of providers, especially in rural areas, 2) Lack of guidelines, lack of decisional tools, plans of care, educational resources 2) Carriers valued a holistic approach, described "not being seen as a whole person" (p.77) 4) Need for emotional support, online sources such as FORCE identified as valuable (especially in rural regions)	JBI Checklist (2017) Overall Appraisal (Include) Strengths: Good adherence to qualitative phenomenology Efforts to minimize researcher bias Voluntary, informed consent Limitations: Participants were self-reported BRCA, carrier status not confirmed Participants self-selected Sample lacked socioeconomic diversity

Study/Design	Methods	Key Results	Comments
Authors: Pezaro et al. (2012) Design: Descriptive, cross-sectional study Purpose: To provide pilot data on the long-term consequences & guide optimal management requirements following pre-menopausal RRSO	N: (n=150 women)72% contacted completed questionnaire (n=143 women w/ BRCA 1/2 mutation, no prior history of OC, no intact ovaries) (n=14 still had ovaries insitu) Country/setting: Australia, participants from single site familial cancer center registry in East Melbourne Data collection:. Eligible women sent study pack which included invitation, consent letter and questionnaire containing data about RRSO, menopausal status at time of RRSO, risk factors for osteoporosis, participation in post-operative follow-up programs Outcomes: Prevalence of current menopausal symptoms rated using menopausal rating scale (MRS) (Specific menopause symptoms rated on a scale of 0-4; score 3-4 'severe' Demographic variables associated with MRS score examined	Variables associated with uptake of RRSO Current age > 50: (OR 28.0; p <0.0001) Personal history of breast cancer: (OR 8.0; p < 0.0001) Variables associated with persistent severe menopausal symptoms Pre-menopausal at time of RRSO: (OR 1.98; 0.9-3.8; p =0.1) Women who reported "persistent severe" menopausal symptoms linear correlation with age at RRSO (p=0.002) 73% (n=104) women reported no on-going follow up with Gyne surgeon after immediate post-surgical review	Strength of Design: Weak Quality: Moderate Adequate response rate (72%) Only 4% of missing data in the questionnaires Some variables associated with persistent severe menopausal symptoms did not receive statistical significance Findings: Women not receiving adequate follow-up support for the symptoms associated with RRSO Opportunity for genetics team to initiate long term follow-up of BRCA previvors pre/post RRSO Need for optimal clinical guidelines for the care of women post RRSO

Study/Design	Methods	Key Results	Comments
Authors: Rauscher & Dean (2017) Design: Qualitative Purpose: To identify BRCA previvor's strategies for communicating about family planning (FP) post testing positive for the BRCA PV	N: (n=20) females w/ confirmed BRCA 1/2 PV who were ≥18 years old, with a committed romantic partner, and had at least one conversation about FP with that partner Country/setting: USA, participant recruited from annual FORCE hereditary cancer conference Data collection: Telephone semi-structured interviews (24-68 min) audio recorded, transcribed by professional transcription company Data Analysis: Constant comparative method Use of open-coding, axial coding Research memos taken as an audit trail	Key Participant Themes: Three themes identified 1) Importance placed on a 2-way dialogue about both BRCA previvor and their spouse's feelings about FP (spouse often overlooked) 2) Importance given to information seeking from multiple sources, supportive resources, weighing options 3) Emphasis placed on using emotions in FP decision making "It's not a statistic it's what they feel in their heart is the right thing to do" (p. 491)	JBI Checklist (2017) Overall Appraisal (Include) Strengths Evidence that measures were taken to ensure credibility, transferability, and consistency of findings Final sample was based on achievement of theoretical saturation Rigorous adherence to qualitative methodology Limitations: Claims not generalizable due to nature of data Inherent bias of self-selection Sample demographically homogenous; well-education, relatively affluent women Recommendations Care providers should provide individuals at increased risk of HBOC a list of questions to start conversations with their partner about FP vis a vis being a HBOC previvor Instead of individualized approach to genetic counselling, involve couples and families

Study/Design	Methods	Key Results	Comments
Authors: Phelps & Hughes (2010) Design: Qualitative Purpose: To explore the acceptability of, and preferences for, a support facility amongst male and females identified as having a BRCA1/2 PV	N: (n=17) women Inclusion criteria: 18 years old+, living in the Cardiff and Vale NHS Trust region Country/setting: Wales, UK, recruited from South Wales genetic registry Data collection: Three focus groups, semi-structured interview guide Interviews tape recorded and transcribed verbatim Data Analysis: Coding completed by 2-member team Ambiguity in coding resolved in discussion with team members	Key Participant Themes 1)Reactions to Learning Carrier status Feeling different from the rest of the population Seeking someone to relate to 2) Psychological support needs Feared stigma of seeking informative support, were more likely to attend event framed as 'informative support' Feelings of guilt in families, concern about disclosing PV carrier status The need for anonymity in groups and interventions 3) Information Support Needs The need for an updated trustworthy source of medical and research updates pertinent to BRCA The need to confirm accuracy of information Emphasis on an expert/informal support combined intervention modality	JBI Checklist (2017) Overall Appraisal (Include) Strengths Voices of participants accurately represented Summaries provided to participants over course of discussion to confirm accuracy of data Limitations Participants may be reluctant to attend groups to discuss sensitive issues Lack of representation from male BRCA PV carriers despite being invited to participate

Study/Design	Methods	Key Results	Comments
Authors: Watkins et al. (2011) Design: Qualitative, grounded theory Purpose: To explore how confirmed Lynch Syndrome (LS) carriers experience disease management, explore the quality of interactions with overall health care system	N: (n=23) individuals w/ confirmed MSH2 mutation (n=14 female, n = 9 male) Country/setting: Canada, NL provincial medical genetics registry Data collection: 60-90 min interview in participants homes or conference rooms, use of open-ended questions Data Analysis: Interviews transcribed verbatim Inductive approach, emphasis placed on describing social/psychological processes	Key Participant Themes: 3 Main Barriers Identified 1)Person-centered barriers (psychological aspect of going through continuous testing, logistics of testing i.e. commute/use of sick leave) 2)Provider-centered barriers (HCPs discounted young age of PV carrier, lack of knowledge/skill of HCPS surrounding testing recommendations) 3) Health care system barriers (lack of continuity of care, lack of collaboration between primary/specialty care sectors, inconsistencies in medical opinions among specialists/primary care providers)	JBI Checklist (2017) Overall Appraisal (Include) Strengths: Information confirmed with participants Transcripts independently perused by 3-member team Independent consultant reviewed emergent data and theory to enhance credibility & accuracy Limitations: Small sample size Inherent bias of participant recall Recommendations: Overhaul of current uncoordinated physician dependent of screening of individuals with LS in Canada Need for more genetic counsellors to improve timely access to genetic services Existing counselling/disease management inadequate to meet demands of LS

Appendix B

Study/Design	Methods	Key Results	Comments
Authors: Cohen et al. (2018) Design: Case Series Purpose: To evaluate an iPhone application for BRCA screening adherence, examine the usefulness from a user perspective	N: (n=86) Inclusion criteria: women, ≥ 18 yrs old, + for a BRCA PV, with at least 1 ovary or breast, must own an iPhone Country/setting: USA, recruitment from online support groups and genetic counsellors Methods: Baseline data collected from (n=86) participants, at the end of the survey, (n=69) participants given a code to download an iPhone application to assist BRCA+ PV carriers to manage surveillance Data collection: Self-administered questionnaires w/ 50 questions using psychometric scales collected at baseline prior to intervention (T1) (18-month follow-up data pending) Analytic data on iPhone app download over 21-month period Outcomes: (calculated using descriptive statistics) *ADQ* (Adherence Determinants Questionnaire) (0-100 % percentage possible) *Comfort with iPhone technology*: modified Comfort with Technology scale (scores ranging 1-4; 1 being mostly uncomfortable, 4 being mostly comfortable) *Perceived usefulness of iPhones:* The Technology Acceptance Model (scores ranging 1-5; 1 being strongly disagree, 5 being strongly agree)	**ADQ scores** Intention to engage in surveillance plan: 94.3% ±8.4 Perceived support for surveillance: 72.6± 15.9 **Technology Acceptance Model scores** 3.4±1.1 to 4.1±0.7 (majority agreed or strongly agreed apps had a positive impact) **Comfort with Technology Scores** 3.5±0.92 to 3.8±0.82 (majority of respondents reported comfort with iPhone tasks) 50% (n=37) of respondents reported difficulty remembering when to schedule next screening appointment 20% (n=15) reported they rely on their physician to schedule **Download data** Downloaded app codes: 68/69 # Times app accessed: range 2–57; mean=6.28	Strength of Design: **Weak** Quality: **Low** Issues: • Follow-up data not yet published • Convenience sample recruiting from support groups and genetic counsellors • Given recruitment sources, respondents are more likely to be aware of screening recommendations and assume an active role in their care

Study/Design	Methods	Key Results	Comments
Authors: Corines et al. (2017) Design: Cross-sectional Purpose: To report on participant satisfaction and utility of continuation educational interventions for Lynch Syndrome (LS)	N: (n=53-75) participants annually with confirmed LS PV, or family member of individual with a confirmed LS PV Country/setting: USA, single site familial cancer clinic recruitment Establishment of annual Lynch Syndrome Educational Workshop (LSEW), which then lead to the establishment of quarterly Lynch Syndrome Patient Advocacy Network (LSPAN) quarterly participants ranged from (n=2-13) Data collection: Participant survey data from both LSEW and LSPAN collected and summarized using descriptive statistics	**LSEW Participants Years 2-5** ≥96% reported overall satisfaction w/ LSEW >82% verbalized interest in online support ≥87% satisfied with technical aspects of LSEW **LSPAN participants over 11 meetings** 100% reported meetings helpful (58% reported very helpful, 36% helpful, 6% somewhat helpful) 91% found the information clear 67% stated the presence of a genetic counsellor at LSPAN was helpful	Strength of Design: **Weak** Quality: **Low** Issues: • Measurement instruments not based on standardized, reliable outcome measurements • Potential bias as individuals attending LSEW and LSPAN are the LS PV carriers with greatest support needs • No data collected on effect of LSEW/LSPAN on disease knowledge or psychosocial well-being, or compliance with screening/risk-reducing surgery • No collection of demographic data

Study/Design	Methods	Key Results	Comments
Authors: Kwiatkowski et al. (2019) Design: Uncontrolled-Before- After Purpose: To test the cognitive/psychosocial impact of a psychoeducational intervention on young women at increased risk of HBOC	N: (n=7) Inclusion criteria: women aged 18-30, childless, w/ a confirmed HBOC PV, no personal history of breast/ovarian cancer Country/setting: France, single-site recruitment, 2-day psychoeducational workshop at a spa hotel Data collection: Self-questionnaires collected at baseline (T1), end of workshop (T2), after 6 months (T3), and after 12 months (T4) Outcomes: Measure: (Psychometric Instrument) Hopefulness: Herth Hope Inventory (HHI) Self-Esteem: (SES) Anxiety: (STAI-B) Coping: (WCC) Perceived control (IPC) Anxiety State (STAI-B) Quality of Life: (WHOQOL)	**Global HHS scores at T4** 24% increase over 12 months: (p=0.07)* **Increase in psychometric measures of Hope, Self-esteem, QOL over 1 year** (p= 0.00032)* **Coping by focus on problem** Increased at T2 (p= 0.011)* then returned to baseline at T3 **Coping by focus on emotion** Decreased steadily at T2, T3 (p=0.021)*	Strength of Design: **Weak** Quality: **Medium** Issues: • This study was initially pitched as an RCT with the waitlist acting as a control but insufficient interest to form 2 groups • Limited sample size • Lack of socioeconomic diversity of sample

Study/Design	Methods	Key Results	Comments
Authors: Landsbergen et al. (2009) Design: Controlled Before/After Purpose: To determine the effect of an educational support group on proceeding with initial preference for surveillance or RRM	N: (n=151) Inclusion criteria: women newly diagnosed w/ a BRCA PV, no personal hx of breast cancer, no current psychiatric disorder Country/setting: The Netherlands, recruited from single site genetics department IG: (n= 79) women participated in at least 6/8 2.5 hr psychosocial and medical information sessions q 4-6 weeks UCG: (n=84) women did not participate in the psychoeducational groups Data collection: At Baseline, participants' preference for risk reduction methodology reported in both groups, Medical records checked 2 years following in two groups to follow up, predictors for risk reduction	**Women who proceeded with initial preference for RRM after 2 yrs in:** IG: 89% UCG 63% (OR 4.8, p = 0.04)* **Percent of women who preferred RRM at baseline in:** IG: 34% of individuals UCG 19% (p = 0.05*) **Predictors for proceeding with RRM after 2 yrs** Age between 30-50: (OR 9.6, p =0.03) Prior preference for mastectomy: (OR 42.3, p < 0.001)* (R2 = 0.57)	Strength of Design: **Strong** Quality: **Medium** Issues: • Self-selection bias with potential baseline differing attitudes • No evidence if participation in group allowed previvors to better able to cope with their daily lives as BRCA PV carriers, nor their satisfaction with support group interventions

Study/Design	Methods	Key Results	Comments
Authors: Listøl et al. (2017) Design: Uncontrolled Before After Purpose: To evaluate whether anxiety/depression symptoms in BRCA + PV carriers changed following a group patient education (GPE) course, to determine factors associated with anxiety/depression symptoms in BRCA PV + individuals	N: (n=130) women, ≥ 18 yrs old, + for a BRCA PV, with or without history of breast/ovarian ca, able to read Norwegian Country/setting: Norway, single site Intervention: a 1-day standardized GPE course Data collection: Self-administered questionnaires using psychometric scales collected at baseline prior to intervention (T1), and 2 weeks following course (T2) Outcomes: Hospital Anxiety and Depression Scale (HADS) (2 subscales, scores ranging 0-21 with ≥8 as cut-off for elevated symptoms of anxiety and depression) Coping style measured using Threatening Medical Situation Invention (TMSI) Self-efficacy measured by: Bergen Genetic Counseling Self-efficacy Scale (BGCSES) Analysis of correlation of participant variables with anxiety/depressive symptoms using mixed-linear model analysis	**HADS anxiety sub-score (HADS-A)** T1: 6.2 (± 4.4) T2: 5.2 (±3.95) (p= 0.003)* **Correlation with decreased HADS-A scores (anxiety)** Greater time since disclosure of the gene test result:−0.25 (95% CI−0.41, −0.09) (p=0.002)* Higher level of situation specific self-efficacy: −0.96 (95% CI−1.48, −0.45) (p < 0.001)* Women had experienced losing a first and/or second degree relative due to breast or ovarian cancer: −2.25 (95% CI−4.08, −0.43) (p=0.016)* **Correlation with decreased HADS-D scores (depression)** Women had experienced losing a first and/or second degree relative due to breast or ovarian cancer: −2.59 (95% CI−4.17, −1.01) (p=0.002)*	Strength of Design: **Weak** Quality: **Medium** Issues: • Lack of control group • Possible selection bias as participants self-enrolled following invitation • Low baseline depression scores possibly 2 to symptoms of depression such as lack of energy and initiative; may reduce the probability of signing up for a GPE course

Study/Design	Methods	Key Results	Comments
Authors: McKinnon et al. (2007) Design: Uncontrolled Before After Purpose: To evaluate the impact of a 1-day health retreat for BRCA previvors/families on health behaviors	N: (n=41) Inclusion criteria: men/women with personal hx of BRCA PV, VUS, or family member of individual w/ confirmed PV Country/setting: USA, recruitment from single site familial cancer center Participated in 1-day psychoeducational retreat w/ content specific to HBOC Data collection: Questionnaires collected at Baseline prior to intervention (T1) and 6 months following the retreat (T2) Outcomes: Use of the Impact of Events (IES) 15-item scale score to assess the stress experience related to having a PV predisposing to HBOC IE scores ranging from 0-8 subclinical; mild, 9–25; moderate, 26–43; and severe, 44+)	**IES scores** T1: 19.9±13.2 T2: 19.9±14.3 (p = 0.66) **IES Intrusive Thoughts Subscale Scores** T1:8.5 ±6.8 T2: 9.5±7.6 (p =0.66) **IES Avoidance Subscale scores** T1 :10.5± 8.7 T2 :10.4±8.7 (p=0.33)	Strength of Design: **Weak** Quality: **Low** Issues: • Lack of statistical significance reached • Lack of control group • Lack of difference in T1 and T2 IES scores possibly attributable to the high baseline IES scores in some of the participants • Using different research design would have likely yielded different results, participant testimony was overwhelmingly positive

Study/Design	Methods	Key Results	Comments
Authors: van Driel et al. (2019) Design: RCT Purpose: To assess the effects of mindfulness-based stress reduction (MBSR) on the resulting quality of life, sexual functioning, sexual distress after RRSO	N: (n=66 women who had RRSO @ ≤ 52 yrs of age, No prior hx of cancer, chemotherapy or radiation) Country/setting: the Netherlands, single site family cancer clinic Intervention Group (IG): (n=34) received 8-week MBSR training, consisting of weekly 2.5 hr sessions, a 4 hr silent retreat evening, committed to 30-45 min home exercises 6x a week Care as Usual Group (CUG): (n=32) received menopause counselling from specialist nurse during the intake visit.. (An information booklet summarizing this information was provided to participants in both groups, Both groups offered repeat appointment w/ nurse 12 weeks post-randomization) Data collection: Self-administered questionnaires using psychometric scales sent at baseline (T0), 3 months (T1), 6 months (T2), and 12 months (T3) Primary Outcome: Menopause-specific quality of life: (MENQOL) 29-item scale (scores ranging from 1, absence of symptoms, to 8, extremely bothersome) Secondary Outcomes female sexual distress scale (FSDS); female sexual function index (FSI);	**Mean MENQOL scores** **@ T1** IG: 3.5 (95% CI 3.0-3.9) CUG: 3.8 (95% CI 3.3-4.2) (p = 0.04)* **@ T3** IG: 3.6 (95% CI 3.1-4.0) CUG: 3.9 (95% 3.5-4.4) (p = 0.04)* **FSDI scores** No differences between IG and CUG groups reached statistical significance at either T1 (p=0.65), T2 (p =0.77), or T3 (p=0.26) **FSI scores** No differences between IG and CUG groups reached statistical significance at either T1 (p=0.4), T2 (p=0.92), or T3 (p=0.75)	Strength of Design: **Strong** Quality: **High** Issues: • Potential self-selection bias • Impossible to blind participants to IG or CUG group Implications Recommend that healthcare providers advocate MBSR in conjunction with Hormone Replacement Therapy (HRT) MBSR post-RRSO may be especially relevant in settings where HRT is contraindicated (i.e breast cancer survivors)

Study/Design	Methods	Key Results	Comments
Authors: White et al. (2014) Design: RCT Purpose: To assess the effectiveness of a telephone-based intervention in reducing stress among women with a BRCA 1/2 PV	N: (n=207) Inclusion criteria: women responded to baseline questionnaire, ≥18 years old, received genetic results of BRCA PV within 5 yrs, and verbalized interested in receiving peer support Country/setting: Australia, recruitment though 8 familial cancer clinics Random allocation to: Usual Care group (UCG): (n=102) Received usual follow-up care Intervention Group (IG): (n=105) received phone calls from trained peer volunteers over a 4-month period (mean 3.7 calls) Data collection: Questionnaires collected at baseline (T1), at end of intervention (T2), 2 months later (T3) Outcomes: Breast cancer related distress (IES scale) Unmet information needs (UIN 16-item scale) Cognitive appraisal about genetic testing (CAGT 10-item scale) Feelings of isolation (single item questionnaire)	Greater decrease in IES Breast Cancer distress scores in IG vs UCG at T2: -5.96(95% CI, -9.80-2.13; p=.002) T3: -3.94 (95% CI, -7.70 to -0.17; p =.04)* Greater decrease in Reduction in Unmet information needs UIN scores for IG vs UG at T2: -5.17 (95% CI, -7.96 to -2.37; p= < .00)* Not significant at T3: -.67 (95% CI -4.28 to - 0.93; p =0.21) Greater decrease in Cognitive appraisal about genetic testing CAGT stress subscale for IG vs UCG at T2 -.25 (95% CI -2.10 to -0.40 ; p =.004)* Not significant at T3: -0.54 (95% CI -1.35 to -0.27; p =0.19)	Strength of Design: Strong Quality: High Issues: • Participants could not be blinded to IG vs UCG • Drop out rate higher in IG vs UCG • 27% of all eligible women verbalized interest in peer support

Study/Design	Methods	Key Results	Comments
Authors: Yerushalmi et al. (2016) Design: Case series Purpose: To report on a specialized, multidisciplinary follow-up clinic for BRCA carriers	N: (n=318) women seen by the clinic since its inception (n=292 women opted for BRCA testing, included in final report) Country/setting: Israel, single-site familial cancer follow-up center Biannual clinic visits recommended for screening, follow-up, Additional psychosocial support beyond routine visits provided when needed Data collection: Clinic attendee risk surveillance updated in Excel file with each visit Outcomes: Length of follow-up, Disease outcomes, risk reduction surgery uptake Descriptive statistics used to report clinical outcomes	**Length of Follow-up** Women followed by the clinic 5+ years: (n=168) Median follow up: 46 months **Disease outcomes in women attending clinic during 5 yr+ F/U** Women who developed cancer: 7.2% (n=21, 17 breast ca, 1 ovarian ca, 3 additional cancers) % of breast cancer cases diagnosed w/ stage I disease: 94.1% % cancer diagnosed by: *MRI* : 70.6% *Mammography:* 17.6% *Ultrasound*: 5.9% **Uptake of Surgeries** RRSO in women 40+: 87.3% RRSO in women < 40: 0.84% RRM in women 40+: 16.2% RRM in women < 40: 8.4%	Strength of Design: **Weak** Quality: **Low** Issues: • Outcomes of clinic attendees were not compared with outcomes from a family physician-based model of referral • Possibility that the low incidence of malignancy reported to date is attributable to the relatively short median follow-up period (4 yrs) Implications High rate of RRSO before natural menopause in clinic attendees was higher than in most other reported registries and in the literature (8-75%)

Appendix II Consultation and Environmental Scan Report

Policy Proposal for a Novel Hereditary Breast and Ovarian Cancer Navigation and Follow-Up Program

Consultation and Environmental Scan Report

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Policy Proposal for a Novel Hereditary Breast and Ovarian Cancer Navigation and Follow-Up Program: Consultation Report and Environmental Scan

Project Introduction and Background

Currently, there is no programmatic and systemic follow-up in Newfoundland and Labrador (NL) for individuals with pathogenic variants (PVs) that predispose them to hereditary breast and ovarian cancer (HBOC) syndrome (Roebothan et al., n.d.). Following disclosure of genetic results, these individuals are essentially "orphaned by the healthcare system" (Mercer, 2018, para 2). As a result, they are being diagnosed, and in many cases, dying from a disease that we know how to prevent in this population. Therefore, the goal of my MN practicum project is to develop a policy proposal for a coordinated approach to HBOC PV carrier follow-up in NL. At this point in the project, I am referring to this prospective program as a novel HBOC Follow-up and Navigation Program for high-risk individuals. I characterize it as 'novel' because in my current vision, it is most accurately described as a hybrid of patient support, screening and follow-up, and patient navigation programs.

The first patient navigation program was developed in 1990 by Dr. Harold Freeman as a model to reduce the inequities experienced by socioeconomically disadvantaged individuals in their access to early detection and treatment of cancer (Freeman, 2012). Over the course of the past 30 years, the scope of patient navigation programs has expanded "to be applied across the entire health care continuum, including prevention, detection, diagnosis, treatment and survivorship to the end of life" (Freeman, 2012, p. 1614). Patient navigation programs are patient-centered programs with the objective of reducing the fragmentation and barriers that individuals face in navigating the healthcare system (Freeman, 2012). Key principles of the Freeman patient navigation model are relevant in the development of programs to reduce barriers

and improve outcomes in PV carrier high-risk populations. Furthermore, in NL, findings of the Cameron Inquiry into Breast Hormone Receptor Testing "highlighted the importance of communication and coordination of care throughout the cancer journey, recommending patient navigators to assist patients" (Department of Health and Community Services, 2010, p.13).

This MN practicum project was conceptualized primarily as a result of my attendance at two 1-day education symposiums offered at Memorial University of Newfoundland, one on the topic of ovarian cancer and the second with a focus on PVs in the BRCA genes. It was evident at these symposiums that there is a need to improve the provision of services for individuals and families in NL who carry PVs that predispose them to HBOC. I was further compelled to embark on this project based on the lack of genetic content in my own nursing education and the paucity of available genetic opportunities for nurses in Canada (Bortoff et al., 2004; Dewell et al., 2020). I maintain that the lack of nurses in genomics represents a significant missed opportunity to improve the quality of care provided to individuals with PVs, as the nursing profession has long been considered the "holistic, humanistic complement to the biomedical approach" (Bortoff et al., 2004, p. 24). As a Registered Nurse with interest in the topic and an awareness of the urgent need to reform the current HBOC follow-up process, this topic was a natural and obvious choice for my MN practicum project.

An important early step in this practicum project was to conduct an integrative literature review on the psychoeducational needs of individuals with HBOC syndrome, as well as the barriers and facilitators they encounter when seeking support and follow-up care. An additional objective of the literature review was to examine extant interventions that were implemented to meet the unique needs of PV carriers. The findings of the literature review were revealing; first, that in NL and in many Canadian and international health jurisdictions, salient barriers exist for

individuals with HBOC syndrome. They are largely under-identified, under-screened, and under-supported. The literature review findings were suggestive that a more coordinated approach to HBOC follow-up is needed. However, in the second part of the literature review, the findings were promising for the future of HBOC follow-up care. An examination of various HBOC follow-up care models and interventions provided a fair picture of what a basket of primary care services for HBOC PV carriers in NL could look like. This may include but is not limited to a dedicated follow-up clinic, navigational assistance with screening and surgical decisions, individual and health care provider education sessions, and assistance with the family implications of PV carriership. In short, the findings of the literature review were supportive of the need and feasibility for this HBOC follow-up program policy proposal.

The literature review was an important step in the direction towards the development of an effective policy proposal for HBOC PV carrier support and follow-up in NL. The next logical step in the policy proposal development was to engage in consultation with stakeholders and to conduct an environmental scan. Semi-structured interview consultations were conducted with relevant stakeholders, such as genetic counsellors and physicians in NL. This allowed the stakeholders to provide their input on the most salient needs and recommendations for a tailored HBOC PV carrier program in NL. As these stakeholders have important perspectives in providing health care to PV carriers in NL, it is important to confirm that the tenets of a HBOC PV carrier program would be acceptable to them and compatible with the NL health care system. The policy proposal would be futile without their input. To complement this information, an environmental scan was also conducted to examine relevant supportive, screening, and follow-up services offered in other jurisdictions. Together, this information provides a comprehensive

direction for the policy proposal document. This paper provides a report on the stakeholder consultations and environmental scan conducted for my MN practicum project.

Part One: Consultation Report

Objectives of the Consultations

There were four specific objectives for conducting the consultations. First was to confirm with stakeholders that a systematic approach to HBOC PV carrier follow-up is relevant and acceptable to them. Second, was to identify issues in the current process of follow-up care for individuals in NL with HBOC syndrome. A third objective was to explore how the role of the nursing profession could be optimized in the proposed policy, as well as in genetic/genomic care. And the final objective was to provide stakeholders with an opportunity to recommend priority features for a HBOC navigation program policy, from their vantage point. This allowed them to propose alternative strategies that I may not have considered.

Methods

Individuals identified as potential stakeholders for this project were emailed letters explaining the nature and purpose of the consultations. This letter is included in Appendix A of this consultation report. It was made clear to the participants that their participation was voluntary and that they could refuse to participate or omit any question without any coercion or repercussions. In the early interviews, some consultees recommended other individuals who could also provide valuable input for this project and thus, snowball sampling was used to recruit these additional individuals. Participants who were invited to participate in this consultation included: (n=2) genetic counsellors, (n=3) oncologist specialists who provide risk-reduction care to high-risk individuals, (n=2) primary care physicians, (n=2) individuals who were involved in

research pertinent to these high-risk populations in NL, and an (n=1) individual involved in the development of a cancer prevention registry in NL. Ultimately, a total of (n=8) key informant interviews were conducted. As the participants had different vantage points in the NL healthcare system, six different question guides were developed so the questions were most applicable to the participant(s). These question guides are included in Appendices C-H of this consultation report.

Each participant was given the semi-structured question guide in advance of the interview. Given the current COVID-19 pandemic, participants were given the option to either engage in a telephone/online meeting room interview, or to respond directly to the interview questions in writing via email. For the phone/online meeting room interviews, hand-written notes were taken of participant answers. These answers were then typed in a Word document and passworded with access available only to myself and my practicum supervisor. All raw data from the key informant interviews were stored in passworded Word documents on my computer and accessible only to me and to my practicum supervisor as needed. Content analysis was used to analyze the data generated from the interviews. Content analysis is described as "the process of organizing and integrating material from documents, often narrative information from a qualitative study, according to key concepts and themes" (Polit & Beck, 2017, p. 723). The emergent themes from the raw data are presented in this consultation report.

Ethical Considerations

As stated, to safeguard the participant information, the raw data were stored in secured Word documents. Moreover, the e-mail correspondence between myself and the key informants was on a secured university e-mail server. Again, participants were informed that their participation was voluntary and that their names and any identifiable information would not be

published in the consultation report; findings of these consultations would only be reported as general themes of the common barriers and goals for hereditary cancer care in NL. At the end of the phone and online interview meetings, the notes were reviewed with participants to confirm the accuracy and acceptability of these answers. They were also notified that this data would be destroyed upon completion of my degree in December 2020. Prior to engaging in the consultations, it was determined that Health Research Ethics Approval (HREA) was not required because the nature of the data to be used in the consultations fell under one of items 1, 3, or 5 of the HREA authority screening tool. The completed screening tool is included in Appendix B.

Results

Every consultee who I spoke with to date agreed that there was value in implementing a novel dedicated follow-up and navigation program for PV carriers. While the consultees were from a wide variety of health care disciplines and specialty areas, six themes emerged in several of the interviews. These themes included 1) the need for a centralized, coordinated registry, 2) 'one-stop shopping', and 3) the need to address the psychosocial dimensions of PV carriership. Other common themes were 4) considerations for primary care providers, 5) the role of the nursing profession, and 6) funding considerations.

A Centralized, Coordinated Registry

Some of the consultees expressed concern over the lack of central, trusted authority in NL that assumes responsibility for coordinating long-term PV carrier follow-up and risk management. We know that this type of approach is theoretically possible because it is currently in use in provincial cancer screening registry programs, such as the provincial cervical screening initiatives program. In one interview, a participant noted that the use of health registries ensures that affected individuals are not 'falling through the cracks' in the healthcare system. Some

consultees provided examples of these 'cracks' in the primary care system in NL, such as turnover rates of family physicians, wait times to see a primary care provider, and in some cases, lack of primary care provider knowledge and ability to perform the role of managing high-risk individuals. One consultee noted that there is ample research evidence from around the world that confirms the value of a centralized, coordinated follow-up registry for individuals at high risk of inherited cancers. She expressed that a registry of high risk-individuals should ideally facilitate appointment/screening bookings and reminders. Another individual involved in a screening registry program expressed that having a central authority and database for testing follow-up ensures quality control and accountability in the health care system. Of note, a dedicated provincial high-risk breast screening program has also been implemented in Ontario and will be discussed later in the environmental scan report.

'One Stop-Shopping'

Some of the consultees had been involved in research with individuals living with inherited cancer syndromes in NL. A common theme reported in their work with PV carriers was that they wanted 'one-stop shopping'; in other words, carriers wanted a central coordinated clinic that addresses and schedules all their risk-reduction follow-up. PV carriers reported very practical issues with adherence to recommended screening, such as having to travel long distances and take multiple days off work to attend to the various screening and medical appointments. One informant noted that in her work with mutation carriers (and their family members for that matter), she found that 'life gets in the way' of risk management. It was easy for asymptomatic PV carriers to forget or disregard the multitude of screening appointments in the context of hectic everyday life. For some, all these appointments became so overwhelming and mentally taxing that they had to stop for a while. This was particularly common for

individuals with Lynch Syndrome where there is an increased risk of inherited cancer in multiple organs and thus multiple screening modalities are recommended. To this effect, a follow-up navigation program could work with individuals and families to arrange the various screening and follow-up appointments in a coordinated, personalized manner. In contrast with the current fragmented approach where the coordination of recommended screening and appointments is the sole responsibility of the individual and/or their primary care provider. Moreover, some PV carriers cited inconsistencies in the risk management information they were provided by their primary care providers and by various specialists. By having a centralized provincial authority with consistent information, this would reduce the confusion and disillusionment that many PV carriers feel with the current system of risk-management. Several of the consultees noted that a coordinated program for screening appointments should operate on the assumption that 'the individual is directly informed first' of any appointments and results, in addition to their primary care provider. It was also noted by an informant that any such program should be developed in partnership with PV carriers and primary care providers so that the program is informed by the people who will use and ultimately stand to benefit from it.

The Imperative of Psychosocial Considerations

Some informants noted that while a PV carrier registry and the coordination of follow-up testing is altogether necessary, a HBOC follow-up navigation program should not have a narrow focus on risk-management. One informant stated that 'there is an ethical responsibility to address carriers' emotional and mental health needs.' She noted that a novel PV carrier follow-up program ideally should offer PV carriers assistance with communicating genetic results with their family, as well as offer them with counselling services, and the opportunity to network with other carriers and high-risk individuals. Furthermore, it was recommended that a clinic/follow-up

program be delivered by a multidisciplinary team, such as social workers, specialists, psychologists, and nurses, to adequately address the multiple dimensions and considerations of PV carriership. Another informant noted that in her work with PV carriers, higher levels of 'family support and cohesion' were associated with improved psychosocial adjustment to their PV carrier status and improved adherence to the recommended screening and risk-reduction modalities. An effective PV carrier follow-up program would be one where the family system is recognized as a facilitator of psychosocial adjustment and adherence to recommended risk-reduction modalities. Therefore, the participation and involvement of family members in a follow-up program should be welcomed and encouraged. One consultee noted that unaffected family members of PV carriers also experience distress associated with having multiple affected relatives with an inherited cancer predisposition syndrome. Yet, in the current system, their concerns and perspectives are largely overlooked. This total oversight of family centred care should be addressed and rectified in a HBOC follow-up and navigation program.

A former genetic counsellor expressed that while individuals (and their primary care providers) are provided detailed, printed material on the implications of their carrier status at the time of genetic result disclosure, it was evident that individuals are not always psychologically prepared to 'readily absorb' this information at the time of disclosure. Thus, there is value in long term follow-up, to determine how individuals and families are coping with carriership and to provide them with ongoing information, referrals, and support as their needs may evolve. A coordinated program should also be respectful of individuals' right to an informed choice of whether they choose to proceed with genetic testing, surveillance, or preventative surgery. One consultee noted that in some instances after multiple unsuccessful attempts to reach out to a PV carrier, health care professionals must 'take the hint' that an individual is not interested in further

follow-up testing and respect this as their right.

Considerations for Primary Care Providers

Literature review findings were suggestive that there are deficiencies in the genetic knowledge, skills, and ability of primary care providers to act as coordinator of genetic follow-up (Cherry et al., 2013; Leonarcyzk & Mawn, 2015; Watkins et al., 2011); this theme was further probed in the consultations. One consultee remarked that she did not think this was a simple education problem and noted there are 'reams of information out there for GPs' (and primary care NPs). When asked, a primary care provider explained that he completes an annual online family medical refresher course offered through a Canadian University (in another province) that usually features content on genetic screening every 1-2 years. Informants attributed the barriers in primary care to the fact that in NL, there is no central, trusted site or authority responsible for getting this information into the hands of primary care doctors (and NPs). This lack of central authority also means a lack of health care system accountability when these recommended tests for PV carriers are not being ordered. Again, some PV carriers may opt not to proceed with further surveillance for personal or family reasons, as is their right. However, there are instances where individuals are agreeable to the recommended surveillance, but for whatever reason, it is not being ordered by their primary care provider. A primary care provider found in his experiences, patients tend to be over-reliant on their GPs to remember and coordinate all screening appointments. In the context of a busy family practice with no centralized hereditary cancer screening reminders, it is easy enough for a GP to overlook a preventative screening referral. He added that his 'greatest fear was that [he] will miss a screening and early detection will be missed'.

In several existing registry systems in the province, primary care providers are provided

notifications, for example: when their patient is due for recommended screening/follow-up, if the recommended imaging is not in the registry after a certain period following the recommended screening interval, and of the imaging results. It was expressed that a registry should not have a sole focus on assigning responsibility to primary care providers but also on supporting them to meet their professional obligations as a primary health care provider. A former genetic counsellor reported that their encounters with primary care providers were mostly positive, as primary care providers were interested in learning 'what they had to do' vis- a-vis their patients' genetic results, recognizing their professional liability to act on the information they were sent from Provincial Medical Genetics (PMG). Similarly, a primary care provider reported that he 'referred as much as possible to PMG' and tended to adhere generally strongly to their recommendations.

It was recommended that a follow-up program should involve regular communication and support for primary care providers and provided the example of having an identified person in a follow-up program with expertise in inherited cancers who could be readily available to primary care providers for consultations. This may also improve the appropriateness of formal referrals to PMG and the appropriateness of screening, specialist referrals. A primary care provider agreed, and also noted that a central registry with patient/primary care provider notification of when screening is required would 'be ideal' and cited examples of the provincial colon and breast screening programs as prime examples of how a prospective registry could work. There was some variation in opinions of those interviewed as to whether the primary care physician should remain the one to order screening and the role of the registry would be 'reminder' and follow-up. Or, alternatively, if the follow-up registry program would facilitate the ordering and booking of the tests and follow the results up with the family primary care provider

and the individual. This question will need to be examined in greater detail later in the policy proposal development.

The Role of the Nursing Profession

Several informants expressed that there is unmet potential for nurses to become involved, and to ultimately improve the delivery of genetic health care. There are nurses who work as genetic counsellors in Canada who were 'grandfathered in' prior to the advent of the Canadian Association of Genetic Counsellors. Since then, the entry level requirement for certification as a genetic counsellor is a master's degree in genetic counselling. One individual expressed that a master's degree in genetic counselling is rightfully so the entry level requirement to practice as a genetic counsellor but there is still a plethora of potential nursing roles in genetic care. This included, but is not limited to, conducting, and assisting in genetic research, engaging in follow-up and supportive care. Other informants hypothesized how the nursing profession could be expanded in genetic care; one nurse expert noted that ultimately this directive needs to come from 'the top down' in the profession i.e. nursing and nursing education regulatory bodies and professional organizations. However, they expressed that nurses in practice and in research must self-advocate how nurses could improve outcomes in genetic care. The successful implementation of nurse navigation programs in other populations, such as patients with a confirmed cancer, was also referenced to give credence to the argument that a nurse navigator could provide a similar service to PV carriers. The question was also raised if a nurse practitioner associated with a hereditary cancer screening program could be the one to order the recommended follow-up tests.

A nurse with experience in genetic counselling spoke of how in the past, they were often invited to give guest lectures at nursing schools on the topic of genetics. However, in recent

years this seems to have fallen by the wayside. There are unmet opportunities to engage future nurses in the topic of genetics and to stimulate their brainstorming on the future of genetic nursing. This assertion is supported by anecdotal evidence from my own undergraduate nursing education and in informal conversations with nurse practitioners and nurse practitioner students who noted that genetic content in their education programs was minimal to nil. There are clear opportunities for mobilizing genetic nursing in nursing practice, research, education, and in professional governance.

Funding Considerations

When discussing potential barriers to implementing a dedicated follow-up program, interview participants consistently cited the challenges in securing 'dedicated and sustained provincial health system funding'. Some informants expressed uncertainty over whether the will to create and fund such a service exists 'in the current climate of very scarce resources' despite the urgent need. Another informant described the current health care system as a 'sick care system' that is so consumed with expensive disease treatments that there are little resources available for disease prevention. Thus, this downstream approach to 'health' further perpetuates this unsustainable and dysfunctional cycle and inhibits the development of programs, such as this proposed program policy. Some informants noted that in this dysfunction, there is a great case to be made for these programs in terms of cost-effectiveness. They recommended that I examine publications where researchers demonstrated health care value in population-based genetic testing and long-term clinical management of hereditary cancer predisposition syndromes (Manchanda et al., 2018; Shanahan et al., 2020; Yerushalmi et al., 2016). One informant suggested that this program proposal should be framed to policy makers in terms of how it could reduce costs associated with expensive cancer treatments and drugs, hospital admissions and ER

visits associated with complications of cancer. It is important to convince health system decision makers that the question is not 'how are we going to pay to implement this program?' rather, 'how are we going to pay to NOT implement this program?' It was recommended that the foreseeable price tag of this program should be presented to decision makers and contrasted with a cost analysis of the health care system costs of undetected and/or non-surveyed hereditary cancer.

To overcome the challenges in securing program funding, several informants interviewed agreed that a pilot project is an effective way to provide 'proof of concept to health system funders and policy makers.' It was remarked that several successful health care programs and interventions once started as health needs assessments, which led to pilot projects, funded largely through provincial and federal government grants. It was also suggested that a potential pilot program should be evaluated in terms of patient experience and satisfaction, primary care provider experience and satisfaction, health system utilization, cost savings, and clinical outcomes. There was a wealth of data collected in the consultations that both corroborate the literature review findings and provides further direction to proposed program, from the perspective of key NL stakeholders.

Part Two: Environmental Scan

Objectives of the Environmental Scan

The key informant interviews provided insight into the local stakeholder's perceptions of the needs, current gaps, and recommendations for a HBOC follow-up program. It is also important for this policy proposal to determine what works well for PV carrier follow-up programs in other health jurisdictions. To this effect, an environmental scan was conducted. The objectives of the environmental scan were: 1) to gain an idea of what services are common

features of familial cancer programs in areas of Canada outside of NL and globally. 2) To examine breast screening programs in Canada and features of a high-risk breast registry, and 3) to examine the gamut of services offered by HBOC non-for-profit organizations.

Ethical Considerations

The HREA screening tool was completed prior to conducting the environmental scan. It was determined that the environmental scan was exempt from requiring HREA ethics approval because items 1 and 4 of the checklist were applicable (See Appendix B). The entirety of the information collected for the environmental scan was obtained from publicly available websites and there was no expectation of privacy from these sources.

Methods

For the first objective, I scanned the websites of other healthcare facilities in Canada and elsewhere in the world where tailored follow-up programs/clinics for HBOC PV carriers are offered. I also searched for a variety of available websites of international health care facilities that were written in English. To meet the second objective of the environmental scan, I retrieved an existing environmental scan of breast screening programs throughout Canada. This document provided useful information and led me to the website of the High-Risk Ontario Breast Screening Program, a coordinated service that arranges the follow-up imaging for individuals with a hereditary breast cancer predisposition syndrome in the province of Ontario. For the third objective, when I was completing the literature review, non-for-profit HBOC organizations were often referenced by PV carriers as facilitators to follow-up care and psychosocial adjustment. I searched websites of these non-for-profit organizations that were commonly referenced in the literature review findings to determine the services offered.

Results

Familial Cancer Clinics

In many larger centers in Canada and around the world, familial cancer clinics are available as part of the genetic care process. Comparable to the process offered through PMG, genetic counselling and genetic testing referral are offered through many of these centers. This also includes immediate genetic testing follow-up and personalized genetic risk assessment. How these centers differ from PMG is in their approach to surveillance follow-up and support. Many of these centers feature multidisciplinary teams including geneticists, genetic counsellors, nurses, dieticians, gynecologists, oncologists, social workers, and psychologists. In these ways, the multiple facets of PV carriership are addressed. Also, a few of these clinics/programs offer periodic carrier support groups and sessions where PV carriers can liaise with other PV carriers and families and attend support sessions with guest speakers and genetic/hereditary cancer experts. Many of these programs also differ from the current PMG model in that PV carriers are scheduled to visit the clinics annually or bi-annually for surveillance, follow up and supportive care. In the province of Ontario, many of these clinics work with the High-Risk Ontario Breast Screening Program (OBSP) to coordinate breast surveillance of eligible high-risk individuals.

Another important impact of these familial cancer clinics/programs is the contribution to translational hereditary cancer and genetic research. Clinic attendees may be given the option to participate in research that may also be of benefit to them. For example, clinic attendees who are followed long-term may be given the option to participate in trials of new prevention modalities, as the field of genetic medicine continues to rapidly evolve. Similarly, if clinic attendees undergo genetic testing and are found to have variants of uncertain significance (VUS), these variants may later be classified as pathogenic. Having a registry and follow-up system would be an ideal

way to notify individuals if their identified VUS was reclassified as pathogenic, and of the emerging clinical actions recommended for their particular PV. For a full list of familial cancer centers examined in this scan, please see Appendices I & J.

Average and Increased Risk Breast Screening in Canada

The Canadian Partnership Against Cancer (CPAC) (2018) conducted an environmental scan of the breast cancer screening programs available in all provinces and territories in Canada. With the exception of Nunavut, 12 provinces/territories offer organized breast screening programs. All 12 provincial programs offer breast screening services for women at average risk. According to Canadian Task Force on Preventive Health Care (2011), women at average risk for breast cancer should be screened using mammography from ages 50-74 years old, every two to three years. Most provinces recruit women for the provincial breast screening programs via physician referral (n=11) and self-referral (n=11) (CPAC, 2018).

In addition to average risk women, some of these provincial programs manage women at 'elevated risk' and 'high-risk' of breast cancer. There is a distinction in 'elevated risk' and high-risk. CPAC (2018) defined 'elevated risk' women as those who have an above average risk of breast cancer but less than the highest risk group. Risk factors for 'elevated risk' include increased breast density, use of hormone replacement therapy in the past, family history of breast cancer, and increased risk for benign breast disease (CPAC, 2018). In (n=10) provincial programs, women with an elevated risk of breast cancer are screened and managed by the program, including in NL, and these women are recommended to undergo annual mammography starting at age 40-50 (CPAC, 2018). Generally, provincial definitions of 'high risk' of breast cancer include known carriers of pathogenic variants, and first-degree relatives of a known PV carrier who did not opt for genetic testing, among other risk factors (CPAC, 2018). The CPAC

(2018) outlined that a challenge with managing high-risk women is that there are currently no national guidelines for screening this population, therefore there are variations in screening protocols across various Canadian jurisdictions. Most provinces have guidelines for recommended high-risk screening intervals, consisting of annual mammography, MRI and/or ultrasound beginning at age 30, 40 or 50, to stop at age 69-74 (CPAC, 2018). In NL, the Breast Disease Site Group (2017) of Eastern Health (EH) recommends alternating annual MRI and mammography for high-risk women, starting at age 30. What is perplexing is that although most provinces and territories have guidelines for high-risk breast management, only five provincial/territorial breast screening programs manage high-risk women (CPAC, 2018). Despite the Breast Disease Site Group (2017) policy, women at high risk of breast cancer in NL are referred back to their primary care provider for further management (CPAC, 2018).

High-Risk Ontario Breast Screening Program

In the province of Ontario, there is a dedicated follow-up program for women at high-risk of breast cancer. To be enrolled in the High Risk OBSP, a referring physician must submit a requisition form to a designated High Risk OBSP site (Cancer Care Ontario, n.d.) It is implicit in the program requisition form that the ordering physician is requesting future MRI testing and in some cases, image guided biopsies, which under current Ontario regulations requires a physician's signature (Cancer Care Ontario, n.d.) Women who are eligible for the program fall into one of two categories, category A and B. Women in Category A are directly enrolled in the program because they carry a known PV associated with increased breast cancer risk, or are a first-degree relative of someone with a known PV and underwent genetic counselling but opted not to have genetic testing (Cancer Care Ontario, n.d.) Women in Category B require further genetic assessment prior to enrollment in the program; this includes individuals who are a first

degree relative of someone with a known PV who have not been assessed by a genetic counsellor. Following the genetic assessment, it will be determined whether the woman in Category B is enrolled in the High Risk OBSP. Cancer Care Ontario (n.d.) outlined that the high risk OBSP program is operated by high risk OBSP navigators with a list of responsibilities. These include booking the screening and breast assessment appointments, following up on abnormal results, arranging annual recalls, and communicating all imaging results to women and the referring physician. From my assessment, it was clear that this program was an effective method to address some of the common barriers to screening identified in the consultations.

Non-For-Profit HBOC Organizations

In a scan of websites, I discovered three non-profit organizations with a focus on HBOC: Bright Pink, FORCE: Facing Our Risk of Cancer Empowered, and the HBOC society. Both Bright Pink and FORCE are operated out of the United States while the HBOC society is a Canadian non-for-profit, with headquarters in Alberta. All three organizations offer some form of peer support either through connecting PV carriers with other PV carriers. This is achieved through peer navigation and peer mentoring programs as well as through online message boards. All three organizations also offer resources and information to PV carriers and families about risk management, the genetic testing process, and other concerns related to carriership.

Bright Pink (2020) distinguishes its mandate as having a focus on health rather than cancer. Among its programs is an online digital platform that allows women to assess their risk for HBOC and understand how to manage that risk. Bright Pink (2020) identified that over 1.5 million women have used their online risk platform, resulting in over 600,000 women being identified as high-risk. Bright Pink (2020) also offers health provider education initiatives and identified that since the advent of this initiatives program, over 18,000 health care providers have

participated in their online, continuing education, and in-person education sessions on HBOC. FORCE: Facing Our Risk of Cancer Empowered (2018) highlights in its mission statement that they also have a focus on advocacy to represent the interests and concerns of HBOC PV carriers to the health care providers, research scientists, and legislators. FORCE (2018) also has a focus on research advocacy and promotion. Their *XRAYS* program provides up-to-date research findings presented in clear, everyday language. The organization promotes and advocates for research specific to HBOC and provides PV carriers with information on the research studies they may qualify for. In partnership with the University of South Florida, FORCE (2018) offers a patient-powered research registry. This research registry allows PV carriers to offer input on ideas and priority areas for future research, to participate in designing research studies, to participate in research and become involved in disseminating research results. In this way, FORCE ensures that HBOC research is truly patient-informed. It is important to note that these three organizations are non-for-profit organizations and therefore their mandate differs from that of a provincial health policy. Still, a provincial follow-up program may be effective in connecting PV carriers with other PV carriers and promoting their group identity. This may lead to the advent of special interest non-for-profit groups that represent the interests of PV carriers in the province. From my environmental scan, I could not find any local groups or special-interest organizations with local chapters in NL with a focus on HBOC or hereditary cancer. Prospective local HBOC organizations and the provincial navigation program could then work in partnership with one another to improve the quality of care in this population.

Conclusion

The consultations and environmental scan are further evidence that the current follow-up process for PV carriers in NL is underserving the needs of this population. This current follow-

up process is fundamentally wrong; individuals are notified of their carriership but provided with no long-term follow-up or support to navigate and cope with the lifelong implications of their carriership. Essentially, it is as though PV carriers and their families are being brought up stream, then left to navigate for themselves without a paddle. The evidence is indelible, there needs to be a service to help PV carriers navigate the uncharted waters. The consultations reinforce the literature review findings that a novel follow-up navigation program will need to provide a centralized outlet for PV carriers and their primary care providers on information, and recommended screening and medical appointments. Likewise, it was also clear from the consultations that a novel program must go beyond screening and appointments; it must provide ongoing support to carriers, as PV carriership comes with its own unique territory of psychosocial and family implications. There is also evidence to suggest that the nursing profession has unrivaled potential to contribute to the development and delivery of dedicated follow-up programs. Nurses are ideally situated to assume the role of patient navigator for PV carriers.

The environmental scan provided a picture how dedicated follow-up programs are commonly implemented in several health jurisdictions, suggesting they are both feasible and effective interventions. Furthermore, examining the features of the High Risk OBSP provided a model of effective high-risk breast screening follow-up. Roebothan et al. (n.d.) noted that only 41.6% of eligible BRCA PV carriers in NL had undergone the recommended MRI screening in an 18-month period ($p < 0.001$). A prospective program in NL, similar to the High Risk OBSP, would likely improve this low rate of MRI screening uptake in PV carriers. Lastly, it was clear that a dedicated follow-up program for PV carriers could be an invaluable starting point for PV carriers to connect and form a group identity. This could perhaps result in the advent of local

chapters of HBOC non-for-profits organizations and/or support groups. This could allow them as group to self-advocate, promote awareness and education, support one another, and disseminate relevant information.

In a policy framework document on cancer control in NL, the Department of Health and Community Services (2010) outlined the need to "build upon current activity to develop a hereditary cancer screening program ensuring best practice and necessary support, infrastructure (e.g. genetic counseling) and resources" (p.13). It is discouraging that over the past 10 years, it appears as though the NL government has made little to no progress on actualizing this goal. It is clear what needs to be done and how it will improve outcomes and reduce costs associated with cancer treatments. This consultation/environmental scan report adds to the mounting evidence that it is time to act for the PV carriers in NL, to improve and ultimately save their lives.

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Appendix A: Letter to Participants

MEMORIAL UNIVERSITY Faculty of Nursing

July 2020

To Whom it May Concern,

My name is Rebecca Puddester and I am a registered nurse. I am also currently a Master of Nursing student at the Faculty of Nursing at Memorial University of Newfoundland. My nursing career began on a women's health inpatient unit where I provided care to women with gynaecological malignancies. It was there that pointed me in the direction of research related to individuals and families with hereditary breast and ovarian cancer (HBOC). It became clear from the literature that in many health jurisdictions, there are considerable gaps in the care of individuals and families with HBOC predisposition syndrome. This results in missed opportunities to improve and indeed, save their lives. My master's practicum project is to develop a policy proposal for a specialized program in Newfoundland and Labrador (NL) that would provide on-going health care system navigation, follow-up, risk-reduction, and psychosocial support to these high-risk individuals.

Part of this project is to engage with stakeholders and experts involved in the current system of identification, follow-up management, and support of HBOC syndrome individuals and families. In so doing, this will help to identify gaps in the current system, to determine the acceptability of such a program to stakeholders, and to provide direction on useful features of such a program. It will also provide stakeholders with an opportunity to propose alternative strategies or considerations for the management and support of this high-risk population.
You have been asked to participate in this consultation because you have been identified as having an important vantage point in the NL health care system that should be heard and considered prior to the development of any HBOC policy proposal in NL. Your participation in this consultation is voluntary. The findings will be anonymous, and your name will not appear in any report unless otherwise discussed and confirmed with you. The consultation will involve a telephone or online interview with myself, using the semi-structured question guide attached. If preferred, you may answer these questions in writing via e-mail. The content of all the consultations will be analyzed and reported as themes in the consultation report. The interview transcripts will not be included in the final practicum. Moreover, the interview transcripts will be secured in a passworded document accessible only to myself and my practicum supervisor. They will be destroyed at the completion of my degree in December 2020.

If you have any further questions about this consultation, please do not hesitate to contact either myself at rjp823@mun.ca or my practicum supervisor, Dr. Joy Maddigan at n6jm@mun.ca.
Thank you for your time and consideration, it is greatly appreciated and valued.
Rebecca Puddester, BN RN

Appendix B: Health Research Ethics Authority (HREA) Screening Tool

Student Name: Rebecca Puddester

Title of Practicum Project: Policy Proposal for a Hereditary Breast and Ovarian Cancer Navigation Program

Date Checklist Completed: July 3rd, 2020

This project is exempt from Health Research Ethics Board approval because it matches item number 1,3,4,5 from the list below (in bold).

1. **Research that relies exclusively on publicly available information when the information is legally accessible to the public and appropriately protected by law; or the information is publicly accessible and there is no reasonable expectation of privacy.**
2. Research involving naturalistic observation in public places (where it does not involve any intervention staged by the researcher, or direct interaction with the individual or groups; individuals or groups targeted for observation have no reasonable expectation of privacy; and any dissemination of research results does not allow identification of specific individuals).
3. **Quality assurance and quality improvement studies, program evaluation activities, performance reviews, and testing within normal educational requirements if there is no research question involved (used exclusively for assessment, management or improvement purposes).**
4. **Research based on review of published/publicly reported literature.**
5. **Research exclusively involving secondary use of anonymous information or anonymous human biological materials, so long as the process of data linkage or recording or dissemination of results does not generate identifiable information.**
6. Research based solely on the researcher's personal reflections and self-observation (e.g. auto-ethnography).
7. Case reports.
8. Creative practice activities (where an artist makes or interprets a work or works of art).

For more information please visit the Health Research Ethics Authority (HREA) at https://rpresources.mun.ca/triage/is-your-project-exempt-from-review/

Appendix C: Semi-Structured Interview Guide #1

A Consultation with Primary Care Providers in NL

1) Is there a screening tool that you use in your family practice to identify individuals who would benefit from a referral to Provincial Medical Genetics (PMG)?
2) From a primary care provider perspective, what are the most salient barriers to ensuring adequate follow-up and risk management for patients with an inherited pathogenic variant (PV)?
3) What resources are available to you when providing genetic follow-up to patients and guidelines for recommended screening and risk-reduction? Have you used these resources?
4) Without providing any identifiable patient information, can you describe your experiences with managing the recommended screening and risk management for your patients with PVs that are considered "actionable"?
5) How is the topic of genetic risk management incorporated into your continuing medical competency education?
6) If available, would you be interested in participating in a module/workshop about management of actionable PVs if it counted towards continuing competency hours?
7) If there was a coordinated cancer registry with a PV carrier navigation program in NL, do you think such a program would be beneficial? If so, what are some key features that you think such a program would entail?

Appendix D Semi-Structured Interview Guide #2

An Interview with a Genetic Counsellor in Provincial Medical Genetics

1) What are some salient barriers to follow-up following genetic testing for individuals with pathogenic variants (PV) and variants of uncertain significance (VUS) in the current genomic care paradigm?
2) Can you describe your role from the point of intake of a referred individual to PMG, to the disclosure of their genetic results and follow-up?
3) How long do you follow-up with an individual if they have an identified PV?
4) Do you have continued follow-up with their primary care provider?
5) What is your role in their adherence to recommended screening/risk modalities for actionable PVs?
6) From your perspective, would a PV/VUS registry/follow-up program with a coordinated approach to risk-reduction/psychosocial support be a beneficial program? If so, what would be some key features of that program in terms of the basket of services that it should offer?
7) What would be some barriers to the development of establishing such a program from your perspective?
8) Some health jurisdictions implement nursing care in genetic programs and follow-up. Has this been the case in NL and is there potential for an expanded nursing role in genetic care?
9) To your knowledge, are there any other individuals who are relevant stakeholders who you recommend I should consult as part of this practicum project?

Appendix E: Semi-Structured Interview Guide #2

An Interview with a Professor of Clinical Epidemiology, specializing in Ethical, Legal, and Social Issues Pertinent to Genetics

1) From your perspective as an expert researcher and professor of clinical epidemiology, what are some of the most salient barriers to adequate follow-up for individuals in NL who test positive for a known pathogenic variant (PV)?
2) Do you think there is a need for a coordinated approach to follow-up care/risk reduction adherence/psychosocial support for individuals in NL with PVs and VUS? If so, what do you believe should be key services that such a program should offer?
3) From your perspective, what would be some barriers to implementing such a program?
4) What would be some important ethical and legal considerations to account for when developing a program model for follow-up care in NL?
5) Are there research publications that you recommend I review that are relevant to the focus of my policy proposal?
6) Hypothetically, if in the future, such a program policy proposal was implemented, could this program be a feasible basis for a pilot study where outcomes were measured and/or qualitative data was collected about participant satisfaction with the program?
7) Are there other relevant stakeholders who you recommend that I should include as part of my consultation plan?

Appendix F: Semi-Structured Interview Guide #4

An Interview with a Nurse Involved in the Development of a Registry for a Cancer Screening Program

1) What lead you to become involved in the development of the provincial screening program?

2) What was/is the role of the program?

3) What were common barriers in adherence to recommended screening?

4) What were barriers/facilitators to developing the program?

5) What was the process for rectifying when primary care providers did not order the recommended screening modality based on the algorithm in the provincial screening program?

6) From your perspective, what are key features that should be incorporated in any screening registry/navigator program?

7) How did you secure funding when you proposed the cancer screening registry?

Appendix G: Semi-Structured Interview Guide # 5

A Consultation with a Nurse with Experience in Research Pertaining to Individuals with an Inherited Cancer Predisposition Syndrome

1) From your research with individuals carrying pathogenic variants (PV) what were the most salient barriers to their adherence to recommended screening barriers? What were some facilitators that positively influenced their adherence?
2) Did you conduct any interviews/consultations with primary care providers or tertiary care providers about what barriers they are experiencing providing primary care to individuals living with pathogenic variants?
3) Have you discussed with other individuals in Canada, how NL compares to the follow-up and counselling paradigm for individuals with PVs in other areas in Canada?
4) Given your experience with the topic, if there was a PV carrier navigation program in the province, do you think this would be a beneficial intervention? What are some key features you think such a program should include?
5) Your PAHD psychometric scale was validated in a population of Lynch Syndrome PV carriers in NL. As the co-developer/primary author of the scale, do you think this scale could be used to evaluate the impact of a systematic program for PV carriers in a prospective before/after study +/- a control group? Has this scale been validated in other populations such as the ARVC PV carriers?
6) Do you have any recommendations for individuals who I should contact as stakeholders both provincially and elsewhere as I go forward in developing the policy proposal?
7) Canada is trailing behind other developed countries including the US and the UK with respect to how nurses are incorporated in the genetic/genomic health care paradigm. This

includes a lack of outlined genomic nursing competencies and lack of inclusion of genetic content in nursing education. In your opinion, as a profession and a discipline, regulatory bodies (CNA, CASN), how do we rectify this? Should this be a priority?

Appendix H: Semi-Structured Interview Guide #6

An Interview with an Oncologist with Experience in Inherited Cancer Predisposition Syndromes

1) From the perspective of oncologists/surgeons who perform risk-reduction surgeries, what barriers are they experiencing to ensuring adequate identification, and follow-up of individuals with PVS predisposing them to HBOC?

2) In your opinion, how does NL compare to other jurisdictions in Canada in terms of follow-up, management, and support to individuals with cancer predisposition syndromes? How does Canada compare with other developed countries?

3) In other jurisdictions outside of NL, what aspects of their PV carrier follow-up programs/strategies have been most impressive to you? In other words, what have you seen in other provinces that they are doing right in their management of these high-risk individuals?

4) From your perspective, what factors facilitate adequate adherence to risk-reduction modalities and screening for HBOC PV carriers?

5) If there were to be a systematic program for follow-up, management, and support of PV carriers, what would be the most important features that such a program should entail?

6) What do you think would be the greatest barriers/obstacles to implementing such a program?

7) Do you think there is a potential to expand the nursing role in the development and implementation of dedicated follow-up and navigation programs for PV carriers?

8) Is there anything else you would like to add/mention that is relevant to the focus of this project that has not been discussed in this interview?

Appendix I: Websites of Canadian Genetics Centres Offering Multidisciplinary Care and Follow-Up (Outside of NL)

Atlantic Canada

Maritime Medical Genetics Centre, IWK
Dr. Richard B. Goldbloom Research and Clinical Care Pavilion **IWK Health Centre** **5825 South St, Halifax, NS B3H 1V** **Canada** **Services Offered (according to website)** "Our team of Medical Geneticists, Genetic Counsellors, Nurses, Dietitian & Administrative staff provides genetic services to Nova Scotia, New Brunswick and Prince Edward Island" "In genetics, we help people learn about diseases in their families and help to determine if there is a genetic cause. Our knowledge of genetics has increased tremendously over the past few years, and there have been many new developments in testing techniques and availability. A genetic assessment can aid families understand the hereditary aspects of diseases in their family." ***No discussion of long-term high-risk follow-up of PV carriers on website** **Website URL:** http://www.iwk.nshealth.ca/childrens-health/services/#/mmgs

Quebec

Cancer Genetics Service, McGill University Health Centre
Montreal Children's Hospital **1001 Boulevard Décarie** **Montréal, QC, H4A 3J1** **Canada *also in partnership with sister hospital, Jewish General Hospital, Montreal QC** **Services Offered (according to website)** "Our multidisciplinary team of cancer genetics specialists provides personalized cancer risk assessments based on medical and family history, and when indicated, help individuals navigate through the process of genetic testing" "Through supportive counseling and education about hereditary cancers, we help empower individuals to use genetic information to make informed decisions about cancer screening and primary prevention. We also provide recommendations to other medical specialists working in surgery, oncology, gastroenterology, and gynecology, to ensure that individuals at "high-risk" for developing cancer have access to the appropriate surveillance and follow-up" **Website URL** https://muhc.ca/med-genetics/clinical-genetics
High Risk Breast Clinic, Jewish General Hopsital
High Risk Breast Clinic **JGH Stroll Cancer Prevention Centre** **E-Pavilion, Room E740** **3755 Ch. de la Cote Ste Catherine** **Montreal, QC, Canada** **Services Offered (according to website)**

"The objective of the High-Risk Breast Clinic is to provide screening and clinical surveillance to women who are at an elevated risk of developing breast cancer. The clinic is also designed to provide supportive counselling and prevention strategies that can help lower risk in high-risk patients"
"Depending on your personalized risk assessment and level of risk, you may require further appointments and/or regular follow up visits. Most patients have one visit per year or two visits per year"
Website URL: https://www.mcgill.ca/cancerprev/clinical/hrbc

Hereditary Gynecologic Surveillance Clinic, Jewish General Hospital

Jewish General Hospital
3755 Côte-Sainte-Catherine Road
Room E-740
Montreal, QC, H3T 1E2
Canada
Services Offered (according to website)
"The objective of the Hereditary Gynaecologic Surveillance Clinic is to promote prevention and make best detection services available to women who have been identified as being at a higher risk for developing gynaecologic cancers. The focus of the clinic is to offer long-term medical and emotional support to these women"

"Depending on your discussion with the staff members of the Hereditary Gynaecologic Surveillance Clinic and the medical tests requested, you might need more appointments. The Clinical Care Coordinator handles all of the appointment scheduling. Most patients either have one visit per year or two visits per year"
Website URL: https://www.mcgill.ca/cancerprev/clinical/hgoc

Ontario

Cancer Genetics and High-Risk Program, Sunnybrook Health Sciences Centre

Louise Temerty Breast Cancer Centre
2075 Bayview Avenue,
M-wing, 6th floor
Toronto, ON M4N 3M5
Canada
Services Offered (according to website)
"Offers cancer risk assessment, genetic counselling and/or genetic testing to eligible individuals and families who may be at risk for hereditary cancer"

"At Sunnybrook, female BRCA carriers may be followed yearly in our Breast High Risk Program. This may start at age 25 where visits consist of clinical breast exams until age 30. At age 30, women are eligible for mammogram and breast MRI imaging through the High Risk OBSP"

Website URL: https://sunnybrook.ca/content/?page=occ-cancer-genetics

Cancer Genetics Clinic, Medical Genetics Program of Southwestern Ontario

London Health Sciences Centre
Victoria Hospital
London Regional Cancer Program
800 Commissioners Road East
London, ON, N6A 5W9
Canada
Services Offered (according to website)
"The Cancer Genetics Program (CGP) is a collaborative effort between the London Regional Cancer Program (LRCP) and London Health Sciences Centre (LHSC). The program offers genetic counselling and testing for families at risk for inherited forms of breast, ovarian or colorectal cancer"
"Following services available to individuals and families: genetic counselling, testing, assessment, education, surveillance"
"A patient support group has been formed to provide emotional support for individuals and families. Meetings consist of group discussion as well as occasional guest speakers"
Website URL: https://www.lhsc.on.ca/medical-genetics-program-of-southwestern-ontario/cancer-genetics-clinic

Clinical Genetics Program, Thunder Bay Regional Health Centre

980 Oliver Road
Thunder Bay, ON, P7B 6V4
Canada
Services Offered (according to website)
"The genetic counsellor, genetics nurse and geneticist provide information and counselling to those at risk of having a hereditary cancer syndrome. In collaboration with the Regional Cancer Care Northwest, a consultation with an oncologist is also available as needed"

"Geneticists, genetic counsellors and genetics nurses are part of a team of health care professionals who provide genetic services. Geneticists are medical doctors who have special training in the area of genetic conditions. A genetic counsellor has post graduate training in genetic counselling. A genetics nurse is a nurse with extra training in genetics. Together, they can provide information about genetic conditions and explain how they are passed from one generation to another"
Website URL: http://tbrhsc.net/programs-services/prevention-and-screening-services/genetics-program/

The Charlotte and Lewis Steinberg Familial Breast and Ovarian Cancer Clinic, North York General Hospital

North York General Hospital, General site
4001 Leslie Street, 3rd floor
Toronto ON, M2K 1E1
Canada
Services Offered (according to website)
"Provides genetic assessment and counselling for individuals and families at increased risk for inherited breast and/or ovarian cancer"

"In our multidisciplinary team setting, we are joined by a family doctor with a special interest in breast and ovarian cancer surveillance and prevention, and a gynecologist. Some patients who are at high risk for inherited breast and ovarian cancer are seen on a regular basis by this multidisciplinary team for surveillance and support. In some cases, genetic testing is available"

The Centenary Genetics Clinic (Rouge Valley Centenary
2867 Ellesmere Road, Toronto**,** ON M1E 4B9) is a satellite clinic of the North York General Hospital Genetics Program.
Website URL: https://www.nygh.on.ca/areas-care/genetics/clinical-services

Familial Cancer Centre, Princess Margaret Cancer Centre

Princess Margaret Cancer Centre
M740-610 University Avenue
Toronto, ON, M5G 2M9
Canada
Services Offered (according to website)
"The Familial Cancer Clinic provides genetic counselling and risk assessment services to people with a personal or family history of cancer. We can talk with you about whether the cancer in your family may be hereditary and discuss how you can manage your risk of developing cancer. We can also let you know if you are eligible for high-risk breast cancer screening (OBSP)"

"Familial Cancer Clinic includes world-class health care professionals working for you and with you. Your team can include doctors, genetic counsellors, students, volunteers and many others dedicated to helping you and your family"
Website URL:
https://www.uhn.ca/PrincessMargaret/Clinics/Familial_Breast_Ovarian_Cancer/#tab1

Familial Breast Cancer Clinic, Mount Sinai Hospital

Marvelle Koffler Breast Centre
Mount Sinai Hospital
Joseph & Wolf Lebovic Health Complex
600 University Avenue, 12th Floor
Toronto, ON
Canada
Services Offered (according to website)
"Offers a comprehensive counselling and referral service for individuals with a personal and/or family history of breast cancer, or other cancers suspicious for a hereditary predisposition."

"Facilitated by genetic counsellors who provide breast cancer risk assessment to determine eligibility for high-risk breast cancer screening through the Ontario Breast Screening Program. Also, the genetic counsellor determines eligibility for genetic testing. Following an assessment, referrals are made to members of the team in the departments of medicine, nursing, psychiatry, social work, and nutrition as required."

"Individuals who are found to carry a hereditary predisposition may be offered ongoing clinical follow-up through our high-risk clinic"

Website URL: https://www.mountsinai.on.ca/care/cancer/cancers-we-treat/marvelle-koffler-breast-centre

Familial Oncology Program, Kingston General Hospital

25 King St. West
Kingston, ON K7L 5P9
Canada
Services Offered (according to website)
"The Familial Oncology Program (FOP) offers genetic counselling and testing to you and your family if you are at risk of inherited forms of breast, ovarian, colorectal or other cancers"

"Cancer screening and preventative treatment options available to you and your family. To do this, we work closely with the High Risk Cancer Clinic and breast cancer screening programs.
Website URL: https://kingstonhsc.ca/cancer-care/genetics-familial-oncology-program

Genetics/Hereditary Breast Cancer Clinic, Women's College Hospital

Women's College Hospital
76 Grenville Street
Floor 5
Toronto, ON M5S 1B2
Canada
Services Offered (according to website)
"Provides comprehensive genetic counselling services for individuals with a personal and/or family history of cancer"

"Testing services are available to those who are eligible"

"Families may receive other clinic services or long-term follow-up"

"We can also let you know if you are eligible for additional breast cancer screening through either the WCH High Risk Breast Screening clinic or the Ontario Breast Screening Program (OBSP)"

Website URL:https://www.womenscollegehospital.ca/care-programs/henrietta-banting-breast-centre/genetics-and-hereditary-breast-cancer-clinic

Hereditary Cancer Clinic, Children's Hospital of Eastern Ontario

Regional Genetics Program, CHEO
401 Smyth Road
Ottawa, ON K1H 8L1
Canada
Services Offered (according to website)
"We specialize in the assessment of children, adults, and families with familial or hereditary forms of cancer predisposition. We offer genetic counselling and genetic testing services if indicated. Through partnership with the Ontario Breast Screening Program (OBSP) we also provide women with a personal or family history of breast and/or ovarian cancer with a personalized breast cancer risk assessment"

"Our goal is to identify individuals and families who may be at an increased risk of developing specific types of cancer and to inform them about the appropriate prevention and screening recommendations available"

"Our services are provided by a team consisting of genetic counsellors, geneticists and administrative/clerical staff"'
Website URL: https://www.cheo.on.ca/en/clinics-services-programs/hereditary-cancer-clinic.aspx

Manitoba

Hereditary Breast Cancer Clinic, Health Sciences Centre Winnipeg

FE-229 Community Services Bldg, 820 Sherbrook Street
Winnipeg, MB R3A 1R9
Canada

Services Offered (according to website)

"Consisting of geneticists, genetic counselors and laboratory technicians who provide education, counseling, risk assessment gene testing, screening recommendations and links to other support services"

"We work with colleagues in CancerCare Manitoba and the WRHA Oncology Program to develop a multidisciplinary approach to patient information sharing, health and preventative medical care, risk assessment and counselling. This service works closely with the DNA Diagnostic Laboratory at the Health Sciences Centre"

Website URL: https://sharedhealthmb.ca/services/breast-health-centre/services/genetics/ and https://wrha.mb.ca/genetics-and-metabolism/hereditary-cancer-service/

Western Canada

Allard Hereditary Breast and Ovarian Clinic, Lois Hole Hospital for Women

Lois Hole Hospital for Women
10240 Kingsway Avenue
Edmonton, Alberta
T5H 3V9
Canada

Services Offered (according to website)

"Provides care for women who have a high risk of hereditary breast or ovarian cancer"
"Offers assessment, surveillance, and follow up for women who have a high risk of hereditary breast or ovarian cancer including BRCA1 and BRCA2 mutations"

"Ongoing breast Surveillance and referrals for Gyne-Oncology for Risk Reducing Surgery. Support services are offered to clients and their families"

Comments

"Clients must be between 25 and 70 and meet the eligibility listed on the referral form. Clients will also be considered if they have:

- a recommendation from a genetics clinic
- BRCA1 or BRCA2 mutation
- a first degree relative (a parent, sibling, or child) with BRCA1 or BRCA2 mutation
- had radiation to their chest before age 30
- a strong family history of breast or ovarian cancer
- must meet the referral criteria for acceptance in the clinic

Website URL: https://www.albertahealthservices.ca/findhealth/service.aspx?Id=1062103

Hereditary Cancer Program (BC & Yukon)

(2 Clinic Locations)
Hereditary Cancer Program
600 West 10th Avenue
Vancouver, BC V5Z 4E6
Hereditary Cancer Program
32900 Marshall Road
Abbotsford, BC V2S 0C2
Canada

Services Offered (according to website)

"Provides genetic counselling and genetic testing for BC/Yukon residents who may have inherited an increased risk for specific types of cancer. Similar services are available across Canada and in other countries"

"The HCP team includes specialized physicians, genetic counsellors, nurses and support staff. Working together, we provide hereditary cancer risk assessment, genetic counselling and genetic testing. Our team also provides education (link to other section) about hereditary cancer topics"

Website URL: http://www.bccancer.bc.ca/our-services/services/hereditary-cancer

Appendix J : Scan of Websites of Genetics Centres Offering Multidisciplinary Care and/or High-Risk Follow-Up (Countries Outside of Canada)

BRCA Multidisciplinary Clinic, Rabin Medical Center's Davidoff Cancer Center

Country: Israel

Services Offered (according to website)

"The **BRCA Multidisciplinary Clinic** was created to serve patients with a high risk of cancer at Rabin Medical Center's Davidoff Cancer Center. Since its commencement in 2011, the clinic has served thou - sands of women. Women who carry the BRCA gene mutation have up to an 80% chance of developing breast and/or ovarian cancer. More than 200 mutations have been identi - fied, three of which are typical to Ashkenazi Jews."

"At the Rabin Medical Center BRCA Clinic, patients receive counseling, screening and follow-up care"

"BRCA Multidisciplinary Clinic also treats men with prostate cancer. Research data indicates that men carry mutations in the BRCA 1 and 2 genes, similar to the syn - drome for breast and ovarian cancer in women"

Website URL: https://afrmc.org/brca.php
https://afrmc.org/newsarticle/340

Cancer Genetics Unit, Royal Marsden NHS Foundation Trust

Country: UK

Services Offered (according to website)

- Familial cancer risk assessment
- Diagnostic genetic testing (testing of one or more genes known to associated with cancer predisposition)
- Predictive genetic testing (testing for a specific mutation in a gene, already identified within a family)
- Advice and referral for cancer screening
- Discussion of cancer risk-reducing management options
- Assistance with decisions on cancer treatment options
- Offer of enrolment in genetic research studies
- Long term open access follow-up for patients with a known gene mutation on the RM Carrier Register

"Our aim is to promote cancer prevention, early detection and to help in some cases with management decisions. We assess personal and family history of cancer to decide whether it is likely that there is a hereditary cause. We use this assessment to decide whether a genetic test might help an individual to clarify their own risk"

Website URL: https://www.royalmarsden.nhs.uk/our-consultants-units-and-wards/clinical-units/cancer-genetics-unit

Cancer Genetics and Prevention Program, Dana Farber Institute

Country: USA

Services Offered (according to website)
"A visit to the Cancer Genetics and Prevention Clinic usually includes time with both a physician and a genetic counselor, both of whom have expertise in all forms of inherited cancer syndromes. We educate patients about cancer risk and its implications for them and their family members. Working with referring physicians, we design individualized programs to monitor for the earliest signs of cancer — diagnosing if it occurs, and, in many cases, preventing it from arising."

"Dana-Farber Cancer Institute's Center for Cancer Genetics and Prevention includes a team of expert health professionals — medical oncologists, gastroenterologists, geneticists, gynecologists, psychologists, surgeons, nurses, and genetic counselors — who provide cancer risk assessment and comprehensive recommendations for managing cancer risk"
"Dana-Farber's clinical psychologists work with patients from hereditary cancer families. We regularly consult with individuals who are considering, or have had, genetic testing for a variety of adult and pediatric cancer."
Website URL: https://www.dana-farber.org/cancer-genetics-and-prevention/

Hospital General Universitario Gregorio Marañón, Heredofamilial Cancer Unit

Country: Spain

Services Offered (according to website)
"The Heredofamiliar Cancer Unit provides care for cancer patients and / or healthy people, whose tumor, personal or family aggregation characteristics may lead us to suspect that we are dealing with a hereditary cancer syndrome. The Heredofamiliar Cancer Unit is made up of a multidisciplinary team, which includes hospital professionals from the services of medical oncology, gynecology, general surgery, pathological anatomy, GI specialists, pediatric onco-hematology, and oncological nursing. A nurse affiliated with this unit provides support by managing/following the patient cases. Since its creation in 2010, the center has provided care to approximately 300 new patients a year, who can be referred from any hospital service or from the primary care and providers and specialists from the Gregorio Marañón General University Hospital.

"Its mission is to identify patients at risk of hereditary cancer and provide regulated genetic counseling that, through preventive and early diagnosis strategies, reduces the risk of these patients and their families. On the other hand, through its own clinical studies and collaborations with other institutions, it is intended to continue advancing in the field of clinical and translational research in hereditary cancer"

Website URL: https://www.comunidad.madrid/hospital/gregoriomaranon/profesionales/relacion-especialidades/oncologia-medica
(website translated)

Familial Cancer Program, Genetic Services of Western Australia

Country: Australia

Services Offered (according to website)

"We offer a range of services to people with a personal and/or family history of cancer. We can:

- provide you with information about familial cancer conditions (conditions that may run in your family which put you at an increased risk of developing cancer)
- assess your risk of developing an inherited cancer based on your personal and/or family history
- assess whether genetic testing may be an option for your family
- recommend risk management options
- discuss lifestyle strategies to encourage good health and reduce your risk of developing cancer
- address any questions you may have about familial cancer
- provide support and short-term counselling
- support and facilitate familial cancer research."
-

"We are a team of health professionals who can provide advice about your familial cancer risk, and ways to manage that risk. Our team includes genetic counsellors and geneticists. We provide information and support to people and families who have concerns about familial cancer"

Website URL: https://healthywa.wa.gov.au/Articles/F_I/Familial-Cancer-Program

Appendix III Policy Proposal

Navigating Inherited Cancer Risk

A Policy Proposal for a Dedicated Follow-Up and Navigation Program

Rebecca Puddester

Memorial University of Newfoundland Faculty of Nursing

NAVIGATING INHERITED CANCER RISK

Policy Proposal for a Novel Follow-up and Navigation Program

Rebecca Puddester | Memorial University Faculty of Nursing

Executive Summary

Background to the Issue: Individuals who carry certain inherited pathogenic variants (PVs) have an increased lifetime risk of developing hereditary breast and ovarian cancer (HBOC). There is hope in mitigating increased risk in asymptomatic HBOC PV carriers through risk-reduction modalities. Risk-reducing salpingo-oophorectomy reduces the risk of ovarian cancer by 80% and breast cancer risk by 50% in asymptomatic BRCA PV carriers (Rebbeck et al., 2009). Risk-reducing mastectomy in asymptomatic BRCA PV carriers almost entirely reduces breast cancer risk (Li et al., 2016), while annual breast magnetic resonance imaging and annual mammography alternating every six months, is > 90% effective in detecting stage I breast cancer in high-risk PV carrier groups (Warner, 2018).

Despite the effectiveness of risk reduction modalities, many PV carriers in Newfoundland and Labrador (NL) remain unidentified. This also reflects a global trend; up to the year 2014, only an estimated 2.6% of BRCA PV carriers in the general population had been identified (Manchanda et al., 2018). Moreover, for known PV carriers in the province, there is no systemic support available to them in their long-term risk management. This fragmented approach is resulting in reduced adherence to risk reduction modalities. In an NL study, only 41.6% of BRCA PV carriers in NL had undergone the recommended annual MRI screening in an 18-month period ($p < 0.001$) (Roebothan et al., n.d.). PV carriers have reported that their primary care providers were insufficiently prepared to coordinate their follow-up care and informational needs (Cherry et al., 2013; Leonarcyzk & Mawn, 2015; Watkins et al., 2011). There is no

centralized outlet in NL that provides ongoing support, information, or screening reminders to PV carriers, or to their primary care providers, as they navigate the lifelong implications of carriership. Therefore, these high-risk individuals and families are not receiving the information and support needed to live a long, healthy life vis-à-vis their carrier status.

Project Overview: A policy proposal to address the gaps in HBOC PV carrier follow-up care in NL was conceptualized as my Master of Nursing practicum project. The purpose of the project was to outline recommendations for a HBOC PV carrier navigation and follow-up program in NL and to make sub recommendations for features of this proposed program. In the first practicum course an integrative literature review, key informant consultations, and an environmental scan were conducted. The focus in the first practicum course was to determine the main barriers and facilitators in the current approach to PV follow-up care, and to identify what interventions and follow-up programs for PV carriers have been successfully implemented in other regions; thereby confirming what features of extant programs were pertinent to the proposed program. Input was also obtained from (n=8) relevant NL health care stakeholders on priority issues and recommendations in PV follow-up care. In the second practicum course, all the data collected in the first practicum course was analyzed. Informed by this data, a policy proposal document was developed for a novel follow-up and navigation program for PV carriers in NL.

Key Policy Proposal Recommendations: The establishment of a novel, dedicated HBOC PV carrier navigation and follow-up program in NL is recommended. The five key recommendations for this prospective program are:

1) A central HBOC PV carrier registry in NL

2) The establishment of a nurse navigator position to coordinate PV carrier surveillance and follow-up.

3) Coordinated involvement of a multidisciplinary HBOC team

4) A Person and family centred approach to care

5) Virtual and electronic infrastructure to support delivery of the program

The following is a brief explanation of the proposed program and the significance of the five, recommended program features. Individuals carrying pathogenic variants, likely pathogenic variants, and variants of uncertain significance would be entered into a PV carrier registry following the disclosure of their genetic testing results. This new registry would be a sub-registry of the existing Newfoundland and Labrador Cancer Care Registry. The data in the carrier registry should include demographic information, information on the pathogenic variant, information on the testing panel ordered, and information on the recommended clinical actions for the variant. The registry data should also be updated as PV carriers undergo the recommended screening and risk-reduction modalities. The registry data would be accessible to all approved multidisciplinary team members in the PV carrier follow-up program. PV carrier participation in the follow-up program would be voluntary.

A central feature of the follow-up and navigation program will be the nurse PV carrier navigator role. The nurse navigator would be responsible for maintaining ongoing follow-up with PV carriers, coordinating and scheduling screening appointments, connecting them with other multidisciplinary team members, and assisting with discussing genetic testing results with family members. Therefore, the nurse navigator will rely on the carrier registry data to facilitate booking and screening reminders and to develop a personalized plan of care. Moreover, under the prospective program, PV carriers will be connected to expert multidisciplinary care providers as needed including gynecologists, oncologists, breast surgeons, social workers, psychologists, dieticians, and genetic counsellors as needed, following ongoing assessment with the nurse navigator.

A person-centered approach is recommended for this program when working with PV carriers to deliver a follow-up care plan that is psychologically suited to their needs, preferences, and individual life circumstances. Given these individuals are generally asymptomatic and lead active lives, this program should have flexible delivery options. Moreover, given that the follow-up will be long-term, it is important to offer flexible program delivery modes for PV carriers such as evening and weekend appointments and different communication options, such as in person appointments and/or video and telephone conferencing. The framework recommended to guide the delivery of this program is Hartrick Doane and Varcoe's (2015) relational inquiry. Relational inquiry is an approach to nursing practice where nurses are encouraged to "look beyond superficial clinical situations and recognize the impact of various contexts in nursing practice" (Younas, 2017, p. 340). The principles of relational inquiry are relevant when delivering

person-centered follow-up care that acknowledges all the systemic, familial, and individualized factors influencing adherence and appraisal of risk mitigation in PV carriers.

A family centered approach is also a key feature recommended for this proposed program. Both affected and unaffected family members of PV carriers may experience psychologic distress emanating from the awareness of their own risk and/or the worry of their relatives' increased risk of cancer. In the current PV carrier follow-up processes, the perspective of the family is largely overlooked. The prospective program would address this oversight by encouraging the participation and involvement of family members in a follow-up program. PV carrier families would also be supported by the follow-up program in family genetic results disclosure sessions. There, HBOC follow-up team members would be present to help explain the implications of PV carriership to PV carriers and to their potentially at-risk family members. The program would also support at-risk relatives who wish to pursue genetic testing by connecting them with Provincial Medical Genetics.

The final key recommendation is that virtual and electronic delivery methods should be offered for this program. This will include telehealth and virtual appointment delivery to ensure PV carriers provincewide have equitable access to quality follow-up services, regardless of their geographic location. PV carriers should also be able to opt for electronic text appointment reminders and online information. The proposed program should also be compatible with the province-wide electronic medical record and electronic health record software so that relevant information pertaining to their PV

carriership can be clearly communicated and shared to all relevant providers in the circle of care, ensuring continuity and consistency of care.

Interpretation: The genomic era of health care is upon us and with this comes the responsibility to translate genomic knowledge into improved health outcomes. As clinical genetics and genomics become increasingly integrated in routine health care, PV carriers will require support to understand and apply this knowledge in their lives. This proposed PV carrier navigation and follow-up program is a viable, promising strategy to improve outcomes and save lives in PV carriers. Similar programs have been shown to increase PV carrier satisfaction, improve their adherence to risk-reduction recommendations, facilitate the cascade genetic testing of other at-risk relatives, and to contribute to health care system cost-savings. This policy proposal also highlights an opportunity for a key nursing role in genomic care delivery through the proposed PV carrier nurse navigator role. There are compelling arguments described in the policy proposal to pilot this proposed program in collaboration and consultation with relevant stakeholders. Evaluation of a program pilot project will provide proof of its value to the NL health care system going forward.

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Improving Follow-Up Care for Individuals with Hereditary Breast and Ovarian Cancer Syndrome: A Policy Proposal for a Dedicated Follow-Up and Navigation Program

Pathogenic variants (PVs), such as PVs in the BRCA 1 &2, MLH, and RAD51C genes, are associated with an increased risk of developing hereditary breast and/or ovarian cancer (HBOC). Risk-reduction modalities recommended for PV carriers have been shown to significantly improve HBOC-associated morbidity and mortality. As a result, these and other PVs are considered clinically actionable. Despite this, many PV carriers remain unidentified and/or under-surveilled and as a result, many are missing critical opportunities to benefit from lifesaving risk-reduction procedures, chemoprevention, and surveillance. In the Newfoundland and Labrador (NL) healthcare system, primary care providers are responsible for the initiation of genetic referrals and appropriate PV carrier follow-up. Many PV carriers do not receive sufficient follow-up, screening, and support to adequately manage their risk and cope with the implications of PV carriership. Although there are dedicated PV carrier follow-up models and clinical guidelines implemented in some areas of Canada and internationally, overall, there is a paucity of follow-up programs that address the health considerations of this high-risk population. As the identification and long-term follow-up of PV carriers provides an unrivaled opportunity for disease prevention, the current follow-up process for PV carriers is unacceptable. In this policy paper, issues in the current PV carrier follow-up process in NL will be

presented and recommendations for a novel follow-up and navigation program for PV carriers will be outlined.

In partial fulfilment of the requirements of the degree of Master of Nursing, data from an integrative literature review and from a consultation plan/environmental scan were compiled for this practicum project. Together, the data served as the basis of program policy recommendations outlined in this proposal. The establishment of a provincial navigation & f/u program is the primary recommendation to improve health outcomes for the PV population in NL. Five sub-recommendations identify the essential program characteristics needed to remedy the current approach to PV carrier follow-up care in NL: 1) the establishment of a centralized, coordinated registry for all known PV and VUS carriers in the province who opt for genetic testing. 2) The development of a nurse navigator position that would have responsibility for coordinating screening and follow-up appointments, providing ongoing individual and family support, and connecting individuals with the necessary health care services. 3) The coordinated involvement of multidisciplinary healthcare providers in the provision of PV carrier follow-up. 4) The adoption of a person-centered, family approach to care and, 5) electronic and virtual infrastructure to support the delivery of this service, including remote service options for province-wide delivery of this service. Considerations surrounding the cost-effectiveness of the program, the suitability of this proposed program within the Canadian health care system, and methods for program evaluation will also be presented in this policy proposal.

Background to HBOC Syndrome

It has been nearly thirty years since the discovery of BRCA1, a tumor suppressor gene associated with hereditary breast and ovarian cancer (HBOC) susceptibility (Beamer, 2019). Since that time, there have been significant advances in research and clinical genetics. The completion of the mapping of the entire human genome (International Human Genome Sequencing Consortium, 2004), allowed for increased speed and accuracy of clinical genetic testing, while costs of testing have also significantly reduced (Bingham, 2012). As the area of genomics continues to rapidly advance, the identification and surveillance of PV carriers is an emanating priority health issue. This is particularly relevant in the context of HBOC syndrome, for which there are several options available to affected carriers, proven to reduce their risk of developing HBOC. With increasing availability of multi-genome sequencing technologies, health care policy makers must determine how to utilize genetic HBOC testing effectively and responsibly. They must also provide adequate guidance to affected carriers as they navigate the management of their risk. Thiruchlevam et al. (2018) accurately noted that "genetic testing can be life-changing and indeed life-saving, but it is crucial that it comes with all of the facts and appropriate professional support to enable individuals to live and plan for a healthy life" (p. 2091). It is an underlying assumption in this policy proposal that PV carriers can be identified, surveyed, and supported in a more suitable and cost-effective manner than the current process in the NL health care system.

'HBOC syndrome' refers to a predisposition to developing cancers of the breast and/or ovaries, including the peritoneum and fallopian tubes due to an inherited pathogenic variant (PV) with an autosomal dominance inheritance pattern (Gabai-Kapara et al., 2014). While all cancers occur due to genetic mutations, hereditary cancer occurs due to PVs that offspring inherit through one or two parents (National Comprehensive Cancer Network, 2019). HBOC syndrome is most often associated with PVs in the BRCA 1 & 2 tumor suppressor genes but has also been associated with up to 24 various inherited PVs (Nielsen et al., 2016). It is estimated that more than 20% of cases of ovarian cancer are associated with a hereditary predisposition syndrome (Susyznska et al., 2020). In a study of subjects with ovarian cancer, 24% of the ovarian cancer cases were associated with germline PVs; 18% were attributable to BRCA1 or BRCA2 and 6% were associated with PVs in the BARDI, BRIP1, CHEK2, MRE11A, MSH6, NBN, PALB2, RAD50, RAD51C genes (Walsh et al., 2011). Increased lifetime risk for ovarian cancer also occurs in the context of PVs associated with Hereditary Non-Polyposis Colorectal Cancer or Lynch Syndrome (LS) (Stuckless et al., 2007).

The estimated lifetime risk of cancer in PV carriers varies depending on the specific PV. For example, women who carry PVs in the BRCA1/2 genes have a 51-72% lifetime risk of breast cancer and an 11-44% lifetime risk of ovarian cancer (Kuchenbaecker et al., 2017; Rebbeck et al, 2015). This is in contrast with a 12% lifetime risk of breast cancer and a 1.3% lifetime risk of ovarian cancer before age 70 in the general population (National Cancer Institute, n.d). In another example, individuals who are carriers of PVs associated with Lynch Syndrome (MLH1 and MSH2) have a 4-20%

likelihood of developing ovarian cancer before age 70 when compared to an estimated 1-2% lifetime risk in the general population (Kohlman & Gruber, 2018).

Prevalence of HBOC PVs in Canada and NL

It is difficult to fully estimate the total prevalence of all PV carriers as genetic aberrations are continually being discovered and reclassified as pathogenic (NCN, 2019). The prevalence of BRCA 1 and 2 PVs in the general population is estimated at approximately 1 to 300 to 1 in 500 (Nelson et al., 2014). The Hereditary Breast and Ovarian Cancer Society (2018) estimated that 354,965 Canadians have been or will be diagnosed with a hereditary breast or ovarian cancer and as many as 709, 930 Canadians carry a known PV that predisposes them to hereditary breast and/or ovarian cancer. Roebothan et al. (n.d.) identified a total of 276 BRCA PV carriers in NL since the introduction of genetic testing in NL. While the BRCA PV carrier prevalence rate of 0.05% in the general NL population was lower than expected, they cited the current opportunistic genetic testing paradigm, resulting in the under-identification of BRCA carriers in the province, as a possible cause for their findings. Using population risk estimates of 1 in 300, Roebothan et al. (n.d.) estimated the prevalence of BRCA 1 & 2 PV carriers in NL to be upwards of 1,700. The low prevalence of BRCA PV carriers in NL could also be attributable to the fact that NL is considered a founder population, primarily of Irish and English descent (Zhai et al., 2016). Despite the low prevalence of BRCA PV carriers, NL has the

highest rate of breast cancer mortality and the fourth highest rate of ovarian cancer mortality in the country (Canadian Cancer Statistics Advisory Committee, 2019).

High rates of breast cancer mortality in NL may be related to findings of a recent case control study on molecular genetics of HBOC in NL. Dawson et al. (2020) performed multigene paneling on five female probands with a personal history of breast and/or ovarian cancer who tested negative for known high and moderate risk HBOC variants. However, they all shared a variant of uncertain significance (VUS) in the RAD51C gene. The results of a controlled haplotype analysis indicated a 52-fold increase of the RAD51C VUS in the NL population versus a general Caucasian population control (0.165% vs 0.0032%). From this, Dawson et al. (2020) concluded that the RAD51C(NM_058216.3: c.571 + 4A > G) variant is pathogenic. They suggested that this and other, yet unknown, variants may be responsible for the increased incidence and mortality associated with HBOC in NL (Dawson et al., 2020). The unique genetic composition of NL is characterized by the historically isolated nature of the island, an increased inbreeding coefficient, and reduced heterozygosity (Zhai et al., 2016). This has made the NL population more susceptible to the disease expression effects of founder mutations. As new evidence emerges surrounding the pathogenicity of HBOC genetic variants in both NL and in global populations, this will further the need for programmatic follow-up for these high-risk individuals in NL.

Incidence and Impact of Breast & Ovarian Cancer

Breast cancer is the most diagnosed cancer in females in Canada, accounting for 25% (26,900) of new diagnoses annually (Canadian Cancer Statistics Advisory Committee, 2019). Breast cancer accounts for 4,992 deaths annually in Canada (12.9% of cancer-related deaths in females) (Canadian Cancer Statistics Advisory Committee, 2019). While ovarian cancer is far less prevalent than breast cancer (3,000 new cases annually in Canada), it is the most lethal gynecologic malignancy and accounts for approximately 1,896 deaths annually (Canadian Cancer Statistics Advisory Committee, 2019). It is often described as a "whispering disease" (Martin, 2000, p.8) due to its insidious symptoms, with only 25-30% of women with ovarian cancer diagnosed in the early, highly treatable stages (Baldwin et al., 2011). Additionally, there has been no proven method of primary ovarian cancer screening effective in reducing ovarian cancer morbidity and mortality in the general population (Buys et al., 2011; Jacobs et al., 2016). It is because of the lack of primary ovarian cancer screening, coupled with the insidious onset of the disease, why it crucial to identify women with a high inherited risk for ovarian cancer and allow them to avail of targeted prevention strategies (Walsh et al., 2011).

HBOC Risk-Reduction Modalities

Much attention has been given to HBOC in the field of cancer genomics as there is strong evidence of the favorable impact of risk-reduction modalities on morbidity and mortality in asymptomatic BRCA PV carriers. Risk-reducing salpingectomy oophorectomy

(RRSO) has been associated with 80% reduction in ovarian/fallopian tube cancer risk and a 50% reduction in breast cancer risk in asymptomatic women with BRCA1 & 2 PVs (Rebbeck et al., 2009). Risk-reducing mastectomy (RRM) is also discussed with PV carriers as a potential risk-reduction intervention which has been shown to essentially eliminate the risk of breast cancer in asymptomatic female BRCA 1 &2 PV carriers (Li et al., 2016).

There are also breast screening recommendations for this high-risk population. Annual breast magnetic resonance imaging (MRI), alternating every 6 months with annual breast mammography has been shown to have a combined sensitivity of > 90% for detecting early stage breast cancer and is therefore recommended in this population (Warner, 2018). The Breast Disease Site Group (2017) of the Eastern Health (EH) Regional Health Authority (and NL tertiary care provider) recommended alternating annual MRI and mammography for women with an increased risk of breast cancer, starting at age 30. The Breast Disease Site Group (2012) also established a policy stating that premenopausal women ≥ 35 and postmenopausal women with a high risk of hereditary breast cancer should be offered oral Tamoxifen (a selective estrogen receptor modulator) once daily for five consecutive years. While this strategy may appear promising, many BRCA-associated breast cancers are estrogen-receptor negative, and therefore would not respond to Tamoxifen (Stadler & Kauff, 2009). Use of oral contraceptive medication for six or more years has been associated with decreased risk of developing ovarian cancer in BRCA1 &2 carriers (OR 0.62, 95% CI 0.35-1.09) (Whittemore et al., 2004). While oral contraceptives reduce the risk of ovarian cancer, there is evidence that the use of oral contraceptives may

increase breast cancer risk (Narod et al., 2002). In essence, decisions surrounding HBOC risk management are complex and can cause psychological distress to PV carriers and their families. Coupled with burdensome treatment decisions, PV carriers may struggle with alterations in self-image, the disclosure of this information to family members, and with the family-planning implications of having a hereditary condition (White et al., 2014). With the increased availability of genetic testing, health care providers must provide support and guide PV carriers make risk-management choices most suited to their life circumstances (Landsbergen et al., 2009).

Issues with the Current HBOC Follow-up Process in NL

Referrals to the NL provincial medical genetics program (PMG) are made by a primary care provider such as family doctor or nurse practitioner, or by a specialist physician when there is a suspicion of HBOC syndrome. Criteria for referral may include: a strong family history of disease, a first-degree relative of a known PV carrier, or other clinical features that are suggestive of a hereditary condition, such as early age of onset or breast tumors that test negative for estrogen, progesterone, and HER 2 receptors (triple negative) (Morrison et al., 2016). Ovarian cancer patients are also offered genetic testing, as the confirmation of a hereditary predisposition would confirm the indication for poly-ADP ribose polymerase (PARP) inhibitors as a treatment option. PARP inhibitors have proven effectiveness in the treatment of BRCA 1 and BRCA 2 associated ovarian cancer (Fong et al., 2009). Once an individual is referred to the PMG, genetic testing is

exclusively delivered by medical genetics specialists after in-person counselling (Adams & Etchegary, 2015). There are significant wait times in NL to be seen by PMG. Hynes et al. (2020), conducted a quality improvement project in NL to improve the wait times to be seen by PMG for investigation of potential cancer predisposition syndromes. Using a novel model of group genetic counselling plus "mini" individualized sessions, they successfully reduced wait times from nearly three years down to one year. Still, with a backlog of referrals to PMG, any chance of long-term supportive care from genetic counsellors in the current provincial financial climate is low. With no care provider systemically following up on how PV carriers are understanding and applying the genetic information they have been provided by PMG, it is difficult to ascertain to what degree this information has been beneficial to them. This is regrettable as the purpose of offering genetic testing and counselling is to empower people to lead a healthier, longer life vis-à-vis their carrier status.

Following the immediate disclosure of genetic results, the navigation of annual screening, complicated treatment decisions, and family planning considerations are left entirely in the hands of the individual and their primary care provider (Roebothan et al., n.d.). Dr. Lesa Dawson, a gynecologic oncologist and Associate Professor of Women's Health and Genetics at Memorial University of Newfoundland, described BRCA PV carriers in the province as being "orphaned by the healthcare system" (Mercer, 2018, para 2). Other than the work done through Dr. Dawson's gynecologic oncology inherited cancer prevention clinic at Memorial University and a small group of medical oncologists who offer annual follow-up, there is no systematic follow-up for these women in the

province, resulting in a significant missed opportunity. In a retrospective study, Roebothan et al. (n.d.) noted that only 41.6% of eligible BRCA PV carriers in NL had undergone the recommended annual MRI screening in an 18-month period ($p < 0.001$). While proven effective to prevent disease, the recommended modalities are still largely underutilized by HBOC PV carrier populations who stand to benefit from them. In the literature review and key informant interviews, several themes emerged to suggest that in the province, and in other health systems around the world, many of the current PV follow-up processes are inadequate. These themes included: carriers' unmet information needs, primary care provider centered barriers, lack of coordination of care, lack of quality assurance, carriers' unmet psychosocial needs, and issues surrounding disclosure to families. These issues are translating into adverse and avoidable health outcomes in PV carriers. Furthermore, these issues are resulting in complex, advanced cancers that require costly, aggressive treatments. These themes are examined in depth in this section.

Unmet Information Needs

A prevailing finding in the literature review was that many HBOC PV carriers around the world are not getting their information needs met by the current HBOC follow-up process. In one example, HBOC PV carriers made erroneous statements when asked about their perceptions of risk-management (Cherry et al., 2013; Hughes & Phelps, 2010). PV carriers noted the lack of a central, up-to-date research hub where they could retrieve reliable medical and research updates about HBOC, such as a e-newsletter, or

other type of online resource (Hughes & Phelps, 2010). While these findings were not universal among all the studies, they highlight the reality that many women are not given the clear information to make a truly informed decision about HBOC risk-management.

The decision to undergo RRSO comes with its own gamut of considerations; to undergo RRSO with or without hysterectomy, hormonal replacement therapy versus nonhormonal treatments for symptoms of premature menopause, as well as interventions to optimize bone and cardiovascular health (Walker et al., 2019). The medical and psychosocial needs of PV carriers considering RRSO and RRM are complex and these women often require personalized support in their risk management. Yet, in a study by Etchegary et al. (2015), conducted in an NL population, premenopausal women who underwent RRSO reported that they did not have an adequate understanding of the full extent of surgical menopause prior to surgery. As a result, they felt overwhelmed and unprepared when these menopausal symptoms occurred. It is important to note that while RRSO can be lifesaving, health care providers and health care delivery systems must take the necessary steps to minimize the potential iatrogenic harms of these procedures.

In the key informant interviews, another common issue reported with respect to unmet information needs is that the current follow-up process does not account for how PV carriers' needs change over time. Genetic counsellors and specialists noted that PV carriers do receive detailed information with a printed copy for their records as part of the immediate genetic testing results disclosure. Still, individuals are not always psychologically prepared to 'readily absorb' this information at the time of disclosure. Their information needs may also change over time; for example, if women have

completed their childbearing years since the time of carrier status disclosure, their readiness for risk-reducing surgery may have changed (Manchanda et al., 2012). It is also difficult to fully ascertain the individual's level of health literacy and understanding of the information provided in a single session with a genetic counsellor. If an individual does not fully comprehend the information they are presented with, this will influence their value appraisal of and adherence to risk reduction recommendations.

With no dedicated program following these individuals and no readily available resource person available to answer their questions, the information provided at the time of genetic results disclosure may be cast aside or poorly understood when it could be in use to empower individuals to live a longer, healthier life.

Primary Care Provider-Centered Barriers

Reiteratively, in many current Canadian health care systems, the navigation of considerations specific to their PV carrier status becomes the responsibility of the individual and/or their primary care provider (Roebothan et al., n.d.). Yet in literature review findings, several study participants voiced that they felt as though they were the ones guiding their primary care provider in their HBOC journey, as the information provided by their primary care provider was not always accurate nor reliable (Cherry et al., 2013; Leonarcyzk & Mawn, 2015; Watkins et al., 2011). PV carriers reported challenges in obtaining relevant HBOC risk-management information from their primary care

provider, especially about subjects considered taboo such as the potential adverse sexual implications of HBOC risk management (Cherry et al., 2013).

This theme was further probed in the consultations that were conducted with key stakeholders prior to the development of this policy. Informants attributed NL primary care barriers to the fact that there is no central, trusted site or authority responsible for getting this information into the hands of primary care doctors (and NPs). This lack of central authority also means a lack of health care system accountability when these recommended screening tests for PV carriers are not being ordered. Some PV carriers may opt not to proceed with further surveillance for personal or family reasons, as is their right. However, there are instances where individuals are agreeable to the recommended surveillance, but for whatever reason, it is not being ordered by their primary care provider. A GP reported that in his experiences, patients tend to be over-reliant on their primary care providers to remember and coordinate all screening appointments. He added that his 'greatest fear was that [he] will miss a screening and early detection will be missed'. In the context of a busy family practice with no centralized hereditary cancer screening reminders, it is easy enough for a GP to overlook a preventative screening referral. An oncologist agreed that with busy family practices and screening/surveillance recommendations for PV carriers that change rapidly, it is unrealistic to expect the onus of arranging specialized follow-up care to lie completely with the primary care provider. It was also noted that in NL there are high turnover rates of primary care providers and many individuals in the province are without a primary care provider. In essence, the

systemic overreliance on GPs to coordinate follow-up creates an environment in which there are many cracks in the system through which PV carriers can slip.

The Lack of Coordination of Follow-Up Care

In the literature, there was confusion reported by PV carriers about inconsistencies in medical advice and surveillance recommendations from various members of their health care team (Caita-Zufferey et al., 2015; Cherry et al., 2013; Watkins et al., 2011). This caused PV carriers to feel overwhelmed and frustrated by the sometimes-conflicting advice they received (Caita-Zufferey et al., 2015). A similar concern was echoed in a focus group of breast care providers who noted the high probability of HBOC PV carriers being missed in the disintegrated lines of communication involving multiple health care providers (Komatsu & Yagasaki, 2014). Watkins et al. (2011) noted that breakdowns in lines of communication in Lynch Syndrome management were most obvious between medical specialists and primary care providers in NL. It was also reported that scarcity of health care providers, particularly in rural regions, posed challenges to adherence to recommended screening modalities (Leonarcyzk & Mawn, 2015). Moreover, with limited resources, breast care providers noted that they had limited time to focus on preventative measures when they were dealing with active of cases breast cancer and thus hereditary breast cancer prevention was placed lower on their list of priorities (Komatsu & Yagasaki, 2014).

In the consultations, some informants had been involved in research with individuals living with inherited cancer syndromes in NL. A common theme reported in their work with PV carriers was that carriers desired a coordinated, 'one-stop' clinic that addresses and schedules their risk-reduction follow-up. PV carriers reported very practical issues with adherence to recommended screening, such as having to travel long distances and take multiple days off work to attend to the various screening and medical appointments. One informant noted that in her work with PV carriers (and their family members for that matter), she found that 'life gets in the way' of risk management. It was easy for asymptomatic PV carriers to forget or disregard the multitude of screening appointments in the context of hectic everyday life. For some, all these appointments became so overwhelming and mentally taxing that they had to stop for a while. This was particularly common for individuals with Lynch Syndrome where there is an increased risk of inherited cancer in multiple organs and thus multiple screening modalities are recommended (Watkins et al., 2011).

Lack of Quality Assurance

Closely related to issues in care coordination is the absence of quality assurance measures in place for known PV and VUS carriers. In the absence of an electronic health registry to monitor PV carriers and coordinate surveillance programs through systemic tracing, an important opportunity is lost. Marrow et al. (2013) conducted a systematic analysis of the impact of registration and screening on colorectal cancer incidence and

mortality in familial adenomatous polyposis (FAP) and Lynch syndrome (LS). They concluded that there was high quality evidence of the significant benefits of colorectal cancer morbidity and mortality rates in individuals who participated in hereditary colorectal cancer registries. They also outlined the plethora of organizational, patient-focused, and researched focused benefits of hereditary colorectal cancer registries. Currently, there is no genetic registry of this kind in NL.

In a policy framework document on cancer control, the NL Department of Health and Community Services (2010) proclaimed the need to "develop a hereditary cancer screening program ensuring best practice and necessary support, infrastructure (e.g. genetic counseling) and resources" (p.13). Patient registries are currently in use in the Provincial Cancer Care Program. The Newfoundland and Labrador Cancer Care Registry (NLCCR) is an authorized registry under the Provincial Cancer Care program. The NLCCR includes five registry subprograms: breast screening, cervical screening, colon screening, tumor surveillance, and chemotherapy surveillance (Eastern Health, 2016). Despite the existence of the NLCCR and its mandate to improve cancer outcomes, a hereditary cancer PV carrier registry has not been included in this provincial cancer registry; this is an egregious missed opportunity.

Another common scenario that is being overlooked in the current system is when individuals undergo genetic testing and are found to carry a variant of uncertain significance (VUS). In clinical genetics, gene variants are classified on a five-item tier, ranging from benign to pathogenic (Richards et al., 2015). VUS fall in the middle of the tier and to date, neither the pathogenicity nor the benign status of the variant can be

confirmed. It is currently contraindicated to recommend any clinical actions in the context of a VUS result from genetic testing (NCCN, 2019). Still, empirical evidence surrounding variant pathogenicity is likely to expand as more gene sequencing and testing is being carried out. This will likely lead to reclassification of certain variants; either as more likely pathogenic or benign (Richards et al., 2015). Informants from PMG reported that if a person is found to have a VUS, thy are encouraged to follow-up in two to three years' time to see if the status of their VUS has changed. This opportunistic approach leaves no process in place to monitor these individuals or to inform them if the status of their VUS has changed. If VUS carriers were included in a carrier registry, this could streamline them with up-to-date information and connect them with the recommended clinical decision-making tools, should they become available. Relying on individuals to follow-up on their own variant status is ineffective and leaves far too much to chance.

Unmet Psychosocial Needs of PV Carriers

Beyond issues of medical surveillance is the consideration that PV carriers are holistic beings with unique psychosocial needs. In the consultations, it was noted by an informant that 'there is an ethical responsibility to address carriers' emotional and mental health needs.' Yet, qualitative data in the literature review revealed that in their interactions with health care providers, many PV carriers felt they were "not being seen as a whole person" (Leonarcyzk & Mawn, 2015, p.77). To narrowly focus on risk management

and screening does not address the full complement of implications of being a PV carrier. An informant explained that many HBOC families experience severe psychological trauma, having witnessed many of their family members die at an early age while facing the uncertainty of their own inherited risk. Yet, in the current system, their unique concerns and perspectives are largely overlooked. There is no program or healthcare professionals available to assist them with communicating genetic results with their families, counselling services, or the opportunity to network with other carriers and high-risk individuals.

It is interesting that a consultee noted that in her work with PV carriers, higher levels of 'family support and cohesion' were associated with improved psychosocial adjustment to their PV carrier status and improved adherence to the recommended screening and risk-reduction modalities. Yet there is no outlet in the current follow-up process to encourage the participation and involvement of family members in risk management and psychosocial adjustment.

Issues Surrounding Family Disclosure

As it currently stands in NL, individuals with a confirmed PV are responsible for notifying their at-risk relatives that they may also be at increased risk. Several informants in the stakeholder interviews expressed that this responsibility is an unreasonable burden to place on a PV carrier. Moreover, the reliance on PV carriers to recruit other at-risk relatives has created significant gaps in the way that at-risk individuals are notified, and

many at-risk individuals are likely being missed in this line of family communication. Some PV carriers reported feelings of guilt and/or resentment when one family member carried the gene and another did not and opinions often greatly varied among family members about whether or not PV carriership should be disclosed (Hughes & Phelps, 2010). It has been reported that the unearthing of this information can cause tensions between parents and children. (Dwyer et al., 2020). Probands have described feeling conflicted in their sense of duty to inform relatives of their potential risk with their desire to protect family members from the worry stemming from the disclosure of that information (Croster & Dickerson, 2010). Evidently, there are several social, ethical, and legal factors to consider when addressing these gaps. There must be a balance between the risk mitigation and the privacy, confidentiality, and right to autonomy of both probands and their family members. A master's student in the clinical epidemiology program at the Faculty of Medicine at Memorial University in Newfoundland is currently working on a research project to explore optimal means of family outreach in high-risk families. This will provide insight on how health care delivery systems should facilitate family risk disclosure and/or support PV carriers in the family risk disclosure process. It is an assumption in this policy paper that the findings of that research could have implications for the novel PV carrier follow-up program. Based on the findings, it could provide direction for how the PV carrier navigator and the multidisciplinary team could work with families to discuss their risk status and the options available to them, as well as provide psychosocial support to the unique needs of at-risk families.

A Policy for a Dedicated HBOC Follow-Up and Navigation Program

In examination of the evidence collected for this MN practicum project, it is safe to conclude that there are glaring and detrimental gaps in PV carrier surveillance and support in NL. It is put forward in this proposal that the best way to address these needs is through formulation of a dedicated follow-up and navigation program for PV carriers in the province. Informed by the evidence, there are several key features for this novel program that are recommended:

1) ***A provincial centralized, coordinated PV and VUS carrier registry***
2) ***Establishing the role of a nurse PV carrier navigator***
3) ***A coordinated multidisciplinary team approach to the care of individuals and families in the program***
4) ***A person and family centred approach to follow-up care***
5) ***Electronic and virtual infrastructure to support provincial delivery of the program***

It is important to note that several features of the proposed program have been influenced by several extant programs that function successfully. These include patient navigation programs, patient registries, the Ontario High-Risk Breast Program, and dedicated multidisciplinary hereditary cancer follow-up clinics. To understand the applicability of these features in the conceptualization of the dedicated follow-up and navigation program, this section begins with an overview of these existing programs.

Patient Navigation Programs

Patient navigation programs were introduced 30 years ago as a model to reduce the health system inequities experienced by socioeconomically disadvantaged individuals in their access to early detection and treatment of cancer (Freeman, 2012). Since that time, the scope of cancer patient navigation programs has expanded to include prevention, detection, diagnosis, treatment, survivorship, and end of life care. (Freeman, 2012). One of the main implications from the Cameron Inquiry Report into Breast Hormone Receptor Testing in NL was the "importance of communication and coordination of care throughout the cancer journey, recommending patient navigators to assist patients" (Department of Health and Community Services, 2010, p.13). There is currently a cancer patient navigation program in the province of NL (Eastern Health, 2018). In this program, cancer patient navigators are nurses with specialized training in oncology, with offices located in seven areas throughout the province (Eastern Health, 2018). They offer a variety of services to cancer patients such as providing ongoing support and information, coordinating appointments, and connecting patients with relevant specialists and community resources. Patients can be self-referred to the program, or by their family doctor, other care provider, or cancer specialist (Eastern Health, 2018). Three other patient navigation programs are also offered in NL for palliative care patients, indigenous peoples, and individuals with mental health and addictions issues, in order to meet their unique needs and reduce the barriers they encounter (Eastern Health, n.d.; Eastern Health, 2019; Eastern Health, 2020). The principles of patient navigation, coupled with the ways that these programs are in use for

other groups in the province, demonstrate how this model is applicable to a PV carrier sub-population. Patient testimonials on the cancer patient navigation program in NL have been overwhelmingly positive (Eastern Health, n.d.). Nurse navigators provide patients with accurate information that is easily understood, facilitate continuity of care, and provide a mentally and emotionally supportive health care experience (Baileys et al., 2020; Winstead, 2012). For breast cancer patients in particular, the nurse navigator model of care has been shown to empower patients, to encourage them to self-advocate, and to practice self-care in their breast cancer journey (Trevillon et al., 2015).

Patient Registries

The Newfoundland and Labrador Cancer Care Registry (NLCCR) received authorized registry status in 2017 ("Newfoundland and Labrador Cancer Care Registry Launched by Eastern Health", 2017). The NLCCR includes five registry subprograms: breast screening, cervical screening, colon screening, tumor surveillance, and chemotherapy surveillance (Eastern Health, 2016). When it was introduced, Eastern Health (EH) CEO David Diamond stated that the NLCCR was an important part of EH's ongoing efforts to provide "access to information, education and tools that would lead to preventing disease before it starts and detecting it earlier for the benefit of our patients" ("Newfoundland and Labrador Cancer Care Registry Launched by Eastern Health, 2017, para 5). According to Eastern Health (2016), the aims of the NLCCR include to improve clinical decision making and the delivery of care and screening programs. Given these

mandates and the wealth of research evidence in favor of PV carrier registries, a hereditary cancer PV carrier registry is an unmistakable fit for the NLCCR. Hereditary cancer registries are in use around the world and the evidence of their effectiveness is strong (Barrow et al., 2013). The benefits of hereditary cancer registries to the health system include the easily retrievable storage of genetic and demographic information, an efficient way to trace and recruit other at-risk relatives into surveillance programs, the coordination of screening, and the monitoring of PV carrier follow up (Barrow et al., 2013). Barrow et al. (2013) also noted several patient-focused benefits of carrier registries such as the continuity of care, psychosocial benefits of involvement, screening reminders, and opportunities to participate in clinical trials. On that note, PV carrier registries also provide an unrivaled opportunity to promote and conduct research and to improve the current screening and risk-reduction protocols (Vasen et al., 2016).

Ontario High-Risk Breast Screening Program

As part of the environmental scan conducted for this project, the Ontario High-Risk Breast Program (High-Risk OBSP) was examined. The High-Risk OBSP is the only provincial breast screening program of its kind for high-risk women in Canada. To be enrolled in the High-Risk OBSP, a referring physician must submit a requisition form to a designated High-Risk OBSP site (Cancer Care Ontario, n.d.) It is implicit in the program requisition form that the ordering physician is requesting future MRI imaging and in some cases, image guided biopsies, which under current Ontario regulations requires a

physician's signature (Cancer Care Ontario, n.d.) Women eligible for the program fall into category A or B. Women in Category A are directly enrolled in the program because they either carry a known PV associated with increased breast cancer risk, or are a first-degree relative of someone with a known PV and underwent genetic counselling but opted not to have genetic testing (Cancer Care Ontario, n.d.) Women in Category B require further genetic assessment prior to enrollment in the program; this includes individuals who are a first degree relative of someone with a known PV who have not been assessed by a genetic counsellor. Following the genetic assessment, it is determined whether the woman in Category B is enrolled in the High-Risk OBSP. Cancer Care Ontario (n.d.) outlined that the High-Risk OBSP program is delivered by High-Risk OBSP navigators with a list of responsibilities. These include booking the screening and breast assessment appointments, following up on abnormal results, arranging annual recalls, and communicating all imaging results to women and the referring physician. The effectiveness of the High-Risk OBSP has been reported and confirmed in the research literature (Chiarelli et al., 2014).

Multidisciplinary Follow-Up Clinics

Outcomes in dedicated HBOC multidisciplinary follow-up clinics have been reported in the literature (Bancroft et al., 2010; Firth et al., 2011; Pichert et al., 2010; Yerushalmi et al., 2016), and Engel et al. (2012) provided an overview of the program structure of a dedicated BRCA follow-up clinic in the US. Yerushalmi et al. (2016) found

that of the BRCA PV carriers who attended a dedicated BRCA follow-up clinic for biannual screening and follow-up appointments, only 7.2% developed cancer (17 were breast cancers, one ovarian cancer, and three were additional cancers.) Of the 17 cases of breast cancer, 94.1% of those cancers were detected at stage I disease when treatment outcomes are generally far more encouraging; 70.6% of those cancers were detected by MRI and 17.6% were detected by mammography (Yerushalmi et al., 2016). Clinic outcomes were not compared with outcomes from a matched control of a family physician based BRCA follow-up model. Therefore, it is indeterminable if the low incidence of malignancy occurred as an exclusive result the dedicated follow-up clinic. Still, the rate of RRSO uptake at age 40+ at the clinic in Yerushalmi et al. (2016) was high at 87.3%. This high uptake rate of RRSO in clinic attendees was higher than in most other reported registries and in the literature (Yerushalmi et al., 2016). The median age at the time of RRSO in the multidisciplinary clinic (46.5, 33-68) was also lower than the median age at time of RRSO of 49.6±9.7 reported in NL BRCA PV carriers (Roebothan et al., n.d.). Yerushalmi et al. (2016) provided a strong case that a "dedicated, multidisciplinary clinic for BRCA mutation carriers provides a home for a unique population with unmet needs" (p.551). PV carriers have reported high levels of satisfaction with multidisciplinary BRCA follow-up clinics (Firth et al., 2010). BRCA PV carrier participation in a dedicated follow-up clinic was associated with significantly greater participation in related clinical trials ($p < 0.001$) (Pichert et al., 2010).

As part of the environmental scan, several websites of familial/hereditary cancer clinics were examined. These clinics were comparable to the multidisciplinary clinics

reported in the literature (Bancroft et al., 2010; Engel et al., 2012 ; Firth et al., 2011; Pichert et al., 2010; Yerushalmi et al., 2016). Comparable to PMG, genetic counselling and genetic testing referral are offered through many of these centers. Also consistent with the services at PMG, immediate genetic testing follow-up and personalized genetic risk assessment are also offered. How the multidisciplinary clinics differ from PMG is in their approach to surveillance, follow-up, and support. Many of these centers feature multidisciplinary teams including geneticists, genetic counsellors, nurses, dieticians, gynecologists, oncologists, social workers, and psychologists. With this wide variety of medical professionals, the multiple facets of PV carriership are appropriately addressed. A few of these clinics/programs also offer periodic carrier support groups and sessions where PV carriers can liaise with other PV carriers and families. They also have opportunities for PV carriers and families to attend support sessions with guest speakers and genetic/hereditary cancer experts. Many of these programs also differ from the current PMG model in that PV carriers are scheduled to visit the clinics annually or bi-annually for surveillance, follow up, and supportive care. In the province of Ontario, many of these clinics work with the High-Risk Ontario Breast Screening Program (OBSP) to coordinate breast surveillance of eligible high-risk individuals.

In short, features of these four existing programs have been borrowed in the development of this program policy proposal. This will be evident going forward as the five recommended components of the novel follow-up and navigation program in NL are delineated below:

1. A Centralized PV Carrier Registry in NL

Under the prospective program, the processes following disclosure of genetic testing results will be significantly modified. Individuals who undergo genetic testing through PMG and are found to carry pathogenic variants, likely pathogenic variants, or VUS will be entered into a provincial registry. A PV carrier registry is well-suited to fall under the umbrella of the NLCCR, as it fits with the mandate of the NLCCR and under the umbrella of cancer prevention. Consent to use information is not collected within the current five subgroups of registries in the NLCCR; this ensures the accuracy and completeness of the data so it is being used to properly inform clinical decision making and health care delivery (Eastern Health, 2018). It should be reviewed with the health authority ethics review board if non-identifiable PV carrier data can be collected in this registry with presumed consent, as is the case with the other subprograms of the NLCCR. Invariably, the exception to this would be, as is the case with the cancer screening programs currently under the NLCCR, individuals would have the option not to participate and/or receive communication from the PV carrier follow-up program. This will be further discussed later in the proposal. Individuals in the PV carrier registry will also have the option to be notified if they wish to participate in research studies and clinical trials. Participation in these studies and trials would be completely voluntary.

As is the case with current programs under the NLCCR, custodians of the personal health data in the PV carrier registry would be held accountable to the legally binding obligations of authorized registries under the Personal Health Information Act (2008) (PHIA). Custodians of the PV carrier registry data would be held to stringent expectations

as they pertain to the collection, storage, and access to personal health data in the registry. However, these expectations would be comparable to the expectations of current data custodians of the NLCCR and of other NL Health data IT systems users. Still, prior to launching this registry, transparent and clearly described policies are needed regarding the collection and use of genetic data. A transparent governance structure and clearly described policies are necessary to ensure that the expectations of privacy and confidentiality can be forthright regulated. A key informant recommended that all policies and patient documents should also be informed by ethics experts, as well as by the literature in recent years related to genomic medicine that 'begins to highlight patient data concerns that can be identified and mitigated.'

Health care professionals involved in the delivery of the PV carrier follow-up program would be included under custodians of the data in the PV carrier registry. A registry system would also be a quality assurance measure for PV carriers who are otherwise being missed for screening in the current family-physician dependent follow-up model. It would be impossible to deliver a HBOC follow-up program without access to the registry data and the registry data should be compatible with provincial electronic medical records. Part of the role of the PV carrier navigator would be to periodically index the registry to ensure that the classifications of the variants are up to date, as are the recommended clinical actions associated with the variant. This should be done as part of team of genetics and hereditary cancer experts. The registry must be subject to periodic reviews and quality/audit controls. For carriers participating in the follow-up program, the registry must be updated as PV carriers complete the recommended screening

intervals and/or opt for risk-reduction surgery. The information included in the registry should include demographic information, information on the pathogenic variant, information on the testing panel ordered, and information on the recommended clinical actions for the variant. The data in the registry should be compatible/accessible within the electronic medical record software that will be used by the nurse navigator when providing care to individuals enrolled in the follow-up program. This data should also be linkable with electronic pedigree data collected from PMG. Specific recommendations on the health IT infrastructure needed to deliver the program will be explored in further detail in subsection five.

2. Establishing the Role of a Nurse PV Carrier Navigator

During genetic results disclosure at PMG with a genetic counsellor, PV carriers would be given the option to enroll in a follow-up program for PV carriers. PV carriers would have the option to withdraw from correspondence with the program at any time and/or return without any repercussions or coercion. Central to this policy proposal is the need to establish a position for a Registered Nurse or Nurse Practitioner who assumes the role of PV Carrier Navigator for individuals in the province. This individual should have experience and/or certification in cancer genomics to be ideally suited to take on this position. Komatsu and Yagasaki (2014) proposed that specialized cancer nurses are ideally suited to utilize genetic health data in electronic health records and to coordinate communication among multidisciplinary HBOC team members, including the

coordination of multidisciplinary team meetings. Komatsu and Yagasaki's (2014) postulate is consistent with this key policy proposal recommendation. The primary contact person/administrator of this program will be the nurse PV carrier navigator. (S)he will have access to the data in the registry and will be responsible for establishing annual or bi-annual contact with PV carriers. This contact will be arranged either via telephone, online video conferencing, online messaging, or in-person depending on the carrier's geographic location and/or personal preference.

In the annual/bi-annual outreach to carriers, the nurse navigator would discuss with them and assess how they are coping with the multiple dimensions of their carrier status. This would include assessing their readiness for risk-reduction surgery if applicable, providing education and follow-up on their inherited risk, and risk management. The nurse navigator would also be responsible for notifying individuals when new therapies, recommendations, and clinical trials are available to take part in that may be relevant to them. The nurse navigator would also assess the psychosocial impact of carriership on individuals and families and provide them with support and appropriate referrals as needed.

As stated, a research project is currently underway in the province to determine how to appropriately notify a proband's relatives of their potential inherited risk. The challenge of this process, known as cascade testing, is balancing the privacy and confidentiality of the probands with the sense of duty to inform other at-risk relatives. Moreover, some individuals may not want to know about their inherited risk. Facilitators and barriers to cascade testing have been explored in the literature (Dwyer et al., 2020;

George et al, 2015; Schwiter et al., 2018). The consensus is that probands seek and value the input of health care professionals in the cascade testing process when communicating this information with their relatives (Schwiter et al., 2018). Similarly, key informants in the consultations expressed that it is unreasonable to place the entire responsibility on the probands to notify relatives of their risk. Several informants expressed that a dedicated follow-up program, as described in this proposal, would be ideally suited to assist with at-risk relative recruitment/notification as part of its gamut of services offered. For this program, it is put forward that family risk disclosure sessions should be facilitated by the nurse navigator and a social worker or psychologist. In the family disclosure process, the role of the nurse navigator is to provide families with information on the implications of their relative's carrier status and what it means to them in a way that is easily understood. If desired by at-risk relatives recruited through the program, the nurse navigator will notify a genetic counsellor at PMG to coordinate cascade testing and genetic counselling. The electronic medical record systems in use should be compatible between the nurse navigator, PMG, and other HBOC team members to allow a seamless flow of communication.

Another key role of the nurse navigator will be to coordinate and facilitate breast screening appointments in eligible PV carriers. Currently, women at high risk of breast cancer in NL are referred back to their primary care provider for further management (Canadian Partnership Against Cancer, 2018). Under the proposed policy, this responsibility will be re-delegated to the nurse navigator. This service will draw heavily on the model used in the province of Ontario for the High-Risk Breast Ontario Breast

Screening program. The nurse navigator will determine if an individual is eligible for the annual magnetic resonance imaging (MRI) recommended for individuals at high risk of breast cancer. As per the Eastern Health Breast Disease Site Group (2017) policy, to be effective, breast MRI must be timed within days 5 to 13 of the menstrual cycle in premenopausal women and must be alternated every six months with mammography. Therefore, this takes coordination, and these women must be streamlined and prioritized in the health care system. The nurse navigator will send a requisition form either to a breast specialist or to the individual's primary care provider to sign off on. The completed requisition form is then sent back to the nurse navigator with the assumption that the ordering care provider has requested future MRI imaging to be facilitated and coordinated by the nurse navigator. The nurse navigator would also be responsible for booking patients with follow-up breast assessments with medical oncologists following abnormal screens, informing PV carriers of the results of the screening, and arranging annual recall for women who are due for their next scheduled MRI. Ideally, the nurse navigator will be in contact with the PV carrier in the few months leading up to when she is due for her MRI. This will allow the nurse navigator to facilitate the annual check in with the woman, confirm the timing of her menstrual cycle if applicable, and assess her readiness for screening based on her life circumstances.

3. A Multidisciplinary Approach to Care

There was overwhelming evidence from the literature review, consultations, and the environmental scan that PV follow-up care must be delivered from a multidisciplinary standpoint. While the nurse navigator will be the consistent, primary point of contact for individuals enrolled in the program, (S)he will also connect carriers with other multidisciplinary team members as needed. For example, the navigator will connect women with breast specialists in the event of an abnormal screening result. The navigator will also connect individuals with surgeons such as gynecologists, breast surgeons, or plastic surgeons if they indicate their interest in risk-reduction surgery. For individuals with Lynch Syndrome, this may include referrals to gastroenterologists and general surgeons. Moreover, if women are experiencing issues with the symptoms of surgical menopause following risk-reduction surgery, the navigator will connect them with medical professionals who can prescribe therapies to alleviate these distressing symptoms. Once an individual is referred to these team members, it is at the discretion of the physicians whether they continue to see the patient on a regular basis. Consistent communication between all HBOC team members will be essential. The nurse navigator may use their discretion to determine when an individual may benefit from a specialist referral, provided they consent to do so. Some familial cancer clinics listed in the environmental scan also connect PV carriers with dieticians and nutritionists. As it is established that maintaining a healthy weight is a protective factor against cancer, dietician referrals may be appropriate for some PV carriers as part of a holistic, comprehensive cancer prevention strategy. In this model, the nurse navigator may be

ideally situated to facilitate referrals to other programs, for example, the smoker's help line.

The nurse navigator will also connect individuals with appropriate multidisciplinary team members who focus primarily on their psychosocial needs. If there are issues adhering to the recommended screening and risk-reduction modalities due to social and economic circumstances, the navigator will connect PV carriers and families with social workers and social work assistants. As an example, social work team members may arrange transportation/accommodation assistance with travel to specialist and screening appointments for PV carriers who may otherwise be unable to attend. They may liaise with the PV carriers' existing social workers in the community where applicable. In these ways, these multidisciplinary team members reduce fragmentation and barriers that women may encounter in their PV carrier journey.

Social workers and psychologists may also work with the nurse navigator in the family disclosure process. There is a wide body of evidence that PV carriers desire the input of health care professionals when disclosing this information with family members. Croster and Dickerson (2010) noted that the 'receivers' require sensitivity to the impact of learning about their inherited risk, time to process the information, and supportive and informational resources. Otherwise, this can be an extremely distressing experience for them upon realizing they are in a high-risk family. The role of the nurse navigator in the family risk disclosure process was outlined in a previous section. The role of the social worker/psychologist in the disclosure process will be to moderate and focus on how this information is being received, to encourage expression of emotions, and to assess and

encourage support systems in place. In a study by McInerney et al. (2005), individuals who underwent BRCA PV testing and were randomized to a client-centered counseling intervention reported a decrease in conflict among family members (p=0.006). Having a social worker/psychologist present with the nurse navigator is an ideal way to communicate risk while mitigating the potential iatrogenic distress that the revelation of this information may cause. The psychologist/social worker may use their discretion to determine if further family counselling and supportive sessions are required.

As part of providing a multidisciplinary approach to care, periodic group psychoeducational sessions for PV carriers and families should be offered as part of this program. In psychoeducational sessions, multidisciplinary team members present new research findings and updates and there is also opportunity for PV carriers and families to seek formalized peer support. Psychoeducational group sessions for PV carriers have been widely examined in the literature (Corines et al., 2017; Esplen et al., 2004; Kwiatkowski et al., 2019; Landsbergen et al., 2009; Listøl et al., 2017; McKinnon et al., 2007). These group interventions featured both psychosocial and medical content pertinent to HBOC PV carriers, such as breast reconstruction surgeries, genetic insurance discrimination, and family communication about genetic testing. Some of these groups occurred as a one-time retreat, as multiple sessions over several months, or annually. Quantitative measures of participant satisfaction, percentage of unmet information needs, and psychological distress improved following participation in HBOC psychoeducational groups. Several PV carriers have reported that both professional and peer support was vital to them when making decisions about their HBOC management. (Cherry et al., 2013; Phelps & Hughes,

2010; Rauscher & Dean, 2017). Bringing PV carriers together for these psychoeducational groups allows them to establish a sense of normal through shared experiences, the opportunity to learn through the experiences of others, as well as improved coping (Landsbergen et al., 2010).

In pre-pandemic circumstances, an annual ovarian cancer educational symposium takes place at Memorial University of Newfoundland that is largely funded by the Belles with Balls charitable organization and by community sponsors. Belles with Balls supports the ovarian cancer research and education fund (OCRE) at the Faculty of Medicine at Memorial University (Belles with Balls, 2020). The 2019 symposium focused primarily on BRCA and hereditary cancer modules. Multidisciplinary team members spoke about medical and research findings and a BRCA PV carrier shared her personal experiences as a carrier. It is asserted in this proposal that the Belles with Balls are a vital community partner for this follow-up program to facilitate multidisciplinary education sessions. If content relevant to PV carriers is included in future symposiums, the nurse navigator should extend invites to PV carriers in the follow-up program to attend these symposiums. In so doing, PV carriers can benefit from the relevant information, medical updates, and peer support offered at these symposiums.

4. A Person and Family Centered Approach to Care

Administrators of the follow-up program must take measures to mitigate the commonly reported experiences of PV carriers feeling as though they are not being "seen

as a whole person" (Leonarcyzk & Mawn, 2015, p.77). It is important for a follow-up program to be *person-centered.* Team members in the follow-up program must acknowledge that there is no one size fits all approach to PV carrier care. The role of the nurse navigator and multidisciplinary team members is not to push interventions on PV carriers. Rather, to ensure they have all the appropriate facts to make an informed decision, and to empower them to select an approach to risk management suited to their life circumstances. PV carriers have reported they felt pushed by medical professionals to adhere to recommended risk-reduction modalities (Caiata-Zuffery et al., 2015). Despite the evidence of the survival advantage offered by risk-reduction surgery, there is nothing simple about the decision to undergo these surgeries and it is ill-advised for health care providers to treat it as such. This was perhaps most apparent in an interview with a woman in the BRCA documentary film, *In the family* (Rudnick & Kartemquin Films, 2008). Linda, who put off undergoing risk-reduction surgeries and at the time of the interview was battling terminal BRCA1-associated breast cancer, described her experience as this:

> This is 2005, There has to be a better way. The only thing you can do is remove body parts? I have no breasts, I have no hair, I have no ovaries, I have no uterus, I have no hormones. To say, 'Oh, it doesn't matter' is a lie. It sucks. But in spite of how awful it is to feel less than female, being alive is what matters. And in retrospect, if I could have turned the clock back, I would have had all those [preventative] surgeries. It may not be the ideal life that you want, but it's life. You don't mess with that (Rudnick & Kartemquin Films, 2008).

Linda's honesty and vulnerability captures the complexity of the decisions that women face in a hereditary cancer journey. Health care providers must always remain cognizant that on the receiving end of the services is a holistic being who is more than a survival statistic. A being who deserves a supportive, non-judgmental environment where (s)he is able to receive accurate information, free from coercion and medical paternalism. A genetic counsellor noted that there comes a point after a certain number of attempts to reach out to a PV carrier with no response, care providers must 'take the hint' that the individual is not interested in further communication about risk (and risk management). This should be respected without any undue pressure exerted on the individual. The principles of holistic care must remain in the minds of every health care provider working with this population.

From a theoretical nursing perspective, the nurse navigator should be encouraged to adopt a relational inquiry approach to providing nursing care (Hartrick-Doane & Varcoe, 2015). Using a relational inquiry approach when working with HBOC PV carriers, the nurse navigator acknowledges the complexities of the interpersonal, intrapersonal, and contextual factors that influence their risk management decisions. The relational inquiry approach was influenced by critical theory, a philosophical movement where disparities in sociopolitical structures are highlighted and there are calls for action to mitigate the lasting effects of disadvantageous socioeconomic, political, and historical ideologies (Polit & Beck, 2017). Using relational inquiry is a way to provide an emancipatory approach to HBOC care. It is recommended that the nurse navigator be familiarized with Hartrick-Doane and Varcoe's (2015) *How to Nurse: Relational Inquiry*

with Individuals and Families in Changing Health and Health Care Contexts as it is an appropriate framework and tool for the nurse navigator when designing and delivering the program.

Another way that this program should be *person-centered* is that team members must acknowledge that asymptomatic PV carriers are not 'sick'. In fact, in this proposal, there is refrainment from referring to PV carriers as 'patients' as this already labels them as sick, which does not instill much hope for their futures. However, it is important to appreciate that the fact PV carriers are considered 'well' may be a barrier to their risk-reduction adherence. Many of these women lead active, busy lives and cancer prevention may be appraised as lower on their list of priorities when they do not 'feel sick'. This barrier can be overcome by designing a flexible, user-friendly follow-up program that strives to meet PV carriers where they are in their lives. For example, many PV carriers may be younger and middle-aged women who are still working and have various family commitments. It may be unreasonable for them to take time off work or other commitments during the middle of the day to have their appointment with the nurse navigator. The nurse navigator should be able to offer flexible hours, such as a weekly or bi-weekly availability for evening and/or weekend appointments. This way, PV carriers would not have to use sick time or family leave, that they may not even have at their disposal, to attend a routine appointment. As the nurse navigator will also be the one to facilitate screening appointments and specialist appointments, efforts should be taken to find an appointment date and time that is acceptable for the PV carrier. Though this may

not always be possible, the nurse navigator should consult the individual with what times/dates are preferrable when booking them with their appointments.

A follow-up program can also be person-centered by giving PV carriers options in their preferred method of communication with the program. Some may prefer to come see the nurse navigator in person while others, especially in rural regions, may prefer to communicate via telephone, online teleconferencing, or email/online chat platforms. Providing PV carriers options in their preferred method of communication is likely to have a positive effect on their comfort levels and sense of satisfaction, which in turn will likely have a positive effect on their participation in the program. The nurse navigator should also be readily available to PV carriers when they may have further questions or concerns either via telephone or email.

Intertwined with the concept of person-centered care is *family-centered* care. It is established that "approaches to care that acknowledge all relevant people and effects will result in more efficient allocation of resources than do piecemeal approaches" (Wittenberg & Prosser, 2016, p. 1806). While this statement is true in all health spheres, it is especially accurate for PV carrier populations. On an obvious level, being in a high genetic risk family confers psychological distress about one's own increased risk for cancer. A personal family history of ovarian cancer in PV carriers has been shown to be a significant factor in the decision to undergo RRSO (Bradburry et al., 2008; Etchegary et al., 2015; Metcalfe et al., 2008; Schwartz et al., 2003). PV carriers who lost close family members to HBOC have reported that the process of their own HBOC risk management decision-making was triggering to their feelings of loss (Caita-Zuffery, 2015). Some

asymptomatic PV carriers have reported their strong sense of moral obligation to both their ancestors and their dependents to make use of the genetic and medical information at their disposal and to stay healthy for their loved ones (Caiata-Zufferey et al. 2015). A key informant noted that PV carriers who reported higher levels of 'family support and cohesion' experienced improved psychosocial adjustment to their PV carrier status, as well as improved adherence to the recommended screening and risk-reduction modalities. In an effective PV carrier follow-up program, the family system is recognized as a facilitator of psychosocial adjustment and adherence to recommended risk-reduction modalities.

One consultee noted that unaffected family members of PV carriers also experience distress associated with having multiple relatives affected by an inherited cancer predisposition syndrome. Yet, often, these unaffected family members are not included in the care circle despite the unique psychological trauma they may experience. A fitting example of this was in a scene in the documentary, *In the family* (Rudnick & Kartemquin Films, 2008) when three young sisters were given their BRCA-1 genetic testing results. While two of the sisters tested positive, it was the sister who tested negative for the BRCA 1 PV who broke down in tears and said that this was the exact opposite outcome she had been hoping for. It is in these heartbreaking moments we are reminded that the concepts of 'family' and 'health' in hereditary cancer care cannot be separated. Therefore, the participation and involvement of family members in a follow-up program should be welcomed and encouraged. Within the boundaries of privacy and confidentiality, PV carriers should be offered follow-up sessions where family

members/support persons can attend, even if they themselves do not carry a PV. Provided that PV carriers consent in advance to discussing their information with family, family members should also be able to ask questions and seek supportive care as this inherited syndrome also affects them greatly.

5. Electronic and Virtual Infrastructure to Support Delivery of the Program

It is unrealistic to expect clinicians to manage the complexities of genomic information without the assistance of health information technology (IT) (Welch et al., 2014). In NL at PMG, pedigrees are currently completed by hand during the genetic counselling process. It was discussed with key informants that PMG is exploring options with IT management to enlist electronic pedigree drawing software. As part of this practicum project, a brief internet search was conducted of the available pedigree drawing and genetic risk assessment software products. One product of note was the Progeny pedigree and risk assessment software. According to the Progeny Genetics (2020) website, their software offers many benefits: individuals can answer questions about family history online prior to their appointment, the system is able to run automated risk assessment models, order and track genetic testing, and integrate structured pedigrees into electronic medical records. It is a premise of this proposed policy that a pedigree drawing system should have the capacity to allow a seamless flow of pedigree and risk data to both the provincial electronic health record (EHR) and to the provincial medical record system (EMR). Further discussion between health care stakeholders and the

different pedigree software and EMR vendors is warranted to determine the compatibility of these systems with the health IT systems already in place in NL.

It is important to distinguish the difference between electronic medical records (EMR) and electronic health records (EHR). EMRs are digital versions of patient medical charts contained in physicians' offices, while the EHR is a province wide health record that contain information from various health services and health authorities (NLMA, 2018). In NL, the current provincial EMR is the eDOCSNL EMR. As of 2019, 291 family physicians, 76 specialists, 25 nurse practitioners, 114 fee-for-service clinics, and 33 RHA primary health clinics in NL were using eDOCSNL (eDOCSNL, 2019). The software product used to deliver eDOCSNL is the *MedAccess* EMR manufactured and distributed by TELUS health (NLMA, 2018). There are many benefits to the eDOCSNL EMR that are beyond the scope of this paper. However, it is important to note here that a significant benefit of the eDOCSNL system is its compatibility and transferability with the province wide EHR (HEALTHe NL) (NLMA, 2018). Preferably, all HBOC team members involved in the delivery of the program would use the eDOCSNL provincial EMR system. The *MedAccess* EMR product allows patients to schedule their own appointments and complete forms online, use online messaging, and benefit from automated appointment reminders (Telus, 2020). These product features could be used to facilitate coordinated, person-centered PV carrier follow-up care. There are also features of the *MedAccess* EMR that allow communication between health care providers on the platform (Telus, 2020). This would allow seamless communication between the nurse navigator and multidisciplinary team members. As the data in eDOCSNL is compatible with HEALTHe

NL, it can be accessed not only by team members with a focus on inherited cancer prevention, but also by other providers in a individuals' care circle. This includes PV carriers' primary care providers, and other providers with access to HEALTHe NL. In the future, HBOC program administrators should also work with the EMR vendor to customize clinical decision support (CDS) tools for the PV carrier program where applicable. The use of CDSS allows the patient data to be matched to clinical practice guidelines and software algorithms (Berner, 2016). This can be used to alert the navigator who delivers the PV follow-up program when a PV carrier is due for a recommended screening test/and or annually scheduled follow-up.

Technology must also be integral in how the follow-up program reaches PV carriers provincewide. Being a provincial service, the PV navigator program will span four regional health authorities over a wide geographic area with a dispersed population. It is unpractical to expect individuals to travel large distances to tertiary care centers for routine follow-up appointments. This is not a new issue; NL was one of the first provinces to offer facility-based, face-to-face telehealth appointments for individuals living in rural regions in the province (Newfoundland and Labrador Centre for Health Information (NLCHI), 2017). Through this service, individuals living in rural and remote regions have equitable access to health care in over 100 health care facilities throughout the province (NLCHI, 2017). Under this prospective program, the nurse navigator and HBOC multidisciplinary team members who had yet to do so, would complete the application form to use provincial telehealth services. With this facility-based service, hypothetically, a high risk BRCA PV carrier on Fogo Island could have a face-to-face appointment with

the nurse navigator in St. John's without having to leave her community. The use of Telehealth services will allow PV carriers all over the province to connect with HBOC follow-up care team members when it would not otherwise be possible.

Another virtual delivery option for the HBOC follow-up program is through the virtual care or home-based telehealth offered to Newfoundlanders and Labradorians through the NL Centre for Health Information. Social distancing precautions during the COVID-19 pandemic necessitated the delivery of health care appointments from remote locations. As a result, many health care providers began offering virtual care appointments where patients could connect with their healthcare provider without leaving their home. To do so, patients require only an MCP card, a personal e-mail address, an internet or wi-fi connection, and a tablet, computer or smart phone with a camera and speaker (NLCHI, 2020). In the province of NL, health care providers use either Cisco Jabber, Telus EMR Virtual Visit, or Telus EMR Health Myself Solution as videoconferencing tools in the delivery of virtual care (Newfoundland and Labrador Medical Association, 2020). These virtual platforms could be invaluable tools in the delivery of the follow-up navigation program. For eDOCSNL *MedAccess* EMR users, virtual care options are integrated into the EMR and they can sign-up for virtual patient visits through the core application as well as through the TELUS Health Myself Virtual Visits application (NLCHI, 2020). If the HBOC follow-up and navigation program were to utilize the *MedAccess* EMR software, virtual visits could be embedded in the PV carrier's eDOCSNL EMR profile.

Aligning the Program with Canadian Health Promotion Frameworks

To understand how a HBOC follow-up program is well aligned with Canadian healthy public policy frameworks, a revisit of historic Canadian health promotion documents is warranted. The first government publication in the global sphere to challenge traditional notions of 'health' was the Canadian Lalonde (1974) report, *A New Perspective on the Health of Canadians*. Lalonde (1974) challenged the notion of 'health' as the sheer biomedical capacity to treat the sick, but rather it is determined by four interdependent health fields: human biology, environment, lifestyle, and health care organizations. Since Lalonde's (1974) first conceptualization of health field determinants, the number determinants of health have been updated and expanded several times. There are currently 12 determinants of health outlined by the Public Health Agency of Canada (PHAC) (n.d.), including 'biology and genetic endowment' (PHAC, n.d.). Lalonde (1974) laid the groundwork for other fundamental Canadian documents on health policy and health promotion that followed. In *Achieving Health for all: A Framework for Health Promotion*, Epp (1986) went a step further and purported that a) health inequities exist among individuals and groups due to nuances in their health determinants, and that b), a paradigm shift from disease treatment towards prevention is needed. He recommended three population health strategies to achieve this: fostering public participation, strengthening community health services, and coordinating healthy public policy (Epp, 1986). In the same year, the World Health Organization (WHO) (1986) released the *Ottawa Charter for Health Promotion.* In the charter, 'health promotion' was defined as

means of achieving health equity and reducing the differences experienced by groups and individuals. The WHO (1986) acknowledged that people require "a secure foundation in a supportive environment, access to information, life skills and opportunities for making healthy choices. People cannot achieve their fullest health potential unless they are able to take control of those things which determine their health (p. 1).

All three of these frameworks have pertinency to this proposed policy. 'Biology and genetic endowment' is included in PHAC's (n.d.) list of 12 determinants of health. There is sound evidence that because of their genetic endowment, HBOC PV carriers are at an exponentially greater risk of breast and ovarian cancer when compared to the general population. Determinants of health occur simultaneously and are often interrelated; an individual may be affected by multiple determinants of health that are associated with poorer health outcomes (PHAC, n.d.). For example, if PV carriers have a lower level of health literacy, are on a fixed-income, and have limited social support networks, these factors will also increase their likelihood of developing HBOC (and other chronic conditions for that matter). The PV carrier navigation/ follow-up program is a strategy to ensure that PV carriers with lower levels of health literacy, education, as well as those living in rural areas are given an equitable opportunity to benefit from hereditary cancer prevention guidelines. The proposed program reduces barriers to accessing follow-up care, provides PV carriers with information, and promotes positive individual and family coping. This is approach consistent with the conceptualizations of health promotion strategies by both Epp (1986) and WHO (1986). Without such supportive programs in place, this will result in widening health disparities among the privileged and

underprivileged subgroups in PV carrier populations (Sayani, 2019). As PV carriers have an unequivocally increased risk of cancer, it is unjust to treat them the same as people of average cancer risk. In an equitable health system, PV carriers deserve to be identified, prioritized for screening and intervention, and supported as they navigate life as a PV carrier. This follow-up program is well-suited to facilitate these obligations.

Cost-Effectiveness of the Follow-Up Program

It was aptly noted that "the current ideological climate of neoliberalism reflects and reinforces short-term policy interventions that favour continued and increased funding to address perceived urgent problems of today, rather than investments to create better health outcomes in the future." (Collins & Hayes, 2007, p. 341). It is this pervasive view of health funding that has roadblocked the development of preventative health programs, such as the one proposed in this policy. For health system funders to invest in these long-term projects comes at the potential sacrifice of campaign donations and voter support (Collins & Hayes, 2007). It is unfortunate that this ideological climate has created a culture of disunion between evidence and health policy. In this proposal, policy makers are invited to challenge these ideological misgivings, and to invest in a healthy public policy for HBOC PV carrier follow-up that is not only lifesaving, but also cost saving. In this section, evidence is presented how this program can be cost saving to the health care system in the long-term.

As cancer genetics is a relatively new area of medicine, there is a paucity of data on long-term follow-up outcomes for PV carriers. At this current juncture, outcome and cost-analyses of real-world clinical simulation models offer the best evidence on which to base the clinical management decisions for PV carriers. In a recent microsimulation model study out of Australia, the authors validated the cost-effectiveness of long-term BRCA PV carrier follow-up. Petelin et al. (2020) used input data from a real-world clinical database of 983 BRCA PV carriers. They compared the estimated long-term health system costs for both BRCA PV carriers who attended a multidisciplinary high-risk BRCA clinic and those who did not. Petelin et al. (2020) found that in asymptomatic BRCA PV carriers aged ≥ 20 who attended the high-risk clinic, there was a cost-effectiveness ratio (ICER) of $32,359 to $48,263 per quality adjusted life years (QALY). As their findings are based on Australian data, this study has implications for the Canadian health care system as both countries have publicly funded health care systems. This study is especially relevant because Petelin et al. (2020) reported on the projected cost savings of a follow-up clinic with features comparable to the program proposed in this policy paper.

There are limited publications on the cost-effectiveness of long-term management strategies in PV carriers. There is, however, substantial evidence that the risk-reduction options available to PV carriers are cost-effective. Petelin et al. (2018) conducted a systemic review of cost-effectiveness of HBOC cancer risk management strategies. In the studies examining the cost-effectiveness of RRSO in BRCA PV carriers, Petelin et al. (2018) noted ICERs ranging from AU $1,876 to AU $5,789 per QALY gained. In the same study, Petelin et al. (2018) pooled cost analyses of breast screening in PV carriers. Breast

screening consisted of the recommended annual MRI alternating with mammography as per the high-risk screening guidelines for PV carriers. Adherence to this high-risk screening recommendation was shown to have a QALY cost-savings ranging from AU $28, 273 to AU $236, 644, when compared to either MRI or mammography alone (Petelin et al., 2018). These findings have implications for this proposed program. In eligible BRCA PV carriers in NL, Roebothan et al. (n.d.) found that carriers who had been assessed by a gynecologic oncologist were more likely to be compliant with high-risk MRI screening recommendations compared to those who had not (68.9% versus 31.1%; p = 0.006). Eligible PV carriers who had been assessed by a medical oncologist were also likely to be more adherent with MRI recommendations versus PV carriers who did not (71.4% versus 28.6%; p = 0.041) (Roebothan et al., n.d.). There were also statistically significant associations between uptake of RRSO in PV carriers who had been assessed by: a gynecologic oncologist (83.5% versus 16.5% p = 0.012), assessed by a medical oncologist (86.4% versus 13.3%; p = 0.003), or the by University-based Inherited Cancer Prevention Clinic (83.9% versus 16.1%; p = 0.038) (Roebothan et al., n.d.) As stated, a key role of this proposed follow-up navigation program will be to streamline PV carriers to these multidisciplinary specialists. Taking into consideration that specialist assessment has been shown to increase uptake of cost-effective risk-management options in PV carriers, it is a logical inference that this follow-up program may contribute to cost savings.

Another aspect of cost-effectiveness that must be considered is the cost savings incurred from identifying more PV carriers in the population. Up to the year 2014, only an estimated 2.6% of BRCA PV carriers in the general population had been identified

(Manchanda et al., 2018). As costs of testing depreciate further, it is highly likely that HBOC population-based genetic screening will become a routine component of universal health care in the future (Beitsch et al., 2019). Manchanda et al. (2018) determined that population-based genetic testing for HBOC variants, BRCA1, BRCA2, RAD51C, RAD51D, BRIP1, and PALB2 in unselected, general population women is more cost-effective than any clinical criteria/family history-based testing. It is also more effective than testing for BRCA1/BRCA2 variants alone. The prospect of population-based genetic testing offers an unrivaled opportunity to transform the current cancer care paradigm to a "predictive, preventive, personalized, and participatory (P4) medicine strategy for cancer prevention" (Manchanda et al., 2018 p. 715). Despite this, uncertainty remains about the indication for population-based genetic testing HBOC variants due to the low prevalence of PVs in the general population. This is due to the relatively lower risk of developing HBOC in women without a familial history, the direct and indirect costs of testing, as well as the potential psychological and clinical consequences of testing positive (Lippi et al., 2017). In a long-term follow-up of population-based AJ BRCA testing, Manchanda et al. (2020) found that testing did not adversely affect long-term psychological wellbeing or quality-of-life in PV carriers. Rather, Manchanda et al. (2020) noted that population-based testing decreased their anxiety and could identify an additional 150% of BRCA PV carriers.

While a consensus has not been reached to date on the indication and sustainability of population-based testing, it is safe to conclude that a) an insufficient number of PV carriers are being identified and b), if population-based HBOC PV testing becomes integrated into routine care, there will be an increased demand for professional

support for the influx of PV carriers who require assistance to interpret and apply their newfound knowledge. Until a consensus is reached on the indication for population-based testing, cost-savings must be incurred by identifying a greater number of PV carriers in the population. Currently, the best way to do this is through cascade testing. In other words, through the process of informing at-risk relatives of known PV carriers of their risk who then also undergo genetic testing (Griffin et al., 2020). Cascade testing has not reached its full potential in identifying PV carriers. In the literature, percentages of family members who pursued cascade genetic testing ranged from 50% to 9% (Finlay et al., 2008; Trottier et al., 2015; Suthers et al., 2006). There is economic value in optimizing cascade testing in at-risk relatives. In a Canadian patient-level simulation study, Hurry et al. (2020) compared costs in two groups: 1)individuals who did not undergo genetic testing and underwent treatment if cancer developed, and 2) BRCA PV index patients who were tested and cascade testing occurred in their first-/second-degree relatives and all opted for risk-reducing surgery. For group two, Hurry et al. (2020) noted a cost savings ICER of CAD $14,942 per QALY when compared to the costs incurred in group one. Moreover, their model predicted 127 fewer ovarian and 104 fewer cases of breast cancer with twenty-one fewer all-cause deaths. This Canadian data is extremely encouraging. It is also demonstrative that from an economic and sustainability perspective, the process of cascade testing must be improved. Professional support has been identified as a facilitator of cascade testing (Griffin et al., 2020). This follow-up program will support families in the genetic results disclosure process and support at-risk relatives who wish to pursue

cascade testing. This is another means by which the proposed program will contribute to health care system sustainability.

Evaluation: Program Pilot Project

As this is a novel3 program, the best way to provide proof of concept to health system funders and decision makers is through a program pilot project. There is a strong case to be made for a pilot project of this program to receive health research agency funding. A compelling argument for this is made by revisiting two critical documents on the future of health care in Canada, the Kirby report (Kirby & Senate Standing Committee on Social Affairs, Science and Technology (SSCSAST), 2002), and the Romanow report (Commission on the Future of Health Care in Canada(CFHCC) & Romanow, 2003). In both reports, the committees determined that health research is essential to the quality and sustainability of the Canadian health care system. Kirby and SSCSAST (2002) noted that priority health research areas were health determinants, individual and population level disease prevent strategies, and primary care delivery. Likewise, Romanow and CFHCC (2002) cited the areas of health promotion, genomics & proteomics, and interprofessional collaboration as health research priorities in Canada. This proposed program policy fits well with these recommendations in both the Kirby report (Kirby & SSCSAST, 2002) and the Romanow report (CFHCC & Romanow, 2002). Furthermore, on the website of the federal health research funding agency, the Canadian Institute of Health Research (CIHR) (2020), personalized health and personalized medicine are listed

as current, priority research areas. This proposed program is also well-aligned with this CIHR research priority mandate as it takes a creative, novel approach to managing an individual's risk of cancer based on their personal genetic information. For the above reasons, it is asserted that there are credible grounds for awarding health research funding to pilot the delivery of this program. The outcomes of this proposed pilot project in NL will likely have implications for how comparable programs can be implemented in Canada and around the world. Several outcomes of the pilot project should be measured: qualitative and psychometric measures of carrier and family experiences and satisfaction, health system utilization, cost savings, and clinical outcomes.

There are several quantitative and qualitative tools that are well suited to measure outcomes of the follow-up program. To measure PV carrier and family satisfaction, program evaluators should seek qualitative feedback from PV carriers and families who use the program through semi-structured questions. The psychosocial impact of the follow-up program should also be measured using quantitative instruments. Before-and-after psychometric measures such as the Impact of Events (IES) scale, and the Hospital Anxiety and Depression Scale (HADS) have been used to assess the impact of PV carrier interventions (Esplen et al., 2004; Kwiatkowski et al., 2019; Listøl et al., 2017; McKinnon et al., 2007). There is also a unique opportunity to use a psychometric instrument developed in an NL Lynch Syndrome carrier population. The Psychosocial Adjustment to Hereditary Diseases scale (PAHD) (Watkins et al., 2013) is a validated psychometric scale designed to identify psychosocial adjustment challenges in PV carriers (Watkins et al., 2013). PV carrier PAHD scores should be collected prior to and after one to two years of

participation in the program. The PAHD scale can be used to determine if there was an improvement in psychosocial adjustment following participation in a dedicated follow-up program.

A comparison should be made between the retrospective data collected by Roebothan et al. (n.d.) on risk-reduction modality adherence in BRCA PV carriers and in comparable data of PV carriers who participated in the novel follow-up program. In the long-term, clinical outcomes should be reported including the number of cancers detected and the cancer stage at time of detection. A comparison should also be made of the number of family referrals for cascade testing at PMG before and after the advent of the follow-up program. There is a unique opportunity in NL to determine if the proposed program could facilitate the identification of more individuals with the newly identified RAD51C NL founder mutation. Therefore, the number of RAD51C PV carriers detected through participation in the program should also be reported in a program evaluation.

As stated, to convince policy makers of the long-term value of the program, its cost-effectiveness must be clearly demonstrated. The Canadian study by Hurry et al. (2020) can be used as a reference model to design a cost-savings analysis specific to the NL health care system. The costs of delivering follow-up program should be contrasted with the costs of a non-surveyed hereditary cancer and ICERs per QALY should be calculated. Cost-savings to the health care system as well as improved outcomes must be clearly demonstrated to garner the continued provincial government funding required to deliver the follow-up program over the long term. Without proof of concept, policy makers and health system funders will likely be unwilling to support the continuation of

the program. It is therefore critical to present a detailed, high quality evaluation and analysis of the short-term program outcomes.

Discussion

The postulates of this program proposal are the result of several months of researching the literature, consulting with relevant stakeholders, and scanning existing services available both in NL and other regions. To the best of my knowledge, this is the first document developed in a NL setting that proposed detailed policy direction for a much-needed reform of PV carrier follow-up care. It is acknowledged and accepted that there may be variance in opinion of how this follow-up program should be optimally delivered. It would be misguided to assume that this proposal is a stand-alone basis on which to develop this novel program. Ideally, a group of knowledgeable multidisciplinary experts in genetics, health ethics, cancer, and health system delivery should review this document and deliberate on the efficacy and feasibility of its proposed features. PV carriers should also be given an opportunity to provide their input on the features of a proposed program. This is the best way to ensure the comprehensiveness and the acceptability of the proposed follow-up program to all relevant stakeholders. While this document provides detailed policy direction, it is not uncompromising, and it also serves as an invite to further the conversation around improving PV carrier follow-up care in NL. It is hoped that this paper will generate a robust, much-needed discussion towards the

goals outlined in this policy proposal. It is only through collaboration with all relevant stakeholders that these goals can be achieved.

Instating this program will inevitably come with its challenges. The program would be the first of its kind in the province and as is the case with any novel program, there is no existing 'instruction manual' to guide its delivery. It should begin as a pilot project and be subject to evaluation and revisions, as necessary. With the ever-evolving world of science and technology, the delivery of the program will change over time as PV carrier needs evolve. As personalized medicine continues to expand, more than likely, there will be less invasive ways to manage HBOC risk. Until then, the current risk-management options offer PV carriers the best chance of long-term survival and PV carriers have a right to be aware and make use of these options. Hopefully, in the future, the program will be involved in providing PV carriers with options for cutting-edge and minimally invasive risk- management.

There will more than likely be obstacles to securing sustained provincial government funding. However, these obstacles are not insurmountable, nor are they reason enough to halt attempts to develop innovative health care delivery strategies. Indeed, any meaningful progress in health care innovation began with people who challenged the conventionality of the time. We cannot ignore that the world is on the cusp of a scientific revolution in the genomic era. There is mounting evidence in favor of population-based genetic testing for hereditary conditions (Manchanda et al., 2020). Canada has a responsibility to stay abreast of trends in genomic medicine, and to develop pragmatic, innovative ways to ensure that its citizens can avail of the best available

medical and technological discoveries. We also cannot overlook that these genetic and technological advances are meaningless without the necessary support systems in place to help PV carriers apply this information to their life circumstances.

It is warranted to acknowledge that in this proposal, many other individuals with high-risk of inherited disease have been overlooked. This includes individuals with high risk for colorectal cancers such as familial adenomatous polyposis, and for other autosomal dominant inherited conditions such as polycystic kidney disease (PKD). This is not to suggest that these individuals do not require supportive care and improvements in PV carrier follow-up. Rather, given that this is a novel program concept, it was decided to focus exclusively on HBOC risk-management in the early stages of program development. Optimally, as the follow-up program expands, individuals affected by a wider variety of PVs would be surveyed by the program. Alternatively, this program can be used as a prototype to develop other specific, dedicated programs for PV carriers, such as a dedicated PKD or cardiogenetic follow-up program.

Finally, it is acknowledged that the integration of precision health care (such as this proposed program) will only be enacted and sustained through a paradigm shift in public priorities and perception around genetic care (Dewell et al., 2020). Precision health care addresses the unique complement of biological, environmental, behavioral, and other information relevant to the health an individual (Chambers et al., 2016; Feero, 2017). Health care providers must communicate the value of precision/genetic health care in a way that is aligned with public perceptions of health care priorities (Dewell et al., 2020). Cancer is the second leading cause of death globally (WHO, 2018) and most people will

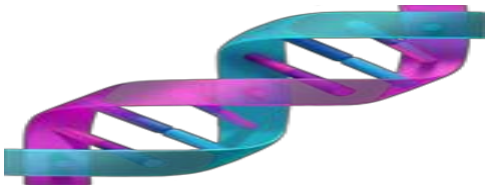
be affected by cancer in their lifetime, either themselves or through the diagnosis of a loved one. Thus, few would argue against improved cancer care as a health care priority. Cancer, in its very basic definition, is a disease of atypical genes and gene expression (Kiernan & Vallerand, 2016). Health care providers and researchers have an important role to play in communicating the link between genetic care and cancer outcomes to the public and stakeholders (Dewell et al., 2020). Public awareness shapes the narrative of priority research funding and health system utilization. This was evidenced in 2013 when Hollywood actress Angelina Jolie disclosed her BRCA PV carrier status and there was an exponential increase in referrals for genetic testing for HBOC around that time (Evans et al., 2014). The narrative must continue in the public sphere that gene sequencing will offer an unrivaled opportunity to improve cancer outcomes now and in future generations (Taylor et al., 2017).

Conclusion

18 years ago, it was projected that with rising health care costs and equally rising public expectations, the Canadian health care system was facing a grave situation if its focus remained on disease treatment in lieu of prevention (Kirby & SSCSAT, 2002). It is discouraging that this prediction has held true of in the case of the current Canadian cancer care system. An aging population, aggressive and costly cancer therapies have further contributed to the unsustainability of cancer care in Canada (Roebothan et al., n.d.). This situation must be urgently addressed. In this proposal, a robust cancer

prevention strategy is presented. Establishing this proposed PV carrier follow-up and navigation program is an invaluable opportunity to save both lives and money. Women with HBOC PVs should not be getting breast and ovarian cancer; we decisively know how to prevent it. There is an ethical responsibility of the health care system to allow PV carriers to make an informed choice about their risk-reduction and to support them and their families as they navigate the lifelong realities of PV carriership. The current approach to PV follow-up care can be likened to an analogy of leading PV carriers upstream in a river, then leaving them to navigate for themselves without a paddle. As a health care system, we have a duty to provide them with an oar and compass, to empower them to go where they need to go in their lives. Undoubtedly, it will be called into question whether we can afford 'oars and compasses' for every PV carrier. Yet the mounting costs of repeated search and rescue missions in the river for 'lost' PV carriers is never questioned; many missions ending in unfortunate outcomes. It is time to change the narrative, from 'how can we afford this? to 'how can we afford NOT to do this?' This follow-up and navigation program will serve as the oar and compass that PV carriers need and that they ultimately deserve.

This proposal would not have been possible without the faculty support of my supervisor, Dr. Joy Maddigan, the expertise of Dr. Lesa Dawson, Dr. Kerri Smith, and all of the individuals who graciously agreed to participate in the key informant interviews. I am deeply appreciative of you all. – Rebecca Puddester



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