EXPLORING PERCEPTIONS AND READINESS FOR NURSING CONTRIBUTIONS TO GENOMIC-INFORMED CANCER CARE IN NEWFOUNDLAND AND LABRADOR: A CONVERGENT MIXED METHODS STUDY

By © Rebecca Jennifer Puddester A dissertation submitted to the School of Graduate Studies in partial fulfillment of the requirements for the degree of

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ABSTRACT

Background: Newfoundland and Labrador (NL) has a high burden of genetic cancer predisposition syndromes (CPS), with many affected or at-risk persons reporting related unmet healthcare needs. Many genomic applications demonstrated to improve cancer outcomes are now indicated in routine cancer care. While nurses have central roles in cancer care, overall, the adoption of genomics-informed practices has lagged across the global nursing profession.

Purpose: In anticipation of an upcoming provincial clinical translational genomic service, this study explored the perceptions and readiness of oncology nurses and individuals with a CPS in NL, to inform potential directions for nursing contributions to genomic-informed clinical care.

Methods: A convergent design and a patient-oriented research approach were used in this mixed method study. In the quantitative stream, 50 NL oncology nurses completed a cross-sectional survey with validated measures of nurses' genomic knowledge (the GNCI©), and their attitudes, practices, and influence of the social system related to genomic-informed practices (questions from the GGNPS). In an interpretive description (the qualitative stream), 37 persons with a CPS in NL were interviewed about their experiences and perceptions of genomics-informed health and nursing care. Each study stream was analyzed and reported separately. Subsequently, integration analysis was guided by an interpretive descriptive theoretical scaffold. The Pillar Integration Process (PIP) was the technique used to develop mixed method inferences.

Results: Three overarching inferences were generated: (1) Genomic testing and related patient inquiries are occurring in cancer care, yet nurses and patients face uncertainty about where to turn next for answers. (2) While not the current status quo, nurses and patients share a recognition of the value of applying a genomics lens to existing nursing roles, including family history collection, navigation, and supportive care. (3) Both groups endorsed a prospective longitudinal high-risk hereditary cancer service, with patients reporting that specialist oncology nurses could contribute to this service.

Conclusion: These findings provide patient and provider perspectives on potential avenues for nursing roles in person-centred genomics care that are responsive to the needs of this patient population. These avenues can be facilitated through workforce development, policy initiatives, and future patient-oriented, participatory research involving multiple end-users.

Keywords: Genomics-informed care; cancer nursing; mixed-methods; hereditary cancer; cancer predisposition syndromes (CPS)

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[1] *'The light within me honours the light within you.'*

DEDICATION

This dissertation is dedicated to my mom, Judy Gard-Puddester, and to all the Moms who support their quirky children in being their authentic selves and, in doing so, give them the courage to share their perspectives with the world.

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List of Abbreviations

ACMG	American College of Medical Genetics
ANA	American Nurses Association
ASIGN	Assessment of Strategic Integration of Genomics Across Nursing
CAGC	Canadian Association of Genetic Counsellors
CANO/ACIO	Canadian Association of Nurses in Oncology/Association Canadienne des Infirmières en Oncologie
CCP	(Provincial) Cancer Care Program
CIHR	Canadian Institutes of Health Research
CPS	Cancer Predisposition Syndrome
CTG	Centre for Translational Genomics
DNA	Deoxyribonucleic Acid
DOI	Diffusion of Innovation
DP	Dialectical Pluralism
ELSI/GE3LS	Ethical, Legal, Social, Issues (US)/ Genomics and its Environmental, Economic, Ethical, Legal, and Social aspects (Canada)
EOC	Epithelial Ovarian Cancer
EUZ	Eastern Urban Zone
FAP	Familial Adenomatous Polyposis
FHH	Family Health History
GC	Genetic Counsellor
GGNPS	Genetics Genomics Nursing Practice Survey
GNCI©	Genomic Nursing Concept Inventory©
G2NA	Global Genomics Nursing Alliance
HBOC	Hereditary Breast and Ovarian Cancer Syndrome
iKT	integrated Knowledge Translation

ID	Interpretive Description
LS	Lynch Syndrome
MM	Mixed Method(s)
NCCN	National Comprehensive Cancer Network
NHGRI	National Human Genome Research Institute
NL	Newfoundland and Labrador
P/LPV	Pathogenic/Likely Pathogenic Variant
PARPi	Poly adenosine diphosphate ribose polymerase [PARP]-inhibitor
PCP	Primary Care Provider
PIP	Pillar Integration Process
P/LP(V)	Pathogenic/Likely Pathogenic (Variant)
PMGP	Provincial Medical Genetics Program
POR	Patient-Oriented Research
RRSO	Risk-Reducing Salpingo-oophorectomy
TFGT	Treatment-Focused Genetic Testing
VUS	Variant of Uncertain Significance

CHAPTER 1. Introduction, Literature Review and Overview of the Study

Exploring Perceptions and Readiness for Nursing Contributions to Genomic-Informed Cancer Care in Newfoundland and Labrador: A Convergent Mixed Methods Study

Cancer imposes a substantial societal burden from its significant associated mortality and considerable physical, psychological, and economic impacts (Bergerot et al., 2024; Bray et al., 2024; Garaszczuk et al., 2022). Cancer is a genomic disease, caused by alterations in DNA structure or expression (National Cancer Institute, n.d.). While most DNA changes resulting in cancer are acquired randomly or due to environmental exposures over the lifespan, 10% of cancers are a result of inherited gene variations linked to cancer susceptibility, also known as cancer predisposition syndromes (CPS) or 'Hereditary Cancer Syndromes', but the former term will be used primarily throughout this dissertation (Garutti et al., 2023). With advancements in molecular biology, there has been a pivot from single-gene molecular testing toward multi-gene panel and even whole genome testing (Santos Simmaro, 2022). These advancements, decreasing costs of gene testing, and ongoing clinical research have culminated in a growing number of evidence-based genomic applications in cancer prevention, diagnosis, treatment, and survivorship (Suehnholz et al., 2024; NCCN 2024a; 2024b). Thus, genomics has and can further offer new hope in the fight against one of the most feared and burdensome diseases in society (Subbiah et al., 2025).

For these applications to reach the patients and families who could benefit from them, a shift is required from genomics being seen as a niche, specialized field, to being recognized as an integrated component of routine cancer care and healthcare services (Horak et al., 2016; Stark et al., 2019; Subbiah et al., 2025). In recent years, alternative service delivery models have emerged where genomic testing and related clinical care are integrated into cancer care services for specific disease sites (Bokkers et al., 2022; Mackley et al., 2025; Canadian Association of Genetic Counsellors [CAGC], 2025). Although this approach shows promise, overall, complex patient, provider, and system-level barriers limit the uptake of evidence-based genomic testing and related cancer control applications (Bokkers et al., 2022; Dusic et al., 2022; Mittendorf et al., 2021; Smith-Uffen et al., 2021).

While multiple strategies are needed to overcome these complex barriers, it is held that, when equipped with genomic competencies, nurses in cancer care settings are well positioned to make valuable contributions to the integration of genomics-informed cancer care that is person and family centred (Beamer et al., 2013; Eggert, 2017; Flynn et al., 2019; Milani et al., 2023). Nurses play central roles in cancer care delivery and frequently interact with patients and families along their extended journeys across the care continuum (CANO/ACIO, n.d; Naito, 2024; Prip et al., 2022). Nurses working in cancer care settings promote patient and family agency and engagement and improve patient satisfaction with their care (Azarabadi et al., 2024; Prip et al., 2022). Yet, little empirical research has examined the genomic competency of nurses working in oncology care specifically, none within the province of Newfoundland and Labrador (NL), which was the setting for this study (Cuthill et al., 2025; Hines-Dowell et al., 2024; Hébert et al., 2022). However, from over two decades of global research across the general nursing profession, it has been found that significant gaps in nurses' genomic competencies remain (Dante et al., 2025; Thomas et al., 2023; Wright et al., 2018). These competency deficits are particularly concerning in the face of the growing scale of evidence-based genomic applications shown to improve healthcare safety and quality (Calzone et al., 2024). A seemingly evident solution is more education for nurses on the topic; however, educational interventions targeting nurses' genomic competency have demonstrated little, if any, sustained progress in advancing nurses' readiness for and adoption of genomics-informed practices (Dante et al., 2025; McLaughlin et al., 2024; Thomas et al., 2023). Innovative strategies are required to effectively mobilize and engage the nursing workforce as the number of evidence-based genomic applications continues to grow (Thomas et al., 2023).

One proposed strategy is for the nursing workforce to collaborate with people with relevant lived experience to co-develop solutions to strengthen nursing workforce preparation and clinical nursing roles in genomics-informed services (Thomas et al., 2023; Tonkin et al., 2020a). Patient perspectives are of utmost value in shaping the design and delivery of genomics-informed cancer care and healthcare services that are relevant to their needs, expectations, and values (Aronson et al., 2025; Daack-Hirsch & Campbell, 2014; Murtagh et al., 2021; Rand et al., 2019; Snow et al., 2024). In research examining the lived experience of people with a CPS, these individuals frequently report unmet health needs and noted that they often assumed the 'expert' role in educating their healthcare providers about their condition (Diez de los Riez de la Serna et al., 2024; Hennig et al., 2018; Hynes et al., 2023; Warner & Grokke, 2022). Research exploring the clinical experiences and perspectives of individuals with CPS has revealed valuable insights into the acceptability and utility of genomics-informed care, as well as potential psychological, provider, and system-level barriers to its implementation (Campbell-Salome et al., 2021; Kim et al., 2021; Kohut et al., 2023; Mittendorf et al., 2021; Pollard et al., 2023; Watkins et al., 2011; Warner & Grokke, 2023). These findings presented implications not only for longitudinal hereditary cancer service delivery but also cancer care more broadly as eligibility criteria for genomic testing continue to expand in cancer care settings (Smit et al., 2024). Yet, there is limited research examining patients' experiences and perceptions of genomics-informed cancer nursing care (De Los Riez de la Serna et al., 2024).

Examining the lived experiences and perceptions of people with CPS in relation to genomics-informed nursing can provide helpful insights in efforts to overcome persistent barriers and to advance genomics-informed nursing care in the precision oncology era writ large (Thomas et al., 2023; Tonkin et al., 2020b). However, while new approaches are needed to mobilize the nursing workforce with genomics, expert consensus is that validated measurements of nurses' genomic competency are required to assess change and measure progress over time (Thomas et al., 2023; Tonkin et al., 2025). These measures can also help identify barriers and facilitators associated with nurses' genomics competency. Review of these current gaps informed the mixed-method (MM), patient-oriented research (POR) approach taken in this study; I sought to explore the perspectives of both patients with lived experience and nurses related to nursing competency and care delivery in cancer genomics.

This study was conducted in NL following the announcement of a new provincial Centre for Translational Genomics (CTG). The proposed CTG will address the needs for in-house genomic testing, expanded genetic and genomic clinical services, and will enable and support translational genomics research. A component of the expanded genetics clinical services is a new High Risk Cancer Clinic, with goal of addressing the longitudinal needs of individuals and families with CPS. This novel service was recommended in a recent report for 10-year health system transformation in NL, commissioned in response to alarming provincial health outcomes (Health Accord, 2022). In anticipation of this upcoming service delivery change, I sought to generate contextually relevant insights to advance nursing contributions under this proposed cancer genomics service that are acceptable and responsive to patients' and families' perceived care needs. Therefore, the aim of this convergent MM POR study was to explore what insights can be learned from the perspectives of cancer nurses and individuals with CPS living in NL related to genomics-informed health and cancer nursing care, with the fundamental goal to inform recommendations for integrating genomics into cancer nursing practice that optimizes patient care experiences and outcomes.

Outline of the Dissertation

This MM dissertation is presented in five chapters and follows the manuscript-based dissertation format. Chapter 1 introduces the research problem and provides a narrative review of background literature, thereby identifying gaps to situate the rationale for this study. It also presents an overview of the research questions, methodology, and methods. Detailed results of this convergent MM study are summarized in Chapters 2, 3, and 4. Chapter 2 contains findings of the qualitative study stream, exploring patients' perceptions of cancer genomic health and nursing care as published in the *European Journal of Oncology Nursing*. Chapter 3 contains the results of the quantitative study stream, a cross-sectional study examining NL oncology nurses' competency and readiness to implement genomics, which was accepted for publication in the *Canadian Oncology Nursing Journal.* Chapter 4 presents unpublished integration findings synergizing the perspectives of nurses and patients related to genomics-informed care, prepared for submission to the *Journal of Mixed-Methods Research*. In Chapter 5, the conclusion chapter, key findings of the overall study are summarized, with its strengths, limitations, and contributions discussed. Implications of the study findings with recommendations for practice, policy, administration, education, and future research are also presented. Each manuscript-based chapter (2, 3, 4) of the dissertation is self-contained, including references, and presented in the format as prepared for the journal submission requirements. References cited in Chapter 1 and 5 are included in a reference list located at the back matter of the dissertation.

Background and Literature Review

To situate the study, in this section, a narrative review of the background literature was presented (Greenhalgh et al., 2018). First, I provided an overview of key advancements in molecular genomics, and cancer genomics specifically, to situate the current state of evidence-based applications in cancer control. I then offered a high-level overview of the person-centred and ethical, legal, and social considerations related to genomic applications. This segued into examples of common genomic applications available across the cancer care continuum, informed by high-quality evidence and best practice guidelines. I then discussed the lags in uptake of cancer genomics applications, including system, provider, and patient-level factors that influence their optimal clinical translation. Next, I focused on the nursing disciplinary discourse to posit that the cancer nursing workforce is well-positioned to address some of these unmet information and supportive care needs among those seeking and receiving genomics-informed cancer care and the research genomics-informed nursing care and competencies. This was accompanied by an exploration of the philosophical worldview, theoretical ideas and professional, personal perspectives that I drew on to inform the research questions, and methods.

The Current Genomic Era of Healthcare

‘Genetics’, the field of study concerning how the basic units of heredity (genes) contribute to health and disease, is certainly not new. The collection of a medical family health history as a clinical screening and decision-making tool dates to the time of Hippocrates, and genetic counselling as a clinical health discipline dates back to 1947 (Resta, 2019). Over 75 years ago, Watson and Crick (1953) discovered the molecular structure of deoxyribonucleic acid (DNA), the chemical code in which our genes are written. Leveraging insights from the (largely uncredited) X Ray crystallographic data of female chemist, Dr. Rosalind Franklin, they determined that DNA is a double helix molecule, consisting of two strands with four chemical bases (nucleotides): Adenine (A), Guanine (G), Cytosine (C), and Thymine (T) with sugar phosphate backbones (Mukherjee, 2017). Four years later in 1957, Francis Crick proposed the 'central dogma of biology'- the notion that genetic material is present in every cell of every living thing, and its primary function is to "to control (not necessarily directly) the synthesis of proteins" (Crick, as cited in Cobb, 2017, p.2). Crick's hypothesis was pivotal to our current understanding that these four nucleotides of DNA (A, G, C, T) function as an alphabet; the 'order' (i.e., the sequence) of these letters codes for a set of instructions for what 'makes us' biological beings. This process occurs when DNA from cells is transcribed by molecules known as messenger ribonucleic acid (mRNA), which then code for the formation of new proteins (Haseltine & Patarca, 2024).

The breakthroughs of Crick, Watson, Franklin, and several other scientists guided humanity toward developing technologies to 'read' or 'sequence' the order in which these DNA nucleotides occur in a person's *genes*, the basic units of DNA which serve a particular function and occupy a specific locus in the human genome. This information provided insight into how variations in these gene sequences (i.e., genotype) influence observable traits and disease risk (i.e., phenotype) (Hutchison, 2007). These efforts were catapulted by the Human Genome Project, a $3 billion, 13-year international initiative completed in 2003 with the ambitious aim to sequence the entire human genome (Lander et al., 2001; Nurk et al., 2022); some 20,000 genes or 3.2 billion base pairs or 'letters' of DNA. The Human Genome Project represented a transformative moment in science and human history, providing a reference human blueprint. The Human Genome Project also accelerated the utility of a technology known as next-generation sequencing, or 'massive parallel sequencing'. Owing to this, by 2022, the cost of sequencing an entire human genome fell from its initial cost in 2001 of $100 million USD to approximately $1,000 USD, with projections that this cost will depreciate to as low as $100 USD (Penninisi, 2022; Wetterstrand, 2023).

With these technological advancements and decreasing costs of DNA sequencing technologies, the field has quickly pivoted from sequencing single genes in isolation towards multi-gene panel testing as the standard of care for many conditions (Sabour et al., 2017; NCCN, 2024a; 2024b). Now, in some jurisdictions, molecular testing of the whole exome (protein-coding regions of the genome), and even of the whole genome are becoming first-line tests for the diagnosis of rare diseases, including rare cancers (Bagger et al., 2024). Whole-genome sequencing is showing promise in reducing the 'diagnostic odyssey'- a prolonged, often agonizing, period with multiple specialist consultations and a battery of tests while waiting for a diagnosis of a rare genetic condition (Scocchia et al., 2019).

Thus, while the classic field of 'genetics' is not new, it is the amount and availability of clinically informative genomic information that can now be interpreted within a reasonable time frame that is the current 'game changer' (Bombard et al., 2013; Pennisi, 2022). Note that genetics refers to the study of genes and their role in health, disease and the inheritance of traits (National Human Genome Research Institute [NHGRI], 2018). 'Genomics' may be a term less familiar than genetics to many but refers to the field of biology focused on studying an organism's genome, including identification and characterization of all the genes and functional elements in an organism, as well as how they interact with the environment (NHGRI, 2018). While there is a distinction between the terms 'genetics' and 'genomics', there is a shift in the nursing and health literature towards using 'genomics' in lieu of 'genetics' as a more encompassing term reflecting the growing amount of clinically relevant DNA-based information and larger gene panel tests (Calzone et al., 2024). Therefore, ’genomics’ will be used frequently throughout this dissertation to refer to anything that encompasses genes, including their interactions with one another and anything that regulates them, for example, the environment. However, I will also use the term ‘genetics’ interchangeably where appropriate, especially when referring to original source material.

Cancer: A Genomic Disease

With advances in DNA-based technologies, cancer has been revealed as a fundamentally genomic disease. It is characterized by molecular changes or ‘variants’ (also referred to as ‘mutations’) in genes that occur due to aging, environmental or chemical exposures, radiation, hormones, other factors in the body, or inherited gene variants present in the germline cells. If a cell acquires a number of DNA changes, it can affect how cells in the body grow and divide, causing them to grow uncontrollably, incurring a selection advantage over neighbouring cells, resulting in cancer (National Cancer Institute, n.d.) (see Figure 1.1). While all cancer is ‘genetic’, a minority (~10%) are considered hereditary, i.e., the result of an inherited gene change associated with a CPS (Garutti et al., 2023). CPS are characterized by a significantly increased lifetime cancer risk (reaching up to 100% lifetime risk in some genes such as *APC*), usually with multiple anatomical sites affected (See Table 1.1) (NCCN, 2024b).

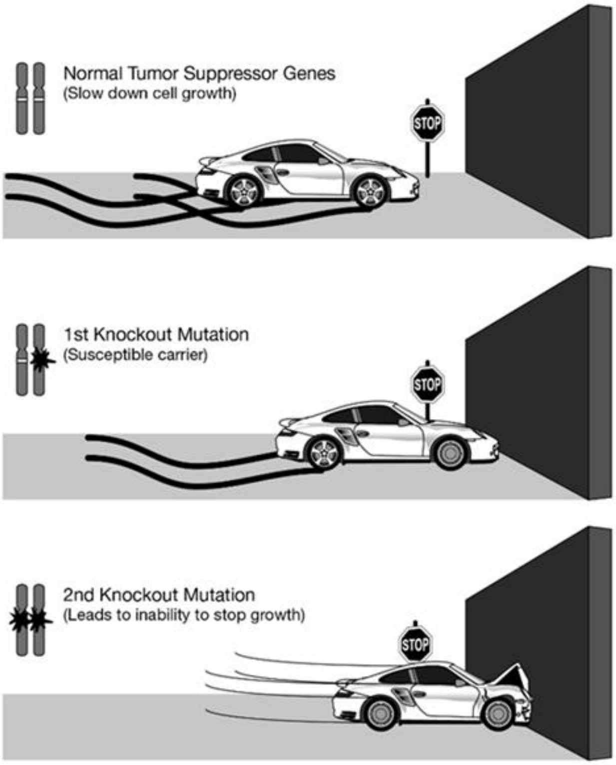
Figure 1.1

Molecular Progression of Cancer

For example, people have two copies of a tumor suppressor gene, one inherited from each biological parent. The normal function of these genes is to prevent uncontrolled cell growth, often likened to cell ‘brakes’. If there is a loss of function in these two gene copies in a cell, often referred to as ‘two hits’, cancer can arise (Chial, 2008). Most cancer is a result of two acquired (somatic)‘hits’ in cancer susceptibility genes, present in the tumour alone which cannot be passed down to biological offspring (NHS England, 2024). People with an inherited gene variant linked to a CPS have a significantly increased lifetime risk of certain cancers because they are born with one copy of the gene in every cell without its tumour suppressing function. See figure 1.2.

Figure 1.2

Two-Hit Hypothesis for Cancer Development in Tumour Suppressor Genes



NHGRI (2000) (available in the public domain).
https://www.genome.gov/sites/default/files/tg/en/illustration/tumor_suppressor_gene.jpg

This genomics-driven conceptualization of cancer and its implications for cancer control strategies have generated hope and enthusiasm in needed efforts to combat the significant global, national, and local burden of cancer (Horak et al., 2016; Seed, 2021).

Cancer Incidence and Mortality

The most recent data from the Global Cancer Observatory indicated that 20 million people globally were diagnosed with cancer in 2022, with the global annual new cases of cancer expected to increase to 35 million by 2050 (Bray et al., 2024). A worrisome global trend is the increase in incidence of early onset cancers (<age 50) and associated mortality since 1990 by

79.1% and 27.7%, respectively (Zhao et al., 2023). It is presumed that a combination of diet, lifestyle, environmental exposures as well as their possible interactions with underlying genomic predispositions, are responsible for the increase in many early-onset cancers (Akimoto et al., 2021; Ugai et al., 2022).

Two in five Canadians are expected to be diagnosed with cancer in their lifetime and one in four will die from the disease (Canadian Cancer Statistics Advisory Committee et al., 2023). While improvement in cancer survival indexes has been observed, cancer remains the leading cause of death in Canada, responsible for an estimated 88,100 deaths in 2024 (Brenner et al., 2024). In the province of NL, the second-highest overall cancer incidence in Canada, as well as the highest incidence and mortality rates for colorectal cancer, are observed (Canadian Cancer Statistics Advisory Committee et al., 2023). As indicated, the catalyst for the NL provincial health system to unveil the proposed clinical translational genomics services was in response to findings of the NL Health Accord (2022), which found unacceptable provincial health outcomes, including cancer outcomes, relative to the rest of Canada.

Incidence and Prevalence of Hereditary Cancer

It is estimated that 10% of cancers are related to an underlying genetic susceptibility associated with a CPS (Garutti et al., 2023). The two most common CPS are Lynch Syndrome and Hereditary Breast and Ovarian Cancer (HBOC) syndrome; approximately 1 in 279 people have a gene change that is associated with Lynch Syndrome, and approximately 1 in 400 have an inherited gene linked to HBOC syndrome (Garutti et al., 2023; Win et al., 2017). It is estimated that 3-4% of endometrial cancers are related to Lynch Syndrome, 5-10 % of breast cancers, and as many as 25% of ovarian cancers are due to HBOC susceptibility genes (Flaum et al., 2020; Garutti et al., 2023; Samowitz et al., 2015). Although there are over 100 CPS-associated genes outside of those associated with HBOC and Lynch Syndrome most CPS-associated genes are individually rare (Garutti et al., 2023). For example, Birt-Hogg-Dube Syndrome, a CPS associated with predisposition to renal tumours, has an estimated prevalence of two per million people (Muller et al., 2021).

CPS in NL

The historical English and Irish settlement patterns observed in the island portion of the province and its geographic isolation in the Eastern North Atlantic resulted in a unique population architecture of the province with evidence of ‘founder’ effects from the European settlement on the traditional, unceded territories of the First Peoples of NL (Gilbert et al., 2023; Zhai et al., 2016). As a result, NL has long been considered a ‘gold mine’ for research in genetically linked health conditions (Rahman et al., 2004). Several CPSs were identified in NL (e.g., Lynch Syndrome, Hereditary Diffuse Gastric Cancer, Multiple Endocrine Neoplasia, and HBOC associated with *BRCA 1/2* and the *RAD51C* genes) (Dawson et al., 2020; Green et al., 2007; Kaurah et al., 2007; Olufemi et al., 1998; Roebothan et al., 2023; Stuckless et al., 2007; Zhai et al., 2016). Table 1.1 presents examples of CPS identified in NL as reported in peer-reviewed publications, including the genes typically involved and organs affected. It is held that the genetic architecture of the NL population is a contributor to the overburden of cancer observed within the province (Dawson et al., 2020; Green et al., 2007; Shanahan et al., 2022). Further, it is reported that the NL population has the highest rates of diabetes, tobacco use, and pro-inflammatory diets (Shanahan et al., 2022). For individuals with non-modifiable (i.e., genetic) risk factors, the impact of modifiable factors (e.g., obesity, alcohol use, sedentary behaviors) on cancer risk can be even more pronounced (Bruno et al., 2021; Lazzeroni et al., 2023). In short, NL is a population that stands to benefit from translational genomic research and evidence-based genomic-informed clinical care.

Table 1.1

Examples of CPS Reported in NL and Associated Genotype and Cancer Risk (Adapted from NCCN 2024a,b)

CPS	Genes involved	Anatomical Site Associated with increased Cancer Risk
Lynch Syndrome	*MLH1, MSH2, MSH6, PMS2, EPCAM*	Colorectal, Endometrial, Bladder, Renal Pelvis, Brain Pancreas & Others
HBOC Syndrome	*ATM, BARD1 BRCA1,2, BRIP1, CHEK2, NF1, PALB2, RAD51C, D*	Breast, Epithelial Ovarian, Prostate, Pancreas & Others
Hereditary Diffuse Gastric Cancer	*CDHI*	Stomach, Breast
Multiple Endocrine Neoplasia Type 1	*MEN1*	Parathyroid adenomas, Neuroendocrine tumors, Pituitary adenomas, gastric carcinoids, lung/thymic carcinoids,
Familial Adenomatous Polyposis (FAP)	*APC*	Colorectal, Desmoid tumours, and others

Other Impacts of Cancer

Further to the mortality associated with cancer, the multifaceted societal burden of both the disease itself and its associated intensive treatments are wide-reaching. The global costs of cancer from 2020 to 2050 are projected to reach 25.2 trillion international dollars (Chen et al., 2023), with recent Canadian estimates at 26.1 billion Canadian dollars annually, with many of these costs incurred by patients and their families (Garaszczuk et al., 2022). Cancer treatments can pose distressing and long-term physical and psychological symptoms that severely compromise quality of life (Batra et al., 2021; Bubis et al., 2018; Desheilds et al., 2014). Anti-cancer treatments can cause life-threatening complications, including secondary cancers; in a cohort of 60,949 patients with uterine cancer, 7,428 developed secondary solid tumors, 11% linked to prior radiation therapy (Lönn et al., 2010). Another example of the iatrogenic harm imposed by cancer treatment is tumour lysis syndrome (TLS), a life-threatening metabolic emergency related to treatment of some high-volume (generally hematologic) malignancies (Molyneux et al., 2025). TLS-related hospitalizations in the U.S. rose from 28.6 to 44.3 per 100,000 between 2016 and 2019 (Ghimire et al., 2022), and further increased to 54.7 per 100,000 by 2020, with an associated 22.1% in-patient mortality rate (Imhoff et al., 2025). In a Canadian single centre chart review, TLS occurred in 25% of patients receiving a standard acute myeloid leukemia treatment between 2022-2024 (Saiyin et al., 2025).

Nearly half of individuals with cancer experience clinically significant distress during their cancer journey (Carlson et al., 2019; Kirk et al., 2021; Mehnert et al., 2018). It is estimated that anxiety and depression in people who received cancer treatment is double that of general population, while one in three cancer survivors experience a mental health condition (Mental Health Commission of Canada, 2021). Psychosocial burdens often extend to the loved ones and informal caregivers of persons with a cancer diagnosis (Junkins et al., 2020; Sweileh, 2024; Thana et al., 2021). Even for people without a personal history of cancer, cancer is one of the most fear-provoking phenomena in society (Vrinten et al., 2017). The word ‘cancer’ has been used in historical and contemporary language as a metaphor for grave and urgent threats (Potts & Semino, 2019), and it has been reported that merely hearing the word ‘cancer’ can arouse negative affective responses (Donovan et al., 2003). When considering these immense impacts of cancer, the growing enthusiasm for genomic applications in cancer control to change this narrative is understandable and warranted.

Conceptualizing Genomic Actionability

More than any other clinical specialty, oncology "has served as a paragon for the application of clinical genomics for the diagnosis and treatment of disease" (Zehir et al., 2017, p. 703). As a result, there are a steady number of somatic (tumour) and germline (constitutional) genomic test results which have clinically *actionable* implications in cancer control strategies (Dixon et al., 2020; Ngeow & Eng, 2016). The Global Alliance for Genomics and Health (2021) broadly defined 'clinically actionable' genomic research results as those that

> indicate risk for or the presence of a condition...for which clinicians can provide prevention or treatment, but also results that allow a participant to take steps (such as change in diet or health surveillance) that can prevent or ameliorate a genetically-based disease or disability (pp. 1-2).

As our understanding of the genome evolved, the term 'variant' is now preferred over 'mutation', in recognition that a molecular test revealing a gene variation can be an inconclusive or neutral result in terms of its clinical actionability (Richards et al., 2015). Classification systems and databases now exist for categorizing both germline and somatic DNA changes according to the strength of the scientific evidence surrounding their implications for disease susceptibility and treatment (Landrum et al., 2025; Li et al., 2017; Richards et al., 2015; Sondka et al., 2024).

Actionability of Germline Variants

The prevailing system for classifying germline variants is the five-tiered scale developed by the American College of Medical Genetics (ACMG) and the Association for Molecular Pathology (AMP) (see Table 1.2) (Richards et al., 2015). When there is strong evidence that a germline variant contributes to disease risk, it is classified as pathogenic or likely pathogenic (P/LP). As more comprehensive genomic tests such as whole exome and whole genome sequencing enter clinical and translational research settings, an important consideration is that while these are among some of the comprehensive tests available, they carry higher potential to reveal results where the evidence related to its disease contribution is inconclusive (often referred to as ‘variants of uncertain significance’ [VUS]), or results which may have health implications, but for which no preventative treatment is currently available. According to the latest ACMG clinical practice guidelines, more than 100 genes are classified as ‘clinically actionable’ if discovered as secondary genomic findings. In other words, if a person underwent whole exome or whole genome sequencing and an unexpected P/LPV was identified in one of these genes, the ACMG recommends that these results should be disclosed to the patient based on the related available treatment or prevention modalities (Lee et al., 2025).

Table 1.2

ACMG AMP Classification of Germline Variants (adapted from Richards et al., 2015)

	Pathogenic (P)	**Likely Pathogenic (LP)**	**Variant of Uncertain Significance (VUS)**	**Likely Benign**	**Benign**
Interpretation	There is conclusive evidence that this sequence change directly contributes to the development of the disease	There is a high likelihood (greater than 90% certainty) that this sequence change is disease-causing	Insufficient evidence currently to support the classification of the disease	Not expected to have a major effect on disease, but the scientific evidence is currently insufficient to prove this conclusively	The variant does not cause disease. Additional evidence is not expected to alter the classification of this variant.
Clinical Significance (*Can it inform clinical decision-making*?)	Actionable	Actionable	Non-actionable (but *may* be prudent for additional monitoring)	Non-actionable	Non-actionable

Gene Penetrance. Another nuance in the classification of germline variants is related to gene penetrance. Only a minority of germline P/LPV associated with cancer risk have 100% penetrance, meaning that for most people, having a molecularly confirmed P/LPV does not mean that they will definitively develop cancer (National Cancer Institute, n.d.). Rather, they have a significantly increased relative risk of cancer when compared to the general population. P/LPV associated with CPS can be classified as 'highly penetrant' (associated with a relative risk of cancer > 4), or 'moderately penetrant' (associated with a relative of cancer of 2-4) (Garrett et al., 2025). Tables 1.3 and 1.4 illustrate the increased risk of breast and colorectal cancer, respectively, with variants in various CPS-associated genes. Table 1.4 shows the average age of onset of colorectal cancer among different P/LPV associated with colorectal cancer susceptibility, which can vary for P/LPV gene involved, even in genes associated with the same CPS.

Table 1.3

Lifetime and Absolute Risk of Breast Cancer by Population and Gene Variant (adapted from NCCN 2024b)

Population / Known +P/LPV	Gene Penetrance	% Lifetime /Absolute Breast Cancer Risk
General Population	-	12–13%
ATM	Moderate	21–24%
CHEK2	Moderate	23–27%
BARD1	Moderate	17–30%
RAD51C / RAD51D	Moderate	~20%
BRCA1	High	60–72%
BRCA2	High	55–69%
CDH1	High	37–53%
PALB2	High	32–53%
TP53	High	>60%
STK11	High	32–54%
PTEN	High	40–60%

Table 1.4

Cumulative Risk of Colorectal Cancer and Average Age of Onset According to Genotype (adapted from NCCN 2024a)

Population /P/LPV Gene	Cumulative Risk of Colorectal Cancer	Estimated Average Age of Onset
General Population	4.1%	70 (Male), 72 (Female)
APC (Familial Adenomatous Polyposis)	Nearly 100%	39
MLH1 (Lynch Syndrome)	46–61%	44
MSH2 & EPCAM (Lynch Syndrome)	33–52%	42–69
MSH6 (Lynch Syndrome)	10–44%	42–69
PMS2 (Lynch Syndrome)	8.7–20%	61–66
MUTYH	70–90%	Not provided

Actionability of Somatic Variants

Tumour testing for somatic genomic changes or gene overexpression (as an example of 'biomarker testing') can match individuals to approved and experimental anti-cancer therapies (Suehnholz et al., 2024). While in some cases, somatic genomic test results can signal the possibility of an underlying germline predisposition driving the somatic change in the tumour cells, in themselves, somatic variants cannot be passed on to offspring (NHS England, 2024). In oncology, 'clinical actionability' carries both unique and plural interpretations, due to a) the presence and interplay of two distinct genomes (the human genome [germline] *and* the cancer genome [somatic]), and b), the integration of experimental research with clinical care, driven mainly by the routine use of experimental clinical trials in the treatment of advanced cancer (Chin-Yee & Plutynski, 2024; Nelson et al., 2013). Thus, in the context of oncology, genomics can also be 'actionable' by virtue of how it can be used to "generate predictive relationships between genetic information and drug therapies" (Nelson et al., 2013, p. 405). This description contrasts with the traditional biomedical conceptualization of actionability in the setting of an inherited genetic disorder (such as a CPS), where the aim is to establish risk for a disease by making a causative link between genotype and phenotype. As a result, unique classification systems and databases are used to interpret somatic cancer variants based on how they affect cancer diagnosis, prognosis, and treatment decisions. The most recognized somatic classification system globally is the four-tiered system developed by the AMP, American Society of Clinical Oncology (ASCO), and College of American Pathologists (Li et al., 2017) (see Table 1.5)

Table 1.5

Evidence-Based Classification of Somatic Variants According to their Diagnostic, Prognostic and Therapeutic Implications, Adapted from Li et al. (2017)

Tier	Level of Clinic/Experimental Evidence	Example	Implication
Tier I (Strong Clinical Evidence)	Level A: FDA-approved therapy or included in clinical guidelines	EFGR p.L8 somatic variant in small-cell lung cancer	Predictive of a response to approved EGFR tyrosine kinase inhibitors (e.g., erlotinib, osimertinib)
	Level B: Well-powered studies, expert consensus in the field	*BRAF* V600E	96% response rate to vemurafenib observed in two multicenter phase 2 clinical trials
Tier II (Variants of Potential Clinical Significance)	Level C: FDA-Approved therapy Multiple smaller studies with some consensus	*JAK2* somatic variant in acute lymphoblastic leukemia	FDA-approved JAK inhibitor ruxolitinib is an FDA-approved drug being trialed in children with lymphoblastic leukemia
	Level D: Pre-clinical trials or case reports without consensus		May influence prognosis or indication for experimental treatment
Tier III (Variants of Unknown Clinical Significance)	Not observed at a significant allele frequency in the general or specific subpopulation database No convincing published evidence of cancer association	Atypical variants in *KRAS* (e.g., A146T)	Limited data on treatment response Not used in clinical decision-making
Tier IV (Benign or Likely Benign)	Observed at a significant allele frequency in the general or specific subpopulation databases	Benign polymorphism in *TP53* p. Pro72Arg	No clinical impact

Complexities of Actionability: System, Person-Centred, Ethical, Legal, and Social Considerations

While a somatic or germline variant may be considered actionable in accordance with evidence classification systems and databases, this does not necessitate that the genomic test and/or corresponding treatment will be available as part of publicly funded healthcare services (Husereau et al., 2023). In Canada, provincial/territorial health system leaders determine policies for adoption and universal coverage of genomic tests and matched treatments with guidance from a federal agency responsible for comprehensive drug and health technology assessments (Canada's Drug Agency, 2025). When a health technology assessment is conducted for a novel genomic test or treatment, multiple factors are considered beyond its clinical validity and utility (Bombard et al., 2013; Husereau et al., 2023). Bombard et al. (2013) summarized seven key precepts, related aims, and evaluation methods regarding health technology assessment of cancer genomic applications in the Canadian context. An adapted version of this summary is presented in Table 1.6.

Table 1.6

Health Technology Assessment Precepts and Key Questions with Examples of Evaluation Methods, Adapted from Bombard et al. (2013)

HTA Precept	**Key Question(s)**	**Examples of Evaluation Methods**
Analytic Validity	How accurate is the health technology in detecting genetic changes?	Lab proficiency tests, Analytic specificity, and sensitivity
Clinical Validity	How accurate is the test in identifying patients with a particular disorder of interest?	Genotype/phenotype relationships, predictive values
Clinical Utility	How will this health technology significantly improve health-related outcomes (e.g., morbidity, mortality, clinical outcomes, quality of life)?	Morbidity, mortality, clinical outcomes, and quality of life data
Personal Utility	How will this health technology contribute to personal empowerment and behavior change? Enable life and reproductive planning? Relieve uncertainty surrounding risk?	Participation rates may be indicators, psychometric measures of satisfaction, empowerment, perceived utility, and qualitative data
Economic Evaluation	What is the value for money of the test vis-à-vis health gain benefit relative to costs and resource utilization compared with available alternatives?	Cost/benefit analysis, cost-effectiveness analysis, cost-utility analysis
Health Service Impact	How feasible is adopting or implementing this genomic health technology or screening program into the health system?	Operational and economic feasibility, net budget impact
Ethical and Social Values	Is adopting this new health technology consistent with the prevailing moral and societal values?	Primary research, expert advice/report, public engagement

A key point evidenced in this list of considerations in Table 1.6 is that it is not just the sheer size and scientific complexity of genomic data that makes it a complex form of knowledge. There are many inherent personal considerations and, ethical, legal, social issues (ELSI) with respect to genomics, making the concept of ‘genomic actionability’ highly contextual (Goddard et al., 2022). Some of these considerations are summarized below.

Person-Centred Considerations

Not specific to genomics health care, there is recognition of the importance of incorporating lived experience data into health workforce preparation, healthcare delivery models, and resource allocation (Rand et al., 2019; Greenhalgh et al., 2019). Since the introduction of predictive genetic testing for HBOC syndrome in the 1990s, a significant body of qualitative and quantitative research has focused on the psychosocial implications and experiential knowledge related to genomic test results in individuals with or at risk for a CPS (Broadstock et al., 2000; Eijzenga et al., 2014; Ringwald et al., 2016; Carlsson et al., 2022). This research has provided insights into how these populations conceptualize utility and actionability in cancer genomic applications (Goddard et al., 2022). A critical consideration revealed from research surrounding the lived experience of CPS is related to 'perceived' risk, one's subjective evaluation of their risk (Ferrer & Klein, 2015; Heshka et al., 2008; Tilburt et al., 2011). It has been shown that individuals' perceptions of their risk do not always align with numerical 'biomedical' risk calculations, such as the percentages of cancer risk outlined in the above NCCN (2024a; 2024b) guidelines, which can impact uptake of risk-reduction measures and behaviors. In a systematic review, Tillburt and colleagues (2011) identified factors associated with heightened cancer risk perception in individuals with CPS, such as the awareness of family history and the ability to process numeric information. Younger age and emotional propensities, such as distress and anxiety, can heighten perceived cancer risk (Tilburt et al., 2011). Tillburt et al. also reported that accurate risk perception is associated with personality traits such as vigilance and a surveillance-oriented coping style.

Generally, the literature suggests that while short-term psychological stress can occur commonly following receipt of positive cancer genomic test results (i.e., a P/LPV), this distress subsides typically to a level similar to that of non-affected persons (i.e. true negatives) in the long-term (Hirschberg et al., 2015; Meiser, 2005). However, in a recent cross-sectional study of participants with a CPS, 66% reported clinically relevant distress well above the average in non-affected persons (Kastner et al., 2023). In a literature review which included 25 qualitative studies of CPS patients' experiences, Eijzenga et al. (2014) reported broad categories of psychosocial issues experienced by people with a CPS: practical issues (such as insurance and employment considerations), family issues (such as risk-communication) children-related issues, and concerns related to the burden associated with living under the shadow of elevated cancer risk, including anxiety, fear, and guilt, especially related to the possibility of passing on a P/LP to relatives.

What has been gleaned from 30 years of lived experience research in people with inherited cancer risk is that receiving this information carries the potential to be life changing (Esplen & Kohut, 2025). Understanding the nuances of person-centred preferences and values can inform tailored, risk communication strategies and supportive care that minimize psychosocial burden and possible iatrogenic harm arising from actionable genomic findings (Clausen et al., 2024; Kohut et al., 2024; Kohut et al., 2023; Morton et al., 2022). There is an identified need for spaces where people can longitudinally access person-centred healthcare related to their risk decision-making and management (Esplen & Kohut, 2025; Manuel & Brunger, 2015). As stated by a person with lived experience in a study co-designing a Lynch Syndrome patient decision aid, "A good decision is the one that feels right for me" (Kohut et al., 2024 p. 9).

ELSI Considerations

The ethical and societal considerations highlighted in the list of seven health technology assessment precepts above are not exclusive to the assessment of genomic health technologies and drug applications (Bombard et al., 2013). Some experts argue that ‘genomic exceptionalism’ (where genomic data is treated as inherently different than other forms of sensitive information) is unnecessary and slows delivery of the benefits of science across society (Evans & Burke, 2008; Garrison et al., 2019). Yet, it cannot be overlooked that many genomic applications that society enjoys today are founded on knowledge derived through investigations that were some of the most egregious human rights violations in history (Da Silva & Hubbard, 2024). A poignant example is when genetics was used to erroneously justify political ideologies of racism, ableism, and xenophobia under the auspices of research in ‘selective breeding’ carried out through horrific Nazi medical experiments in the 1930s and 1940s (Liscum & Garcia, 2022). In acknowledgement of the uncomfortable chapters in history which demonstrated the potential for harm arising from genomic investigations, it was recognized that careful deliberation is needed in genomics policy and health service delivery. It is held that the perspectives of multiple interest holders and disciplines are critical in these deliberations to enable the responsible and just integration of genomic applications across society (Bilkey et al., 2019; McGuire et al., 2008; Murtaugh et al., 2021). This imperative of multi-interest holder engagement was recognized from the beginning of the Human Genome Project when part of its project budget was set aside specifically for interdisciplinary health services and policy research related to its potential ethical, legal, and social impact (known as ELSI research) (Green et al., 2020; Dolan et al., 2022). Following several decades of interdisciplinary ELSI research (referred to as GE^{3}LS research in Canada), several key ELSI/GE^{3}LS issues have been examined at length.

Commonly Researched ESLI/GE^{3}LS Considerations. High-level examples of these commonly researched topics include: 1. genomic data privacy protections given the sensitive, permanent, and identifiable nature of germline genomic information (Bonomi et al., 2020). 2. Concerns related to potential for genetic discrimination from employers, insurers, the justice system, or across society broadly (Cowan et al., 2022; Joly et al., 2013; Joly et al., 2022). 3. The ethical tensions that can arise from the ambiguity from a legal perspective between the duty to uphold privacy and confidentiality and the duty to warn at risk relatives (Rothstein, 2019; Suter, 2020). 4. The balance between harm and benefit, with the potential for psychological distress stemming from receiving the predictive genetic test results in asymptomatic individuals (Fulda & Lykens, 2006). The full gamut of ELSI/GE^{3}LS considerations is beyond the scope of this dissertation. However, as genomics becomes more integrated into routine care, both familiar and newer ELSI/GE^{3}LS considerations will arise in cancer settings (Smit et al., 2024). Some experts advocate universal, multi-gene panel germline testing as standard of care for all cancer patients with solid tumours regardless of family history (Esplin et al., 2022; Shore et al., 2024). Healthcare administrators and providers working in cancer control must be aware and engaged in conversations about the associated ELSI/GE^{3}LS considerations with genomics, as it crosses the chasm from niche to necessity. As use of testing expands to broader populations, there is a benefit to understanding ELSI/GE^{3}LS implications, such as informed consent processes, through examination of how these applications have previously been used in CPS populations (Smit et al., 2024).

Evidence-Based Genomic Applications in Cancer Control

Broad use of whole genome sequencing in cancer treatment has already been occurring within research contexts (Pleasance et al., 2022). For example, Pleasance et al. (2022) found that of 507 patients in British Columbia with advanced cancer and diverse tumour pathologies (with no known family history of cancer) who underwent whole genome sequencing, 248 were matched to whole genome sequencing-informed treatments, with 46% who experienced increased clinical benefits. As universal whole genome sequencing-informed cancer treatment and many other exemplars are occurring in experimental contexts across cancer treatment settings (Farncombe et al., 2023; Pleasance et al., 2022), they are beyond the scope of 'evidence-based genomic applications' for this dissertation. Nonetheless, given the convergence between research and clinical care in oncology, genomic research applications are likely to be frequently encountered in cancer settings. In the following paragraphs, examples are presented of germline and somatic cancer genomics applications backed by strong evidence, and/or clinical practice guidelines currently implemented as the standard of care for cancer treatment and primary or secondary prevention.

Genomic Applications in Cancer Treatment

Using both germline and somatic genomic information as a personalized approach to cancer treatment improves treatment efficacy by providing insight into treatment sensitivities and responses, while avoiding possible side effects associated with traditional chemotherapies and treatments that would be unlikely to yield benefit, yet have burdensome and sometimes lasting side effects (Kato et al., 2020; Kinnersley et al., 2024). Common examples of somatic and germline applications in treatment are summarized below.

Somatic Applications in Cancer Treatment and Prognostication. There are currently at least 20 Health Canada-approved targeted cancer therapies available for specific disease sites, requiring a patient's somatic genomic data for the clinical indication (Liu et al., 2022; Yip et al., 2019). For example, genomic testing for somatic *EGFR* gene variants can be used to match patients with advanced non-small cell lung cancers to drugs known as *EGFR* tyrosine kinase inhibitors (National Cancer Institute, 2021). Another example of a somatic application is microsatellite instability testing (MSI). Microsatellites are small repetitive segments of DNA nucleotide (A, C, T, G) present throughout the genome. When there is an inconsistency in the number of these nucleotide repeats in the cancerous tissue compared to normal tissue, the tumour is 'MSI-high' (MSI-H). Approximately 15-20% of colorectal tumours and 20-30% of endometrial cancers exhibit MSI-H (Vilar & Gruber, 2010). Universal somatic MSI testing of colorectal and endometrial tumours is recommended in Canadian and international guidelines (Aronson et al., 2025; NCCN 2024b). MSI testing has prognostic value, as tumours that exhibit MSI-high typically respond well to immunotherapies (Aronson et al., 2025). MSI-H occurs most commonly due to somatic (acquired) inactivation of the *MLH1* mismatch repair gene. However, an MSI-H result can also be suggestive of a possible underlying germline P/LPV in *MLH1 MSH2, MSH6 or PMS2*, the genes associated with Lynch Syndrome and can prompt further germline testing (Aronson et al., 2025).

Germline Genomic Applications in Cancer Treatment. Benefits of germline testing (via blood or buccal samples) in cancer treatment settings have also been established. For example, in 2010, germline *BRCA* gene testing to inform selection of PARP-inhibitor (PARPi) therapy (e.g., Olaparib) became the recommended standard of care for all individuals with invasive ovarian cancer regardless of family history (Daly et al., 2010), as use of PARPi is associated with improved progression-free survival in individuals with germline *BRCA* 1/2 P/LPVs (Lee et al., 2021). In 2019, the National Comprehensive Cancer Network (NCCN) recommended offering universal germline genetic testing for all individuals with pancreatic cancer (Pilarski et al., 2021), and in 2023, the Canadian Urological Association released best practice guidelines for the offering of universal germline testing in all individuals with metastatic prostate cancer to inform the indication of PARPi therapy (Rendon et al., 2023). Notably, the efficacy and tolerance of several somatically targeted therapies, which are now routinely used in people without inherited cancer susceptibility, were first established in individuals with a known CPS (Glaire et al., 2017; Mastrodomenico et al., 2023).

Germline Genomic Applications in Cancer Prevention, Screening, and Survivorship

Predictive genomic testing to confirm a CPS before the onset of a cancer allows a window of opportunity for targeted prevention and risk reduction strategies. Even beyond the two most well-known CPS (Lynch and HBOC Syndrome), evidence-based, expert panel consensus-based (≥85%) clinical practice guidelines, such as those of the NCCN (2024a; 2024b), now exist for cancer risk management recommendations in individuals with several highly and moderately penetrant CPS. When a CPS is identified through treatment-focused genetic testing (TFGT) in people with a cancer diagnosis, these guidelines are also valuable for informing cancer survivorship care to prevent cancer recurrence and the development of other cancers, as CPSs typically affect multiple organs (Garutti et al., 2023). TFGT also opens a window to initiate cascade testing. As most CPS P/LPV follow an autosomal dominant inheritance pattern, cascade testing involves offering genetic testing to unaffected family members of the initial patient found to carry a P/LPV (i.e. the proband) (Garutti et al., 2023). This extends the benefits of evidence-based prevention and risk reduction guidelines across families and generations.

Some of these evidence-based genomics-informed cancer prevention strategies are discussed below.

Enhanced Screening Protocols. NCCN (2024a; 2024b) high-risk management guidelines typically recommend earlier and enhanced cancer screening protocols in people with known CPS. The benefits of endoscopy surveillance in high-risk populations are well established (Barrow et al., 2013). In their systematic review, which included 43 cohort observational studies, Barrow et al. (2013) found that adherence to registry-initiated screening resulted in a reduction in colorectal cancer incidence and mortality for people with Familial Adenomatous Polyposis (FAP) in 33 of 33 studies, and with Lynch Syndrome in 9 of 10 studies. For example, colonoscopic surveillance in individuals with Lynch Syndrome has been associated with a 65% ($p=0.03$; RR = 0.348; 95% CI: 0.12–0.90) to 72% ($p < 0.025$; RR = 0.28, 95% CI: 0.20-0.59) reduction in colorectal cancer deaths (Dove-Edwin et al., 2005; Jarvinen et al., 2000). In individuals with Lynch Syndrome and other colorectal cancer–associated CPS, the recommended age to begin colonoscopy screening depends on the gene involved, based on its associated phenotype and penetrance. Table 1.7 summarizes the recommended ages for colonoscopy screening based on the gene involved. Depending on the gene/CPS involved and its associated phenotype, for example, in Lynch Syndrome, risk management often entails screening of multiple organs. In HBOC management, a high-risk breast screening protocol, involving annual MRI and annual mammography (at six months intervals), is an option for people with *BRCA* 1/2 variants, as this high-risk protocol has a higher sensitivity than mammography alone for detecting breast cancer and is linked to improved quality-adjusted life years (NCCN, 2024b).

Table 1.7

Recommended Age to Initiate High-Risk Colonoscopic Surveillance According to Genotype as per NCCN (2024b) Guidelines

Population/Gene	**Recommended Age to Initiate Colonoscopy**
General Population (average risk)	Individualized based on clinical factors
APC (FAP)	10–15
MLH1 (LS)	20–25
MSH2 & EPCAM (LS)	20–25
MSH6 (LS)	30–35
PMS2 (LS)	30–35
MUTYH (biallelic)	25–30

Risk-Reducing Surgery. The preventative benefits of prophylactic surgery (i.e., in the absence of a current cancer diagnosis) in persons with HBOC syndrome were established through large meta-analyses and multi-centre cohort studies (Rebbeck et al., 2009; Domcheck et al., 2010; Marchetti et al., 2014). In two meta-analyses, risk-reducing salpingo-oophorectomy (RRSO) or surgical removal of the ovaries and fallopian tubes in unaffected persons with a *BRCA* 1/2 P/LPV was associated with a 79% (HR = 0.21; 95% CI: 0.12–0.39) to 80% (HR = 0.19; 95% CI 0.13 – 0.27) reduction in ovarian cancer risk (Marchetti et al., 2014; Rebbeck et al., 2009). Rebbeck et al. (2009) also reported a 51% reduction in breast cancer risk (HR = 0.49; 95% CI: 0.37–0.65) associated with RRSO. In a multi-centre prospective cohort, RRSO was associated with lower breast cancer mortality (6% vs. 2%; HR = 0.44, 95% CI: 0.26–0.76), ovarian cancer mortality (3% vs. 0.4%; HR = 0.21, 95% CI), and lower all-cause mortality (10% vs. 3%; HR = 0.40, 95% CI: 0.26–0.61) in individuals with HBOC syndrome (Domchek et al., 2010). Instead of high-risk breast surveillance protocols, individuals with *BRCA* 1/2 P/LPVs also have the option of risk-reducing mastectomy, shown in a meta-analysis to reduce breast cancer risk by 93% (HR 0.07; 95 % CI 0.01–0.44; p = 0.004) (De Felice et al., 2015). The recommended age for these preventative surgeries is also gene specific based on its associated penetrance (NCCN, 2024a). While the clinical and/or survival benefits of these risk-reducing surgeries are established, there have been reports of associated psychological distress and decisional regret. RRSO and risk-reducing mastectomy can impose body-image related adverse sequelae (Bai et al., 2019; Gopie et al., 2012; Isselhard et al., 2023). Further, some who elect prophylactic RRSO experience distressing physiological and psychosexual symptoms related to its surgically induced menopause (Graziottin et al., 2010; Rocca et al., 2018). International consensus guidelines exist for the care of premenopausal women who undergo RRSO to mitigate the sometimes-distressing vasomotor symptoms, sleep disturbances, and sexual dysfunction they experience, as well as to prevent the risk of bone and cardiovascular disease associated with RRSO (Nebgen et al., 2023).

As one example of surgical CPS management outside of HBOC, the removal of the stomach, or risk-reducing total gastrectomy, is recommended in best practice guidelines for individuals with highly penetrant P/LPVs in the *CDH1* gene linked to hereditary diffuse gastric cancer (NCCN, 2024a). In individuals with a *CDH1* P/LPV who undergo risk-reducing total gastrectomy, microscopic carcinoma is often found incidentally on their surgical pathology (van der Post et al., 2015). With this information and the known low cure rate in the setting of symptomatic gastric cancer (around 10%), risk-reducing total gastrectomy is generally regarded as a life-saving measure (Koea et al., 2000; van der Post et al., 2015). However, many *CDH1* P/LPV carriers who undergo preventative gastrectomy experience complications and long-term nutritional sequelae that impact quality of life (Gallanis et al., 2024). Thus, in guidelines for all risk-reducing surgeries, informed surgical decision-making and timing of surgery are made on an individualized basis with an overview of the risks and benefits, including potential impact on quality of life, family, and reproductive planning (NCCN 2024a, 2024b).

Chemoprevention. A protective effect has been established from oral medication use in several CPS populations. For example, in 20-year follow-up data from an international double-blinded placebo-controlled trial, individuals with Lynch Syndrome who took daily aspirin for two years had a 35% reduction in colorectal cancer risk (HR = 0.65, 95% CI, 0.43–0.97; p = 0.035) (Burn et al., 2020). In multi-center trials, the use of oral contraceptive medication was shown to reduce ovarian cancer risk in individuals with *BRCA*1/2 P/LPVs (Iodice et al., 2010; van Bommel et al., 2023). However, it is recommended that the decision to prescribe these medications for cancer prevention is made on an individualized basis, factoring in other potential risk factors (Serrano et al., 2022; van Bommel et al., 2023).

The Gap Between Cancer Genomic Evidence and Adoption

Despite the explosion of evidence-based genomic applications in cancer treatment and prevention which are shown to reduce cancer morbidity and mortality, a gap persists in the translation of these applications into routine clinical care (Ahsan et al., 2024; Shore et al., 2021; Mittendorf et al., 2021; Muessig et al., 2022; Linfield et al., 2022; Snow et al., 2024; Tindale et al., 2022). Firstly, cancer genomic testing is underutilized (Ahsan et al., 2024; Shore et al., 2021; Drohan et al., 2012; Linfield et al., 2022; Menko et al., 2019). It is estimated that only 10% of people with a germline P/LPV in *BRCA1 and BRCA 2* are aware that they have the P/LP variant and associated risk (Drohan et al., 2012). In a US retrospective cohort study, of the 14,688 patients found to be eligible for cancer genetic counselling based on family history, only 21.9% of those eligible for genetic counselling were in fact referred (Linfield et al., 2022). Although the NCCN recommends germline testing for all people with metastatic, castration resistant prostate cancer, it was found in a US study that only 13% of eligible individuals had undergone genetic testing (either germline or somatic) (Shore et al., 2021). In a systematic review, uptake of pre-symptomatic (cascade) testing of relatives ranged from 21 to 44% (in persons with HBOC Syndrome) and 41 to 94% (in persons with Lynch Syndrome) (Menko et al., 2019). A more recent systematic review and meta-analysis found that uptake of cascade genetic testing in hereditary cancer among eligible biological relatives was only 41%; 95% CI 34-48% (Frey et al., 2022). In NL, while over 600 individuals with a CPS have been identified since the advent of clinical genetic testing in the Provincial Medical Genetics Program (PMGP), personnel within the PMGP estimate that more than 1500 individuals with a CPS remain unidentified in NL (Etchegary et al., 2022).

The second key point about actionability and uptake of genomics is that a molecular test result revealing a P/LPV in a cancer susceptibility gene, strictly clinically speaking, only has utility if it leads to uptake of evidence-based risk-reduction strategies and health behavior changes (Goddard et al., 2022). Compared to the volume of research surrounding the uptake of genomic testing, there is comparably less research examining the uptake of recommended risk strategies in affected persons (Butkowsky, 2024). However, evidence suggests that the recommended cancer risk-reducing strategies are underutilized in individuals with a confirmed CPS (Mittendorf et al., 2021; Mesa-Chavez et al., 2024; Roebothan et al., 2023). In a narrative review, Mittendorf et al. (2021) found that uptake of recommended enhanced CPS screening protocols ranged from 52%-85% depending on surveillance type, and recommended risk-reducing surgery uptake ranged from 9% and 65%. It was found in a NL retrospective study that almost 40% of eligible individuals with *BRCA* P/LPVs were not undergoing their annual recommended enhanced breast screening over 18 months (Roebothan et al., 2023).

Barriers to the Uptake of Evidence-Based Cancer Genomic Applications

As evidenced from the above literature on health technology assessment and ELSI considerations in genomics and the multiple factors involved in CPS decision-making, genomics applications are inherently complex. It is therefore unsurprising that the factors contributing to the gap in evidence-based genomic applications and their integration into routine cancer care are similarly complex, occurring at the system, provider, and patient levels (Best et al., 2025; Husereau et al., Mittendorf et al., 2021; Mesa-Chavez et al., 2024; Smith-Uffen et al., 2021; Snow et al., 2024). Across many studies describing these barriers, it is reported that the barriers across all three levels interact with the wider social and structural determinants of health. Consequently, barriers to accessing cancer genomics services are disproportionately experienced among groups facing intersecting forms of marginalization, including structural racism, living in medically underserved geographical areas (e.g. rural, remote areas), socioeconomic disadvantage, and those with limited health literacy (Lau-Min et al., 2023; Mittendorf et al., 2021; Snow et al., 2024). Thus, attention to these equity considerations is needed; otherwise, genomics carries the potential to widen existing disparities (Khoury et al., 2022; Curtin et al., 2022; Genome Canada, 2021).

Health System Level-Barriers

In Canada, healthcare services are administered at the provincial level; thus, there is no harmonized national approach to cancer genomic service delivery. A recent environmental scan and interest holder consultation study was conducted with the provincial leads for hereditary cancer programs across Ontario, British Columbia, and NL (Sam et al., 2023). The informants provided descriptions of the available services in these provinces, revealing three primary system-level causes of fragmented and inequitable hereditary cancer care across Canada.

Inconsistencies and Inefficiencies in Genetic Testing Referral and Eligibility Criteria. While a pan-Canadian agency leads standardized health technology assessment processes, each province is ultimately responsible for implementing its health and drug technology coverage policies (Canada's Drug Agency, 2025). Most provinces offer multi-gene panel testing for HBOC, Lynch Syndrome and other CPS, but gene panel content can differ, meaning that some gene panel coverage is being missed (Snow et al., 2024). Eligibility criteria for cancer genomic testing can vary by province and have traditionally been limited to those with significant family history and other clinical criteria suggestive of a hereditary phenotype (Tindale et al., 2022); For example, due to differences in age cut offs or tumour subtype criteria, a woman with a breast cancer diagnosis at age 45 would qualify for publicly funded genetic testing in the province of Ontario but would not in NL or Alberta (Government of Alberta, 2025; Hereditary Cancer Testing Eligibility Working Group, 2024; NL Health Services, 2025). It has been argued that current restrictive eligibility criteria result in the failure to identify an estimated 50% of individuals who harbor P/LPVs associated with HBOC (Li et al., 2019). There have been calls to offer germline testing for all individuals with solid cancer, based on emerging evidence that universal testing may result in improved identification of actionable germline results when compared to family history and guideline-based criteria (Jones et al., 2023; Samadder et al., 2021). In a recent cost-effective analysis, it was proposed that when compared to the current approach to testing by referral criteria, population-based screening for *BRCA 1/2* in Canada would be cost-effective and prevent 39,228 breast or ovarian cancer cases and 4,619 breast and or ovarian cancer deaths (Sun et al., 2024). These referral and eligibility restrictions also carry the potential to widen inequities, often referred to as the 'genomic divide', as individuals of socioeconomic advantages can pay out of pocket for this testing (Grant et al., 2023).

Shortage of Specialty Genetics Workforce. Traditionally in Canada, individuals are referred to specialty genetics services and seen by a genetic counsellor before publicly funded genetic testing is offered (CAGC, 2025). Genetic counselling services provided by a genetic counsellor are associated with increased patient knowledge, improved personal control, risk perception accuracy, as well as uptake of positive health behaviors in people who underwent cancer genomic testing (Braithwaite et al., 2006; Madlensky et al., 2017; Yuen et al., 2020). In Canada and globally, many genetics centres struggle to meet the rising demands for hereditary cancer testing and services (Dragojlovic et al., 2023; Dragojlovic et al., 2020; Stoll et al., 2018). Highly variable, yet unacceptably long wait times to be seen by specialty genetics services for pre-counselling and testing have been reported across Canada (e.g. reported waitlist highs of seven years for ’non-urgent’ cases in NL) (Cancer Care Ontario, 2018; Singer, 2022). Most centers do not have service capacity to offer long-term follow-up for people with a confirmed CPS post-results disclosure (Butkowsky, 2024).

Interaction with genetic counsellors therefore tends to be limited to two consultations; pre and post-test counselling, this fragmentation of access to expert care does not allow for spaces where people with autosomal inherited disorders can have their care needs met that reflect the fluid nature of lifelong engagement with risk perception and risk management (Esplen & Kohut, 2025; Etchegary et al., 2011; Manuel & Brunger, 2015). Several oncology clinics across Canada and globally have pivoted to ‘mainstream’ models of oncogenetics services where oncology prescribers order tests directly without genetics clinic referrals for certain cancers (Barnhardt et al., 2023; Bokkers et al., 2022; Lapointe et al., 2021; Rauw et al., 2022). This alternative service delivery model shows promise to enhance testing access; however, it is not consistently implemented nationwide, and there is significant heterogeneity in the features of mainstreaming models across Canada and globally (Bokkers et al., 2022; Mackley et al., 2025).

Inconsistent/Lack of Provincial High-risk Screening and Follow-up Programs. There is variation in approaches to high-risk surveillance and available follow-up care for those with a confirmed CPS across Canada. While some provinces offer high-risk breast screening programs and or hereditary cancer follow-up clinics, others do not, and there currently are no provincial high-risk prostate, ovarian, and pancreatic screening programs (Canadian Partnership Against Cancer, n.d.; Butkowsky, 2024). Most high-risk individuals are referred to their primary care provider to coordinate their long-term surveillance and risk management (Canadian Partnership Against Cancer, n.d.; Tindale et al., 2022). In a recent qualitative study of 73 Canadians living with HBOC or Lynch Syndrome, participants likened the experience of this prevailing, fragmented approach to CPS follow-up care as "navigating a lifelong road without a map" (Butkowsky, 2024, p. 32). Individuals living with a CPS in NL have reported a desire for 'one stop shopping'- a centralized place to access their CPS-associated healthcare (Hynes et al., 2023; Watkins et al., 2011).

Provider-level Barriers

It has been found that even when evidence-based recommendations are available for genetic testing and related follow-up care across healthcare systems, barriers at the provider-level, such as knowledge deficits, and inefficient communication can considerably impact uptake of testing and risk management recommendations.

Lack of Knowledge Among Non-Genetics Providers. Despite new mainstream/TFGT models in oncology settings, several studies including systematic reviews have reported that both oncology and primary care providers, for example, often have limited knowledge and low confidence related to genomics-informed care, including awareness and interpretation of tests that can be ordered and awareness of evidence-based risk management guidelines (Bishop et al., 2024; Bokkers et al., 2022; Febbraro et al., 2015; Laforest et al., 2019; Loeb et al., 2021; Mikat-Stevens et al., 2015; Snow et al., 2024).

In studies where patients described their experiences with their lifelong CPS risk management across NL, Canada, and globally, many participants perceived that healthcare providers lacked knowledge about their CPS (Campbell-Salome et al. 2021; Mooney et al., 2023; Etchegary et al. 2015; Park et al. 2023; Watkins et al. 2011; Rauscher et al., 2018; Warner & 2022; Warner et al., 2022). In several qualitative studies, patients with a CPS described how the lack of provider knowledge or ineffective communication contributed to their distressing experiences when navigating the healthcare system (Campbell-Salome et al. 2021; Dean & Davidson 2018; Strømsvik et al., 2022; Warner & Groarke 2022). These findings are concerning when considering that many patients with CPS consistently note the importance of accurate, clear information from providers in their genetic test decision-making (Jacob et al., 2018; Mallen et al., 2020), as well as in their decision-making related to risk management (Arts-De Jong et al., 2015; Mittendorf et al., 2021; Park et al., 2023). Numerous cases have also been reported where non-genetic providers inaccurately interpreted genomic test results, leading to low-value care and even iatrogenic patient harm such as unnecessary surgeries (Bonadies et al., 2014; Brierley et al., 2012; Farmer et al., 2019).

Other Provider-Level Barriers. In a systematic review that included 38 studies, in addition to the lack of knowledge and access to continued education, primary care providers identified the lack of time and concerns surrounding patient anxiety related to cancer risk as barriers to their adoption of genomics into their practice (Mikat-Stevens et al., 2015). Provider-level barriers also intersect with system-level barriers; in the absence of dedicated high-risk follow up programs, uncertainty has been reported among generalist and specialist providers as to which providers are ultimately responsible for CPS care coordination, leading to gaps in care experienced by patients who were then left to navigate this ambiguity on their own (Mittendorf et al., 2021; Watkins et al., 2011; Hynes et al., 2023).

Patient-Level Barriers

Limited Awareness/Knowledge. The literature suggests that patients are highly interested in cancer genomic testing for its implications for themselves and their families, especially when it is actionable (Smith-Uffen et al., 2021). Many patients are unaware that genetic testing is available in cancer care, or that it can guide their treatment and prevention options (Snow et al., 2024; Wolyniec et al., 2020; Smith-Uffen et al., 2021). In a systematic review that included (n=9) studies of the experiences of people with cancer most participants lacked understanding about TFGT and desired more information (Wolyniec et al., 2020). Concerningly, in a study of 85 patients with a cancer diagnosis who underwent genetic testing in an embedded oncology clinic, 30% surveyed did not recall having had genetic testing (Wing et al., 2021). Of those who did recall being tested, nearly half did not accurately recall their specific gene or variant-level results. In a pan-Canadian qualitative study of 73 persons with a CPS, many had limited knowledge of the Canadian Genetic Non-Discrimination Act and its legal protections (Gopalakrishnan et al, 2024, Government of Canada, 2017). The *Genetic Non-Discrimination Act* is a federal law instituted to reduce barriers to appropriate use of testing and protect Canadians from potential genetic discrimination by making it unlawful for any person providing services or entering a contract with someone to require them to undergo or disclose results of any molecular genomic test.

Attitudes and Perceptions. Patients' lack of perceived utility is another commonly reported patient-level barrier (Tasnim et al., 2025). While perceptions of limited utility can be linked to knowledge deficits, as discussed, it can also be connected to risk perception which is influenced by a myriad of contextual factors which can change over time; this means that individuals may engage differently with risk perception and management as circumstances change in their life such as the death of a loved one (Etchegary, 2011; Manuel & Brunger, 2015). It has been found that some people delay or avoid potentially unwanted information related to disease risk, often either as a coping strategy for regulating emotions, to avoid taking unwanted actions in response to this information, or if this information is perceived as being potentially at odds with their fundamental beliefs (Case et al., 2005; Emanuel et al., 2015; Taber et al., 2015; Sweeny et al., 2010).

Family Considerations. Family communication about genetic risk impacts individual perceptions of risk, utility, and decision making (Campbell-Salome & Rauscher, 2020). In the current cascade testing model, the person in the family first found to have the genetic condition (proband) is ultimately responsible for communicating genomic risk information with potentially at-risk biological relatives, despite studies indicating that this is not effective (Ahsan et al., 2024). Probands have described this task as burdensome and family members can react with distress or avoidance, which can hinder cascade testing uptake (Levine et al., 2024). Further, the permanent implications for family planning associated with certain surgical risk management options, such as RRSO, can lead to decision uncertainty and avoidance or delayed uptake of risk-reducing surgery for individuals of child-bearing age and capacity (Fine et al., 2022; Rauscher & Dean, 2017).

Access and Practical Barriers. Practical barriers can further limit engagement with genetic testing, such as scheduling conflicts and competing life responsibilities, which may reduce the ability to attend genetic-related appointments (Borle et al., 2025; Etchegary, 2011). Concerns about the potential insurance discrimination have been identified by patients as barriers to uptake of hereditary cancer testing (Kahn et al., 2022; Tasnim et al., 2025). Patients have also identified the costs of testing or follow-up care not fully covered by healthcare systems as barriers to genomic testing and related care (Grant et al., 2023; Borle et al., 2025).

One Strategic Entry Point for Closing the Genomic Adoption Gap: Nursing

As evidenced by these complexities contributing to the observed lag in translating genomics into routine health and cancer care, effectively integrating genomics into healthcare requires a “whole-of-system change” (Best et al., 2025, p. 1739). Thus, examining it from a single sectoral or disciplinary perspective is likely insufficient. However, against the backdrop of these genomic applications and related challenges, it is widely recognized that enhanced genomics health workforce preparation is an essential piece of genomics-informed health system transformation (Best et al., 2025; CIHR, 2022; Stark et al., 2019).

With the increase in mainstream/TFGT testing models in oncology, there have been calls for enhanced medical education about genomics, as well as genomics education for medical oncologists in Canada (Snow et al., 2024; Yip et al., 2019). It is crucial that oncology prescribers understand the implications of genomics, as they are often now tasked with ordering these tests in these mainstream models (Bokkers et al., 2022). However, Rahman et al. (2022) included oncologists and oncology nurses in their scoping review of oncology providers’ genomics education needs, on the basis that both cancer care providers are positioned to provide patients with genomic information. Nurses in cancer care settings have prominent roles in delivering cancer care and frequently interact with patients and families; they provide comprehensive health assessment, supportive care and patient and family teaching, support decision-making, and facilitate system navigation for patients and families across their extended cancer journeys (CANO/ACIO, n.d.; Naito, 2024; Prip et al., 2022). Research has shown that cancer nurses promote patient and family agency, engagement, and satisfaction with care (Azarabadi et al., 2024; Prip et al., 2022). Core disciplinary concepts emphasized across historical and contemporary nursing theoretical literature and professional codes of ethics include: holism, complexity, person-centred care, patient advocacy, and the social determinants of health (Clancy, 2024; Carper, 1978; Fowler, 2016; International Council of Nurses, 2021; McCormack & McCance, 2006; Peplau, 1991; Schim et al., 2007). Thorne (2015) summarized what nursing uniquely contributes to healthcare as follows:

> Whether we nurse at the level of populations or people, the capacity to apprehend the impact of conditions upon individual experience is and always will be at the heart of nursing. We recognize that experience as inherently holistic or integrated, such that changes to any part can be felt across all aspects of the whole. We seek out knowledge that will help us understand commonalities in that experience, but we are always simultaneously mindful of the variations. We know that it is in those margins of patterned human experience that the most vulnerable reside, and therefore, that the potential for harm is the greatest. Our philosophical commitment to both justice and equity keeps our moral compass directed toward those places where we run the greatest risk of missing the mark in our covenant for service. In that light, although we enthusiastically endorse the powerful contributions of science to our understanding of human health and illness experience, we also know that the standardizing tendencies of evidence-based practice can run roughshod over the uniqueness of individual and cultural needs, preferences and rights (p. 284).

Juxtaposing this proclaimed disciplinary contribution with the unmet person-centred supportive care needs reported by people with CPS (Campbell-Salome et al. 2021; Dean & Davidson 2018; De Los Riez de la Serna et al., 2024; Strømsvik et al., 2022; Warner & Groarke 2022) illuminates a potential synergy between the care that nurses are poised to provide and the care that persons with a CPS report that they need. Nurses in cancer care settings occupy a uniquely valuable position to potentially provide genomics-informed cancer care, as patients and families with cancer are typically followed closely by cancer care teams during treatment and receive ongoing follow-up for several years following treatment (Canadian Cancer Society, n.d.). Thus, cancer care settings are a possible option to meet the reported need for relational, longitudinal spaces in healthcare settings, where people with genetic disorders can receive person-centred, supportive care that accounts for changes in risk perception and engagement with risk-management over time (Etchegary, 2011; Esplen & Kohut, 2025; Manuel & Brunger, 2015).

Widespread recognition of this synergy and potential contribution of cancer care nurses to genomics-informed care is evidenced across the several non-empirical publications in oncology nursing journals (e.g., discussion and clinical resource papers) (Aiello, 2015; Beamer et al., 2013; Blix, 2014; Boucher et al., 2014; Burkett, 2024; Calzone et al., 2002; Calzone & Masny, 2004; Carlsson & Limoges, 2022; Cassells et al., 2003; Dickman et al., 2025; Dimond et al., 1997; Dodson, 2017; Eggert, 2017; Flynn et al., 2019; Greco, 2000; Jenkins, 2011; Jenkins & Masny, 2003; Kerber & Ledbetter, 2017; Lea, 1997; Loescher, 1999; Lopez, 2018; Loud et al., 2002; Macdonald, 1997; Middleton et al., 2002; Milani et al., 2023; Peters et al., 2001; Santos et al., 2013; So-Shan Mak & Leong-Tat Chan, 2024; Vorderstrasse et al., 2014). However, what was less clear from initial scans of the oncology nursing literature was whether oncology nurses possess the requisite genomic competencies to enact these disciplinary contributions in genomic-informed care. This gap pointed toward exploration of the literature related to nurses' genomics competencies, which is the focus of the remainder of the literature review.

Genomic Competency Development in Nursing

The College of Registered Nurses of Newfoundland and Labrador (2019) defined competencies broadly as nurses' ability to integrate the "knowledge, skills, abilities and judgment required to practice nursing safely and ethically" (p. 1). Nursing is a competency-based profession; 'readiness' for entry-to-practice is determined based on mastery of entry-level competencies (College of Registered Nurses of Newfoundland and Labrador, 2019). Competencies are communicated as statements of direction usually from professional nursing authorities, such as nursing education and practice regulators and professional associations. They can vary in scope and specificity ranging from entry-level to highly specialized competencies, but this core definition of competency remains stable across various contexts in nursing.

Following the completion of the Human Genome Project, the US and United Kingdom (UK) governments invested heavily in genomics capacity building to enhance healthcare delivery (Department of Health, 2003, NHGRI, 2024). Alongside these efforts, national nursing leadership in both countries worked to develop competency statements for nurses across all practice areas to set the perimeters for the competencies required of nurses to provide genomics-informed care (Calzone et al., 2018; Jenkins et al., 2005; Jenkins & Calzone, 2007; Kirk et al., 2003). Genetic nursing competency frameworks were first introduced in the UK in 2003 (Kirk et al., 2003), revised in 2014 (Kirk et al., 2014), and most recently again in 2023 (NHS, 2023). Similarly, the American Nurses Association (ANA) convened a panel of experts in 2005 to develop genetic and genomic competency statements for baccalaureate-prepared nurses (Consensus Panel on Genetic/Genomic Nursing Competencies 2006), now in its third edition (ANA, 2003); this was followed by the release of the genomic competencies for graduate level nurses (Greco et al., 2012), also currently being updated and revised (Tonkin et al., 2025).

Canadian Context

During the same period of genomic nursing competency development in the US and the UK, a core Pan-Canadian nursing leadership group led a CIHR-funded planning forum and related research to develop strategic directions for Canadian nursing practice in the genomic era. (Bottorff et al., 2004; Bottorff et al., 2005). One key recommendation of this research was establishing Canadian genetic nursing competency statements (Bottorff et al., 2004). Canadian research in recent years revealed that no comparable stand-alone genomic nursing competencies have been developed in Canada (Limoges et al., 2025; Puddester et al., 2023). However, genomic content was included in the CANO/ACIO (2006) general standards and competencies document for the specialized oncology nurse, including that oncology nurses understand genetic risk factors, assess family history, provide patient education related to genetics in care, and help refer patients to genetic counseling and resources.

Assessing Genomic Competency Across the Nursing Profession

Since the release of genetics/genomics nursing competencies, several integrative, systematic, and scoping reviews have been conducted to synthesize the global literature assessing nurses' competency in genomics and related outcomes across all domains of practice (Anderson et al., 2015; Cao et al., 2025; Dante et al., 2025; Keels et al., 2024; Kirk et al., 2012; Laaksonen et al., 2023; Thomas et al., 2025; Rahman et al., 2022; Wright et al., 2018). In this section, common instruments used to assess nurses' genomic competencies, as frequently reported in the literature, will be described. Key findings of knowledge syntheses will be summarized according to three main categories: 1. Descriptive assessments of nurses' knowledge, attitudes, and competencies concerning genomics (i.e. 'provider-focused' outcomes); 2. Outcomes of educational interventions targeting nurses' genomic competency; 3. and patient-outcomes related to the provision of genomic-informed nursing care. In addition to summarizing findings of synthesis studies conducted across all areas of nursing globally, I also highlight findings of primary studies specific to genomics in oncology nursing.

Instruments Measuring Nurses' Genomic Competency

Across the global research surrounding nurses' genomic competency, some studies used validated, robust measures, while others utilized researcher-developed tools that had not undergone comprehensive psychometric evaluation (Dante et al., 2023; Thomas et al., 2023; McLaughlin et al., 2024). In an evidence review, Anderson et al. (2015) retrieved and critically appraised six psychometric instruments measuring nurses' genomics competency. In a more recent scoping review, Laaksonen et al. (2023) identified 19 unique instruments that measure nurses' genomic competency. Current consensus is that the two most widely used validated instruments, the Genomic Nursing Concept Inventory (GNCI©) (Ward et al., 2014) and the Genetics and Genomics Nursing Practice Survey (GGNPS) (Calzone et al., 2012; Plavskin et al., 2019; Plavskin et al., 2023), are the gold standards for assessing nurses' genomic knowledge and competency (Dante et al., 2025; Thomas et al., 2023; Tonkin et al., 2025). Although no validated tools were found assessing genomic competency which were specific to oncology nursing, both the GGNPS and the GNCI© have been utilized in studies that included nurses practicing in multiple practice areas, oncology among them (Alloubani et al., 2023; Zhao et al., 2022).

GNCI©

The GNCI© is a concept inventory; a multiple-choice, psychometric tool, containing distractors designed to distinguish respondents who understand the underlying concepts (Ward et al., 2014). The GNCI© consists of 31 multiple-choice questions assessing knowledge across four topical categories: (a) genome basics, (b) mutations, (c) inheritance, and (d) genomic health care.

The GNCI© items were mapped to the US *Essential Nursing Genetic and Genomic Competencies* for baccalaureate-prepared nurses. The GNCI© has been used in several global studies to measure genomic knowledge among practicing nurses and nursing students (Adejumo et al., 2021; Connors et al., 2022; Dewell et al., 2020; Lim et al., 2025; Kronk et al., 2024; Parviainen et al., 2023; Read & Ward, 2016; Ward et al., 2016; Wright et al., 2019). It was recently translated in Greek and validated (Giakoumidakis et al., 2025).

GGNPS

The GGNPS is another commonly used, validated discipline-specific instrument (Calzone et al., 2012). The GGNPS differs from the GNCI© in that it not only assesses nurses' genomic knowledge, but also other dimensions of competency, including their attitudes/receptivity, decision/adoption of genomics into nursing practice (using family history and referral to genetics as benchmarks of adoption), as well as the influence of wider social systems on their use of genomics in practice. These operational domains of the GGNPS are mapped to the conceptual domains of Rogers' (2003) Diffusion of Innovation (DOI) theory (attitudes, knowledge, influence of the social system), as well as confidence) which are considered antecedents of the adoption of innovations, such as genomics. An adapted version of the GGNPS, the 'GGNPS-CA', was distributed recently to nurses practicing across Canada as a nationwide assessment of genomic competency (Limoges et al., 2024). The GGNPS has been cross-culturally adapted and validated for use in Turkey (Yeşilçinar et al., 2022), Hong Kong, Taiwan, and mainland China (Chow et al., 2023; Wang et al., 2023; Zhao et al., 2022). The benefit of validated, adapted versions of the GGNPS is that they make it possible to assess nurses' competency on a global scale through data aggregation and possible meta-analyses (Tonkin et al., 2025).

Describing Nurses' Genomic Competency: Findings Across Multiple Reviews

There is consistent evidence from multiple knowledge syntheses that nurses have limited knowledge and clinical skills related to genomics. Skirton et al. (2012) conducted one of the first synthesis studies of nurses' genomic competency in their integrative review, which included 13 cross-sectional studies. Overall, they concluded that, while many nurses recognized the relevance of genetics to their role, most did not possess the theory-based knowledge of genomics or the clinical skills needed to meet the expected level of genomic competency outlined in the genomic competency statements. Five years later, Wright et al. (2018) published an updated mixed-methods integrative review on the topic, which included an additional 12 studies (10 cross-sectional and two qualitative studies) published since the original review by Skirton et al. (2012). They concluded that there was minimal evidence of global progress related to nurses' genomic competency development over the five years (Wright et al., 2018).

Knowledge synthesis studies conducted in recent years are suggestive that deficits in nurses' genomic competencies persist globally (Dante et al., 2025; Thomas et al., 2023). Thomas et al. (2023) reported on 126 studies examining nurse-oriented outcomes related to genomics (i.e., knowledge, attitudes, and receptivity) from a systematic scoping review of 232 studies on the topic of genomics nursing over 10 years (2012–2022). While nurses appear to demonstrate positive attitudes towards genomics-informed care, they lack the knowledge and confidence to apply it in practice. In most studies included, nurses reported that they perceived genomics as important, but in other studies, nurses identified that they struggled to understand their specific role in genomics-informed care. Similarly, another recent scoping review synthesizing 63 global studies assessing genomic literacy among nurses, nursing students, and faculty concluded that nurses' overall genomic nursing competency remains limited (Dante et al., 2025).

Nurse-oriented Genomic Competency Outcomes (Oncology Specific)

There were relatively few empirical studies retrieved that specifically investigated the genomic competency of nurses in cancer care settings (Chiu et al., 2024; Hébert et al., 2022; Hines-Dowell et al., 2024; Seven et al., 2018; Wright et al., 2020). In a cross-sectional survey conducted with 98 oncology nurses in Turkey, Seven et al. (2018) found that the respondents had moderate levels of knowledge about the genetics of HBOC with an average knowledge score of 6.74/13 ± 3.85 (52% correct). However, the respondents endorsed a willingness to engage in further education about genomics. Wright et al. (2020) conducted qualitative interviews with nine oncology nurses practicing in Australia. The nurses reported low genomic competency levels, and overall, they did not perceive genomics as relevant in the day-to-day nursing care they provide.

In a cross-sectional study of (n=199) cancer nurses practicing in the UK, 95% of respondents reported that they predicted genomics to profoundly impact their practice within the next five years and indicated their desire for further education (Cuthill et al., 2025). However, approximately half of respondents rated their confidence with genomics-informed practice as low, and only 39% had reported engaging in interprofessional collaboration with their clinical genetics team members over the past year. In another cross-sectional survey of 153 nurses practicing across different areas in the UK, the 32 cancer clinical nurse specialist respondents had higher knowledge of targeted therapies than nurses in other specialties (Carpenter-Clawson et al., 2023). However, they still reported low confidence distinguishing between germline and somatic variants. Akin to the findings of Carpenter-Clawson and colleagues (2023), Hines-Dowell and colleagues (2024) found that only 55% of respondents correctly identified the difference between a germline and somatic genetic variant in a recent US-study cross-sectional study of RNs (n=132), NPs (n=29) and physicians assistants (n=2) practicing in a pediatric oncology research hospital. Moreover, even though 40% of respondents reported that patients or families either ‘occasionally’, ‘frequently’, or ‘very frequently’ asked them questions about genomics, 72% identified that they felt unprepared to answer such questions (Hines-Dowell et al., 2024). Consistent with findings from both studies indicating that oncology nurses have limited knowledge and confidence in distinguishing between germline and somatic variants, a scoping review exploring the global genomic education needs of oncologists and oncology nurses revealed not only a lack of empirical research on oncology nurses' genomic competencies, but a lack of any publications that specifically addressed nurses' understanding of the somatic aspects of genomics (Rahman et al., 2022). This gap is noteworthy given that both types of testing are now common in cancer care, and the identification of a somatic versus germline P/LPV carries very different implications for patients and their families (NHS England, 2024).

Canadian Oncology Nursing Context

Two empirical studies specific to the Canadian context were found related to cancer nurses’ genomic competency. In a cross-sectional survey of 40 nurses in Quebec who provided cancer care, overall, it was found that they lacked the knowledge and confidence to engage with the topic of hereditary cancer in their practice (Hébert et al., 2022). Their responses to knowledge questions were indicative of common misconceptions about hereditary cancer risk. Moreover, respondents indicated that patients (not providers) were the ones to initiate discussions about hereditary cancer risk. In a recent Pan-Canadian qualitative study, oncology nurses reported that patients increasingly approach them with questions about genomics and they recognized the need for further disciplinary upskilling in genomics (Chiu et al., 2024).

No study was conducted in Canada or internationally, exclusively focused on cancer nurse populations that also used psychometrically validated tools to assess nurses' genomic competency. While some studies using the GGNPS or GNCI© instruments reported oncology nurses' knowledge/competency scores, these were only reported as sub-scores of larger studies that included nurses from multiple clinical areas. For example, in a survey of 751 nurses in Jordan, respondents in oncology settings had significantly higher GNCI© scores (mean = 7.05, SD = 4.24) compared to the respondents working in all other areas (p = < .001) (Alloubani et al., 2023). In contrast, a study using the GGNPS to assess the genomic competency of 2,118 nurses from eight tertiary general hospitals and four cancer hospitals in mainland China found that slightly more cancer hospital nurses compared to the overall sample considered genomics important to their practice (75.6% vs 70.6%, p = 0.010) (Zhao et al., 2022). However, their GGNPS knowledge scores did not differ significantly from those of generalist nurses (8.38 vs 8.21, p > 0.05).

Outcomes of Genomics Competency Interventions for Nurses

While the global empirical research on the topic of genomics nursing has been primarily descriptive, some studies have reported on outcomes following interventions to upskill nurses' genomics competency (Dante et al., 2025; Thomas et al., 2023; McLaughlin et al., 2024). In a recent systematic review of 10 studies of genomics education interventions for nurses and nursing students, no included studies assessed the impact of genomics education on learning outcomes across all three measured domains (cognitive, affective, psychomotor) (McLaughlin et al., 2024). The authors also retrieved no studies that measured the impact of education on nurses' genomics learning in the psychomotor domain. In the 126 studies of a scoping review focused on nurse-oriented genomic outcomes, only 42 were interventional, and most of those (72%) focused on nurses’ genomic knowledge following an educational intervention (Thomas et al., 2023). Similarly, Dante et al. (2025) reported that just 14 of the 63 included studies (22%) on nurses’ genomic literacy were interventional, all of which examined outcomes of educational programs targeting nurses’ genomic literacy or competency. From their comprehensive reviews, Dante et al. (2025) and Thomas et al. (2023) concluded that even when cognitive knowledge and attitudes related to genomics are improved following educational interventions, there is a lack of evidence on how genomics educational interventions impact nurses’ clinical adoption in practice and/or patient outcomes.

In their respective literature syntheses, Dante et al. (2025), Thomas et al. (2023), and McLaughlin et al. (2023) identified that future efforts to address these persistent challenges and build nurses’ genomics competency should involve interventional work that is measured with validated tools to evaluate nursing competency and adoption of genomics in practice. Thomas et al. (2023) noted that the only currently available validated instrument to measure all facets of genomics competency and adoption in practice is the GGNPS. Simultaneously, research findings suggested this may represent a more complex implementation issue. For example, findings from a multi-center controlled study involving 8,150 RNs working across US hospitals indicated that, even with a robust year-long, multi-component genomics educational intervention using validated assessments, the sustained integration of genomics into nursing practice may remain challenging (Calzone et al., 2018). They found no statistically significant differences between nurses who participated in the year-long educational intervention and those in the control group on GGNPS items measuring their use of family history information to guide clinical decision-making, or their frequency of facilitating referrals to genetic services.

Patient Outcomes of Genomic-Informed Nursing

In a multi-arm scoping review of 232 studies on genomics nursing outcomes, only 29% of studies specifically addressed patient and family ‘consumer-oriented’ outcomes (Keels et al., 2024). Of these 67 studies, 58% were non-interventional, and very few included studies reported on patient or family outcomes directly related to nurses’ involvement in their clinical care. A large portion of studies labelled by Keel and colleagues (2024) as reporting on ‘consumer-oriented’ outcomes were those where nurses were involved as authors or investigators in research related to patients’ lived experiences with genomics health, and healthcare implications. Additionally, nearly half (46%) of all the included ‘consumer-related’ outcome studies were those wherein nurse researchers described patients’ and families’ knowledge gaps or unmet information needs regarding their genomics-informed care. This is suggestive that although advancements in genomics-nursing have been observed in the nursing research domain, there are still many untapped opportunities for nurses in the clinical practice domain to improve patients’ experiences with genomics-informed care.

Patient Outcomes of Genomics Nursing (Oncology Specific)

Of the 67 articles in the ‘consumer’ (i.e. patient) outcomes arm of the scoping review (Keels et al., 2024), 20 (30%) were related to genomics in oncology. They included studies where the authors reported that nurse-led educational interventions targeted at persons with increased hereditary risk of cancer were associated with increased patient satisfaction and screening uptake (Salimzadeh et al., 2018; Ingrand et al., 2016; Visser et al., 2015). In another US study, specialist oncology nurse navigators embedded in cancer programs improved turnaround time for somatic testing (*Oncotype Dx*) from 38 days to 20 days and contributed to an increased testing uptake from 26 to 88 % in eligible patients with breast cancer (McAllister &

Schmitt, 2015). It has been reported in some settings that specialist nurses and nurse practitioners increased patients' uptake of genetic testing, as well as hereditary cancer risk management recommendations (Barnhardt et al., 2023; Rauw et al., 2022; Shevach et al., 2023; Yoes et al., 2020). Notably, most of these studies reported on outcomes where patients and families received genomics-informed care from specialist or advanced practice oncology nurses with a specific focus on cancer genomics. There is a lack of reported patient outcomes of nursing care provided by cancer nurses, where cancer genomic care was not their primary role or specialty (i.e. genomics care that had been integrated into existing cancer nursing roles).

Summary of Literature Review, Gaps and Research Problem

In this narrative literature review, it was revealed that there are many genomic test results now considered clinically actionable and available as part of routine care in cancer control settings. It was also revealed that despite these evidence-based applications, uptake of these applications across the cancer care continuum has been slow, meaning that the potentially lifesaving benefits of these applications are unevenly experienced across settings. Genomics is a complex innovation, and the considerable barriers to its adoption in healthcare settings occur at the system, provider, and patient levels. With the current and growing state of genomics applications in cancer control, it is no longer seen as sustainable for genomics-informed care to be siloed exclusively to genetics specialty centres and providers. Particularly in oncology, it is proposed that non-genetics providers, such as nurses, will have to upskill in their genomics competency, as genomics applications become increasingly commonplace in the setting of cancer control.

As nurses comprise the largest group of healthcare providers globally, there has been global momentum to build their genomic competency, both specifically in cancer care settings and across the profession. Despite these efforts, there has been limited progress demonstrated that this focus on nurses’ genomic competency has resulted in sustained uptake of genomics into patient care (Dante et al., 2025; Keels et al., 2024; Skirton et al., 2012; Thomas et al., 2023; Wright et al., 2018). There is a consensus that validated measures will be needed to accurately assess and measure change in this area. To my knowledge, no studies specific to cancer care nurses' genomics competency have been conducted in NL. No Canadian studies specific to an oncology setting have used validated tools to measure nurses’ competency. Concurrently, evidence synthesizing 20+ years of scholarship suggests that pure description and siloed genomics educational interventions are insufficient to drive change. These findings indicate a need for disruptive and novel approaches to address this persistent problem, which go beyond developing competency statements, educational interventions, and robust measurement instruments.

Patients' Unmet CPS Care Needs and Nurses’ Genomic Competency Development: Never the T’wain Shall Meet?

With the longstanding focus on nurses’ genomic competency in the global literature, it seemed prudent to return to a key principle related to competencies as outlined in the RN entry-level competencies document, which states:

> The client is the central focus of RN practice and leads the process of decision-making related to care... Client-centred care reflects that people are at the centre of decisions about their health and are seen as experts, working alongside RNs to achieve optimal health outcomes (College of Registered Nurses of NL, p. 2).

Similarly, in their review, Thomas et al. (2023) recommended that future efforts to strengthen genomics-informed nursing should actively involve individuals with lived experience of genomics-informed care to accelerate the integration of genomics across nursing practice. They noted:

> Future work should ensure that genomic nursing practice is responsive to the needs of patients, communities, and populations nurses serve. Similarly, there are opportunities to engage with community stakeholders to co-create solutions for unmet genomic healthcare needs and bridge disparities in genomic healthcare (p. 13).

Comprehensive literature reviews indicate that several nurse researchers have engaged in scholarship related to patient and family reported outcomes of genomics healthcare (Katapodi et al., 2023; Keels et al., 2024). However, it remains unclear how patients and families perceive and experience nursing involvement in provision of their genomics-informed cancer care, or how nurses, patients, and families have collaborated to test new models of genetics service delivery involving nursing roles.

As described above, many barriers persist for people with or at risk for a CPS when seeking the care they need (Campbell-Salome et al., 2021; Dean & Davidson, 2018; Mittendorf et al., 2021; Strømsvik et al., 2022; Warner & Groarke, 2022). People affected by CPS often report feeling like the experts in their condition, sometimes needing to be the ones who educate healthcare providers (Diez de los Riez de la Serna et al., 2024; Hennig et al., 2018; Hynes et al., 2023; Warner & Grokke, 2022). With these ongoing genomic competency deficits across cancer care nursing, there is an opportunity for nurses and healthcare researchers to collaborate with people with lived experience of CPS in the development of innovative solution-oriented strategies to accelerate genomic integration into nursing care (Thomas et al., 2023). In this literature search, it was found that while people with lived experience of CPS were often asked about their healthcare needs, only one qualitative study conducted with individuals with CPS in

Europe specifically examined the roles patients experienced or envisioned for oncology nurses in this area (Diez de los Riez de la Serna et al., 2024).While interventions addressing nurses' competency in genomics remain a relevant research pursuit, research is needed to understand the significance of nurses' genomics competencies within its situated, relational context (Boer et al., 2024; Tonkin et al., 2025).

Research Problem

To my knowledge, there is a lack of studies where nurses' genomics competency has been considered within its relational context- its impact on patients' experiences. This suggests a missed opportunity to generate an understanding of the needs, capacities, and opportunities to solidify the uptake of genomic-informed nursing in cancer care settings. This gap is what this study aims to address.

Methodology and Methods

Worldview

It was important to situate the research within a worldview that could accommodate the complexity of the phenomena of genomics-informed nursing care. I was guided in my dissertation research by the worldview of dialectical pluralism (DP), proposed initially as a metaparadigm for MM research (Johnson, 2017). Key ontological, epistemological, axiological, and methodological assumptions inherent in this worldview are summarized in Table 1.8.

Table 1.8

Assumptions in Dialectical Pluralism Adapted from Deeb et al. (2023)

Ontology	Epistemology	Axiology	Methodology
Plural reality (Objective, Subjective, Intersubjective)	Dialectic, Dialogic, Hermeneutic	Multiple ethical values Described as a change theory towards social betterment	Mixed-methods research

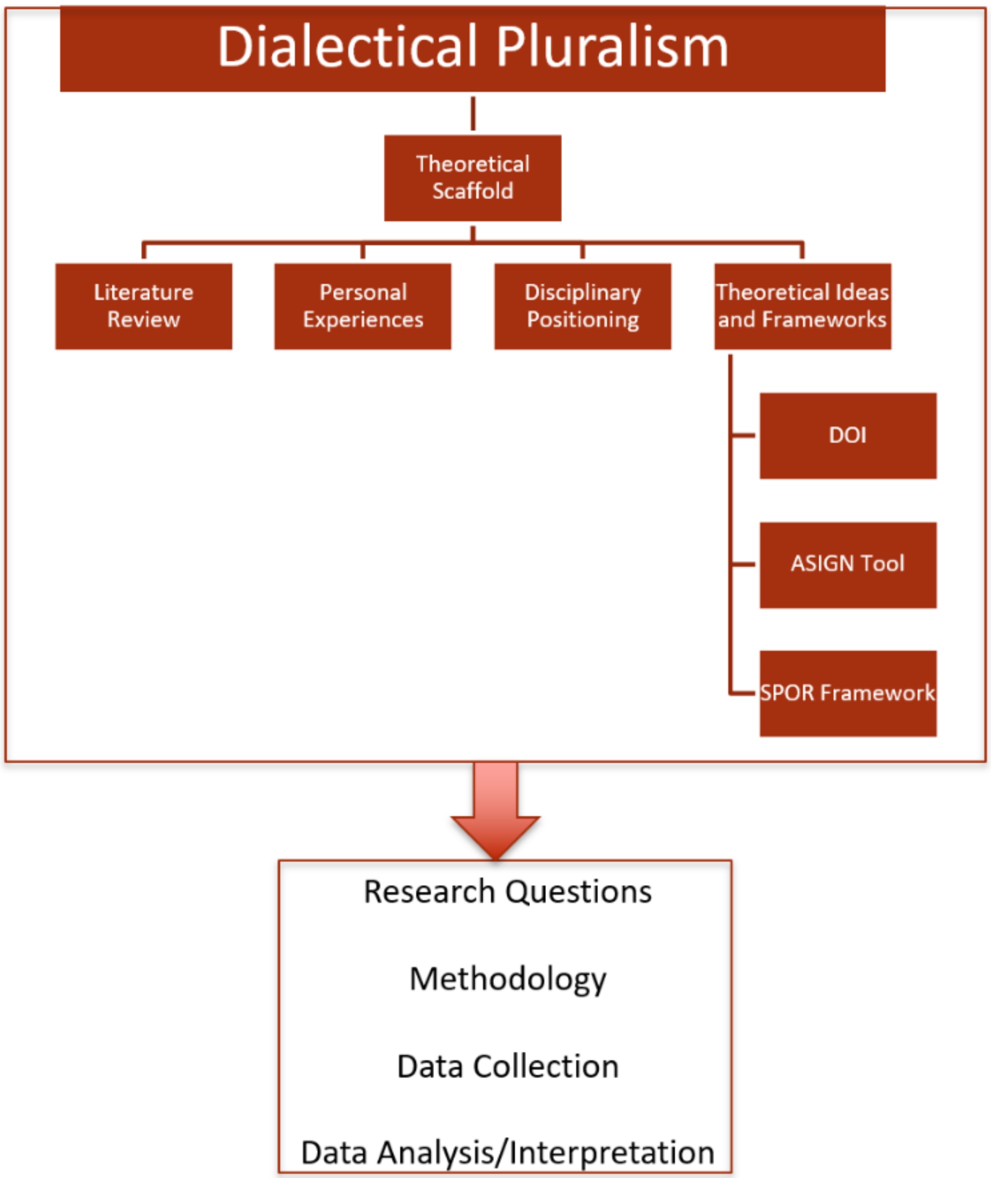
I assumed a worldview of DP because I perceived it to be aligned with my relationship to knowledge as a nurse and now nurse researcher. Nursing is a profession that encounters and acknowledges the existence of multiple forms of reality (subjective, objective, and intersubjective). This involves an appreciation of innovations in evidence-based practice and objective forms of assessment data, while always giving primacy to the patterns and infinite variations in human subjective experiences in the provision of person-centred care. Nursing is inherently intersubjective, a principle I integrated into my research. Knowledge develops relationally between the nurse/researcher ('the knower') and the participant/patient ('what is to be known'), with both influencing each other throughout care and inquiry. I addressed this intersubjectivity by transparently reporting the potential for mutual influence in both qualitative and quantitative study streams. As a nurse in both the clinical and research practice domains, I make my ultimate decisions in practice by weighing the multiple and often dialectical, forms of subjective, objective, and intersubjective realities. In addition to dialects, DP also emphasizes dialogical and hermeneutic logics (including 'epistemological listening') to engage in equal dialogue with the perspectives of many different end-users to produce new syntheses or 'socially-agreed-upon wholes' (Johnson, 2012). Axiologically, DP endorses the goal of societal betterment and justice through mixed research. My commitment to upholding this ethical principle has served as the "raison d'être" for conducting this study and how I hope to disseminate and apply the knowledge generated from this study. This also aligned with my approach as I was guided by the literature related to the considerable ELSI/GE^{3}LS issues associated with genomics and the need to develop solutions that account for the inherent ethical complexities of genomics. DP is commensurate with this dissertation methodology in the following ways: 1. The study involves mixing of qualitative and quantitative paradigms; 2. I have included the dual perspectives of care recipients and providers (nurses) as research subjects while centering my interpretations and implications in patients' subjective experiences. 3. I engaged multidisciplinary members (a medical oncologist, three nurses, and an applied health researcher with expertise in GE^{3}LS) and two individuals with lived experience of CPS as research team members. This approach has allowed for balanced synthesis of perspectives and values through open dialogue and engagement (Johnson, 2017).

Theoretical Scaffold

As a framework to guide this MM study, I enlisted a theoretical scaffold, a design feature of Interpretive Description (ID) (the qualitative approach used here) (Thorne, 2016). The theoretical Scaffold is described as "the explicit articulation of the elements of disciplinary structure that will be brought to bear in shaping and guiding the design elements and applications of each study, and it is these that will ultimately afford it credibility and legitimacy" (Thorne, 2014, p. 9). In other words, it refers to antecedent knowledge, such as the relevant literature, informal and clinical knowledge, disciplinary positioning, and relevant theoretical ideas (Thorne et al., 2016; Chiu et al., 2022). Although developed for qualitative research, I considered its assumptions, including Thorne's notion of disciplinary epistemology, to be philosophically aligned with a DP approach and thus applied it to the entire MM study (Thorne, 2016; Thorne et al., 2016). Thorne also suggested that, rather than defaulting to a single theory to guide a study, it may be more useful to consider several theories relevant to a study. Specific in my theoretical study scaffold was: 1. My disciplinary positioning, 2. my personal experiences, 3. the perspectives of my research team, 4. relevant literature related to the topic, and the DOI, SPOR Framework, and the ASIGN maturity matrix (CIHR, 2014; Rogers, 2003; Tonkin et al., 2020a). I also remained open to exploring other theoretical perspectives throughout the inquiry and analysis.

Figure 1.3

Worldview and Study Scaffold



Disciplinary Positioning

As described above, I view DP as philosophically compatible with nursing disciplinary epistemology. My disciplinary professional ethical mandate emphasizes that nurses' primary responsibility is to the recipients of nursing care and that nurses work in partnership with recipients of nursing care in meeting their perceived health and wellness needs (International Council of Nurses, 2021). Further, as stated, this disciplinary epistemological positioning emphasizes the value of subjective and experiential knowledge, recognizes shared patterns and individual variations, and underscores a moral imperative to ensure that knowledge has practical application (Thorne et al., 2016). This alignment guided my selection of methodology and methods, including the POR approach adopted, and justified placing patient perspectives at the forefront.

Personal Experiences

I occupy situatedness as a RN with previous clinical experience working within a gynecologic oncology surgery setting. There, I interacted with people with lived experience of CPS. They imparted their experiential knowledge of hereditary cancer with me as they were undergoing recommended preventative surgeries. These encounters sparked my interest in cancer genomics. Importantly, they made me question how I, and seemingly other RNs working in gynecologic oncology, knew so little about this crucial topic.

These clinical encounters motivated me to enhance my genomics competency as a nurse and pursue research surrounding genomics-informed cancer nursing care. This was first explored in my Master of Nursing Practicum project. Thus, I also acknowledge that this current study expands on and evolves ideas explored in that project, a policy proposal for a dedicated nurse navigation program. That project led to collaboration with my supervisory committee member, Dr. Etchegary, on her CIHR project grant, *Cancer prevention in cancer predisposition syndromes*: *Testing the feasibility of building a hereditary cancer research registry and nurse navigator follow-up model* (Etchegary et al., 2022). The qualitative interviews in this dissertation were conducted as part of that broader project (see Appendix A; supervisor attestation form). I read further on the topic throughout my doctoral studies and was involved in the Canadian Nursing and Genomics Initiative as a Research Assistant under Dr. Pike. For one of my required PhD institutes, I completed an intensive course in Cancer Genomic Risk Assessment through the City of Hope in Duarte, California. There, I met Dr. Hyde, a local medical oncologist in the provincial CCP, leading the CTG program and related clinical care services. Dr. Hyde joined my doctoral committee and brought a valuable perspective on the advancements of genomics in cancer care settings. From this, there was an evolution in my thinking that the CTG and its reach had implications for all nurses working in cancer care (not just a single nurse navigator). With the support of Dr. Etchegary, amendments were made to the focus of the patient qualitative interviews to also capture patients' experiences and perceptions of cancer genomic nursing more broadly. Collectively, my doctoral research team combines nursing expertise (Dr. Maddigan, Dr. Pike, Dr. Stevens), as well as clinical (Dr. Hyde, myself), research (Dr. Etchegary, Dr. Hyde, Dr. Pike), and lived experience (Vanessa Francis, Mike Warren) with CPS, reflecting a shared commitment to enhancing hereditary cancer care in NL. My previous clinical experiences have invariably influenced my approach to research questions, methodology, data collection, and analysis. With my proximity to the subject matter, I engaged in reflexivity practices during qualitative and integration data analysis. At times I had to take a step back and ask myself if, in interpretations, was I looking for 'what I wanted to see' in the data. I reflected on this at length and adjusted my analysis at times to ensure a variety of perspectives were captured. For example, I paid attention to qualitative participants' accounts of their desire for peer support, involvement of primary care providers, or an advanced practice NP involved in their care.

Examination of Relevant Literature

My methodological approach was guided by immersion in the literature (as detailed above in this chapter) which noted the persistent unmet care needs and barriers to uptake for these two interest holder groups. It was further informed by the state of available research tools to assess the phenomenon. As indicated, while I held vested personal interest in the topic, what evolved in my thinking as a result of my prolonged immersion in this literature was that treatment-focused and mainstreamed genetic testing in cancer care settings were changing the horizon of how individuals could access genomic treatment. Reviewing the relevant literature punctuated a need to focus on validated measures of nurses' competencies, which informed my selection of the instruments used. The GNCI©, along with select, modified items from the GGNPS, were the instruments used in this study as reported in the literature (Calzone et al., 2012; Plavskin et al., 2019; Plavskin et al., 2023).

Theoretical Ideas

Diffusion of Innovation. Rogers' (2003) diffusion of innovation (DOI) is a theoretical framework used to understand key concepts in adopting innovations (such as genomics) across social systems or organizations. As per the DOI, factors that contribute to the adoption of an innovation are described, such as knowledge of the innovation, variables in the social system, as well as the personal attributes of the adopter and their attitudes towards the innovation (i.e., concerning its relative advantage, compatibility, complexity, trialability, and observability). These variables were the theoretical domains used to develop the GGNPS (as a subset of which were included in the quantitative survey package for this study). This theoretical framework was used to analyze quantitative results.

SPOR Framework. The Canadian Institutes of Health Research (CIHR) launched the Strategy for Patient-Oriented Research (SPOR) in August 2011. POR is recognized as a continuum of research that engages patients as research partners, focuses on patient-identified priorities, and aims to improve patient and health system outcomes (CIHR, 2019). CIHR released a SPOR Framework and best practice guidelines for POR (CIHR, 2014). The SPOR framework lists four guiding principles for patient-oriented research (see Table 1.9). These guiding principles informed the approach to patient and public engagement with the involvement of two patient partners alongside all phases of the study.

Table 1.9

Guiding Principles for SPOR, adapted from CIHR (2014) SPOR Framework

Guiding Principle	Explanation
Inclusiveness	POR involves diversity of lived experience, expertise, and perspectives, and their contributions are reflected in the research
Support	Patient participants are supported and offered flexibility to ensure they can contribute to the research process and related decisions. This includes culturally and psychologically safe environments and appropriate compensation for their role on the research team.
Mutual Respect	Researchers, people with lived experience, and other providers acknowledge and value each other’s expertise and experiential knowledge.
Co-Build	Patients, researchers, and practitioners are engaged early in the study process to identify problems and gaps, set priorities for research, and collaborate on developing and implementing solutions.

ASIGN. The Global Genomics Nursing Alliance (G2NA) was formed in 2017 to overcome some of these persistent lags in the adoption of genomics across the global nursing profession and accelerate the integration of genomics across international nursing practice

(Calzone et al., 2018). G2NA developed two practical resources informed by implementation science, including a maturity matrix to benchmark needs, capabilities, and capacities for integrating genomics in nursing and inform strategic integration directions (Tonkin et al., 2020a, 2020b). In the G2NA maturity matrix,'ASIGN' (Assessment of Strategic Integration of Genomics across Nursing),'patient and public involvement,', and 'community and family focused care' are highlighted as key enablers and critical success factors of successful genomics implementation across nursing. In efforts to effectively advance genomics-informed nursing practice, ASIGN indicates that not only the active engagement of nurses is needed, but also patients in genomics integration efforts across nursing, and this assumption also informed my study approach.

Research Questions

This study aims to assess readiness and perceptions of genomics-informed cancer nursing care in NL. The overarching research question to be answered by the MM integration is: *What insights can be learned from the integration of nurses' and patients' perceptions of genomics-informed cancer nursing care to advance its clinical implementation*? The research sub-questions are:

1. What are the genomic-informed healthcare experiences described by individuals in NL living with CPS? (*qualitative stream*)
2. What are the experiences, perceptions, and receptivity of individuals living with CPS related to cancer genomic nursing care? (*qualitative stream*)
3. What are the a) knowledge levels, b) confidence, c) attitudes, and d) practices related to genomics of oncology nurses in NL? (*quantitative stream*)

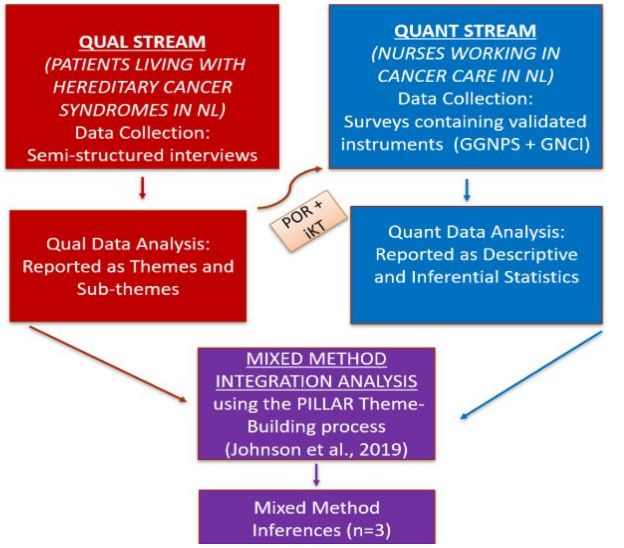
4. Are the social systems/organizational cultures conducive to NL oncology nurses' adoption of genomics? (*quantitative stream*)
5. What demographic and contextual variables are associated with NL oncology nurses' genomics knowledge scores? (*quantitative stream*)

Design

Informed by assumptions of the DP metaparadigm, the methodology employed for this study was mixed methods research. Specifically, informed by DP, I used a convergent MM design (Creswell & Plano-Clarke, 2018). In the qualitative stream of the study, I utilized an ID approach (Thorne, 2016), and in the quantitative stream, I employed a descriptive, cross-sectional survey design. At the integration phase, these data were combined. A study diagram can be found below (Figure 1.3), which emphasizes the parallel, but separate nature of data collection and analysis in each stream, which were integrated in a later phase.

Figure 1.4

Procedural Mixed Method Study Diagram



Aligned with assumptions in DP where the goal is to generate a new product which is "an agreeable package of goals and values that serves multiple important groups and perspectives" (Johnson, 2012, p. 753), the goal of merging these data sets was to combine the perspectives of both nurses and persons with relevant lived experience as recipients of genomic-informed care. In so doing, the overall goal was to generate a more fulsome understanding of the phenomena of genomics-informed nursing care that further elucidates current patterns as well as possibilities for its further implementation.

Provincial Study Setting

NL has a population of approximately 549,900 (Government of NL, 2025). NL's publicly funded healthcare is managed by NL Health Services (NLHS), which is divided into five regional zones throughout the province. The CCP serves the entire population of NL, as does the Provincial Medical Genetics program (PMGP). There are four CCP outpatient clinics located throughout NL: one in the Eastern Zone, two in the Central Zone, and one in the Western Zone (Dowden et al., 2024). The CCP provides outpatient systemic therapy, clinical trials, radiation therapy, screening, and patient outreach services. In-patient systemic therapy is also provided in the EUZ in two inpatient units (adult and pediatric). Although some oncologists in NL have been ordering genomic testing for certain cancers for several years, these tests have historically been processed out of province (Etchegary et al., 2021).

Ethical Considerations

Health Research Ethics Board approval was obtained for the qualitative (Reference#2022.125) (Appendix B), and quantitative study streams (Reference #:2023.122) (Appendix C). Ethical considerations pertaining to each study stream are discussed below.

Patient and Public Engagement

In addition to the guiding principles and strategies in the SPOR framework (CIHR, 2014). Dr Etchegary, a national leader in POR, provided me with expert mentorship in POR best practices throughout my doctoral research. As the findings of the SPOR refresh report were released, I integrated those updated recommendations into my ongoing approach to POR (SPOR Governance and Strategy Refresh Steering Committee, 2025). Two patient partners with lived experience of CPS (Mike Warren and Vanessa Francis) were engaged in this study since its inception. In Figure 1.5 examples of their contributions to the present study are outlined briefly.

Figure 1.5.

Patient Partner Involvement in Study

Qualitative Stream

ID was the methodological approach used to guide the qualitative stream of this study (Thorne, 2016). ID was appropriate as it is intended for qualitative examination of applied health phenomena within a naturalistic context, and it was aligned with my stated disciplinary epistemology, and the aims of research sub-questions 1. and 2. Thorne (2016) also endorsed the use of ID as a qualitative component of MM research.

Sampling and Recruitment (Qualitative Stream)

For the qualitative stream, participants were eligible for the study if they were living in NL, 18 years of age or older, with or without a history of cancer, with any molecularly confirmed P/LPV associated with a CPS. We used purposive and maximum variation sampling to ensure representation of a broad range of lived experiences with CPS (e.g., age, sex, gender, level of education, type of CPS gene involved, length of time since receiving genetic test result, geographic region of the province). Prior qualitative work conducted in NL, examining experiences of individuals with CPS, suggested that approximately 25 interviews were required to capture meaningful patterns in participants' experiences (Etchegary et al., 2015, 2018; Watkins et al., 2011). However, these studies have typically been limited to HBOC and Lynch Syndrome populations. Our inclusion criteria were comparatively broader, encompassing individuals with various CPS-associated genes, each with potentially different considerations. As the perspectives of many of these people were not yet studied in depth in our region, we aimed to recruit upward of 40 participants.

The qualitative study stream was promoted over social media and communication channels of Memorial University, local radio stations, and a local newspaper. Hard copies of study recruitment posters (see Appendix D) were placed in healthcare and community settings (e.g., grocery stores, primary care providers' offices) wherever permissible. With assistance from healthcare providers on Dr. Etchegary's larger research project team, the names of interested patients were provided to a research assistant or myself via secure email.

Data Collection (Qualitative Stream)

Once a participant made initial contact with the research team (either with a Research Assistant or me) to indicate their interest, I conversed with potential participants to explain the purpose of the study. I offered the first chance to answer any questions. During the initial discussion, participants were informed that the interviews would be audio taped for later transcription. A study consent form (see Appendix E) was forwarded to them via e-mail or discussed with them in person during the scheduled interview, as per their preference. Participants were advised of the study's potential risks and benefits. Participants were informed that they could withdraw from the interview at any point and that taking part in the study would not affect their healthcare. Participants were also provided with a list of community resources in the event that discussing some of these sensitive topics during the interview elicited any psychological distress. If following this, they expressed interest in participating in the study, an interview was scheduled at a time as per the participants' stated preference. While no participants dropped out, three eligible individuals who expressed initial interest did not participate after two attempts at follow-up contact.

At the start of the interview, I reviewed the consent forms with participants and answered any questions. Following this, if I they indicated that they still wanted to take part in the study, I asked them to demonstrate consent with a verbal yes to the question "Is this all agreeable to you?" and confirmed they had no further questions. I conducted all patient interviews, which took place either via telephone, the Webex online meeting platform, or in person at a private conference room at the Faculty of Nursing (as determined by the preference of the participant). Participants were offered a small financial incentive ($25.00 e-gift card) to remove any barriers to participating and to show appreciation for their time and contributions. The initial interview guide was drafted based on the study's research questions, literature, the experience of clinical and patient partner team members, and finalized over several iterations (see Appendix F). All interviews were audio-recorded and transcribed verbatim. Audio recordings were deleted once the transcripts were verified for accuracy. The decision to stop collecting data was based on our ascertainment of sufficient information power in the data (i.e. evidence of variation in the sample regarding types of CPS and reported healthcare needs, variation in demographics such as urban versus rural, and early stages of data analysis revealing possible patterns and sufficient richness in the data that could provide illumination to clinical practice and our research questions) (Malterud et al., 2016).

Data Analysis (Qualitative Stream)

Thorne (2016) described general steps in data analysis in ID that I used to guide my process.

1. **Finding patterns among the pieces**. I engaged in prolonged immersion with the qualitative data through two main approaches: a) listening to the recorded interviews before transcription and again after transcription, before deleting the audio recordings, and b) repeatedly reading and engaging with the full interview transcripts before initiating coding and revisiting them throughout the data analysis process, and later during MM analysis. Dr. Etchegary and I independently read and coded the transcripts, assigning codes to segments of quotes i.e. ‘meaning units’ in the individual transcripts. After coding ten transcripts, we met to discuss our findings and early impressions of

patterns observed in the data. We used our institution-hosted secure cloud CAIR drive (hosted by the **C**entre for **A**nalytics, **I**nformatics and **R**esearch, Craig Dobbin Genetics Research Centre, Faculty of Medicine) to view each other’s coded transcripts. Based on consensus, I compiled a list of twelve emerging patterns. I then reviewed all transcripts, color-coded meaning units according to one of the twelve patterns, extracted and hand-sorted 408 meaning units into Excel spreadsheets organized according to these patterns (e.g.,‘knowledge is power’;‘set adrift after test results’).

2. **Making sense of pattern.** This step drew on elements of the constant comparative approach endorsed in Grounded Theory where data from one informant was compared across the data of all others informants in the spreadsheets while engaging in interpretive note-taking guided by questions outlined by Thorne (2016), such as “What variations exist? What does this mean for clinical practice?” This process involved periodically revisiting the transcripts. I also shared examples of meaning units organized according to candidate patterns with patient partners (Warren and Francis) to gather their impressions on what resonated with them. Additionally, I discussed emerging patterns with my primary supervisor, Dr. Maddigan.
3. **Transforming patterns into findings.** With guidance from my doctoral committee, patient partners, and interpretive notes, I considered how the patterns could be collapsed into a cohesive structure that addressed the research question, captured variation in patients’ experiences, and presented a logical narrative reflecting experiences along common points in the genomics healthcare trajectory. From this, I transformed the candidate patterns into the first draft of reportable study findings (themes and sub-themes). I sought feedback on the initial draft from my doctoral committee and patient

partners, revisiting themes and structure as necessary. This iterative process led to slight modifications, resulting in the final presentation of themes (n=2) and sub-themes (n=6), reported in Chapter 2 of this dissertation.

Rigour (Qualitative Stream). Thorne (2016) developed several evaluative criteria for ID studies (e.g., epistemological integrity, representative credibility, analytic logic, interpretive authority) to guide actions to maintain rigour. Epistemological integrity was maintained by ensuring alignment between the stated worldview, disciplinary epistemological stance, and the claims presented, such that they were not reported as 'absolute truths' but rather as context-based and in recognition of the infinite variation of human experiences. I ensured representative credibility through maximum variation sampling to capture a variety of experiences as described above. I ensured that different participants' perspectives were captured in the presented quotes. As I made decisions about data reduction, I kept an audit trail of these decisions to ensure that the criterion of analytic logic was maintained. To assure the criterion of interpretive authority, I engaged in reflexivity and frequent discussions with Dr. Etchegary, Mike Warren, and Vanessa Francis. My supervisor also audited the results of the analysis, and my committee provided feedback. Another strategy employed to ensure rigour in study reporting was in verifying that all relevant criteria of the COREQ (COnsolidated criteria for REporting Qualitative research) checklist had been addressed and reported upon submission to the journal (Tong et al., 2007).

Quantitative Stream

The quantitative stream addressed the overarching research question and research sub-questions 3, 4, and 5. A cross-sectional descriptive survey design was used.

Sampling and Recruitment (Quantitative Stream)

Nurses (RNs, Licensed Practical Nurses, Nurse Practitioners) were eligible for this study if they worked in one of the four outpatient sites of the CCP throughout the province or in one of the two inpatient oncology units within the Eastern Urban Zone (EUZ). The estimated total number of nurses eligible to participate in the survey was 111 (a confirmed 56 in the CCP and an estimated 55 in inpatient settings). This number was calculated based on a figure provided by Dr. Hyde who consulted administrators from her professional networks in the CCP regarding this estimate. Estimations for the in-patient units were provided by members in my professional networks from the local CANO/ACIO chapter. At the 95% confidence level with a margin of error of ±5%, it was determined that a sample size of 86 was needed.

I employed multiple recruitment strategies. Dr. Hyde connected me directly with leaders and the clinical educator within the CCP to assist with recruitment efforts. With institutional approval (see Appendix G), I visited the four CCP outpatient sites and the EUZ adult inpatient unit to provide information about the study and present participants with the link and the QR code to the study survey. I also co-facilitated an integrated knowledge translation (iKT) session at the EUZ CCP outpatient site during one of their regularly held education sessions, alongside Vanessa Francis, a patient partner with lived experience of CPS. This session is described further in Chapter 4, but this iKT approach allowed a bridge between both study streams prior to formal integration analysis. Refreshments were provided during all sessions, and participants were given a QR code to access the survey at their convenience. The CCP clinical nurse educator sent email reminders with the survey URL immediately after the iKT session and again two weeks before the survey closed (see Appendix H). Additionally, posters displaying the survey QR code were placed at all worksites where eligible nurses were based (see Appendix I). Recruitment efforts were further guided by Dillman's (2000) tailored survey methodology, including a "lucky draw" incentive for a $100 Visa e-gift card. Contact information for the prize draw was kept separate from survey responses, and participants could choose whether to enter.

Measurement Instruments (Quantitative Stream)

The survey package comprised 60 items across three sections (see Supplemental Tables in Chapter 2 for a complete description of the research questions and corresponding measurement instruments and items). Section one consisted of seven occupational and demographic questions. Section two included 22 modified, selected items from the original 60-item GGNPS (Calzone et al., 2012), which has various question formats (dichotomous, ordinal, Likert) and measures nurses' attitudes, knowledge, confidence, and practices with genomics, as well as the influence of the social systems on their adoption of genomics in practice. The GGNPS has been shown to have good test-retest reliability (Calzone et al., 2016) as well as content, face, and construct validity (Plavskin et al., 2019; 2023). Of the 22 included GGNPS items, minor modifications were made to some questions to ensure relevance to the Canadian oncology setting.

The entire 31-item GNCI© (as described in the literature review) (Ward et al. 2014) was used to assess knowledge in place of the GGNPS knowledge questions, as the GNCI© assesses knowledge of foundational genomic principles highly relevant to oncology that were not included in the GGNPS. Permission was obtained for use of the GNCI© from the developer of the tool, Dr. Linda Ward (see Appendix J). The GNCI© is a valid, reliable instrument shown to have good internal consistency with reported Cronbach's alpha values ranging from 0.73 to 0.85 (Ward et al., 2014, 2016, 2018). Further extensive psychometric validation of the GNCI© has been completed using item response theory, exploratory factor analysis, and concurrent criterion validity (Ward et al., 2016; 2018).

Data Collection (Quantitative Stream)

The survey package was collected on the Qualtrics (2023) survey platform, where consent was collected electronically (see Appendix K). Individuals interested in participating could follow the web link or QR code to the study, provided in the recruitment materials. The link/QR code brought participants directly to the consent form. Participants were required to read the consent form and could not advance to the survey until they clicked yes, indicating that they had reviewed the consent form. Password-protected digital files containing the data from the demographic questions and survey instruments were stored on a password-protected MUN Computer held by me. Data contained in the lucky draw was deleted following the raffle.

Data Analysis (Quantitative Stream)

SPSS v. 29 (IBM Corp, 2022) was used to complete the analysis. Frequencies and percentages were used to describe categorical variables and means ± standard deviations were used to describe continuous variables. Hierarchical, Multiple linear regression (MLR) was used to identify the predictors for the GNCI© knowledge score (dependent variable). Following consultation with a colleague with advanced statistical training, all possible knowledge predictors (i.e. responses to all seven individual demographic and the 22 GGNPS items) were assessed at the univariable linear regression level ($P \leq 0.2$). Significant variables ($p \leq 0.2$) were entered in the first MLR model, and using a hierarchical approach, variables were removed that were not significant at $p \leq 0.05$. The results were assessed for potential confounding by dropping one variable at a time to check for changes in Beta coefficients (≥ 20%) with no evidence of same. The final model was also tested for effect modification, and no interaction effect was found. As an exploratory test, in exploration of a possible relationship between nurses' practice unit (inpatient vs outpatient), and whether they completed a complete family history over the past three months, family history collection was transformed from an ordinal to a dichotomous variable: those 'who collected history in past three months' (i.e. combining the responses of those who reported that they did so either 'always', 'frequently', and 'occasionally') versus those who 'rarely or never collected family history.' A Chi-Square test examined the relationship between the practice unit (inpatient vs outpatient) and family history practice. Missing data were managed by pairwise deletion for descriptive statistics and listwise deletion for regression analysis. Two-tailed $p \leq 0.05$ was set as the significance level.

Integration Phase

Data from the qualitative and quantitative study streams were integrated to answer the overarching MM research question. The Pillar Integration Process (PIP) guided the integration analysis phase, which draws on joint display tables used in traditional MM approaches (Johnson et al., 2019). To begin the analysis, I created a blank joint display template based on the PIP to facilitate data integration. This template included the following columns (left to right):'qual THEMES and SUB-themes','qual MEANING units' the 'PILLAR' and 'quant DATA POINTS','quant DOMAINS'. Using this template, analysis was guided by the four PIP steps for mixed-methods integration outlined by Johnson et al. (2019): (1) listing, (2) matching, (3) checking, and (4) pillar building (Johnson et al., 2019). I primarily completed the integration analysis with ongoing feedback through each of these steps from my supervisor, who has experience with MM research and previously used the PIP technique in a concurrent, MM study. Initial findings were presented as three MM inferences, with minor refinements made from feedback received from my doctoral committee. As Johnson et al. (2019) described the product of the PIP integration process as ‘pillar building themes’, the integration analysis process was comparable to an additional level of qualitative analysis; we were guided by the theoretical scaffold and drew on guidance from ID to ensure rigour. Further details of the integration process are presented in its corresponding results chapter (Chapter 4).

Significance of the Proposed Research

As NL prepares to launch a new translational genomic cancer service, it is timely to assess oncology nurses’ genomics competencies and work alongside patients to identify barriers to genomics-informed cancer care and identify solutions to address these barriers. The findings from this study will provide contextually relevant insights to guide workforce preparation for cancer nursing practice in this new era. This study employed a validated instrument (GNCI©) and selected components of another (GGNPS) to measure oncology nurses’ genomic competencies. This study contributes to the global literature on nursing competencies and addresses the limited number of oncology-specific studies in this area. This study integrates the perspectives of two interrelated interest groups through a MM design. By combining quantitative and qualitative insights, the research will generate a more comprehensive understanding of opportunities to implement genomics in clinical cancer nursing practice. With the engaged iKT approach employed, findings can promote and inform future interventional studies where nurses and patients (within a shared local context) can co-develop interventions through research and quality improvement initiatives that address unmet needs in CPS care.

Chapter Conclusion

In this first dissertation chapter, I introduced the research problem and reviewed a broad body of literature that substantiated the research questions and provided justification for the study and the approach employed. The research question and detailed methodology were also presented in this chapter. In the subsequent manuscript-based Chapters (2, 3, 4), findings of this study will be presented with some further methodological details, followed by the dissertation conclusion chapter (Chapter 5).

CHAPTER 2: “Anybody who can clarify or humanize the experience would be such a help”: An Interpretive Description of Perceptions of Genomic Health and Nursing Care in Individuals with Cancer Predisposition Syndromes

Rebecca Puddester, RN, PhD(c), [a]

Vanessa Francis, Patient Partner,[b]

Mike Warren, Patient Partner,[b]

April Pike, RN, PhD[a]

Joy Maddigan, PhD, RN, PhD[a]

Angela Hyde, MD, PhD, FRCPC,[c]

Kathleen Stevens, RN, PhD[a]

Holly Etchegary PhD (Senior Author), [b, d]

[a] Faculty of Nursing, Memorial University, St. John’s, Newfoundland and Labrador, Canada

[b] NL SUPPORT Unit, Memorial University, St. John’s, Newfoundland and Labrador, Canada

[c] Disciplines of Laboratory Medicine and Oncology, Faculty of Medicine, Memorial University, St. John’s, Newfoundland and Labrador, Canada

[d] Community Health and Humanities Clinical Epidemiology, Faculty of Medicine, Memorial University, St. John’s Newfoundland and Labrador, Canada

Introductory Statement

Chapter 2 contains a manuscript that presents qualitative interview results (patient participants) from the MM research study. The target audience for this manuscript is oncology nurses globally, other members of the oncology care team, as well as health administrators. This manuscript was published in the *European Journal of Oncology Nursing*. https://doi.org/10.1016/j.ejon.2024.102766.© 2024 The Authors. Published by Elsevier Ltd. This is an open access article under the CC BY license (http://creativecommons.org/licenses/by/4.0/)

CRediT authorship contribution statement

Rebecca Puddester: Writing – original draft, Methodology, Investigation, Formal analysis, Conceptualization. Vanessa Francis: Writing – review & editing, Investigation, Conceptualization. Mike Warren: Writing – review & editing, Investigation, Conceptualization. April Pike: Writing – review & editing, Supervision, Methodology, Conceptualization. Joy Maddigan: Writing – review & editing, Supervision. Angela Hyde: Writing – review & editing, Supervision. Kathleen Stevens: Writing – review & editing, Supervision. Holly Etchegary: Writing – review & editing, Supervision, Project administration, Methodology, Investigation, Funding acquisition, Formal analysis, Conceptualization.

Abstract

Purpose: There is increased use of genomic testing in oncology care. Yet, individuals with hereditary cancer predisposition syndromes (CPS) experience challenges when navigating the lifelong CPS healthcare considerations. The purpose of this study is to describe the healthcare experiences of individuals living with CPS and their perceptions of genomic-informed nursing care. **Methods:** Interpretive description (ID) was the qualitative approach used in this patient-oriented research study, conducted in partnership with two individuals with lived experience of CPS. Participants were recruited to participate in an interview using a variety of public and provider/patient network sampling sources. Inductive data analysis was guided by general steps for ID analysis as described by Sally Thorne. **Results:** Thirty-seven individuals who self-reported a CPS participated in interviews between April and August 2023. Two primary themes were identified: 1), When genomic knowledge is power, and 2), Perceived acceptability of nursing roles in CPS care. Participants described genomic knowledge as a form of personal and family empowerment, but many reported that CPS-related knowledge is inaccessible in the healthcare system. This often resulted in healthcare experiences that were insufficient and fragmented. Although participants reported minimal nursing involvement in their CPS care, there was a consensus that enhanced nursing contributions could be beneficial. They particularly endorsed the potential value of a dedicated oncology genomic nursing role. **Conclusions:** Findings highlight patient-endorsed opportunities for nurses at the generalized and specialized levels to contribute to improved cancer genomic care. Findings can be used to inform novel models of clinical care for individuals with CPS.

Keywords: Cancer predisposition syndromes; Genomics; Hereditary cancer; Nursing roles; Oncology nursing; Patient engagement; Patient-oriented research; Qualitative research

1. Introduction

Cancer is a leading cause of death in Canada, placing a significant economic burden on the healthcare system and affected individuals and families (Brenner et al., 2024). Up to 10% of cancers are linked to hereditary cancer predisposition syndromes (CPS) (Garutti et al., 2023). Many Canadians with cancer can now access both germline (inherited) and somatic (acquired) genomic testing to guide targeted treatment and identify underlying CPS (Liu et al., 2022; Yip et al., 2019). The term 'genomics' is the terminology of the most updated US Essential Genomic Nursing Competencies and Outcome Indicators. While 'genetics' may be used in the manuscript at times to maintain fidelity of participant quotes, we use the term genomics to refer to genes and anything that regulates genes (Calzone et al., 2024). Cancer genomics refers the application of both genetic and genomic data to categorize cancer risk, inform cancer prevention, treatment, or stratify the risk of cancer recurrence (Friend et al., 2021). Long-standing evidence shows that individuals with pathogenic and likely pathogenic (P/LP) variants in *BRCA1/2* (linked to Hereditary Breast and Ovarian Syndrome [HBOC]), and in mismatch repair genes associated with Lynch Syndrome (LS), benefit from CPS risk management strategies which reduce cancer rates and mortality (National Comprehensive Cancer Network 2024a; 2024b). Individuals with confirmed CPS can also inform their asymptomatic relatives, who may then opt for cascade genomic testing and explore management options if confirmed to have the same CPS (Khoury and Dotson, 2021).

While the National Comprehensive Cancer Center (NCCN) has recommended germline multi-gene panel testing in all individuals with invasive ovarian cancer for nearly fifteen years (Daly et al., 2010), with the increased access and affordability of next-generation genomic sequencing, over one hundred cancer-associated genes are now routinely included in oncology trials (Kammula et al., 2022). Cancer risk management guidelines now cover P/LP gene variants beyond those associated with HBOC and LS (e.g., *RAD51C, CHEK2, ATM*) (NCCN 2024a; 2024b); these guidelines vary based on family history and gene involved, but in general, involve earlier and increased screening protocols, and options for preventative surgeries and/or medication, as well as discussions surrounding reproductive health options. Experts predict that multi-gene panel germline testing will be offered as standard of care for all cancer patients with solid tumours (Esplin et al., 2022). This and other advancements in oncology genomics are likely to increase the number of individuals identified with a CPS, who would be missed by traditional family history-based guidelines alone (Samadder et al., 2021).

However, across many Canadian jurisdictions, demands for publicly funded CPS testing and related care are already outpacing the capacity of the medical genetics specialist workforce (i.e. genetic counsellors [GCs] and geneticists) (Dragojlovic et al., 2023; Hynes et al., 2020). Further, while the benefits of long-term follow-up to cancer outcomes have been established in individuals with LS (Barrow et al., 2013; Møller et al., 2017) and HBOC (Lobo et al., 2018; Pichert et al., 2010), in several Canadian jurisdictions (including our study setting), there is a lack of systemic follow-up care for individuals with confirmed CPS (Hynes et al., 2023). This current management approach has contributed to the under-identification of at-risk individuals and sub-optimal uptake of cancer risk management strategies that can be lifesaving (Roebothan et al., 2023; Tindale et al., 2022). For example, Roebothan et al. (2023) found that almost 40% of individuals with HBOC were not undergoing the recommended, annual breast surveillance over an 18-month period.

Canadians with HBOC and LS often face significant barriers from healthcare providers and the system while attempting to manage the lifelong health implications of CPS (Etchegary et al., 2015; Hynes et al., 2023; Snow et al., 2024; Watkins et al., 2011). As genomics increasingly influences cancer care, these challenges may worsen (Tindale et al., 2022). To effectively leverage genomic advancements for better cancer outcomes, innovative delivery models are essential, with a focus on meeting the needs of service users (Dragojlovic et al., 2023; Tindale et al., 2022). One promising approach is genomic "mainstreaming," where oncology teams, including non-genetic specialists, provide genomic-informed care in collaboration with genetics specialists (Bokkers et al., 2022).

Despite the critical role of oncology nursing in cancer care, evidence indicates that oncology nurses often feel unprepared to deliver genomics-informed care (Hébert et al., 2022; Rahman et al., 2022). This is concerning given the rapid pace of advancements in cancer genomics. However, some evidence suggests that nurses in advanced practice and/or specialist roles have enhanced patient access to genomic testing and improved outcomes (Barnhardt et al., 2023; McAllister & Schmitt, 2015; Rauw et al., 2022; Scott et al., 2020). A recent Canadian study emphasized the need to amplify the voices of patients, such as those with lived experience of CPS, to advocate for investments in multidisciplinary workforce training for genomics-informed oncology care (Chiu et al., 2024). Literature underscores the ethical imperative of incorporating lived experience data into health workforce preparation, healthcare delivery models, and resource allocation (Rand et al., 2019). Still, patient perspectives have been largely overlooked in research on nursing workforce contributions to genomics-informed care, while the integration of genomics across nursing practice remains a persistent global challenge (Thomas et al., 2023). Thus, the objectives of this study are to, a) describe the genomics-informed healthcare experiences of individuals living with CPS, and b), to explore their perceptions of cancer genomic nursing care.

2. Methods

Interpretive Description (ID) (Thorne, 2016) was the qualitative methodological approach used in this study. Following the ID approach, we established a 'theoretical scaffolding' (Thorne, 2016) of relevant elements that informed the study from inception through analysis. Components of our study theoretical scaffold included our background literature review, nursing disciplinary epistemological orientation, Canadian-specific frameworks for conducting patient-oriented research, and our positioning as a research team. The lead author's (RP) nursing disciplinary epistemological orientation places value on subjective and experiential knowledge, acknowledges both shared patterns of experiences and individual variances, and emphasizes a moral imperative to ensure that the knowledge generated through ID has practice applications (Thorne et al., 2016). This perspective aligned with our study aims, as participants' experiences with the healthcare system provide insights into their lived reality of having a CPS and its implications for their healthcare needs and preferences. Additionally, we were guided by the Canadian Institutes of Health Research (2014) framework for Patient-Oriented Research, regarding the involvement of two patient partners (VF, MW) with lived and family experience with CPS. VF and MW actively participated as members of the research team throughout all phases of the study. Since this study focused on patient perceptions of nursing roles in genomic care, a key aspect of our theoretical scaffold was our positioning as a research team, which includes the reflexive acknowledgment that RP is a Registered Nurse (RN) with interdisciplinary training in cancer genomic risk assessment. Collectively, our team combines clinical (RP, AH), research (RP, HE, AP), and lived experience (VF, MW) with CPS, reflecting a shared commitment to enhancing hereditary cancer care in our jurisdiction. This study was approved by the provincial Health Research Ethics Board (Reference#2022.125).

2.1. Setting, recruitment, sample

Participants were recruited from the Canadian province of Newfoundland and Labrador (NL) where healthcare services are publicly funded and delivered through five regional health zones. NL offers a unique context for studying hereditary conditions due to the "settler effect" linked to historic Irish and English settlement patterns (Gilbert et al., 2023; Rahman et al., 2004). We choose to use the term 'settler effect' in lieu of what is often referred to as 'founder effect'. As settlers on unceded territory, this is in recognition of the diverse histories of the Beothuk, Mi'kmaq, Innu and Inuit indigenous peoples of Newfoundland and Labrador who have been here on the land since time immemorial. Eligibility for the study included individuals over 18 years old, residing in NL, with or without a personal history of cancer, who self-reported any molecularly confirmed CPS such as HBOC, LS, or P/LP variants in any CPS associated gene (e.g., *CHEK2*, *CDH1*, *PALB2*, *ATM*).

Multiple participant recruitment strategies were employed between April 2023 and August 2023. Study posters were disseminated on social media platforms (e.g., Facebook and 'X') and the study was also advertised on two local radio stations. RP, HE, MW, and VF co-wrote articles for the local newspaper to promote awareness of the study. Members of the research team within patients' circle of care also informed eligible individuals of the study, with a focus on recruiting individuals with P/LP gene variants that were not yet represented in the sample. Finally, study informants and patient partners assisted in snowball sampling by notifying other contacts who were eligible to participate in the study. All eligible participants who were interested in taking part in the study initiated first contact with members of the research team via e-mail or telephone. While no participants dropped out, three eligible individuals who expressed initial interest did not participate after two attempts at follow-up contact.

2.2 Data collection

HE and RP drafted a semi-structured qualitative interview guide, which was refined based on feedback from patient partners (MW, VF) (see supplemental file 1). RP, a female Registered Nurse (RN) and doctoral candidate, conducted all single session interviews. Although she introduced herself as an RN, she had no prior clinical relationships with participants. Informed consent was obtained prior to interviews, and participants were offered a $25 e-gift card as a token of appreciation. Interviews were conducted between April and August 2023, either in person, via online video conferencing (Webex), or by telephone, depending on participant preference. In-person interviews took place in a private conference room at the researcher's faculty. Sessions lasted between 27 and 89 minutes, averaging 50 minutes. Each interview was assigned a unique ID number, audio recorded, transcribed verbatim, verified for accuracy by RP or HE, and deleted after transcription. Data collection was stopped once sufficient information power (Malterud et al., 2016) was achieved, determined by evidence of variation in the sample regarding types of CPS and healthcare needs, as well as contextual demographic factors (e.g., urban vs. rural), and early data analysis revealing patterns that could inform clinical practice.

2.3 Data analysis

Aligned with an ID approach, inductive analysis was informed by general steps outlined by Thorne (2016). 1**.** *Finding pattern among the pieces;* De-identified transcripts were securely stored on an institutional cloud drive and independently read and coded by RP and HE. Segments of quotes within the transcripts (referred to as ‘meaning units’) were assigned initial codes. After both RP and HE coded ten transcripts, they met to discuss their findings, leading to the creation of a list of twelve patterns in coding. This list served as a template of ‘candidate patterns for themes' to organize the data. RP then reviewed the transcripts again to extract and hand-sort a total of 408 meaning units into Microsoft Excel spreadsheets based on the twelve 'candidate patterns' (e.g.,'knowledge is power,' 'set adrift after test results') 2. *Making sense of pattern;* RP analyzed the spreadsheets while engaging in reflexive journaling, guided by questions outlined by Thorne (2016), such as "What variations and associations exist?" and "What does this mean for clinical practice?" This process involved periodically revisiting the transcripts. Additionally, RP shared the meaning units organized according to candidate patterns with patient partners MW and VF to gather their impressions. 3. *Transforming patterns into findings;* Informed by this collaborative, interpretive process, RP transformed the 'candidate patterns' into the first draft of reportable study findings (themes and sub-themes). Following Thorne's (2016) recommendations, member checking was not employed; instead, RP sought feedback on the initial draft of themes from her doctoral supervisory committee (AH, AP, HE, JM, KS) and patient partners (MW, VF), revisiting the data as necessary. This iterative process led to slight modifications, resulting in the final presentation of themes and sub-themes.

2.4 Rigour

Thorne's (2016) four evaluative criteria for quality in ID guided efforts to ensure rigour. *Epistemological integrity* was addressed by ensuring alignment between the research objectives, the theoretical scaffold, data collection and analysis (Thorne, 2016); *interpretive authority* was ensured through RP's use of reflexive journaling during data collection and analysis, ongoing reflexive meetings with HE and patient partners, and prolonged engagement with the data; *representative credibility* was maintained through efforts to ensure the interpretive claims were consistent with the study sampling and data sources; and *analytic logic* was demonstrated through use and revisiting of an audit trail of decisions surrounding data reduction.

3. Findings

In total, (n=37) individuals completed an interview (see Table 1).

Table 2.1

Table 1 Participant characteristics (n=37)

Characteristic	**n (%) / Details**
Age	Mean = 52 (SD = 14.13), Range = 22–75
Sex	Female: 31 Male: 6
Marital Status	Married: 28 Common-law: 3 Divorced/Separated: 2 Single: 4
Children	Yes: 31 No: 6
Previous History of Cancer	Yes: 22 No: 15
CPS-Associated Pathogenic Variant (PV)	LS: 12 *CDH1*: 5 HBOC: 4 *CHEK2*: 3 *ATM*: 3 FAP: 3 Other (*RAD51C, BRIP1*, BHD): 4 Variant of Uncertain Significance (VUS): 3
Highest Education Level	High school: 6 Technical college: 7 Baccalaureate degree: 14 Graduate degree: 6 Other/Not specified: 4
Residence	Rural: 14 Urban: 23

There was significant heterogeneity in the sample with respect to age, level of education, and type of CPS-associated gene. The majority (84%) self-described as ‘female’. Individuals living in rural and urban regions were represented, as well as those with and without a personal history of cancer. The mean participant age was 52 ±14.13 SD, ranging from 22 to 75. During interviewing, for (n=3) participants who initially self-reported a CPS, it was revealed that their genomic results were actually classified as variants of uncertain significance (VUS). While VUS lack official risk management recommendations, it cannot be ruled out that the VUS contributes to increased cancer risk (Friend et al., 2021). As these individuals also represent a subset of individuals with ongoing genomic information needs relevant to the inquiry, it was decided to retain their perspectives in analysis. Findings are reported as two themes with corresponding sub-themes: 1) When genomic knowledge is power, and 2), Perceived acceptability of nursing roles in CPS care.

3.1 Theme 1 When genomic knowledge is power

Many participants described knowing their CPS status as a source of personal empowerment in their cancer prevention and risk management decision-making, as confirmation of CPS status often determined their eligibility for enhanced cancer risk management protocols. For example, it determined eligibility for enhanced endoscopic screening for this participant with LS:

> Getting the genetic testing and being diagnosed with Lynch opens up doors, not enough doors but certainly better than previous, automatically you get this cystoscopy and the colonoscopy every year. I mean every time I go for a colonoscopy they remove polyps (P4)

Participants commonly perceived access to genomic knowledge as both logical and cost-effective within a publicly funded healthcare system. One participant with a *CDH1* PV, linked to hereditary diffuse gastric and breast cancer, emphasized this by stating that access to genomic health information should be considered a "right."

> I just feel if you know you have a gene mutation then you can make the decisions like I did to prevent these illnesses from occurring and as far as I can tell putting a greater strain on the system, it just makes sense to me. I think people are entitled to their genetic information and I think with that information they are entitled to do what they need to do with it (P9)

Participants living with a variety of CPS often perceived that clinical application of genomic knowledge had been lifesaving:

> I feel that I've cheated cancer, or I'm trying to cheat cancer and I don't have to lose my hair, I don't have to die from breast cancer… so now I'm living with scars and emptiness in places where I had organs before, but I won't have that cancer (P31, HBOC)

Similarly, out of the five participants with *CDH1* PVs who were interviewed, four reported that pre or early-stage cancer cells were found on their pathologies when they underwent the recommended preventative gastrectomy. As one participant stated:

> I probably wouldn't be alive right now is the way I look at it. I was on the road to having a horrible short life the way it was going to end and I wouldn't be here for my family and so, I felt very much like I had proof now [from the surgical pathology report] that I did the right thing because up until then I was questioning it (P24, *CDH1*)

While participants viewed genomic knowledge as empowering, it became clear that knowledge is not simplistically equated with "power." There are nuanced, contextual factors that determine when genomic information is empowering—or not—for individuals with CPS. These complexities were explored through the sub-themes of theme 1.

3.1.1: Sub-theme 1.1 Knowledge as power when shared with others

For several participants, especially those with a personal history of cancer, the perception of 'power' assigned to genomic test results was largely related to its value to their biological kin who could then apply this knowledge to make proactive decisions about their health. As evidenced by this participant with LS who reported "my youngest daughter, the one that carries the gene, she goes on a regular basis now and gets scoped also" (P26). Some participants believed their own genomic knowledge was lifesaving to their family members. For instance, a person with a PV in the *APC* gene (associated with Familial Adenomatous Polyposis [FAP]) noted:

> When they sat me down with [son] and the doctor showed me the 3D image of his bowel…there were only a few sections were clear. If we never got the genetic testing done. He was 16 and had that many polyps…what would have happened if I was delayed that much and he was 16 and that full, that fast (P19)

Some participants who underwent testing as part of their cancer treatment, along with their asymptomatic family members, reported that the newfound knowledge of an actionable CPS served as a source of family empowerment during a challenging illness journey. One woman shared her experience of discovering she carried a PV in the *ATM* gene when her mother underwent testing for ovarian cancer treatment.

> It's such a sense of relief and knowledge is power to me. It's just so good to have the information. When you can look for warning signs in a different way because I know for ovarian cancer, most people they don't catch it until stage 4 like my mom (P16)

Further, some participants of reproductive age or their parents indicated that they valued the ability to incorporate genomic knowledge into decisions about the health of future generations with the aid of reproductive technologies. For instance, one participant with a *CHEK2* pathogenic variant (PV) shared how her GC informed her that they could "select [eggs] without the *CHEK2* gene which I thought was quite amazing" (P12).

3.1.2 Sub-theme 1.2 Genomic knowledge is often inaccessible

While most participants viewed knowledge as power and none reported regret about their decision to undergo testing, many identified a significant barrier to applying genomic knowledge: the overall lack of awareness and knowledge of CPS within the healthcare system.

> It's ridiculous how big of a battle it is. It's terrible. And I think what's frustrating is that you've got medical professionals that should know about this and when you mention it to them, 'oh, I've never heard of that' (P10, LS)

Some participants perceived lack of healthcare provider knowledge of CPS as a barrier or delay in their accessing genetic referrals to provincial medical genetics (PMG) services.

> [Family doctor] never said to me at any point in time, well [name] you've had this cancer and you've had this cancer, my dear you need to go and get genetic testing. He didn't say it and neither did any of the other doctors who did the surgeries (P4, LS)

However, once connected with GCs at PMG for pre-test counseling and/or test result disclosure, participants often described these interactions as highly informative and positive. For example, one woman with a *CHEK2* pathogenic variant (PV) reported that "the genetic counsellor that I went to did a really great job laying out what options were there and giving me the information" (P30). Nonetheless, for many participants, leaving PMG with a written copy of their results and management options did not alleviate the challenges of obtaining subsequent, necessary information within the healthcare system.

> The downside I felt was once I gave the blood, once [daughter] talked to Dr. [name] and I talked to the genetics counsellor but once that was done then you're set adrift (P2, *RAD51C)*

Many participants reported challenges in seeking up-to-date knowledge within the healthcare system regarding lifelong management considerations across various CPS. This included needs for information surrounding topics such as management of menopausal symptoms resulting from preventative surgery in HBOC, nutrition following preventative gastrointestinal surgeries, and up-to-date information surrounding CPS-specific screening protocols. For instance, one man with LS expressed uncertainty about where to turn for reliable, up-to-date risk management information.

> Have I reached an age or have other factors contributed to a need for new screening? Like, '[Name] is 42 now, now is the time for him to start getting a urinary scope or whatever the case may be'. Right now, I'm operating under the same set of instructions that I was given when I was 25 (P7)

Uncertainty about where to seek up-to-date genomic knowledge was also reported by individuals in our sample with VUS results, if their VUS result may be reclassified as P/LP or benign. Similar concerns related to access of evolving knowledge were expressed by a participant originally told she had a LP variant in a LS-associated gene that was later reclassified as a VUS.

> I was delighted to know that I didn't have Lynch syndrome, but it was still a bit confusing, you know. I don't know, Like now I'm concerned that I do have Lynch syndrome? (P26, *MSH2* [VUS])

The potential consequences of the lack of accessible genomic knowledge in the healthcare system were highlighted by a participant with FAP, who reported that despite regularly interacting with the health system, it was not until 20 years after receiving positive test results as a minor that their new primary care provider (PCP) informed them that regular colonoscopies are recommended for individuals with FAP starting at age 10-15 (NCCN, 2024b).

> Until [new PCP] was like 'you should be getting scopes', I was like 'oh, should I?' Like I had no idea really (P23)

This participant's narrative was particularly alarming, as they noted that they later developed stage 0 cancer, which would not have been detected without the enhanced screening protocols recommended for FAP. For many participants, the lack of information from local healthcare providers meant that their primary or sole sources of CPS information were the internet and online support groups to "do some reading and hope [they] pick the right site that is legitimate and has more up to date information" (P13, Birt Hogg Dubé syndrome).

3.1.4 Sub-theme 1.3 With new knowledge comes new supportive care needs

While knowledge of CPS was largely perceived as beneficial, it also created new lifelong psychosocial care needs associated with the awareness of living with an increased cancer risk. However, there was little proactive psychosocial care provided to address these needs.

> I think everybody focuses on the increased cancer risk and the testing but think the mental health aspect of it, it wasn't brought up at all (P11, LS)

Several participants found positive peer support through online CPS-specific groups, while others eventually sought counseling services to manage stress related to living with a CPS. For instance, one man with LS disclosed:

> I finally went and I talked to a counsellor. I had to. Because it's stressful knowing that you have got this syndrome and that there is another possibility of cancer showing up again (P6)

For many participants, the lack of genomically-informed healthcare services available to them in their CPS management appeared to exacerbate their psychosocial stressors and contributed to a perception as though they were 'going it alone'. This sense of isolation often weighed heavily on participants without a personal history of cancer, who reported complex psychosocial care needs related to their cancer risk. However, since they had never had cancer, there were no avenues within the cancer care system to address these needs.

> I feel like there are a lot of us out there who have these proactive surgeries that have this upside down world after that nobody really understands and it's so hard to navigate. You don't fit in a box a lot of times (P33*,* FAP)

3.1.2 Sub-theme 1.4 Knowledge as power when care is personalized

Some participants reported positive experiences if their primary or other specialty HCP were willing to co-develop a personalized CPS risk management plan, for example in relation to timing of screening. However, a recurring perception among some participants was that standardized CPS guidelines were sanctioned in healthcare encounters, void of consideration of the contextual factors in participants' lives (e.g., their broader health history, psychological, social and geographical circumstances). This often resulted in disempowering healthcare encounters, especially among younger individuals interviewed who had no personal history of cancer.

> I've had a [specialist] appointment before where the first question they asked me were when am I getting my breasts removed and to me, that was not a good question to ask someone who had just found out they had the *BRCA* gene, so I would just hope that

> whoever you are dealing with, is well informed on all of the options that everyone has and the different paths that people may choose (P29, HBOC)

Participants also had varied expectations of the level of guidance they required from HCPs. Some reported a high degree of self-advocacy and agency with seeking out their own information:

> I'm very literate with respect to the healthcare system and I'm very comfortable navigating the system…I know what I should be receiving, what kind of screening I need to have and I stay on top of all my appointments (P24, LS)

In contrast, others indicated that they needed more ongoing guidance within the healthcare system regarding their CPS management, particularly concerning the frequency and timing of screenings. One participant with LS noted:

> I'm not a medical professional. I don't think I should be worrying, googling all the things that the doctors write down about where these referrals go in…You're kind of relying on these letters...You can't just call someone (P11, LS)

Against the backdrop of these reported unmet knowledge and supportive care needs and person-centered preferences, participants were asked their perceptions of prospective nursing roles to address their CPS-related health needs and expectations. Their perceptions are captured through theme 2 and its sub-themes.

3.2 Theme 2 Perceived acceptability of nursing roles in CPS care

When asked, most participants reported that novel nursing role(s) in CPS healthcare services could be of benefit to them and or their family members. Some individuals considered that nurses would be quite suited for these roles because of their therapeutic communication skills, as one participant with HBOC noted, "Sometimes you can tell the difference between a nurse and [other providers] in a good way" (P35). Yet, for the most part, participants' positive reactions to a proposed CPS nursing role stemmed from the prospect of having access to any healthcare provider who could address the current unmet needs in CPS healthcare.

> Yes, I think anybody who can kind of point to the way or clarify or humanize the whole experience would be such a help. Yeah, I have a family doctor who is supportive and listens, but I definitely think that there is like, gaps in the care. (P9*, CDH1)*

Similarly, a man with LS expressed that a cancer genomic role could be assumed by "*somebody who's a nurse or somebody who's a doctor, LPN, RN, LMNOP, it doesn't make a difference*" (P6).

No significant concerns were raised about nurses providing cancer genomic care; however, several participants expressed an expectation that these nurses would possess extensive knowledge specific to cancer genomics

> … they need to have a knowledge of genetics and the cancers because there are several of them, genetic cancers. They need to know the next steps that need to be taken (P4, LS)

In addition, some believed that in order for nurses' role in CPS care not to be "another administrative loophole" (P27, *PALB2* [VUS]), they would need to have access to someone who could order genomic and/or diagnostic tests as applicable. For similar reasons, some suggested that perhaps a nurse practitioner might be most suitable:

> So you don't need to start over every time to get referrals…hopefully we do get to nurse prescribing but maybe you do need a nurse practitioner (P31, HBOC)

3.2.1 Sub-theme 2.1 Limited exposure to genomic nursing

While there was a general consensus that nurses taking on an enhanced role in CPS care could be beneficial, participants reported limited involvement of nurses in this care to date.

> Nurses, no, not so much. No, it's always been doctors. (P10, LS)

A few participants did report that cancer nurse navigators were involved in their genomic care, such as a participant with HBOC without a personal history of cancer who was referred to a cancer nurse navigator for help with accessing recommended MRI screening, or another participant who was helped by a nurse navigator to access genomic testing:

> I even called the nurse navigator that I had during cancer when we didn't hear back from genetics about me getting tested, I called her and she got things on the ball. So, I found her really helpful – P25, (*MSH2* [VUS])

Although minimal actual nursing involvement in their CPS care was reported, some participants with personal or family experience accessing cancer care services recognized the potential value of the current cancer nurse navigator role if applied to CPS care.

> I know from being a cancer patient the value of nurse navigators, how helpful they are… we need more nurse navigators that work with the genetics department, I think that could be very beneficial (P17, LS)

3.2.2 Sub-theme 2.2 Perceptions of potential nursing contributions in CPS care

Since only a minority of participants had experienced nursing involvement in their genomics-informed care, most could only share their opinions on nurses' *potential* contributions to their CPS care. Nevertheless, participants frequently identified the potential for nurses to meaningfully enhance their care by ensuring the scheduling and completion of evidence-informed screening protocols, as well as providing education and psychosocial support to themselves and/or their family members following genetic counseling.

> To have someone there to have a conversation with, to provide the type of supports that they need, to make sure that they get into the system, are screening, and then their screening is done correctly. That would have been really helpful… and even to help with my husband. There was a lot for both of us to think about right? (P24, LS)

Some participants felt that nurses could follow-up with individuals with CPS for "annual wellness checks" (P35, HBOC), not just for screening and risk management purposes, but also to assess how individuals with CPS are adjusting emotionally to their CPS. A few also saw the potential role of the nurse as being a liaison or a connection between team members, as well as a patient advocate between the different care providers. As one participant noted:

> If you have someone like a nurse navigator? That person has to advocate for you too, cause it can't just be you doing this alone (P22, *CDH1*)

3.2.3 Sub-theme 2.3 A dedicated genomic oncology nursing role

Most participants endorsed the value of a dedicated nursing role in cancer genomic follow-up care. It was reported by participants that if there was an oncology nurse where cancer genomics “became their role specifically” (P30, *CHEK2*), this would support the nurse(s)’ development and maintenance of the high level of genomics knowledge needed to effectively address the complex support and information needs in CPS care. Participants also supported the idea that having a dedicated nurse in this role could enhance care continuity and reduce the commonly reported fragmentation of CPS care. For instance, some indicated that a dedicated nurse could collaborate effectively with PCPs:

> A nurse could really be valuable to have someone that’s kind of like, centralized, looking at all the records and I think that could definitely be done in conjunction with, with the family doctor who kind of also has that, that information and that duty of care to the whole person, cause I think especially with Lynch you go to one person for the digestive system, you go to another person for the reproductive system, like there can be this segmentation of the body instead of looking at the patient as a whole (P28, LS)

Another similarly reported that a dedicated nurse could help them co-develop a person-centered CPS management plan and then “reinforce it to my family doctor or to even some of my specialists” (P6, LS). Others suggested that a dedicated nurse’s role in CPS follow-up could function independently of PCPs, should that be the individual's preference.

4. Discussion

Consistent with extensive research in the field (McAllister et al., 2011; Yuen et al., 2020), participants in our study living with various CPS generally perceived genomic knowledge as a source of empowerment for themselves and their families. Some viewed access to this information as a matter of life and death. However, similar to findings from qualitative studies conducted regionally (Hynes et al., 2023; Watkins et al., 2011) and internationally (Campbell-Salome et al., 2021; Warner and Groarke, 2022; Warner et al., 2022; Wellman et al., 2023), our study highlights that individuals experience significant breakdowns in access to cancer genomic knowledge and support, from the initial process of accessing genomic testing to navigating lifelong CPS management considerations.

While many of the issues presented here are not new (Hall and Olopade, 2005), the use of mainstream genomic testing in cancer care settings is increasing. This was evidenced in our study, as some participants were found to have a CPS when they or their family members underwent genomic testing as part of their cancer treatment. The limited number of participants who had experienced nursing care that included cancer genomics suggests a missed opportunity to receive timely cancer genomic care. Oncology nursing competencies are highly relevant to the healthcare needs identified by participants, such as establishing therapeutic relationships, providing patient teaching and coaching, advocacy, and facilitating decision-making and continuity of care (Canadian Association of Nurses in Oncology/Association Canadienne des infirmières en oncologie, 2006). While few participants in our study had experienced nursing care involving cancer genomics, nearly all were open to the idea of increased nursing contributions to their CPS-related care. Consistent with recent qualitative findings involving individuals with HBOC and LS in Europe (Diez de los Rios de la Serna et al., 2024), many participants spoke enthusiastically about a potential role for oncology nurses in addressing unmet needs in their CPS healthcare.

Our study findings have important implications for guiding strategic preparation as part of a collective, disciplinary professional response to patients' evolving cancer care needs. Firstly, considering that oncology nurses often feel unprepared to provide genomics-informed care (Hébert et al., 2022; Hines-Dowell et al., 2024; Wright et al., 2020), we suggest incorporating cancer genomics content into continuing education for all oncology nurses. Additionally, cancer patient nurse navigators may especially benefit from genomic education, as our findings indicate that some are already facing patients' genomic-related inquiries. While all oncology nurses have a potential role in ensuring the continuity of cancer genomic care, ordering cancer genomic testing and facilitating CPS follow-up care are advanced practice oncology nursing roles in some jurisdictions (Dowling et al., 2024), and positive outcomes have been reported from advanced practice nursing models of cancer genomic care and follow-up (Barnhardt et al., 2023; Rauw et al., 2022; Thompson et al., 2022). Relating these results to our findings, participants were enthusiastic about the prospect of a dedicated, cancer genomic nursing role in CPS-follow-up care. Some participants indicated that a nurse with an advanced scope of practice, such as nurse practitioner, may be most effective in this prospective role. Dedicated and/or advanced practice nurses may also play a crucial clinical leadership role in supporting the genomics proficiency of all oncology nurses also working in their practice areas. The benefits of having a dedicated clinical nurse "champion" as a resource person to support genomics integration in specific practice settings have been previously documented (Calzone et al., 2018). Therefore, incorporating practice-based leadership and education as part of the role of a dedicated genomic oncology nurse may be an important consideration when implementing these new roles.

Our findings add to evidence supporting the potential value of a dedicated, nurse-led follow-up model of care, from the vantage of persons who would be using this service. This may be relevant to leadership in health care organizations and systems when exploring novel models of cancer genomic care delivery. However, piloting and evaluation of the outcomes of dedicated nurse-led models of cancer genomic care are required to confirm our findings. Furthermore, as the need for more person-centred care experiences was highlighted in our findings, we suggest that outcomes should include not only clinical outcomes (e.g., screening adherence, cancer outcomes), but also patient reported outcome and experience measures (PROS/PREMS). For example, PROMS measuring the effect of the service on patient ‘empowerment’ has been identified as an important patient-reported outcome of clinical genetic services (McAllister et al., 2011) and our findings confirm that this is likely a relevant patient reported outcome of cancer genomic nursing care.

Beyond the nursing profession, many interdisciplinary team members in both oncology and clinical genetics settings—such as GCs, geneticists, and oncologists—play a collective role in delivering high-quality mainstream genomic care (Bokkers et al., 2022). Many participants in our study reported positive experiences with GC services; therefore, our findings should not be interpreted as advocating for the replacement of GCs' expertise by non-genetics oncology team members. Instead, they highlight the opportunity for collaboration with genetics specialists to ensure patients receive more timely, integrated approaches to cancer genomic testing and follow-up care. Additionally, our participants expressed varied preferences for the co-involvement of their PCP in a nurse-led model of cancer genomic follow-up care. These findings suggest that a dedicated cancer genomic nurse could work collaboratively with patients and their PCP to co-develop an integrated, person-centered plan for CPS management.

4.1 Limitations and strengths

Our findings should be interpreted with caution. First, because RP identified herself as a registered nurse during data collection, there is a potential for social desirability bias concerning the nursing subject matter. Another limitation is the reliance on self-reported data, including CPS status, meaning that the molecular P/LP variant status of participants was not verified in advance. This inadvertently resulted in the inclusion of three individuals with VUS test results. Additionally, there was potential for selection bias, as individuals who self-identified to participate in the study were likely already highly engaged in their health management. While the theme "When genomic knowledge is power" captures the perspectives of study participants, we acknowledge that there are likely perspectives not represented in the sample, particularly from those who may not view genomic knowledge as empowering. For instance, individuals with serious concerns about privacy and insurance claims related to genomic data may have different views. These concerns have been addressed in prior work by members of the research team, which examined public perceptions of genomics-informed healthcare (Etchegary et al., 2022). Finally, there is limited ethnic diversity within the sample from a population with known settler effect, so it cannot be concluded that these findings are transferrable in other settings.

A key strength to our study is the involvement of two patient partners from study inception through to analysis. Their contributions at all stages ensured not only the relevance of our findings but also that our research process aligned with nursing's professional mandate to collaborate with recipients of care in promoting measures to improve individual and population health (International Council of Nurses, 2021). Additionally, our study included representation from individuals both with and without a personal history of cancer, as well as a broad range of experiences related to different CPS. Traditionally, qualitative research in our jurisdiction has focused solely on HBOC and LS populations; thus, our study provides perspectives on CPS that have not yet been explored in depth.

5. Conclusion

Study findings reveal largely positive attitudes toward genomic knowledge among participants affected by CPS. However, the findings also highlight the ongoing challenges they face in navigating lifelong screening and coordinating their care, including gaps and barriers in accessing ongoing knowledge and supportive care. Participants identified areas where nursing roles could help address breakdowns in the care they receive. This lived experience data can inform strategic efforts to clarify, develop, evaluate and support generalist and dedicated oncology nursing roles in the genomic era that are responsive to patients' needs and expectations.

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CHAPTER 3: Oncology Nurses' Readiness to Implement Genomics-Informed Care: A Descriptive, Cross-Sectional Study in One Canadian Province

Rebecca Puddester, RN, PhD(c), [a]

Angela Hyde, MD, PhD, FRCPC,[c]

Holly Etchegary PhD, [b, d]

April Pike, RN, PhD, [a]

Kathleen Stevens, RN, PhD,[a]

Joy Maddigan, RN, PhD, [a]

[a] Faculty of Nursing, Memorial University, St. John's, Newfoundland and Labrador, Canada

[b] NL SUPPORT Unit, Memorial University, St. John's, Newfoundland and Labrador, Canada

[c] Disciplines of Laboratory Medicine and Oncology, Faculty of Medicine, Memorial University, St. John's, Newfoundland and Labrador, Canada

[d] Community Health and Humanities Clinical Epidemiology, Faculty of Medicine, Memorial University, St. John's Newfoundland and Labrador, Canada

Introductory Statement

Chapter 3 contains a manuscript that presents results from the cross-sectional descriptive survey (oncology nurse participants) from the mixed methods research study. The target audience for this manuscript is oncology nurses in Canada but also in the global oncology nursing community, as well as health administrators. This manuscript was accepted for publication in the January 2026 issue of the *Canadian Journal of Oncology Nursing*.

CRediT authorship contribution statement

Rebecca Puddester: Writing – original draft, Methodology, Investigation, Formal analysis, Conceptualization, Funding Acquisition. Angela Hyde: Conceptualization, Writing – review & editing, Funding Acquisition. Supervision. Kathleen Stevens: Writing – review & editing, Supervision. Holly Etchegary: Writing – review & editing, Supervision, Methodology. April Pike: Writing – review & editing, Supervision, Methodology. Joy Maddigan: Writing – review & editing, Supervision, Project administration, Methodology.

Abstract

Introduction: Cancer care providers need to be equipped to support cancer care recipients in evolving care contexts and genomics is an increasingly common component of cancer care. There is limited understanding of Canadian oncology nurses' readiness to contribute to genomics-informed cancer care. **Purpose:** To describe factors influencing oncology nurses' implementation of genomics in practice in Newfoundland and Labrador (NL) (i.e., knowledge, attitudes, confidence, current practices, and social system influences); and b) to identify predictors of their genomic knowledge. **Methods**: A cross-sectional online survey was administered between Sept 2023 to Feb 2024 to nurses working in cancer care in NL. Variables associated with nurses' implementation of genomics-informed practice were measured using the Genomic Nursing Concept Inventory (GNCI©) and select, modified questions from the Genetics Genomics Nursing Practice Survey (GGNPS). Descriptive and inferential statistics were used to report findings. **Results:** The survey was completed by (n=50) NL oncology nurses. While 46% of participants indicated that patients had initiated conversations about genomics with them in the past three months, their knowledge levels and reported confidence with genomics practices were low overall. Despite this, participants indicated largely positive attitudes toward the benefits of adopting genomics in practice and a willingness to learn more. **Conclusion:** Findings highlight opportunities to support oncology nurses with practice-based education and resources to ensure readiness to meet patients' evolving needs and expectations surrounding genomics-informed cancer nursing care**.**

Key words: genomics; precision oncology; oncology nurses; hereditary cancer

Introduction

Cancer genomics[2] encompasses the application of any germline (inherited) or somatic (acquired or tumor) molecular genomic data to categorize cancer risk, inform cancer prevention or treatment, or stratify likelihood of cancer recurrence (Friend et al., 2021). There is a growing number of evidence-based genomics applications in cancer control strategies shown to improve cancer outcomes (National Comprehensive Cancer Network [NCCN], 2024a, 2024b; Rendon et al., 2023; Yu et al., 2025). Oncology nurses assume vital roles in patient care, including system navigation, teaching and coaching, and health assessment across the cancer care continuum (Canadian Association of Nurses in Oncology/Association canadienne des infirmières en oncologie [CANO/ACIO], n.d.); they are therefore important targets in the adoption of evidence-based clinical innovations, such as genomics. This study examined factors influencing oncology nurses' readiness to adopt genomics-informed care (knowledge, attitudes, confidence, practices, and social system influences) within a provincial setting planning further genomics integration into its Cancer Care Program (CCP).

Background

Treatment-focused *BRCA* gene testing to inform indication for PARP-inhibitor (PARPi) therapy became the recommended standard of care fifteen years ago for individuals with epithelial ovarian cancer (EOC) (Daly et al., 2010), as PARPi is associated with improved progression-free survival in individuals with *BRCA*-mutated EOC (O'Malley et al., 2023; Zhou et al., 2024). In recent years, there is steady growth in the number of Health Canada-approved

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[2] 'Genomics' is the terminology used in the current American Nurses Association (2023) Essential *Genomic Nursing Competencies and Outcome Indicators* (3rd edition) when referring to molecular data involving genes and/or anything that regulates genes (e.g., including single gene, multi-gene and whole exome/genome panel testing). We use this term here although sometimes the word 'genetics' may be used in the manuscript when referring to original source material.

anti-cancer therapies that are indicated by genomic test results (Liu et al., 2022; Yip et al., 2019). At the same time across Canadian healthcare settings, hereditary cancer testing eligibility criteria, as well as the number of genes included in multi-gene panel tests are expanding (Hereditary Cancer Testing Eligibility Working Group, 2024). As genomic testing becomes more widespread in cancer care, there is a greater likelihood of identifying individuals with pathogenic or likely pathogenic (P/LP) germline variants associated with cancer predisposition syndromes (CPS). If a CPS is identified, biological relatives of the affected person then qualify for cascade predictive gene testing. Affected individuals can access evidence-based cancer risk management protocols (e.g. enhanced screening, preventative surgeries) shown to reduce cancer-related morbidity and mortality in these high-risk populations (NCCN, 2024a, 2024b).

The current scale of evidence-based genomic applications in cancer control has increased demands for hereditary cancer testing and services such that demands now exceed the capacity of the genetics workforce (Dragojlovic et al., 2023). As a result, alternative cancer genomic service delivery models have emerged, including 'mainstreaming' models where genomic testing and related care is delivered by non-genetics oncology care providers, with varying levels of involvement of genetics specialists (Bokkers et al., 2022; Byrne et al., 2024; Mackley et al., 2025). It has been reported in some settings that mainstream oncogenomic service models improved access to timely genomic testing (Bokkers et al., 2022; Hamilton et al., 2021). However, it has also been reported that testing uptake remains uneven in Canada; this is attributed in part to variability in oncology providers' knowledge of genomics and in their initiation of conversations with patients about genomics (Snow et al., 2024; Tindale et al., 2022). Further, while mainstream service models may improve testing access, they are likely to also increase the current unmet demands for ongoing follow-up care after genomic test results are disclosed (Tindale et al., 2022). Pre-dating the current and mounting scale of genomic applications in oncology, individuals living with a CPS in Newfoundland and Labrador (NL) reported provider and system-level barriers when seeking the follow-up care needed to manage their lifelong cancer risk (Etchegary et al., 2015; Hynes et al., 2023; Watkins et al., 2011). Low uptake of the recommended high-risk breast screening protocols for individuals with P/LP variants in *BRCA* 1/2 has been reported in NL (Roebothan et al., 2023). As clinical applications in cancer genomics are increasing amid ongoing challenges accessing the related clinical care, it is time to rethink how to effectively integrate genomics across the cancer care continuum.

Despite the evolving paradigm shift toward genomics-informed ‘precision oncology’ and although nurses assume key roles in cancer care delivery, there is limited research assessing Canadian oncology nurses' readiness to provide genomics-informed care (Chiu et al., 2024; Hébert et al., 2022). In a recent pan Canadian study, oncology nurses reported increasingly fielding patients' questions about genomics and recognized a need for additional professional development in this area to adequately address patients’ related informational needs (Chiu et al., 2024). Similarly, nurses who provided cancer care in Quebec reported low confidence with discussing genomic implications of cancer with patients, and their responses to knowledge-based questions revealed misconceptions about hereditary cancer risk (Hébert et al., 2022). Although a recent Canada-wide survey of nurses across all practice areas established a national, profession-specific benchmark of readiness for genomics-informed practice (Limoges et al., 2024), no studies to date have specifically focused on the readiness of oncology nurses in NL to deliver genomics-informed cancer care.

Assessment of oncology nurses' genomic competency is timely in the NL setting. The NL population has some of the highest rates of cancer in the country (Canadian Cancer Statistics Advisory Committee et al., 2023) and is considered a unique resource for studying genetic disorders, including hereditary cancers, due to its geography and historical settlement patterns (Rahman et al., 2004). Treatment-focused genomic testing for certain cancers have been ordered by oncologists in NL for several years, however, traditionally those samples were sent out of province (Etchegary et al., 2021). With the acquisition of genomic sequencing equipment and recommendations of a recent strategic plan for provincial health systems transformation (Health Accord NL, 2022), plans were announced for a clinical translational genomics service which would provide in-house genomic testing and integrated clinical care within the provincial CCP. In anticipation of this service delivery change, for her doctoral dissertation, the lead author (RP) conducted a mixed-methods study aimed at a comprehensive assessment of readiness for genomics-informed cancer nursing care in NL. Qualitative patient reported findings have been previously published (Puddester et al., 2025) and integration findings are forthcoming. Here, we report quantitative study findings that address the following study sub-questions:

Research Questions

1. What are NL oncology nurses' a) knowledge levels, b) confidence, c) attitudes, and, d) practices related to genomics?
2. Are the social systems/organization cultures conducive to NL oncology nurses' adoption of genomics?
3. What demographic and contextual variables are associated with NL oncology nurses' knowledge scores?

Theoretical Framework

The complexity of genomics presents unique challenges and considerations for its integration into professional clinical practice and the translation of evidence-based genomic applications has been a frequent focus in healthcare implementation science (Best et al., 2025). A change theory frequently referenced in implementation science frameworks and the nursing specific genomics implementation literature is Rogers' (2003) Diffusion of Innovations (DOI) (Damschroder et al., 2022; Dwyer et al., 2022; Leach et al., 2016). Per the DOI, an individual's readiness for adoption of an innovation (such as genomics-informed care) is influenced by their knowledge, competency and attitudes/receptivity surrounding the innovation, as well as the social system channels (Rogers, 2003). These key theoretical domains in the DOI theory were the basis of the measurement domains of the Genetics and Genomics Nursing Practice Survey (GGNPS). The GGNPS is a valid, reliable self-report instrument designed to measure the DOI theoretical domains in the context of nurses' adoption of genomics in practice (Calzone et al., 2016; Plavskin et al., 2019, 2023).

Methods

Design

Quantitative study results reported here are from a descriptive, cross-sectional survey. We were guided by relevant criteria of the CHERRIES (Checklist for Reporting Results of Internet E-Surveys) guidelines (Eysenbach, 2004) to report study findings.

Setting and Sample

Publicly funded health care in NL is delivered by the provincial health authority with five regional zones: Eastern Urban (EUZ), Eastern Rural (ERZ), Central (CZ), Western (WZ) and Labrador Grenfell (LGZ). The largest zone (EUZ) provides healthcare to approximately 50% of the NL population (Dowden et al., 2024). There are four CCP outpatient clinics located throughout NL: one in the EUZ, two in the CZ, and one in the WZ. (Dowden et al., 2024). The CCP provides outpatient systemic therapy and clinical trials, radiation therapy, screening and patient outreach services. In-patient systemic therapy is also provided in the EUZ in two inpatient units (adult and pediatric). Nurses (Registered Nurses, Licensed Practical Nurses, Nurse Practitioners) were eligible for this study if they worked in one of the four outpatient sites of the CCP or in one of the two inpatient oncology units within the EUZ. The estimated total number of nurses eligible to participate in the survey was 111 (a confirmed 56 in the CCP and an estimated 55 in inpatient settings). At the 95% confidence level with a margin of error of ±5%, it was determined that a sample size of 86 was needed.

Recruitment

Multiple recruitment strategies were employed. Team member AH (an oncologist in the CCP) connected RP with nursing administrative and clinical education personnel in the CCP. RP then visited the four CCP outpatient sites and the EUZ adult inpatient unit to explain the study's purpose. This included an integrated knowledge translation (iKT) session at the EUZ CCP outpatient site, co-facilitated by RP and a patient partner with lived experience of hereditary cancer as part of the larger mixed-methods study. During all sessions, refreshments were provided, and participants received a QR code to access the survey at their convenience. The clinical nurse educator in the CCP sent email reminders with the survey URL immediately following the iKT session and again two weeks before the survey closed. Additionally, posters displaying the QR code were placed at all worksites where eligible nurses were based. Further recruitment efforts were guided by Dillman's (2000) tailored survey methodology, including a "lucky draw" incentive for a $100 e-gift card.

Data Collection

Survey data were collected on an electronic survey platform (Qualtrics, 2023). The survey was piloted for face validity and comprehension with (n=3) nursing colleagues prior to launching with no changes made. Data collection occurred between September 15th 2023 and February 20th 2024. Participants voluntarily completed the survey on their own time on their electronic devices. Questions were delivered over seven screens, ranging from 1-28 question items per screen. Participants did not have the option to go back and change their responses. No evidence of fraudulent data was detected; timestamps for completed surveys aligned with the estimated minimum completion time (>15 minutes), IP addresses matched eligible geographic regions, and no suspicious email entries were found in the separate raffle.

Ethical Considerations

Health Research Ethics Board approval (HREB #:2023.122) as well as institutional approval were obtained for the study. Participants provided informed consent before starting the survey and were advised that participation was voluntary, with the option of skipping any question they preferred not to answer.

Measurement Instruments

The survey package consisted of 60 items across three sections (See Appendix A for a full description of the research questions and corresponding measurement instruments and items). *Section one* consisted of seven researcher-developed occupational and demographic questions including a priori predictors of nurses' genomic knowledge as well as questions relevant to the NL oncology context. *Section two* included 22 modified, select items (dichotomous, ordinal, Likert) from the original 60-item validated GGNPS (Calzone et al., 2012) which measures nurses' attitudes, knowledge, confidence, and practices with genomics, as well as the influence of the social systems on their adoption of genomics in practice. The GGNPS is publicly accessible https://www.genome.gov/Pages/Health/HealthCareProvidersInfo/GGNPSurvey_2.5%207-2-2014.pdf with the view that it can be modified to context as was recently done in Canada (Limoges et al., 2024). In our study, we used only select (22 out of 60) GGNPS questions, making minor modifications to some questions to ensure contextual relevance to the Canadian oncology setting. In *section three*, knowledge was assessed using the 31-item Genomic Nursing Concept Inventory (GNCI©) which measures the genomic knowledge expected of US-baccalaureate-level nurses across four topical categories (genome basics, mutations, inheritance, genomic healthcare applications) (Ward et al. 2014). Although not created for specific use in oncology, the GNCI© is a valid, reliable instrument which assesses nurses' knowledge of foundational genomic principles highly relevant to cancer (Ward et al., 2014; Ward et al., 2018). As these topical knowledge categories in the GNCI© were not included in the GGNPS, the GNCI© was used in lieu of the GGNPS knowledge questions.

Data Analysis

Data were exported to SPSS v. 29 (IBM Corp, 2022) to complete analysis. Frequencies and percentages were used to describe categorical variables and means ± standard deviations were used to describe continuous variables. Multiple linear regression (MLR) was used to identify demographic and contextual predictors of oncology nurses' genomic knowledge. We first assessed all possible knowledge predictors (i.e. responses to all seven individual demographic and the 22 GGNPS items) at the univariable linear regression (SLR) level ($P \leq 0.2$): Variables that were significant ($p \leq 0.2$) were entered in the first MLR model and using a hierarchical approach, variables were removed that were not significant at ($p \leq 0.05$). The F-Test was used to test the significance of the variables removed when comparing models (with and without the variable). We also assessed for confounding by dropping one variable at a time to check for changes in Beta coefficients ($\geq 20\%$) and found no evidence of same. We also tested for and found no evidence of effect modification. The final model was tested for assumptions and met all linear regression assumptions. In exploration of a possible relationship between nurses' practice unit (inpatient vs outpatient), and whether they completed a complete family history over the past three months, family history collection was transformed from an ordinal to a dichotomous variable: those 'who collected history in past three months' (i.e. combining the responses of those who reported that they did so either 'always', 'frequently', and 'occasionally') versus those who 'rarely or never collected family history'. An exploratory chi-squared analysis was then conducted. Two-tailed $p \leq 0.05$ was set as a significance level.

Results

Given the small total available population and our institutionally supported, diverse recruitment efforts, most eligible nurses likely would have received notification of the survey, but this cannot be confirmed definitively. Nonetheless, the approximate participation rate was 56%. Of the (n=61) who consented to enter survey, 11 submissions were removed due to missing data. This resulted in a final 50 respondents included in analysis. While the target sample size was not met, the approximated response rate was 45% and the achieved sample size aligned with a 90% confidence level and a 10% margin of error (recommended sample size 43), suggesting that these findings are reasonably representative of the target population. Most respondents reported they provided direct patient care (88%) and worked in the outpatient setting (74%) (see Table 1 for participant characteristics).

Table 3.1

Table 1. Participant Demographics

	M ±SD (Range)
Age	38.6 ± 10.1 (22-57)
Years of Experience in Oncology	7.65 ± 8.21 (0-33)
	% (n)
Sex/Gender	
Female	94% (47)
Male	2% (1)
Gender queer/Gender non-conforming	2% (1)
Practice Setting	
Outpatient Urban	44% (22)
Outpatient Rural	20% (10)
Inpatient Urban	30% (15)
Other/Did not specify	6% (3)
CNA Specialty Certification (Oncology)	
Yes	36% (18)
No	64% (32)
Highest Level of Education	
Diploma	30% (15)
Undergraduate Degree	56% (28)
Master’s Degree	14% (7)
Nursing Role	
Direct Patient Care	88% (43)
Education	4% (2)
Other	6% (3)
Prefer not to Answer	2% (1)

The completeness rate (those who clicked the consent to participate versus those who completed the entire questionnaire) was 70%. Missing data was managed by pairwise deletion for descriptive statistics and listwise deletion for regression analysis. Descriptive results for research questions one and two are reported below in accordance with the corresponding DOI domains, i.e., knowledge, attitudes/receptivity, confidence, practices, influence of social systems (see Appendix B for tables of full descriptive results related to these domains). Regression results (research question 3) are also reported below separately.

Knowledge

The mean overall knowledge score (measured by the GNCI©) was 12.02± 3.54 out of 31 or 38.8% of total questions correct, with scores ranging from 4 to 21, indicative of low overall levels of foundational genomic knowledge. On the GNCI© topical category subscales, the lowest performance was observed in the "Genome Basics" category, with an average score of 29% percent of questions correct, followed by 41% in "Genomic healthcare applications", 43% correct in "mutations" and 44% correct in "Inheritance". On a particularly relevant individual GNCI© question, only 13/43 respondents (30.2%) correctly identified a 'red flag' in a family history phenotype concerning for an inherited condition.

Attitudes/Receptivity

Most participants (94%) indicated that they thought it was either very or somewhat important to become more educated about the genomics of oncology, with 68% reporting that they intended to learn more about the topic. Overall, participants reported potential benefits of integrating genomics into their practice such as 'identification of individuals who could benefit from high-risk screening' (98%) and 'improved patient health teaching' (98%). Participants also indicated potential disadvantages of integrating genomics into practice such as: increased patient anxiety related to risk (48%) and the time commitment involved (24%).

Confidence

Across the five items assessing confidence in providing genomics-informed care, the majority of respondents reported that they were ‘not at all confident’ in the following areas: discussing how family history affects screening intervals (54%), discussing the benefits of genomics (54%), accessing current and reliable information about genomics (64%), discussing the risks of genomics (90%) and explaining the limitations of genomic testing to patients (94%).

Influence of Social Systems/Organizational Cultures

Only 40.8% reported that genomics was part of their foundational nursing education and only 14.3% of oncology nurses reported attending education about genomics since licensure. Most respondents indicated that they were unsure if (56%) or did not think (18%) their manager perceived genomics as an important part of their role. Similarly, the majority were either uncertain (44%) or did not think (18%) that senior staff saw genomics as an important part of their role. The majority were either uncertain (64%) or did not think (24%) there would be available time during their work hours to upskill their professional knowledge about genomics.

Practices

Of note, 46% of respondents indicated that within the last three months patients had approached them with questions about genomics. While some reported that they always (4%), often (10%), or occasionally (30%) collected a complete three-generation family health history in the past three months, the majority (56%) indicated that they did so rarely or never. In our exploratory analysis, it was found that outpatient nurses were more likely than inpatient nurses to have conducted a complete family history in the last three months ($p = 0.032$).

Multivariate Analysis of Predictors of Genomic Knowledge

In the univariable linear regression (SLR), seven out of 27 variables were significant predictors of genomic knowledge at the ($P \leq 0.2$) significance level: practice setting; years of oncology experience; genomics content in their undergraduate curricula; perceived support for genomics-informed practices from their manager; family health history collection practices; and responding that the following were disadvantages of integrating genomics: too much time, more professional education required, and potential to increase patient anxiety about risk. In the final MLR model assessing predictors of GNCI© score, the following variables were included practice unit: (inpatient vs outpatient) and identifying a 'need for additional professional education' as potential disadvantage of integrating genomics in practice (see table 2):

Table 3.2

Table 2 Multiple Linear Regression Model of Predictors of GNCI© score

Variable	Coefficient Beta (β)	SE	t	P-value	[95% Conf. Interval]
Practice unit (inpatient vs outpatient)	3.74	.978	3.829	<.001	1.77 – 5.72
Need for additional professional education as a perceived disadvantage	-2.59	1.059	- 2.477	0.019	(-4.74) – (-0.45)
Adjusted R^2 =*.323*	F = 10.56				

There was no relationship identified between GNCI© scores and variables identified in previous studies using the GGNPS or GNCI©, such as education level (Wright et al., 2020) or attending a previous continuing education course in genomics (Connors et al., 2022). An unexpected finding was that working in an inpatient setting (compared to the outpatient setting) was associated with a significantly higher GNCI© score (β= 3.74 p < .001; 95% CI 1.77 – 5.72) and reporting the 'need for additional professional education' as a perceived disadvantage of integrating genomics in practice was associated with a significantly lower GNCI© score (β = -

2.59 $p = 0.019$ 95% CI -4.74 to -0.45). The overall model was significant $p < 0.001$ and the predictors accounted for 32.3% of the variance in the GNCI© knowledge score.

Discussion

Overall, in our study, we found that the NL cancer nursing workforce lacks readiness for genomics-informed care, as their genomic knowledge scores were low, and they reported a lack of confidence to provide genomics-informed care. Our findings are comparable to prior Canadian (Chiu et al., 2024; Hébert et al., 2022) and international findings (Hines-Dowell et al., 2024; Seven et al., 2017; Wright et al., 2020). For example, Hines Dowell et al. (2024) found that only 55% of nurses working in a US pediatric oncology setting could correctly identify the difference between a germline and somatic genetic variant.

Further interpretation of our results reveals several implications for practice, policy advocacy, continuing education, and future research. It is warranted to emphasize that most respondents expressed positive views about the benefits of integrating genomics into their practice. When this is considered in conjunction with respondents' low reported confidence, limited knowledge of, and minimal use of genomics in practice, these gaps appear less likely to stem from nurses' attitudes related to genomics. Rather, our results appear to reflect other contributing factors. For example, nurses who responded that the 'need for additional professional education' was a potential disadvantage of adopting genomics in their practice had significantly lower GNCI© scores. We also found that many nurses reported a lack of confidence in their ability to access up-to-date genomic information. Most respondents were either uncertain if or did not think there would be protected time in their organization for them to learn more about genomics. Therefore, respondents' limited genomics knowledge and low reported rates of consistently collecting a complete FHH likely reflect, at least in part, the limited resources and structural supports available to nurses for the implementation of genomics in their practice**.**

While nurses in outpatient settings reported collecting a complete FHH slightly more frequently than those in the inpatient settings, the observed knowledge scores were higher among the inpatient nurse respondents. Therefore, our findings suggest that while some nurses working in outpatient settings may be collecting comprehensive FHHs, this practice may be of limited value in the absence of the foundational genomic knowledge (e.g. inheritance patterns and red flags in a phenotype suggestive of a CPS) that illuminates the clinical value of a FHH. Thus, our findings also possibly indicate opportunities in the CCP to identify high-risk persons who could potentially benefit from genomic testing which are currently being missed. Further, in the CANO/ACIO (2006) specialty oncology nurse competences, an expected standard of practice is that specialized oncology nurses will “apply knowledge of the role of genetics in disease-associated variations to assess genetic family history information” (p. 34). Considering this, our findings underscore that more organizational and system level supports are needed to ensure that specialized oncology nurses can meet their existing practice standards and competencies pertaining to genomics-informed care.

Based on our findings, we highlight opportunities in the policy and professional advocacy arena for nursing unions, professional nursing organizations, and nursing and health system leaders to support nurses’ genomics-informed practices. Persons serving in such roles and organizations can advocate for protected time, educational opportunities and resources for oncology nurses to meet their expected and evolving standards of practice in the delivery of genomics-informed care. An encouraging development in this area is that CANO/ACIO (2025) recently released a position statement on genomics-informed oncology nursing. This position statement affirms a unified pan Canadian stance about the relevance of genomics in oncology nursing and the need for enhanced foundational and continuing nursing education about genomics which is also supported by our study findings.

In addition to advocating and ensuring protected time for nurses to upskill their genomic competency, other efforts to optimize oncology nurses' FHH collection practices could include research and quality improvement, informed by robust implementation frameworks and co-developed with nurses, to develop and implement FHH and other tailored clinical decision support tools within electronic health record system (Lau-Min et al., 2023; Zorn et al., 2022). Implementation research surrounding the barriers and facilitators to oncology nurses' uptake of other evidence-based practices may also provide useful insights to efforts to integrate evidence-based genomics applications across oncology nursing. As an example, in a recent scoping review underpinned by implementation frameworks, barriers and facilitators of oncology nurses' uptake of evidence-based symptom management guidelines were examined (Teggart et al., 2024). It was found that in some cases, managers and advance practice nurses assumed 'opinion leader' roles that positively influenced oncology nurses' uptake of evidence-based symptom management guidelines across their organizations. Juxtaposing this finding against our results reported here, no high-level managers or advanced practices nurses responded to our survey. Interestingly, in some jurisdictions, advanced practice oncology nurses assume dedicated genomic risk assessment and follow-up roles in cancer care settings (Dowling et al., 2024). A consideration for future genomics implementation efforts may be to engage advanced practice nurses working in oncology who have an interest in genomics to take on clinical leadership roles that promote genomics competency development among nurses within their practice settings. High and mid-level managers can also play a role by ensuring institutional support for nurses to engage with genomics, by ensuring protected time for genomics education and facilitating access to genomics resources and expertise.

While respondents' attitudes towards adopting genomics in practice were mostly positive, nearly half indicated that they perceived its potential to increase patient anxiety as a disadvantage to adopting it in practice. Primarily, oncology nurses interact with people actively receiving cancer treatment who have multiple, complex and sometimes competing care needs (e.g., physical, informational, spiritual) (Fitch, 2008). Thus, it is plausible that nurses may hesitate to adopt genomics into their practice, not only due to their limited confidence in providing genomics-informed care, but also due to concerns of overwhelming patients with information related to cancer risk during their cancer treatment journeys. Another possible interpretation is that nurses may perceive that discussing genomics with patients serves little practical purpose when there is a lack of patient referral pathways to a dedicated hereditary cancer follow-up service, as was the case in our study setting at the time of data collection (Hynes et al., 2023).

Irrespective of any reservations oncology nurses may have about adopting genomics, aligned with recent findings of Chiu et al. (2024), our results indicate patients are already approaching oncology nurses with genomics-related questions. Forty-six percent of respondents in our study reported receiving patient questions about genomics in the past three months, in comparison to 19% who reported the same in a recent nationwide survey of nurses practicing across all areas (Limoges et al., 2024). This underscores how patient interest is driving the relevance of genomics, especially in oncology nursing practice settings (Chiu et al., 2024; Limoges et al., 2024). Thus, we suggest further research about nurses' implementation of genomics in the context of cancer nursing practice. Firstly, we suggest further qualitative examination of nurses' perceived barriers and facilitators of adopting genomics into their practice. We also suggest future in-depth examination related to the types of questions oncology nurses frequently receive from patients about genomics. Examining the types of patient-driven informational needs could inform patient centered and contextually relevant professional genomics education and clinical implementation strategies.

A final consideration is that only 32.3% of the knowledge scores were explained by the regression model, thus it is probable that other unmeasured variables influenced oncology nurses' genomic knowledge scores. Notably, multidisciplinary influences were not captured in our measures of social systems influences. As oncology care is incredibly interdisciplinary, other healthcare professionals likely influence nurses' knowledge and adoption of genomics. A multidisciplinary approach to building clinical genomic competency may offer advantages over nursing discipline specific efforts, which have yet to result in strong evidence of sustained nursing practice change globally (Thomas et al., 2023). A potential strategy used in Italy is the co-development of multidisciplinary core competencies in cancer genomics to promote role clarity and care continuity in genomics healthcare (Hoxhaj et al., 2021). Similarly, in other international settings, implementation science frameworks have been used to guide the clinical translation of genomics in healthcare systems in collaboration with multidisciplinary non-genetics healthcare teams (Best et al., 2025; Ma et al., 2025).

Limitations and Strengths

Our findings should be interpreted cautiously as there are several limitations to our study. Firstly, as data were self-reported by nurses, our findings carry potential for social desirability bias. While we used only select, modified questions from the GGNPS in an effort to minimize response burden, the survey was still quite long which could have contributed to the observed participant attrition. Our modifications and removal of some items from the original GGNPS limited our ability to make standardized comparisons with other studies that used the full instrument. We also did not conduct psychometric testing on the modified GGNPS questions used here. Finally, while the GNCI© is a valid, reliable instrument, it was not designed for specific use in oncology settings and there are some oncology specific knowledge considerations which were not captured.

Despite these limitations, as an exploratory study, its strengths include a response rate of 45% with representation of oncology nurses across different geographic areas of the province where cancer care is delivered. Further, data was collected at a time when there is a clear provincial health system mission alignment for enhanced cancer care services that include genomics (Health Accord NL, 2022).

Conclusion

As publicly funded genomic testing becomes more widely available in Canadian oncology settings for more clinical indications, the cancer care workforce, including nurses, must evolve to meet the changing needs of individuals affected by cancer. In this study, NL oncology nurses recognized the benefits of genomics and reported interest in learning more, but they have not yet been adequately supported to confidently apply genomics in patient care. However, as nearly half of the oncology nurses reported that patients already are approaching them with questions about genomics, our findings add further empirical support to claims that oncology nurses are well positioned in cancer care settings to meaningfully contribute to the delivery of evidence-informed cancer genomic care to patients and families. Developing and implementing strategies to translate evidence-based genomics applications into high quality cancer care will require organizational and leadership support, collaboration across many disciplines, and the active engagement of many interest holders. Our findings and implications may be relevant to oncology nurses and nursing leaders, advanced practice nurses, and health system administrators as they plan and execute efforts to support genomics implementation in NL and in other oncology settings.

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Supplemental Tables in Journal Article

[ARTICLE] Appendix A: Research questions, survey instruments, items and analysis

Research Question and Variable DOMAINS	Measurement Instrument + # of individual items	Question Item #s in Survey Package (+ *How They Are Reported in Results*)
1. What are NL oncology nurses' genomic: a) knowledge levels? **[KNOWLEDGE]**	GNCI© (31 items) Multiple choice questions across four topical categories (a) *genome basics* (13 items); (b) *mutations* (3 items); (c) *inheritance* (8 items); and (d) *genomic health care applications* (7 items)	Qs.30-60 (*reported as a mean/SD /percentage of total GNCI© out of 31 and for the four GNCI© subscales)*
b) attitudes related to implementing genomics in practice? **[ATTITUDES/ RECEPTIVITY]**	GGNPS[MSQ] (10 items)	Qs. 8, 14-20, 25, 27 (*reported as frequencies/percentages, for the individual questions*)
c) confidence levels? **[CONFIDENCE]**	GGNPS[MSQ] (5 items)	Qs 9-13 *(reported individually as frequencies/percentage of total respondents for the individual questions)*
d) practices? **[PRACTICES]**	GGNPS[MSQ] (2 items)	Qs 21, 22 *(reported individually as frequencies/percentages for the individual questions)*
2. Are the social systems/organization cultures conducive to NL oncology nurses' adoption of genomics? **[SOCIAL SYSTEMS]**	GGNPS[MSQ] (5 items)	Qs 23, 24, 26, 28, 29 *(reported as frequencies/percentages for the individual questions*)
3. What demographic and contextual variables are associated with oncology nurses' knowledge scores? [**ALL DOMAINS]**	Demographic Questionnaire [Predictors] (7 items) GGNPS [MSQ] [Predictors] (22 items) GNCI© [Outcome] (31 items)	Qs 1-7, 8-29 [Predictors] Qs 30-60 [Outcome] (*reported as a regression model with total GNCI© score as outcome variable*)

MSQ = Modified, Select Questions

[ARTICLE Appendix B]: Descriptive Data Tables

Genomic Knowledge (as measured by the GNCI©)

	% correct; M ±SD (Range)
Total GNCI© Score out of 31	38%; 12.02 ± 4.02 (4-21)
GNCI© Subscales Scores	
i.Genome Basics (out of 13)	29%; 3.8 ± 1.85 (0-8)
ii.Mutations (out of 3)	43%; 1.30 ± 0.83 (0-3)
iii.Inheritance (out of 8)	44%; 3.53 ± 1.68 (0-8)
iv.Genomic Healthcare applications (out of 7)	41%: 2.88 ± 1.54 (0-6)

Attitudes/Receptivity About Genomics

	(n), %
Importance of Oncology Nurses' Increased Knowledge of Cancer Genomics?*	
Very Important	(33), 68.8%
Somewhat Important	(14), 29.2%
Not sure/Don't Know	(1), 2%
Intention to Learn More about Genetics?	
Yes	(34), 68%
No	(1), 2%
Don't know	(15), 30%
Would you attend a course on your own time?	
Yes	(32), 64%
No	(3), 6%
Don't Know	(15), 30%
Potential Advantages and Disadvantages of Integrating Genomics into Oncology Nursing Practice	
Better Treatment Decisions:	
No Advantage	(5), 10%
Advantage	(45), 90%
Improved Services to Patients (e.g., Health teaching):	
No Advantage	(1), 2%
Advantage	(49), 98%
Better Uptake of Clinical Recommendations:	
No Advantage	(1), 2%

Advantage	(49), 98%
Identification of Individuals Involved in High-Risk Screening Programs	
No Advantage	(1), 2%
Advantage	(49), 98%
Would take too much time*	
No Disadvantage	(35), 72.9%
Disadvantage	(13), 27.1%
Need for Additional Professional Education	
No Disadvantage	(38), 76%
Disadvantage	(12), 24%
Increased Patient Anxiety Around Risk	
No Disadvantage	(26), 52%
Disadvantage	(24), 48%

*= 48 respondents for the item

Confidence

	(n), %
Discuss how family history affects screening intervals	
Not At All Confident	(27), 54%
Confident	(23), 46%
Access Reliable and Current Information	
Not At All Confident	(32), 64%
Confident	(18), 36%
Provide Patients Information about Risks of GT	
Not At All Confident	(45), 90%
Confident	(5), 10%
Provide Patients Information about Limitations of GT	
Not At All Confident	(47), 94%
Confident	(3), 6%
Provide Patients Information About Benefits of GT	
Not At All Confident	(27), 54%
Confident	(23), 46%

Practices

	(n), %
How often in the last three months have you collected a complete, 3 generation family history?	
Always	(2), 4%
Often	(5), 10%
Occasionally	(15), 30%
Rarely or Never	(28), 56%
In the past three months, has any patient initiated a discussion about genetics?	
Yes	(23), 46%
No	(27), 54%

Influence of the Social Systems

	(n), %
Did your nursing curriculum include genetics content?**	
Yes	(20), 40.8%
No	(29), 59.2%
Since licensure, have you attended any continuing education that included genetics as a major component?**	
Yes	(7) 85.7%
No	(42) 14.3%
Do you think managers see genetics as an important part of your role?	
Yes	(13), 26%
No	(9), 18%
Don't Know	(28), 56%
Do you think senior staff members see genetics as an important part of their role?	
Yes	(19), 38%
No	(9), 18%
Don't Know	(22), 44%
Would you be able to attend a genomics course during work hours?	
Yes	(18), 36%
No	(12), 24%
Don't Know	(20), 40%

** = 49 respondents for the item

CHAPTER 4: Nurses and Patients' Experiences and Perceptions of Genomics-Informed Cancer Nursing Care: Inferences from the Integration of a Convergent, Mixed Methods Study

Rebecca Puddester, RN, PhD (c),[a]

Holly Etchegary PhD [b, d]

April Pike, RN, PhD, [a]

Kathleen Stevens, RN, PhD, [a]

Angela Hyde, MD, PhD, FRCPC, [c]

Joy Maddigan, RN, PhD, [a]

[a] Faculty of Nursing, Memorial University, St. John's, Newfoundland and Labrador, Canada

[b] NL SUPPORT Unit, Memorial University, St. John's, Newfoundland and Labrador, Canada

[c] Disciplines of Laboratory Medicine and Oncology, Faculty of Medicine, Memorial University, St. John's, Newfoundland and Labrador, Canada

[d] Community Health and Humanities Clinical Epidemiology, Faculty of Medicine, Memorial University, St. John's Newfoundland and Labrador, Canada

Introductory Statement

This unpublished manuscript reports integration findings of the qualitative and quantitative study streams. It is prepared for submission to the *Journal of Mixed Methods Research*. It is intended for the global audience with an interest in genomics nursing implementation. This manuscript also targets a global audience of mixed methods researchers who may find the interpretive description–informed integration approach of particular interest.

CRediT authorship contribution statement

Rebecca Puddester: Writing – original draft, Methodology, Investigation, Formal analysis, Conceptualization, Funding Acquisition. Holly Etchegary: Writing – review & editing, Funding Acquisition, Supervision, Methodology. April Pike: Writing – review & editing, Supervision. Kathleen Stevens: Writing – review & editing, Supervision. Angela Hyde: Conceptualization, Writing – review & editing; Joy Maddigan: Writing – review & editing, Supervision, Methodology.

Abstract

Background: Although genomics is increasingly integrated across cancer care settings, its integration into nursing practice remains limited. Existing global genomic nursing implementation frameworks highlight the importance of including the perspectives of nurses and patients with lived experience within local contexts to support implementation; however, few studies have reported on both perspectives. This study addressed that gap by exploring patient and nurse perceptions of genomics-informed cancer nursing care during the pre-implementation phase of a clinical genomic service change in a Canadian provincial setting.

Methods: A convergent mixed-methods design was used, integrating findings from a cross-sectional survey with validated measures of oncology nurses' (n=50) knowledge, confidence, attitudes, and practices with genomics, and a qualitative interpretive description of patients with cancer predisposition syndromes (CPS) (n=37), in the Canadian province of Newfoundland and Labrador (NL). Patient partners and nurse knowledge users were engaged at different phases of the research process, following Canadian guidance for best practices for patient-oriented research and integrated knowledge translation. Integration analysis was inductive, guided by the Interpretive Description qualitative approach. The four-step Pillar Integration Process (PIP) was followed as an analytic technique to develop mixed-method inferences.

Results: Three inferences were generated: (1) Although genomic testing and related patient questions are emerging in routine cancer care, both nurses and patients often remain uncertain about where to turn next for answers. (2) Not the current status quo, but a shared recognition among cancer care nurses and patients about the benefits of adopting a genomics lens to existing nursing roles**,** such as accurate family history collection, assisting with health system navigation and providing supportive care. (3) Both groups endorsed the idea of a dedicated high-risk follow-up service for individuals with hereditary cancer predisposition while patients indicated that specialist cancer nurses could play key roles in a dedicated hereditary cancer follow-up program. **Conclusion:** Using a novel interpretive description–informed mixed-methods approach to integrate diverse perspectives on genomics-informed nursing (including provider measures and patient perceptions) within the same healthcare context, this study identified both the need and opportunity for new approaches to address persistent challenges in the adoption of genomics across nursing, in ways that are responsive to patients' current expectations and unmet care needs.

Key words: Genomics-informed care; cancer nursing; mixed-methods; interpretive description, patient-oriented research; hereditary cancer; cancer predisposition syndromes (CPS)

Introduction

Cancer is driven by DNA changes (variants) acquired randomly or due to environmental exposures throughout the lifespan, although approximately 10% of cancers stem from constitutional (inherited) variants in the 100+ known cancer susceptibility genes; also referred to as hereditary cancers or cancer predisposition syndromes (CPS) (Garutti et al. 2023; National Cancer Institute, n.d.). 'Genomic cancer control applications' refers to the clinical use of germline (inherited) or somatic (tumor) genomic data, ranging from single gene to whole genome testing, to assess cancer risk, guide prevention, inform treatment, and predict recurrence (American Nurses Association, 2023; Friend et al., 2021). A growing number of evidence-based genomic cancer control applications improve progression-free survival, reduce cancer incidence, and/or lower mortality (e.g., genomic matched anti-cancer therapies).

In many regions, multi-gene testing is now recommended for cancers like ovarian and pancreatic, even without a family history of cancer, to guide treatment with oncologists who can now order this treatment-focused genetic testing (Alberta Precision Laboratories, 2022; Hereditary Cancer Testing Eligibility Group, 2024; Rendon et al., 2023). While this was the case for a long time in the Canadian province of Newfoundland and Labrador (NL), this study took place in NL ahead of a planned expansion of genomic-informed cancer care.

Genomic Service Change Context in Newfoundland and Labrador

The widely dispersed NL population (~ 549,900/1.4 per km^2) has some of the highest provincial cancer incidences, including the highest colorectal cancer incidence and mortality rates in Canada (Canadian Cancer Statistics Advisory Committee et al., 2023; Government of NL, 2025; Statistics Canada, 2021). The NL population also bears a high burden of modifiable and non-modifiable cancer risk factors (Shanahan et al., 2022). Among these is an increased prevalence of genetic predispositions to cancer (Rahman et al., 2003). Canada's universal healthcare is provincially administered, with some variations. In NL, a single provincial health authority administers healthcare across five regional zones. For several decades within this health authority, the Provincial Medical Genetics Program (PMGP) has provided tertiary genetics counselling and services to all zones in NL. Historically, Canadians only accessed hereditary cancer testing ordered by medical geneticists following specialty counselling in tertiary genetic centres (Tindale et al., 2022). Some oncologists in NL have been able to order 'mainstream' genetic testing for certain cancers (e.g., ovarian cancer) for several years. However, samples were processed out-of-province (Etchegary et al., 2021), and even when mainstream testing occurred, individuals identified with a CPS were referred to PMGP for post-test counselling. Beyond results disclosure, no formal long-term high-risk follow-up service existed for CPS patients in NL, leading to unmet care needs and missed care (Etchegary et al., 2015; Hynes et al., 2023; Watkins et al., 2011). Moreover, many with a CPS remain unidentified and are unaware of their cancer risk or evidence-based prevention options (Etchegary et al., 2022).

As NL has long been a paragon for genetic research discoveries, provincial cancer outcomes remain poor and unmet genomics information and supportive care needs are reported (Canadian Cancer Statistics Advisory Committee et al., 2023; Hynes et al., 2023, Rahman et al., 2004). In 2015, this prompted large-scale private and public investment in a research Centre for Translational Genomics (CTG) equipped with genomic sequencing capabilities. In response to a recent health system reform report, the NL government announced plans to repatriate genetic testing to this CTG, expand access to translational genomic testing, and further integrate clinical cancer care settings through a new high-risk cancer service located in the outpatient cancer clinic.

This anticipated genomics cancer service change in NL requires preparation of cancer providers to deliver and support expanded genomics-informed services. This includes oncology nurses who play key roles in NL's cancer care services. Though NL's context drives urgency, these considerations are relevant to nurses practicing across many settings where evidence-based genomics applications already are present in routine care. Competency statements for genomic-informed nursing practices were developed in some countries over 20 years ago, and validated measures have since been developed to measure nurses' knowledge/competency and use of genomics in practice (ANA, 2023; Plavskin et al., 2023; Ward et al., 2016). While the evidence base is modest, studies have identified contexts where nurses enhanced uptake of genetic testing and/or hereditary cancer management recommendations (Shevach et al., 2023; Yoes et al., 2020). Yet, concerningly, despite 20+ years of nursing leadership efforts and a growing number of evidence-based genomic applications, global reviews of studies measuring nurses' knowledge, competency, and use of genomics in practice suggest that little progress has been made in this area (Calzone et al., 2018; Wright et al., 2018).

Global Genomics Nursing Implementation Frameworks

The Genomics Nursing Alliance (G2NA) was formed in 2017, recognizing that novel, harmonized leadership approaches were needed to overcome challenges and drive the successful integration of genomics across nursing practice worldwide (Calzone et al., 2018). To advance this aim, the G2NA convened nursing leaders from 19 countries to create two tools to support nursing and health system leaders in integrating genomics within their organizational and local contexts. The G2NA implementation roadmap is based on the Consolidated Framework for

Implementation research. It outlines phased strategic guidance for genomic implementation. The second companion tool, ASIGN, enables nurses to benchmark the readiness of their regions/organizations to integrate genomics across 19 key enablers of successful implementation. ASIGN also highlights core genomic competencies as a key enabler of genomics nursing. Still, others included ‘clearly defined patient outcomes’, active patient involvement and empowerment’, and positive’ attitudes within the nursing workforce towards genomics, which are locally sensitive and build on broader public awareness’ (Tonkin, 2020a). Despite this, limited research has explored the relationship between nurses’ attitudes, knowledge/competency, and practices and patients’ perceptions, experiences, and expectations for genomics-informed cancer care.

Considering this gap and the anticipated genomics service delivery change in NL, this mixed-method integration aimed to explore perceptions of genomics-informed nursing, of nurses and patients[1] with lived experience receiving cancer genomic test results and navigating their subsequent care in the NL healthcare system. The ultimate aim of this mixed-methods integration was to generate a more complex, contextually relevant understanding of the needs, capacities, and opportunities for genomics-informed nursing care in cancer care settings. A secondary aim of this article is to illustrate how features of the Interpretive Description qualitative approach were used to inform integration analysis of this convergent mixed-methods study.

Methodology and Methods

Philosophical and Theoretical Positioning

Dialectical Pluralism

The dialectical pluralism (DP) metaparadigm was the worldview guiding this study, undertaken as a doctoral dissertation by the lead author (RP) (Johnson, 2012, 2017). The ontological assumptions of DP are that reality is plural (subjective, intersubjective, and objective). The interrelated DP epistemological stance is that complex phenomena can be known through a dialogical, reflexive, relational process of engaging with plural realities and interest holder perspectives to create a holistic understanding of the phenomena. DP's axiological assumptions hold that knowledge production aims to contribute to societal betterment and justice. DP endorses methodological integration of qualitative and quantitative data to produce comprehensive understandings (Johnson, 2012, 2017).

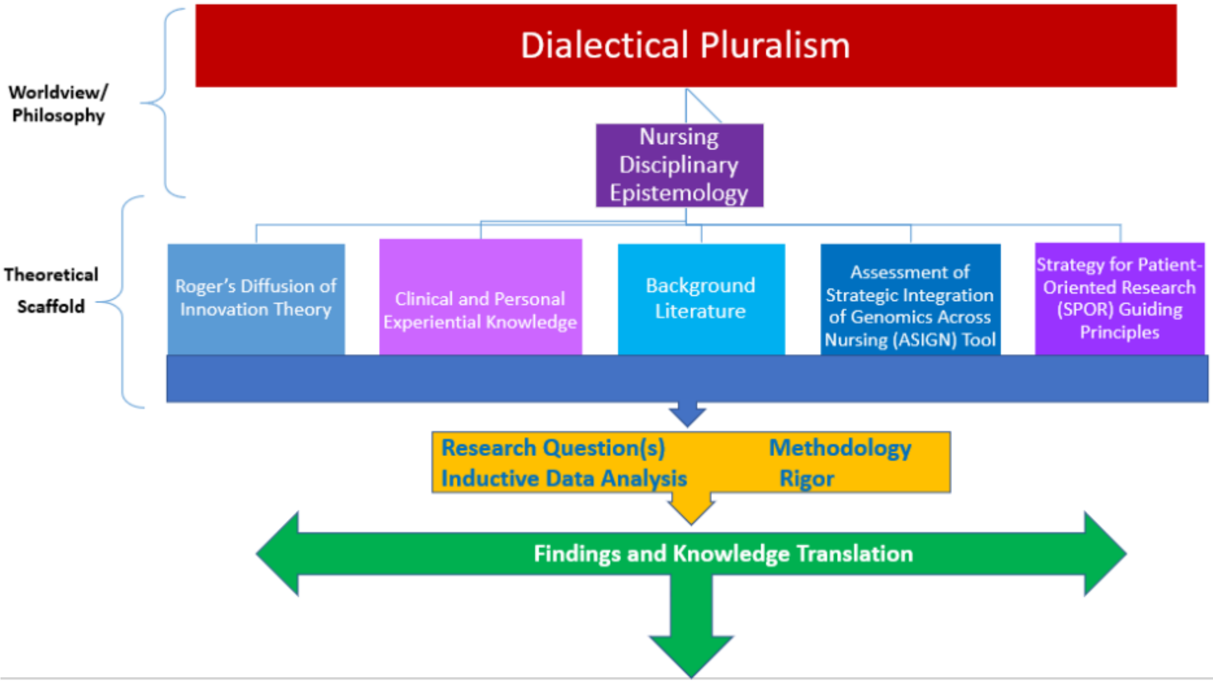
Theoretical Scaffold

Coined as a feature of the Interpretive Description (ID) qualitative approach, a 'theoretical scaffold' was adopted for this study (Thorne, 2016). Instead of selection and rigid adherence to a singular theoretical or conceptual framework, a 'theoretical scaffold' is a combination of any or all of the following elements that provide a flexible, non-prescriptive framework to guide the design logic, analysis, and reporting of a study: background literature, personal and clinical experiential knowledge, disciplinary epistemology, and any relevant theoretical ideas.

In addition to the background literature outlined in the introduction, specific to the theoretical scaffold in this study were: the lead author's clinical background as a RN with clinical experience in gynecologic oncology surgery; the lead author's disciplinary epistemological positioning which is philosophically compatible with the DP metaparadigm (e.g. attention to the intersubjective, subjective, and evidence-based practice; attention to patterns and individual variances, and the imperative to produce actionable, practical findings). Other theoretical and conceptual ideas in the scaffold included the diffusion of innovations (DOI) (Rogers 2003). This theory explains factors influencing how innovations (such as genomic technologies) are adopted in a social system over time. The DOI underpinned implementation science frameworks, such as Consolidation Framework for Implementation Research (CFIR) which was the basis of the G2NA tools (Damschroder et al., 2022; Tonkin et al., 2020a), including the ASIGN maturity matrix. Lastly, the study scaffold also included guiding principles for patient-oriented research (POR). POR is a term widely used in Canada, as described by the Canadian Institutes of Health Research (CIHR) (2014) in its Strategy for POR (SPOR) framework, to describe a continuum where patients and members of the public are actively engaged in the research process, rather than just subjects, to ensure both the relevance, acceptability and uptake of research for people who stand to be affected by it.

Figure 4.1

Visual Diagram of the Worldview and Theoretical Scaffold Informing the Study



Mixed-Method Integration Question

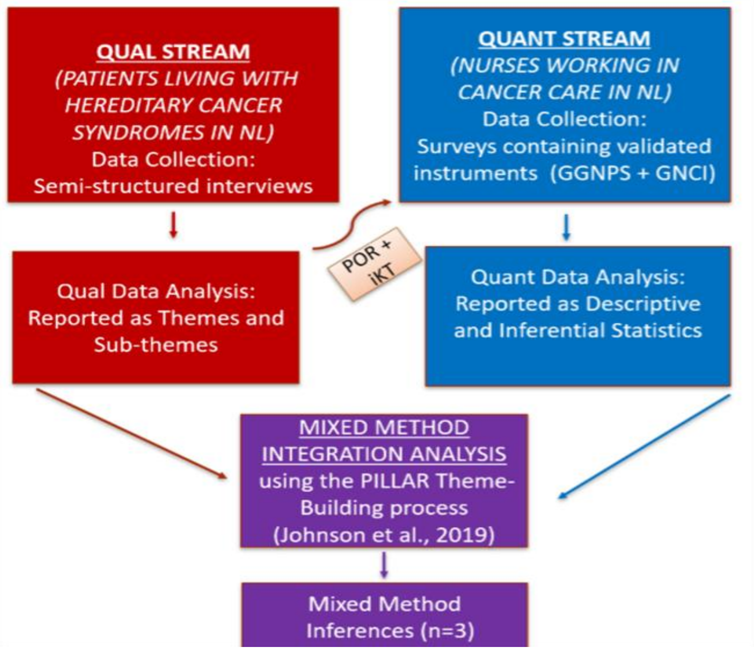
What insights can be learned from the integration of patients' and nurses' perceptions of genomics-informed cancer nursing care to inform further clinical implementation efforts?

Design

This mixed-methods study drew on patient-oriented research (POR) and interpretive description (ID) approaches (CIHR, 2014; Thorne, 2016). A convergent mixed-method design was used, with qualitative and quantitative data collected, analyzed, and reported separately prior to the integration (Creswell & Plano-Clarke, 2018; CIHR, 2014; Puddester et al., 2025; Puddester et al., in press). (see Figure 4.2 for a visual diagram of the phase of mixed-methods integration).

Figure 4.2

Mixed Method Study Design and Streams



A high-level summary of previously reported qualitative and quantitative study streams, including methodology, samples, and key findings, is presented in Table 4.1. Health Research Ethics Board approval was granted for both study streams (Reference #s 2022.125; 2023.122). While formal integration did not occur until after separate qualitative and quantitative analysis, there was early crossover between the streams using POR and integrated knowledge translation (iKT) (see Figure 4.2). This is described further in the following paragraph.

POR-iKT Approach (Patient and Public Engagement)

Following CIHR (2014) SPOR guidance, two patient[3] partners with lived experience of CPS were members of the research team, involved during the earliest stages of the qualitative study conceptualization, recruitment, data analysis, and reporting, as previously detailed in an article in which they co-authored (Puddester et al., 2025). Like POR, iKT involves research engagement and partnership, extended to many knowledge users (including healthcare providers) who are seen as partners in the research process. Guidance developed for POR also informed integrated knowledge translation (iKT) with the nurses in the quantitative stream. A research team member employed in the CCP (AH) connected the lead author with several key nurse informants in the CCP, including the clinical nurse educator. The lead author consulted the clinical educator on how to best engage with local oncology nurses (target population in the quantitative stream). Based on their recommendations, the lead author and patient partner team

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[3] Patient' is a term widely recognized and used in the literature related to lived experiences accessing health services and POR literature. For this reason, we use this term when describing our qualitative study participants and their accounts of their interactions with healthcare professionals and the system in relation to their CPS. However, we acknowledge there are limitations with the use of this word, as it can inadvertently imply that persons living with health conditions are passive recipients of care and thereby possibly devalue their expertise in their own care experiences. It is our position as authors that people affected by cancer and CPS are experts in their own health experiences (Alder & Horsfall, 2008; Starfield, 2016). Thus, elsewhere it is our preference to use person-centred language (e.g. 'person living with a CPS' which is featured elsewhere throughout the manuscript)

member co-facilitated an engagement session with the NL oncology nurses during their regularly scheduled education day. At this session, preliminary impressions from the qualitative stream results were presented. After the session, nurses received the QR code to complete the quantitative survey anonymously on their own time.

ID-Informed Mixed Method Integration Analysis

The ID qualitative methodology guided integration analysis (Thorne et al., 2016). Though distinct from raw data analysis, integration analysis shares similarities with qualitative analysis (i.e., involves constant comparison and interpretation). ID also supported the purpose of mixed method integration, which was to interpret the expanded insights from the combined quantitative and qualitative findings. Integration analysis was completed primarily by RP with ongoing guidance from her doctoral supervisor (JM). The analytic technique applied was the Pillar Integration Process (PIP), a four-step process using elements of traditional MM joint display tables (Johnson et al., 2019). This stepwise process is described below.

Table 4.1

Summary of Individual Qualitative and Quantitative Streams (Previously Reported)

Study Stream/Design	**Quantitative** (Descriptive, Cross-sectional)	**Qualitative** (Patient-Oriented Research, Interpretive Description) (Puddester et al., 2025)
Interest Holder Groups (Participants)	**Nurses** (n=50) of any designation working in inpatient or outpatient settings where cancer care is provided within the province of NL	**Patients** (n=37), > 18 years old, self-reported personal history of any CPS P/LP variant (e.g., *BRCA1/2*, *ATM*, *MSH6*, *CHEK2*) with or without personal history of cancer, within the province of NL
Study Stream Aim(s)	To describe the NL oncology nurses' knowledge, attitudes, confidence, and practices about genomics-informed care To describe the influence of the social systems on nurses' adoption of genomics-informed practices To assess what demographic and contextual variables are associated with oncology nurses' knowledge scores	To describe the genomics-informed healthcare experiences of individuals living with CPS To explore their experiences and perceptions of cancer genomic nursing care
Data Collection and Timeline	Electronic surveys (demographic questions, the validated 31-item Genomic Nursing Concept Inventory (GNCI©), along with 22 select, modified items from the validated Genetic/Genomic Nursing Practice Survey (GGNPS), (Calzone et al., 2012; Plavskin et al., 2019; Plavskin et al., 2023) were self-completed by nurses between September 2023 and February 2024	Interviews (with semi-structured interview guide) conducted by RP in person, via online meeting platform, or telephone between April and August 2023
Data Analysis	Descriptive and inferential statistics	Inductive constant comparative approach to analysis, as follows guidance by Thorne (2016)
Key Results	46% reported that their patients had initiated discussions about genomics over the past three months. Yet, this sample's overall knowledge and self-reported confidence in genomics practices were low, and the family health history collection is uneven. Nurses identified several potential advantages to patients of integrating genomics into their nursing practice and expressed receptivity toward receiving further genomic education.	Two primary themes: (1) *When genomic knowledge is power,* and (2) *Acceptability of nursing roles in CPS care.* Participants viewed genomic information as vital for personal and family decision-making, yet found it largely inaccessible within the healthcare system, leading to experiences of fragmented care. While most had limited nursing care involvement, there was support for expanded nursing roles, including introducing a dedicated oncology genomics nurse.

Step 1. Selective Listing

A PIP joint display template was created with columns (left to right) that included: qual THEMES AND SUB-THEMES; qual MEANING UNITS (organized by theme and sub-theme) the 'PILLAR'; quant DATA points and quant DOMAINS (see Table 4.2). Analysis began by listing relevant qualitative themes, sub-themes, and meaning units from the qualitative code book in the left columns of the PIP table. Once all relevant qualitative meaning units were listed in the PIP joint display table, quantitative data points categorized according to the diffusion of innovation theoretical domains of the quant measurements instruments (knowledge, attitudes/receptivity, practices/adoption, etc.) were displayed in the right-side columns of the PIP joint display table.

Step 2. Matching

When listing quant domains and datapoints, quantitative data were placed in horizontal proximity to qual themes, sub-themes, and meaning units with similar relational properties. Not all qualitative data were matched to relevant quantitative data and vice versa. During this stage, colour coding (highlighting) was used to identify possible linkages and relationships between the two data sources, drawing on guidance from the theoretical scaffold when making interpretive memos about possible expanded insights placed alongside the PIP table. The lead author held regular debriefs with her supervisor about emerging insights from the early steps in the integration process.

Step 3. Checking

Once all 'matches' were identified between the two streams, data were reduced in a new PIP template, which included only 'qual MEANING UNITS' and 'quant DATA' with confirmed matches. Codebooks, transcripts, and descriptive tables summarizing all quantitative results were reviewed to ensure no relevant information had been missed. At this stage, initial impressions were shared with patient partners, who reviewed them and confirmed the relevance of these interpretations.

Step 4. PILLAR Building

Through further interpretive memos, qualitative and quantitative data matches remaining in the PIP alongside each side of the blank 'PILLAR' following stage three resulted in the building of the integrated 'PILLAR' inferences. Integration findings are called 'inferences' to distinguish these from the previously reported qualitative themes (Puddester et al., 2025). An excerpt of the PIP joint display table with condensed quotes and datapoints supporting the development of the 'PILLAR' inference 2 is shown in Table 4.2 as a visualization of this analytic process.

Rigour

Integration rigour was maintained per ID quality criteria: epistemological integrity, representative credibility, analytic logic, and interpretive authority (Thorne, 2016). Epistemological integrity was maintained by confirming alignment with the DP metaparadigm and nursing epistemology with the research question and techniques used to develop inferences. Representative credibility was maintained by including qualitative quotes in the main text and qualitative and quantitative results in the PIP joint display table. Analytic logic was demonstrated in the detailed description of the PIP four-step process and joint display table. The lead author addressed interpretive authority through reflexive discussions with her supervisor, feedback from an interdisciplinary doctoral supervisory team (a medical oncologist and genomics health service researcher), patient research partners, and the use of interpretive memos. Further, contextual awareness is included in supplemental 'beyond evaluative criteria in ID' (Thorne, 2016). This was addressed by providing fulsome details on the NL context in the study background to situate the findings with the acknowledgement that they may not hold relevance in other contexts.

Results

Integration analysis generated three mixed-method inferences reflecting nurses' and patients' perspectives on genomics-informed cancer nursing care in NL.

Inference 1. While genomic testing and patients' related questions are arising across routine cancer care settings, many nurses and patients face uncertainty about where to turn next for guidance

It was revealed in inference 1 that genomics-related care discussions are increasingly occurring outside the silos of PMGP within routine oncology care. Many of the patient participants who received their genomic test results in recent years reported that their CPS gene status was identified when either themselves or their biological kin underwent treatment-focused genomic testing within the cancer care setting:

> *My aunt she was diagnosed with ovarian cancer, and they had asked her if she was interested in doing any type of genetic testing…then it came back that she had a rare gene…the ATM gene… so then we all got tested* (Participant 32).

For some of these patient participants, this testing had treatment implications: "*I'm on a PARP inhibitor* [genomic targeted therapy] *right now"* (Participant 3). Patients appeared to be imitating conversations about genomics in cancer care settings, as nearly half of cancer care nurses reported patients had initiated questions about genomics within the past three months. A few patient participants who were found to carry a CPS in more recent years, (both with and without a personal history of cancer) indicated that nurse navigators in the CCP had either helped them access genetic testing during their cancer journey or the recommended CPS screening.

> *The cancer nurse navigator in town did call me, and then she gave me the number to the in case I had any questions… where we are just trying to sort out what I was doing, if I was getting MRIs again* (P35, no personal history of cancer)

Most cancer care nurses cited low confidence discussing many aspects of genomics, including its risks, benefits, and limitations. Nurses' overall limited knowledge of genomics was observed in their low genomic knowledge scores. Nurses' unknowing related to genomics was also triangulated by patients' perceptions of gaps in provider knowledge overall. They reported an overall lack of knowledge of healthcare providers during their interactions with the healthcare system outside genetics specialty centres.

> It's *amazing how so many health professionals have never even heard of it* [ATM gene]. (Participant 16)

The lack of access to sufficient genomic knowledge outside of PMGP in the health and cancer care system where it could be applied, was often described as highly frustrating by the patient participants. One younger participant likened the current follow-up care approach as follows:

> It's *like someone dropped a bomb, but it's then like okay, figure that out, like clean that up on your own, and I'm like, ahh, isn't that your job? Like, I need help with this.* (Participant 30).

Many cancer nurses shared patients' uncertainty about how to proceed with responding to or directing patients' genomics questions. Some nurses expressed concern about integrating genomics into practice, owing to its potential to increase patients' risk anxiety. Nearly all reported a total lack of confidence about where they could access accurate and reliable genomics information to provide to patients and their families. These gaps in genomics care pathways across the cancer care continuum appeared to contribute to situations where several patient participants were living with unmet needs arising from their hereditary cancer risk and management considerations, when no official 'clinical space' existed where these needs could be addressed within the healthcare system*:*

> *I'm like doing my own Google, like research like about stuff like that, so it's like, no I've never had cancer, but I still have a lot of the effects from my preventative measures to not get cancer* (Participant 23)

Inference 2. Not the current status quo, but a shared recognition among cancer care nurses and patients about the benefits of adopting a genomics lens to existing nursing roles

With a few exceptions as described in inference 1, most of the patient participants who underwent genomic testing in recent years reported no involvement of nurses in their genomics health care along the pre- or post-testing continuum (see table for supplemental qualitative quote 1 [Q1]). As the phenomenon of genomics-informed nursing care was not grounded in their experiences, it was a prospect they considered hypothetically. Still, nearly all patient participants identified that nurses could play a key role in improving their genomics-informed care experiences. Though not related to genomics-informed care, many patient participants had previously interacted with nurses in the CCP during their or their family member's cancer treatment journeys. They were able to reflect on these experiences to identify potential opportunities regarding how a 'genomics lens' could be applied in existing cancer nursing roles and the related care they had experienced (e.g. nurse navigators, clinic nurses)

This perception of patients was shared by many nurse participants who also indicated there would be several advantages to patients if they were to integrate genomics into their nursing practice (D, 3, 4). Specifically, patients and nurses noted that nurses have a role in identifying and referring people at risk through an accurate family history and risk assessment (Q4 D3). Despite this, only a minority of nurse respondents indicated that they were consistently collecting a complete 3-generation family health history (D1), further in a knowledge question assessing nurses' knowledge of red flags for a genetic condition in a family history suggested underlying knowledge gaps.

Further, many nurse participants identified that adopting a genomics lens into their existing practice could be beneficial for improving uptake of clinical recommendations and health teaching among patients (D4). Some patient participants expressed similar ideas (Q3, 5). Patient participants suggested that many of the supportive care needs experienced by people living with cancer dually apply to people affected with CPS, regardless of their personal cancer history. Some suggested that the supportive care they received from nurses while undergoing cancer treatment could similarly benefit people living with an increased cancer risk, especially given the shortage of genomics specialist care providers (Q2).

Table 4.2

Excerpt of the PIP process in the generation of Inference 2

QUAL THEMES+ ***Sub-themes***	**QUAL MEANING UNITS Patients →**	**PILLAR Inference**	**QUANT DATA ← Nurses**	**QUANT DOMAINS**
Sub-theme 2.1 Limited Exposure to Genomic Nursing	**Q1:** *Nurses, no not so much, no it's always been doctors* (P10)	**Not yet status quo…**	**D1**: Only 4% of nurses reported taking a complete three generation family history within the last three months	**Practices**
	Q2: *Even if you had someone that [people living with a CPS] could talk to. But this genetics department is so understaffed that they don't have time to talk to people and when I say talk to people, I mean talk to people, like when I call the cancer center nurse you never ever get the impression that she wants to get off the phone* (P15)		**D2:** 54% were not at all confident to explain how family history affects screening intervals	**Confidence**
Sub-theme 2.2 perceptions of potential nursing contributions in CPS care	**Q3:** *I look at it as life and death. And if a nurse could actually navigate and say, okay, [name] you're aware of this. We can probably get you referred to genetics testing quicker. We can probably get you some support* (P3) **Q4:** *[nurses] could play a role at any point even when first discussions with the patient about the genetic history. I know the value from being a cancer patient, the value of nurse navigators, how helpful they are to ...and I think certainly more nurse navigators that work with the genetics department, I think that's something that could be very beneficial* (P17).	**...but shared recognition among cancer care nurses and patients about the benefits of adopting a genomics lens to existing nursing roles**	**D3:** 98% identified 'Identification of Individuals who Can Enroll in High-Risk Screening Programs' as an advantage of integrating genomics into their practice	
	Q5*: I think the nurse navigator that they had in place for [daughter], [daughter] was really pleased to avail of that service. Like I said, information is power. Education is power…You need to empower patients so that they know what kind it is, what the statistics are, what the protocol you need to follow, this is what you've got to do, all of this, all of that information...*" (P2)		**D4**: 'Improved Services to Patients (e.g., health teaching)' and 'better uptake of clinical recommendations' were identified respectively by 90% and 98% of respondents as advantages of integrating genomics into their practice	**Attitudes/ Receptivity**
	Q6*: They [nurses] need to have a knowledge of genetics and the cancers because there are several of them, genetic cancers. They need to know the next steps that need to be taken* (P4)		**D5**: 98% indicated it was either 'very' or somewhat important' for nurses to become more educated about cancer genomics	

Q: Qualitative Patient Participant Quote; D: Datapoint from Quantitative Study

Inference 3. Both nurses and patients see the potential benefit of a dedicated high-risk hereditary cancer follow-up service

In this final inference, it was revealed that patients and nurses perceived the potential advantages of establishing a dedicated high-risk hereditary cancer follow-up program, which at the time of data collection did not exist for individuals in the province at risk of hereditary cancer. Nurses largely reported that integrating genomics into their current practices could be advantageous, as they could support the functioning of a high-risk program by identifying potentially eligible patients who could benefit from this service. Many patients indicated that the current model of CPS follow-up care was far too 'ad hoc' and insufficient for meeting their complex screening, management, and supportive care needs experienced.

> *I really do think that more formalized process of monitoring people when they've been identified if you're not your own advocate under the current system I do feel it's very easy to fall through the cracks without a formalized structured and regular, predictable program that you can be part of and feel like there is somebody else other than you monitoring it* (Participant 7)*,*

Similarly, some patients expressed that it would be helpful if such a program were administered by

"a *central person, who's getting all my reports in, who's ordering my bloodwork, who's somebody that I can touch base with if I have a concern. So, having somebody that is a one point of contact*" (Participant 24)

Due to the nature of the data collection instruments administered to nurses, no measures captured their insights about which providers should be involved in delivering a dedicated high-risk program. Most of the patient participants indicated their receptivity to many providers serving in a dedicated follow-up role, including nurses.

"*I don't care if it's a doctor, LPN, RN, L-M-N-O-P…*" (Participant 6) (in Puddester et al., 2025, p.5)

Several patient participants noted that a dedicated specialist cancer nurse with knowledge about genomics could play a central role in a high-risk follow-up service.

> *I think that would be really great, especially if it became like more of like, I don't wanna say a specialized thing, but like if it became like their role particularly, I would definitely want that person to have some underlying knowledge that would be specific to genetics obviously, and I know like that nurses are trained in all aspects but I would definitely want them to be able to answer the more complex questions* (Participant 30)

While specialist genetic nursing roles currently do not exist within this setting, a few patient participants with a CPS who had received genetic test results several years prior had encountered genetic specialist nurses who were practicing in genetic counselling roles, including in rural areas. Consideration of the historical context during our analysis, these participants received this service before genetic nurses were phased out of official genetic counselling roles across Canada when a Masters degree in genetic counselling became the requirement for entry to practice as a genetic counsellor. Of the participants who encountered genetic nurses in the past, they recalled the genetic nurses' involvement in their care positively:

> "She *[nurse] was fantastic...she suggested what I should do, and so that's what I done. And I felt better. I felt I was doing the right thing as far as screening for myself"* (Participant 1)

Discussion

This mixed-method study was conducted during the pre-implementation of a novel genomics service within a provincial cancer care setting. Study findings provided a benchmark of patient and provider experience related to genomics-informed cancer and nursing care. Specifically, this integration analysis merged measures of oncology nurses' genomic knowledge scores and self-reported attitudes, confidence, and practices with genomics, with patients' experiences, perceptions, and preferences for their genomics-informed care. Integration findings revealed that as treatment-focused testing enters routine oncology, patients and oncology nurses share uncertainty about where to turn next to address patients' questions related to genomics. Further, although not status quo, patients and nurses saw opportunities and benefits of further integrating genomics into existing oncology nursing roles and identified the benefits of a dedicated high-risk hereditary cancer follow-up service.

Many quantitative studies have benchmarked all or some aspects of nurses' genomics competency (e.g., knowledge, confidence, attitudes, and practices), relying exclusively on self-reported data from nurses, with findings comparable to the quantitative results in this study (Limoges et al., 2024; Thomas et al., 2023). It is held that research in this area must shift from deficit-oriented descriptions of nurses' genomic competencies toward interventional studies of genomic implementation in clinical practice (Limoges et al., 2024; Thomas et al., 2023). While we concur, we also note findings of a recent synthesis of 86 systematic reviews of clinical implementation studies (not specific to genomics), where Boaz et al. (2024) concluded that the successful clinical implementation efforts require more than just isolated intervention studies. They suggested what is urgently needed is a more fulsome understanding of the "situated, relational, and organizational capabilities" (p. 28) that support implementation of evidence-based applications across healthcare settings. While there are other factors at play, this integration provided contextually relevant insights which reflect the relational nexus of professional nursing knowledge, which, since the earliest nursing code of ethics, has emphasized nurses' first and foremost duty of responsiveness to the individuals and families who receive nursing care (International Council of Nurses, 2021; Fowler, 2016).

Methodological Contribution

In addition, this integration study demonstrated philosophical compatibility between DP, nursing epistemology, and ID as methodological guide to a convergent mixed method study that brings together different perspectives through integration. The ID theoretical scaffold (drawing on background and clinical knowledge, nursing epistemology and genomic nursing implementation frameworks, and the DOI theory) provided a flexible, pragmatic approach that supported methodological innovation and responsiveness to real-world complexity.

While DP has been used as a metaparadigm for mixed-methods studies, and the use of ID as a qualitative component of mixed methods studies has been discussed, there are limited reports of the ID theoretical scaffolding and quality criteria used to guide mixed methods research and integration. This study also exemplified how the PIP analytic technique can support inductive, ID-informed integration analysis.

Study Implications

We also suggest this integration illuminated the alignments and gaps between provider capacity and patient needs, strengthening the relevance and actionability of the findings for nursing practice, policy development, education, and future research.

While most patients reported minimal contact with nurses in their genomics journey, some positively recalled past involvement of specialist nurses in genetic counselling services in NL. Historically, some NL nurses were grandfathered into these roles prior to the requirement of a Master's degree in genetic counselling for certification as a genetic counsellor. When these nurses retired, satellite genetic counselling clinics in rural areas in NL were closed (Dooley Adams, 2022). Bottorff et al. (2005) anticipated that with this change, the absence of an alternative genomic credentialing route for nurses would hinder the growth of a genomic competent nursing workforce. Since the 2009 certification change, no Canadian pathway has emerged to support nurses seeking genomics credentials, which may explain the current shortage of specialized nurses in this domain (Limoges et al., 2025). While our findings suggest patient satisfaction with nurses who practiced in genetic counselling roles, with nurses who still do so in countries like the UK, other alternative genetic service delivery models such as group genetic counselling and patient decision-aids have been proposed to address genetic counselling workforce shortages. Further, reinstating nurses into genetic counselling roles would not address the reported unmet care needs related to the fragmentation of genomic high-risk follow-up care in the health system outside of genetic counselling sessions.

Variation in cancer care delivery across regional, national, and global contexts means there is likely no “one-size-fits-all” approach to implementing nursing roles in genomics-informed cancer care. Globally, some cancer care centres have introduced nurse-led hereditary cancer clinics or collaborative models involving specialist genomic nurses (Rauw et al., 2022; Scott et al., 2020; Stempel et al., 2023). This may be feasible in some settings depending on demand and funding. Integrating genomics-informed care into existing oncology nursing roles, such as general nurse navigation or advanced practice nursing, may be more practical in other settings. However, these recommendations are not mutually exclusive. In fact, the synergy of dedicated nurse genomic experts with genomic-literate cancer nurses with broader roles may offer the greatest benefit.

There is anticipation that multi-gene panel testing may soon become standard for all patients with solid tumors, regardless of family history (Esplin et al., 2022). Further, in this study, the needs reported by patients affected by CPS also overlapped with broader supportive care needs experienced by patients across the cancer journey (Fitch et al., 2008). For this reason, genomics-informed care should not be viewed solely as a specialized role, but rather as an essential part of high-quality, person-centred supportive cancer care. Future research should prioritize identifying and reporting the specific roles responsibilities of cancer care nurses in genomics practice implementation, as well as the contexts and mechanisms that enabled the implementation of genomics-informed care. This is also an area for future realist reviews, to support informed policy and practice development.

Regardless of the various levels of specializations and responsibilities in genomics-informed cancer nursing, nurses' low knowledge and confidence in this study point to an overall need for enhanced genomics education. Yet, competency-based genomic nursing education interventions have yielded minimal sustained practice change (Thomas et al., 2023) This lack of practice uptake has driven questions about the relevancy of genomic competencies to all nurses if they are not consistently enacted in practice in the context of overloaded nursing educational curricula (Newcomb et al., 2019). Although a decidedly more complex issue, we argue that this is not because genomics is not relevant to nursing practice. Rather, one challenge is that the validated measures do not capture the full breadth of how nurses can apply genomic understanding in person-centred ways that reflect the rapidly evolving contexts of genomic-informed care delivery. For example, the GNCI© used in this study provides static knowledge assessments. Others, like the GGNPS, with sub-questions also used here, rely on family history collection and referral to genetic services as the sole benchmarks of adoption in practice. There is, however, an encouraging development to note in this area, the ACCESS framework, created by the International CASCADE Nursing Consortium based on their research conducted with patients and families affected by hereditary conditions (Katapodi et al., 2024). It outlines a unifying guide to nursing actions to reduce disparities in genomic care across five domains: Advocating, Active Coping, Communication, Cascade Screening, and Surveillance. While it is unclear whether the ACCESS framework is intended as a foundation for developing new measurement tools, our collective findings suggest that such an approach could be valuable, as patients reported needs and expectations for nursing care aligned with these five areas (Puddester et al., 2025).

Our decision to use both qualitative and quantitative streams was also influenced by the state of available research tools to assess the phenomenon. Although quantitative instruments exist to assess patient experiences and outcomes in hereditary cancer genetic counselling (Grant et al., 2019; Cella et al., 2002; DeMarco et al., 2004), none are specifically validated for capturing patient experiences with genomics-informed nursing care. New tools may be needed to assess how nurses perceive and enact genomics-informed practices with companion measures to capture patient experiences with genomics-informed nursing care. For example, the person-centred practice framework formed the basis of two companion validated tools, the person-centred practice inventory (staff) designed to be completed by healthcare providers (Slater et al., 2017) and the person-centred practice inventory (care) to be completed by parients (McCormack et al., 2024). While a considerable undertaking, developing such tools could facilitate the evaluation of genomic nursing education and service delivery improvements and may inform the development of education that is most needed to guide genomics-informed practice in the context of nurses' competing educational priorities (Dewell et al., 2021).

Limitations

This study does have certain limitations. Limitations of the individual qualitative and quantitative study streams were previously reported (Puddester et al., 2025; Puddester et al., in press); however, there are also limitations specific to the mixed-methods integration phase. The differences between data collection instruments made it impossible to compare or link all findings across both datasets. While one of the validated quantitative measurements used to capture nurses' perspectives were based on a theory cited in implementation science frameworks, our exclusive use of these tools meant that the same level of rich qualitative exploration of patients' perceptions of genomics-informed care was not possible with the nurse participants (Damschroder et al., 2022; Rogers, 2003; Tonkin et al., 2020b). Qualitative data from nurses would likely have elucidated deeper insights about the factors impacting their adoption of genomics into their practice.

A strength of this study was the use of participatory research methods of scholarship through iKT and POR with patients and nurses (CIHR, 2014) to promote study relevance and engagement. However, a limitation of this approach was that the nurse key informants were not engaged in the earliest stages of the study, as was the case with the patient partners. It is best practice to engage all study partners in the earliest stages, which is a consideration for future research (CIHR, 2014). During the iKT engagement sessions, the nurses identified helpful ideas which could have been beneficial during the earlier stages of the study. This limitation can be addressed in future experience-based co-design research that involves patients and providers in collaborative solutions for genomics-informed care in research and quality improvement initiatives.

Further, while we drew on the ASIGN maturity matrix as part of this study scaffold, other ASIGN key enablers of genomics-informed implementation were not assessed in this study. For example, ASIGN also highlights interdisciplinary collaboration as an essential key enabler (Tonkin et al., 2020a). Cancer care is incredibly interdisciplinary and several other interest holders impact the successful integration of genomics into cancer care (oncologists, pharmacists, genetic specialists, healthcare administrators, policymakers etc). These relevant interest holder perspectives were not represented in our findings. Future research can address this limitation by using implementation frameworks to study how different professional roles in genomics-informed cancer care are introduced and coordinated. It is also suggested that future research involve participatory research methods (e.g. deliberative dialogues and experience-based co-design) to involve the wide range of people affected by or involved in cancer genomics care.

Conclusion

By integrating both patient and provider perspectives through mixed methods, this study revealed a need for innovative approaches to support the integration of genomics into cancer nursing pathways in ways that align with patient needs and expectations in NL’s evolving genomic care landscape. Their shared recognition of nursing’s potential to enhance genomics-informed care, through optimizing existing roles and new dedicated services points to a promising direction for future service delivery. At the same time, findings also highlight a need to revisit what ‘essential genomic nursing competencies’ may mean in different and evolving contexts and to ensure that priority workforce genomic development efforts reflect nurses’ disciplinary commitment of responsiveness to the complex needs of individuals seeking genomics-informed cancer services. Future participatory research building on these findings should draw on robust implementation frameworks and engage multiple interest holders (e.g. multidisciplinary providers and persons with lived experience) in the co-development of new care models and strategies to improve health outcomes for patients and families affected by hereditary cancer risk. This study also offered methodological innovation through use of ID-informed mixed-method research.

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CHAPTER 5 Conclusion

Introduction

In this concluding chapter, the dissertation research problem, questions, and methodology are briefly revisited. Key findings from the qualitative, quantitative, and integration results presented in Chapters 2, 3, and 4 are summarized, and study limitations are addressed. Implications of these results for practice, policy, administration, education, and future research are also discussed. This chapter ends with a concluding reflection on the dissertation research process.

Summary of Research Problem and Research Questions

Chapter 1 began with an overview of the literature, revealing that genomics is a fast-moving train, traversing across the specialty of oncology with a growing number of evidence-based cancer control applications (Aronson et al., 2025; NCCN, 2024a; 2024b; Rendon et al., 2023). While genomic applications offer considerable opportunities for improving cancer outcomes, persistent barriers at the patient, provider, and system levels hinder their adoption into routine cancer care (Ahsan et al., 2024; Mittendorf et al., 2021; Linfield et al., 2022; Snow et al., 2024; Tindale et al., 2022). While nurses assume essential roles in the delivery of cancer care, evidence suggests that the adoption of genomics in day-to-day oncology nursing practice remains limited (Chiu et al., 2024; Cuthill et al., 2025; Hébert et al., 2022; Hines-Dowell et al., 2024; Seven et al., 2017; Wright et al., 2020; Zhao et al., 2022). Despite concerted global disciplinary efforts to address this issue, adopting genomics across nursing practice is a persistent global challenge (Calzone et al., 2018; Tonkin et al., 2025). Opinion among leading expert global genomics nursing scholars is that patient involvement and perspectives are vital, as is nursing workforce engagement, in future efforts to advance the integration of genomics across nursing (Thomas et al., 2023; Tonkin et al., 2020a; 2020b).

Guided by this position, this study sought to generate insights from the combined perspectives of cancer care nurses and patients with Cancer Predisposition Syndromes (CPS), within a shared local context, to yield *patterns and possibilities* to inform future nursing contributions to genomically-informed cancer care. At the time of the study, this knowledge was particularly relevant in NL as the arrival of a novel provincial clinical translational genomics cancer service was imminent, and NL is a known genetic isolate population which experiences persistently high cancer incidence and mortality rates (Canadian Cancer Statistics Advisory Committee et al., 2023; Green et al., 2007; Rahman et al., 2004).

Overarching Mixed Methods (MM) Research Question

What insights can be learned from the integration of nurses' and patients' perceptions of genomics-informed cancer nursing care to advance its clinical implementation?

Research Sub-questions

1. What are the genomics-informed healthcare experiences described by individuals in NL living with CPS?

2. What are the experiences, perceptions, and receptivity of people living with CPS in NL related to cancer genomic nursing care?

3. What are the a) knowledge levels, b) confidence, c) attitudes, and d) practices related to genomics of oncology nurses in NL?

4. Are the social systems/organizational cultures conducive to NL oncology nurses' adoption of genomics?

5. What demographic and contextual variables are associated with NL oncology nurses' genomic knowledge scores?

Summary of Methodology

Informed by a worldview of Dialectical Pluralism (DP), MM methodology with a convergent design was used to address the dissertation research questions (Creswell & Plano Clark, 2018; Johnson, 2017). See Table 5.1 for a summary of study samples, design, and method components.

Table 5.1

Summary of MM Study Design and Methods

Study Stream/ Design/Methodology	**Qualitative** (Interpretive Description)	**Quantitative** (Descriptive, Cross-sectional)	**Mixed-Method Integration** (Convergent, Parallel, Patient-Oriented Approach)
Interest Holder Groups (Participants)	**Patients** (n=37), > 18 years old, self-reported personal history of any molecularly confirmed CPS P/LP variant (e.g., *BRCA1/2*, *ATM*, *MSH6*, *CHEK2*) with or without personal history of cancer, within the province of NL	**Nurses** (n=50) of any designation working in inpatient or outpatient settings where cancer care is provided within the province of NL	**Nurses** [quantitative] (n=50) + **Patients** [qualitative] (n=37)
Data Collection and Timeline	Semi-structured interviews conducted in person, via online meeting platform, or telephone between April and August 2023	Electronic surveys (demographic questions, the GNCI©, and select items from the GGNPS instruments) were self-completed between September 2023 and February 2024	Data collected in the qualitative and quantitative streams
Data Analysis	Inductive constant comparative approach to qualitative data analysis, following guidance by Thorne (2016). Findings were reported as themes and sub-themes	Descriptive and inferential statistics	Using the PIP integration technique to merge quantitative findings and qualitative findings with MM inferences generated. Integration analysis took place between August 2024 and February 2025

Following guidance from Thorne (2016), I developed a multi-component 'theoretical scaffold' as a basic structure to guide design and analysis decisions throughout the MM study. Elements of the theoretical scaffold included background literature, my nursing disciplinary epistemology, and our professional and personal experiences as a research team related to the topic (Johnson, 2017; Thorne et al., 2016). The theoretical scaffold also encompassed principles underpinning POR and theories, frameworks, and ideas common in the health and genomics implementation literature (CIHR, 2014; Rogers, 2003; Tonkin et al., 2020a' 2020b). These elements were woven together as a basic frame to cohesively connect the various MM components along the study trajectory.

Summary of Key Findings

Chapter 2 (Manuscript 1)

In Chapter 2, results of a qualitative interpretive description (ID) of the perceptions of 37 individuals in NL with CPS were reported; two primary themes, *when genomic knowledge is power*, and *perceived acceptability of nursing roles in CPS care,* were generated to address research sub-questions 1 and 2.

Participants frequently perceived that the knowledge of their CPS gained through genetic testing was a form of personal and family empowerment (e.g., health behavior change, evidence-based risk management, and reproductive planning). They often viewed this knowledge as a matter of life and death. Most reported high satisfaction with the interactions they had with genetic counsellors in the PMGP. However, these encounters were finite (1-2 interactions), and many participants reported that knowledgeable genomics-informed care was inaccessible in the periods before and after their limited interactions with PMGP. This fragmentation engendered systemic conditions where evidence-based care was missed, with respect to both genetic risk identification and adherence to risk management. Many felt ‘left adrift’ following the disclosure of their test results to navigate its lifelong implications; for some, this was highly distressing. Participants reported highly individualized information and supportive care needs related to risk management and decision-making in CPS that were often unmet in the current system.

Most participants identified that nurses were not involved in any aspects of their healthcare related to their CPS or cancer genomic testing. However, they identified specific areas where nurses' further involvement could benefit their hereditary cancer care, such as advocacy, assistance with screening and risk-management, and psychosocial support through the cancer genomic testing journey and beyond. Many participants endorsed the prospect of having a 'go to' nurse focused on CPS follow-up care with a high level of expertise in genomics and with whom a therapeutic relationship could be built in their long-term management of cancer risk.

Chapter 3 (Manuscript 2)

In Chapter 3, results from the quantitative stream, a cross-sectional survey of 50 nurses working in oncology in NL, were reported. These results addressed research sub-questions 3, 4, and 5.

Nurses in the study had low overall genomic knowledge scores. They self-reported low confidence in performing several genomics-related tasks, including accessing up-to-date, accurate genomic information. Nonetheless, most nurses surveyed reported positive attitudes towards adopting genomics, such as improved patient health teaching and identifying individuals in high-risk screening programs. They indicated receptivity toward learning more about genomics. Nurses’ commonly reported barriers to adopting genomics, included concerns related to patient anxiety about risk, the need for additional education, and the time commitment involved. Forty-six percent of nurses indicated they had been approached by patients with genomics-related questions in the past three months. Few nurses identified that they were consistently collecting a complete three-generation family health history (FHH), although nurses in the outpatient setting reported doing so more frequently than those in the inpatient setting.

There was no evidence in the quantitative results to suggest that NL oncology nurses perceived strong organizational or senior nursing leadership support for adopting genomics into their practice. Oncology nurses in inpatient settings had higher knowledge scores than those working in the outpatient setting, which was an unexpected finding. Nurses who identified the need for additional education as a barrier to adopting genomics in practice had significantly lower knowledge scores. No other significant demographic or occupational predictors of nurses' genomic knowledge were identified in the regression analysis.

Chapter 4 (Manuscript 3)

In Chapter 4, findings of the mixed method integration analysis were presented, which merged patients' and nurses' perspectives (qualitative) + (quantitative) on the phenomenon using the Pillar Integration Process (PIP) (Johnson et al., 2019); these results addressed the overarching MM research question.

It was clear from the integrated data that patients' genomics-related questions are becoming increasingly present in routine cancer care settings. Many participants who underwent testing in recent years did so when they or their family members underwent treatment-focused genetic testing (TFGT) offered as routine care in their cancer treatment journeys. Almost half of nurses reported receiving questions from patients about genomics, and a few patient participants reported that they contacted cancer nurses, such as nurse navigators, with questions related to accessing genomic testing and recommended screening. While there is evidence that TFGT and related genomics questions are entering the routine cancer nursing care setting, most patients with a CPS and oncology nurses share uncertainty about where to seek accurate information related to genomics in the current healthcare system.

While most patients interviewed had not encountered cancer nurses in their CPS care, and most oncology nurses were not collecting complete FHH information, both groups saw the potential benefits of further integrating genomics into existing cancer nursing roles to optimize patient outcomes and experience. Some patients with a personal history of cancer experienced the benefits of existing cancer nursing roles during their cancer care journey. They saw opportunities for how this care received could be applied to the context of CPS care. Nurses and patients reported the potential benefits of a dedicated high-risk service for people with a confirmed CPS; oncology nurses saw that in their current roles, they could help identify people who could be referred to a high-risk program, while patients reported receptivity to a specialist nurse serving in this dedicated role in a high-risk CPS follow-up program in a cancer care setting. Notably, a few patients with CPS in our study had historically received hereditary cancer genetic counselling services delivered by RNs who worked in smaller satellite centres throughout the province. While these specialized nurse genetic counselling positions and satellite genetic counselling sites no longer exist in NL, a few patients reported high levels of satisfaction with their interactions with nurses in these roles.

Study Limitations

While the overall study had several methodological strengths as discussed in the previous chapter (e.g., the DP and theoretical scaffold-informed methodology, use of POR), this research also had several limitations to acknowledge. Detailed descriptions of the limitations specific to each study component are reported in each manuscript chapter, but a summary of salient study limitations is noted below, beginning with the limitations to the overall MM study and integration, and then limitations specific to each study stream.

Limitations of the Overall MM Study and Integration

The data collection instruments used in each stream, while conceptually related, were designed for use by two different populations (patients and nurses) for slightly different purposes. As a result, it was not always possible to make linkages between all data points from the two study streams. Data in both qualitative and quantitative streams relied on self-report and were collected at a single point in time, meaning causality cannot be inferred. There was limited ethnic diversity of participants in both study streams, sampled from a single provincial setting with a population considered a genetic isolate. Therefore, it cannot be concluded that findings are transferable or generalizable to other jurisdictions. The use of engaged patient and public engagement methods (iKT, POR) was a strength of this MM study. However, CIHR (2014) SPOR best practice for engagement guidelines indicated that engagement should occur in the earliest stages of the study. I did involve the clinical nurse educator at the CCP in consultations about effective and mutually beneficial ways to engage oncology nurses before the study engagement sessions. However, I did not engage these 'nurse partners' in the earliest phases of the study, as I did with the two patient partners, which is a limitation of this approach.

Limitations of the Individual Study Streams

Qualitative. Although outside of our original inclusion criteria, we elected to retain the perspectives of patient participants whose genetic test results were revealed over the course of the interviews to be classified as a VUS (variant of uncertain significance). This decision was made on the basis that they had generously provided their time to participate in the study and represented a sub-population with unique, relevant information needs in the context of the increased use of TFGT in routine care. However, other patient perspectives pertinent to the phenomenon of genomics-informed cancer nursing and healthcare were not represented, such as individuals with cancer who underwent TFGT and received negative results, or those who declined TFGT.

While patient participants were asked in the interviews about other healthcare providers whom they felt should be involved in their CPS care, questions in the interview guide focused primarily on their view of genomic nursing roles and care, which created a potential response bias. Moreover, I conducted all qualitative patient interviews and did not conceal from the participants during the interview that I was an RN. While I did not have previous clinical relationships with the participants, respondents may have felt compelled to respond favorably to the prospect of CPS nursing care, given that they were aware they were speaking with an RN. As previously described, I engaged in reflexivity practices during data analysis to account for these potential response and social desirability biases surrounding the nursing subject matter.

Quantitative. While the entire validated, reliable GNCI© instrument was used to measure nurses' knowledge in this study, the use of only select items from the GGNPS (to measure confidence, attitudes/receptivity, and social system influences) limited the possibilities for inclusion of this study in future data aggregation with other global studies that used the entire GGNPS instrument. Further, while the GGNPS and GNCI© are suited for administration to nurses working across all areas, they were not explicitly designed for the oncology nursing context. There may be nuances and specificities of the genomic competencies required/enacted in cancer care settings that are not captured by the validated measurements currently available. Despite these limitations, this MM study offers several insights about genomics-informed nursing care in local and global contexts.

Study Contributions and Implications

A novel contribution of this study was in merging the self-reported data of patients and nurses (within a shared local context), related to the phenomenon of genomics-informed nursing. Nursing practice is inherently intersubjective and relational; nurses partner with individuals to improve their health status (ICN, 2021). While there is much research assessing nurses' genomic competency levels, with little evidence of their adoption of genomics in practice, some have questioned whether genomic competencies are essential for all nurses (Newcomb et al., 2019). Thus, this study offers a contextually relevant answer to 'so what?' about oncology nurses’ limited genomic competency to drive local change with respect to this issue.

This study also contributed to the global literature related to genomics in oncology nursing, as there are many discussion papers on the topic; however, comparatively fewer empirical studies specific to cancer care settings have focused on assessing oncology nurses' genomic competency (Cuthill et al., 2025; Hebert et al., 2022; Hines-Dowell et al., 2024; Seven et al., 2018). This study also added to the limited global literature where patients were explicitly engaged to inform genomic nursing role development and enactment (Chiu et al., 2024; Diez de los Riez de la Serna et al., 2024). This study was the first assessment of nurses' genomic competency specific to the NL cancer care setting. The use of validated measurements provided a baseline assessment of NL oncology nurses' genomic competency that can be used as a benchmark to examine the impact of future interventional studies targeting nurses' genomics knowledge and practice in the NL setting. The use of patient and public engagement in this study generated momentum and awareness of the topic, particularly among the oncology nurses, which can serve as a foundation to build upon through future local participatory research in the CCP.

This study also highlighted that patients and nurses see the benefits of a dedicated high-risk follow-up program for people with CPS.

Methodological Contribution

There are potential contributions to the MM and ID methodology literature from the methodological approach employed in this dissertation. Others have highlighted the synergy between MM research and ID, including discussion of how rigour is addressed in ID qualitative components of a MM study (Dolan et al., 2023; Thorne, 2025). However, there is a lack of published MM studies where design features of ID have been used to inform the overall MM study (i.e., the theoretical scaffold), and similarly to guide rigour in the integration analysis. This dissertation also highlighted the synergy between the DP worldview, nursing disciplinary epistemology, and MM-ID. This may be relevant to other doctoral students in nursing when conceptualizing and selecting design logic to guide their MM-ID research.

There is also a potential contribution from the approach used to report this MM doctoral manuscript-based dissertation. A common critique in the MM methodology literature is when researchers provide little to no details of the data integration process in MM studies (O' Caithain et al., 2008; Fetters et al., 2013). Interestingly, in my informal scan of the literature, I did not retrieve any publications where qualitative, quantitative, and integration analysis findings of a convergent MM study had been presented as three separate manuscripts, which was the approach taken for this manuscript-based dissertation. Critiques have arisen about the increased popularity of manuscript-based dissertations, in that they may constrain the depth and direction of dissertation research to fit it into formats suitable for publication (Thorne, 2024). Thus, the approach to manuscripts used in this dissertation may be of interest to researchers conducting convergent MM studies while conceptualizing how they will approach reporting their manuscript-based dissertation findings in ways that may allow an in-depth description of all three separate points of analysis.

Implications and Recommendations for Practice, Education, Policy, Administration, and Future Research

The disciplinary implications of this study are discussed below in eight broad recommendation categories, each with implications across several domains of nursing. Recommendations are also summarized according to each of the five domains (practice, education, policy, administration, future research) in Table 5.2

1. *Determine the Most Effective Way Forward for Establishing Genomic Nursing Competency Statements in Canada*

It was clear from study findings that NL oncology nurses have low levels of foundational knowledge of genomics with minimal exposure to pre- and post-licensure genomics education, which is consistent with global findings (Hebert et al., 2022; Hines-Dowell et al., 2024; Thomas et al., 2023; Wright et al., 2020). Canada does not have stand-alone genomic competency statements to guide genomics-informed nursing practice and education (Chiu, Limoges, Pike et al., 2024). However, it is worth reiterating that genomic competency deficits persist even in countries where such competency statements have existed for over two decades (Calzone et al., 2024; Carpenter-Clawson et al., 2023). There is divergence of opinion about whether foundational genomic competencies are necessary for all nurses (Calzone et al., 2024; Newcomb et al., 2019). Concerns about curriculum overload in nursing education and questions related to the relevance of genomic competencies for nurses in practice have been identified as barriers to further integrating these competencies into nursing curricula (Dewell et al., 2021).

Considering these challenges, our findings also support the conclusions of Chiu et al. (2024) that leaders of Canadian nursing organizations should engage both point-of-care nurses and experts with lived experience in genomics-informed healthcare to thoughtfully determine a competency development approach that reflects and responds to nurses' real-world practice contexts and emerging healthcare trends. There are numerous examples of options for approaching genomic competency statement development in the global literature, e.g. 'add on'/integrated' genomics statements in existing competencies, 'stand-alone' genomic competencies, or area-specific competencies, such as those in Europe specifically for cancer care nurses (ANA, 2023; Chiu, Gretchev et al., 2024; Diez de los Rios de la Serna et al., 2022; European Oncology Nursing Society, 2023). Development of interdisciplinary cancer genomics competencies may also represent a practical approach, as was reported in Italy (Hoxhaj et al., 2022). These global examples of general, practice area-specific, and interdisciplinary genomic competencies can be adapted and fit to the Canadian nursing context. This is a potential area for a future Delphi study, involving Canadian nurses, patients, and other relevant interest holder groups in consensus-based genomic nursing competency development.

2. ***At the Point of Care, Embrace Patients' Lived Experience Expert Narratives Related to Genomics***

It was revealed in this research that oncology nurses are receiving patients' questions about genomics, and some patients are already seeking nurses' assistance when navigating aspects of their genomically-informed care. Understandably, oncology nurses may hesitate to engage patients in discussions about genomics, when it is a complex form of knowledge with which they have had little formal education on the topic. However, nursing knowledge remains in constant evolution in tandem with evolving healthcare contexts; it is intersubjective, tacit, and pattern-based, and oncology nurses learn every day through their interactions with patients and their narratives (CANO/ACIO, 2001). Our findings and the existing literature suggest that people affected by CPS often have had to be their own advocates and experts for many years when navigating the healthcare system. Many people with CPS interviewed in this research were highly knowledgeable about their condition and keen to educate others about it. When nurses encounter experts with lived experience of genomics testing in their practice, they can engage in patients' narratives with curiosity and authenticity, to learn about genomics and CPS to inform future patient interactions.

If nurses encounter a patient with a CPS or a VUS test result that they may not be familiar with, they can use this as a brief self-directed teachable moment to learn more about it. In saying this, it cannot be overlooked that oncology nurses working in fast-paced and often understaffed clinical areas have limited time. However, there are several evidence-based, easily digestible point-of-care resources readily available from resource repositories, such as the US Oncology Nursing Society Genomics Taxonomy and the Genetics Education Canada Knowledge Organization (GECKO) to support nurses' 'five-minute teachable moments.' Nurses can also bring forward to their clinical educators and managers the types of questions they encounter from patients related to genomics to inform them of the necessary education and tools needed to support patients, including those related to some of the ethical and legal considerations that may arise with genomics. A future area for qualitative and/or participatory research is to explore the nature of the questions that oncology nurses commonly receive from patients about genomics.

Further, even if nurses feel they do not have answers to complex technical questions about genomics, many of the supportive psychosocial needs reported by people with a CPS in this study mirror those commonly experienced by individuals with a cancer diagnosis, and nurses are accustomed to helping patients navigate their complex supportive care needs (Fitch, 2008). Although the patient interviews in our study involved no provision of clinical care or health information, one woman in this study with Lynch Syndrome remarked that even being able to talk to someone about her journey with Lynch Syndrome through the interview was helpful to her. Thus, oncology nurses at the point of care should not underestimate the power they hold in listening to patients' genomics healthcare and lived experiences, as both they and patients determine how to navigate uncertainty and move forward in the genomics era of oncology together.

3. ***Leverage iKT, and POR, Pivot to Experience-Based Co-Design to Engage Nurses and Patients, in New Solutions to Genomics-Informed Care***

The awareness building and engagement achieved when lived experience expert, Vanessa, and I interacted with nurses during the study iKT/POR engagement sessions can be built upon in future research and quality improvement efforts to improve genomics-informed care for the NL population. An option for going a step further in building on these participatory approaches in future research is experience-based co-design, a form of participatory action research and or quality improvement that brings together patients and healthcare providers to collaboratively develop solutions to improve healthcare (Donetto et al., 2015). As an example, in the UK, experience-based co-design was used to create health interventions for people with CPS, such as the development of patient decision-aids (Kohut et al., 2024). Our study suggested that similar opportunities could be leveraged in NL, as several patient participants in the qualitative stream expressed interest in joint peer and provider support interventions for individuals with CPS.

To name a few future possibilities for experience-based co-designs, first, future complex educational interventions (involving regular, ongoing education sessions and a practice change component) to address the genomic nursing knowledge deficits identified in this study could be co-developed and delivered to oncology nurses in partnership with lived experience experts (O'Shea et al., 2021). An example of a practice change could involve the co-designing of an intervention to enhance nurses' current FHH practices that leverage electronic health records and clinician and or patient decision-support tools. This has been done in other jurisdictions and was an idea proposed by a local oncology nurse during the engagement sessions associated with this study. Further, it is known that there is a high burden of modifiable and non-modifiable cancer risk factors in the NL population (Shanahan et al., 2022). In Europe, consensus-based education guidelines were generated specifically surrounding the education needed by nurses to promote health and wellness behaviors in people with increased hereditary risk of cancer (Diez de Los Rios de la Serna et al., 2022). Drawing on some of these guidelines and patients' preferences and experiences, cancer care nurses and experts with lived experience of CPS could participate in an experience-based co-designed wellness intervention for people with increased risk of cancer in NL.

4. ***Expand on this Study with Multidisciplinary Approaches to Genomics Education, Research, and Practice Implementation***

As indicated, longitudinal, complex genomics educational interventions are likely required to advance practice implementation, as simple genomic educational interventions for nurses have not yielded sustained practice change (Thomas et al., 2023). While interdisciplinary perspectives were not captured in this study, as genomics is increasingly integrated in oncology, there will potentially be steep learning curves for other oncology non-genetics specialist providers, not just nurses. Taking an interdisciplinary approach to future complex educational interventions would reflect the highly multidisciplinary nature of oncology, where nursing does not exist in silos from other healthcare disciplines. Further, a finding highlighted in our qualitative study and in previous research is that breakdowns in communication between providers and specialties concerning genomic-informed care can be distressing for patients. This may be mitigated through education and policies that support interdisciplinary learning, where each discipline has a clear understanding of the other's roles and responsibilities in genomics-informed care.

Patient participants in our study reported high levels of satisfaction with genetic counselling services; their frustrations, instead, lay with the challenges they experienced before and after genetic counselling; thus, there is a need for improved care continuity. Nurses may also be less hesitant to engage in genomics discussions with patients when there is a lack of pathways in the healthcare system to access genetics expertise, either in referring patients or to seek timely guidance related to patients' genomics-informed questions. As mainstreaming and other new models of integrated genomics care emerge, health system administrators, genetics specialty and non-genetics specialty oncology care providers, including nurses, can work together to co-develop new care pathways and negotiate how roles in genomic-informed care can be delegated and shared among interdisciplinary team members.

5. ***Draw on Implementation and Complexity Science to Inform, and Evaluate Genomic Implementation***

As established from the literature reviewed in Chapter 1, genomics is the epitome of a complex intervention (Calzone et al., 2018). There is a benefit to using guidance from the established complexity science and implementation frameworks to ensure the acceptability, feasibility, scalability and sustainability of genomics implementations (Braithwaite et al., 2018; Damschroder et al., 2022; Yakovchenko et al., 2023). A robust implementation framework-informed approach has been used successfully in other countries, such as Australia, when implementing genomics across entire healthcare systems (Best et al., 2025). We did draw on DOI in this study, which is included in robust multi-theory informed implementation science frameworks, as well as logic underpinning the implementation science-informed ASIGN tool as theoretical ideas to guide the study (Rogers, 2003; Tonkin et al., 2020a). However, there are opportunities to conduct genomics implementation research with more rigorous application of theory-informed implementation frameworks and taxonomies that consider broader complexities that influence implementation success. It is recommended that all future research and quality improvement initiatives involving genomics implementation and evaluation in the CCP are underpinned by robust implementation frameworks and taxonomies. Examples of such well-established tools include the Consolidation Framework for Implementation Research, the Expert Recommendations for Implementing Change taxonomy, a compilation of implementation strategies developed through expert consensus, and Proctor's Implementation Outcomes Framework, among others (Damschoder et al., 2022; Proctor et al., 2011; Yakovchenko et al., 2023).

6. ***Bolster, Develop, Implement, and Evaluate Genomic Nursing Roles Across Different Levels of Oncology Nursing***

Both patients and nurses expressed the benefits of implementing a dedicated high-risk follow-up service, while patients indicated a receptivity toward a nurse serving in a specialized high-risk follow-up role. With the recent announcement of funding for a hereditary cancer follow-up clinic as part of the CTG, our findings highlight acceptability from a patient perspective to support specialized nursing roles in this service. However, future possibilities for optimizing genomics into existing oncology nursing roles were also illuminated in our study. For example, a few patients described how they experienced effective genomics-informed care in existing nursing roles, such as assistance with system navigation. Other patients and nurses saw potential for enhancing current nursing practices, such as improved FHH-taking practices. Further, it is recognized that, depending on the available resources in a healthcare system, implementing a 'stand-alone' genomics nursing role may not always be feasible.

When health system decision-makers, administrators, and nursing leaders consider where genomics integration fits into oncology nursing roles, the CANO/ACIO (n.d.) Cancer Nursing Practice and Knowledge Framework may be helpful. It outlines varied cancer nursing practices and cancer knowledge required, categorized in levels according to what is relevant to *all nurses, many nurses, some nurses,* or *few nurses.* Examples of role descriptions and scenarios are provided for each level in the framework (e.g., a nurse caring for patients in community care who may have cancer, versus a clinical nurse specialist in oncology). While the framework is not specific to genomics, health administrators and nursing leaders may find it helpful to determine where various genomics care responsibilities are most aligned with existing cancer nursing role typologies, and therefore what practice changes and education are needed for genomics implementation, depending on the nurses' level of specialization and the nature/frequency of their interactions with individuals with cancer. As an aside, an opportunity exists for CANO/ACIO to develop a practical resource toolkit that reflects the different educational resources needed for genomics-informed care according to the CANO/ACIO Cancer Nursing Knowledge and Practice Framework.

As new nursing roles and practice changes in genomics-informed cancer are implemented, they should also be evaluated and reported in the published literature with detailed descriptions of contexts, enabling mechanisms, and outcomes. It is recommended that outcomes should include patient-reported outcomes, including those related to empowerment, which was identified by participants in our study as an important outcome of their genetic testing experiences. An area for future research is a realist synthesis to determine what nursing roles work best in what contexts and what mechanisms can enable these outcomes. This could allow decision-makers and health funders to decide what nursing roles and responsibilities could contribute to the best outcomes, depending on the health system context.

7. ***Engage Opinion Leaders and Mobilize Champions to Facilitate Genomics-Informed Oncology Nursing***

Guidance from implementation literature highlights the importance of opinion leaders and champions in implementing evidence-based adoptions (Powell et al., 2015). With other implementation of evidence-based practices in oncology, managers and advanced practice nurses have served as influential champions of nurses' uptake of these evidence-based practices (Teggart et al., 2024). A unique challenge in Canada relates to the change to a master's degree in genetic counselling as the entry-to-practice requirement to provide genetic counselling in the early 2000s. When nurses grandfathered into genetic counselling positions retired and left the positions, in the absence of an alternative credentialling pathway for nurses to acquire genomics expertise, limited clinical nursing champions of genomics remained in Canada (Limoges et al., 2025). Rebuilding a pool of disciplinary expertise in genomics will require administrative and policy support that allows specialist nurses to develop these specialty skills and ensure that time is protected for all nurses to acquire complex genomic competencies. Nurses interested in developing special expertise or a new specialty role should have access to high-quality training, such as the City of Hope Intensive Course in Cancer Genomic Risk Assessment. This can also connect nurses to communities of practice where they can ask questions of nurses and other multidisciplinary providers globally about genomics. Specific to Canada, although in its infancy, the CANO/ACIO specialty interest group in Oncology Genomics can facilitate pan-Canadian connections with other nurses in cancer care who desire discipline-specific knowledge or advice related to genomics.

Further, institutional leaders and managers can also be champions of genomics by facilitating the protected time to engage in education to upskill with this genomic competency, and to contribute to research and quality improvement initiatives involving genomics, such as opportunities for experience-based co-design studies previously highlighted.

8. ***Embrace Innovation through Novel Oncology Nursing Research Practice Partnerships***

An opportunity was revealed from the dissertation research process for symbiotic partnerships to advance the mutual interests of nurses in academia and in clinical practice. My connections with nurses in the CCP were facilitated by my supervisory committee member, who works in the CCP, and my professional networks within CANO/ACIO. These connections were critical in nurse recruitment efforts, and this informal partnership also resulted in mutual benefits. I joined the local CANO/ACIO conference planning committee to assist in efforts when the 2024 CANO/ACIO conference was held in St. John's, NL. This informal partnership continued beyond the study period. For example, I facilitated a request from nurses in the CCP to tour our nursing Faculty Learning Resource Centre when they were exploring options for skills practice equipment for a pilot project in the Chemotherapy home infusion program. These examples fall outside the scope of this study, but they are shared here to emphasize the imperative of mutual benefits in such successful collaborations.

While this partnership, initially established for this study, was quite informal, it illuminated the potential value of doing so on a more formal basis. It is recommended that academic and healthcare partner institutions embrace innovation and support novel partnerships between nurses in clinical practice and academia in ways that advance both parties' interests. For example, lack of faculty knowledge of genomics is identified as a barrier to genomics implementation (Tonkin et al., 2025). As genomic nurse champions or experts are instated at the point of care, through research practice partnerships, they could work with nurse faculty to co-teach or integrate this content into teaching with real-life practice exemplars. As in other jurisdictions, researchers can help facilitate nurse-identified priorities for research and quality improvement initiatives that involve genomics or other issues. It is also recommended that health systems leaders and academic administrators think 'out of the box' about dual practice and research/implementation science roles. For example, not specific to genomics implementation research, but other jurisdictions in Canada have introduced joint clinician scientist and clinical nurse research specialist roles in oncology (Chisholm et al., 2024). In other Atlantic Canadian jurisdictions, embedded nurse implementation science roles have been introduced (Campbell & Cassidy, 2025). Given the established complexity of genomics as a barrier to its adoption, embedded implementation roles may facilitate adoption vis-à-vis the persistent reported challenges.

Table 5.2

Summary of Study Recommendations by Domain

Domain	Recommendations
Practice (nurses at the point of care)	• Listen to and learn from the narratives of individuals with lived experience of CPS to inform future care and pinpoint educational and research priorities • Participate in research to provide experience-based input on future initiatives to integrate genomics into nursing practice • Engage in interdisciplinary communication and collaboration between other oncology providers and genetic specialists when encountering patient questions and scenarios • For nurses with a special interest in genomics, embrace opportunities to champion and lead the integration of genomics in their nursing practice settings
Administration	• Facilitate avenues for nurses in clinical practice interested in genomics to act as champions/ resource persons in their practice areas • Support and enable nurses at the point of care with requisite protected time to engage with genomics educational opportunities • Support collaborative partnerships and joint appointments within health systems and academic institutions to advance genomics-informed care • Develop, implement, and evaluate nursing roles in genomics-informed care
Policy	• Ensure protected time for nurses' continuing professional development to engage with this complex competency • Ensure protected time for nurses that allow nurses to be involved in participatory oncology nursing research • Develop organizational frameworks and policies to embed interdisciplinary education and teamwork in genomics • Adopt system-level policies that encourage sustainable cross-disciplinary collaboration in genomics practice and research
Education	• In academia, leverage clinical expertise as genomic champions are instated at the point of care, enabling nursing faculty to integrate genomics into undergraduate education in ways that correspond with real-world practice • Place patients' lived experience at the forefront of developing genomic education for nurses • Judiciously develop genomic competencies to guide practice and education (with involvement of multiple interest holders) • Offer and participate in competency-based genomics education and interdisciplinary advanced training

	opportunities for oncology nurses and other oncology providers at the point of care
Research	• Conduct a Delphi study for consensus on genomic competency statements • Consider future development of new measurements to evaluate nurses' genomic competencies that reflect genomic nursing as enacted and patients' experiences • Involve patients and nurses in the development of new genomics-informed interventions using participatory methods of scholarship, such as experience-based co-design • Use implementation science to evaluate and inform multidisciplinary genomics-informed care in clinical oncology settings (including evaluation of various nursing roles in this care) • Conduct a realist review of genomic nursing roles and outcomes across different cancer care settings • Engage in academic research partnerships and explore opportunities for novel oncology nursing research and implementation roles

Concluding Reflection on the Dissertation

As nurses, we often proudly wear our badge of distinction as the largest and most trusted healthcare workforce. Our nursing knowledge, skill set, and professional code of ethics foster our sense of discernment and ability to recognize when those we serve require *care.* Care that is not contingent on their biomedical markers, their adherence to treatment, or their health behaviors. There is no shortage of published opinion pieces that draw on these disciplinary discourses to posit that nurses hold great power in the provision of holistic genomics-informed care. However, there is limited evidence that the scale of what has been espoused in the genomics nursing discourse has been enacted in clinical practice. This is despite tireless 20+ year efforts of global nursing scholars, such as the G2NA group, who I acknowledge in full measure as having laid the foundational disciplinary structures required for genomics-informed nursing. In my attempts to decipher and reconcile this observed disjuncture between 'nursing as espoused' and 'nursing as enacted' during this dissertation research process, I found myself asking "*If we actually hold this 'power' as a nursing profession, when are we going to harness that power?"*

As a neophyte in the nursing academy, I appreciate that I am being bold in making such a statement. This concluding reflection is not intended to critique the genomics nursing discourse. I remain grateful for the path paved for me and many others by the legacy scholars in this area. Nor is it my intention from where I sit in academia to assign added responsibilities and blame to oncology nurses at the point of care for the limited adoption of genomics in practice. It is not my place to run roughshod over their practice experiences and realities as they face mounting, competing pressures (Kelly et al., 2024). Also, while some nurses in clinical settings may fare better than others depending on their locus in the system(s), many are afforded minimal say in the transformation of their day-to-day professional practice realities. And for many nurses, the systemic and organizational structures in their workplaces were not designed to accommodate the protected time for continuing professional development that is required to acquire the complex competency of genomics.

Despite these challenges they face, oncology nurses continue to show up each day for individuals and families affected by cancer. I say this from personal experience, having become one of those family members during the completion of this dissertation. Similarly, despite my awareness of these real forces and challenges, I haven't relinquished my conviction that nursing holds tremendous power in genomics healthcare implementation. Thus, this conclusion is intended as an open invitation to my fellow nurses across all nursing domains for further critical reflective dialogue. I opened this conversation with a frank acknowledgement of the disjuncture between nursing espousal and enactment; these tensions are not unique to the issue of genomic implementation but rather deeply embedded in the broader realities of healthcare and nursing work. The rationale for my directness is to avoid creating an impression of hollow rhetoric and recommendations. Instead, to invite meaningful, solution-focused dialogue about how we can advance genomics implementation and perhaps address other pressing, complex challenges within our profession. While I lay no claims to any concrete solutions, as I reflect on what I have learned in conducting this dissertation research, I believe the words of Dillard-Wright and Jenkins (2024) offer a helpful clue on the way forward for nursing:

"*Nursing can-under the right circumstances-be a force for liberation, but it requires us to see patterns and possibilities, both in ourselves and the institutions we participate in*" (p.7)

As context, this quote comes from a discursive essay where Dillard-Wright and Jenkins proposed the notion of mutual aid as the way forward for the future of nursing; the idea that people who collaborate to advance each other's interests experience an edge in resilience and preservation over those who do not. Linking this idea to my experiences completing this dissertation, it was consistently through the partnered work with Vanessa and Mike, the experts with lived experience on this project over the past four years where I found helpful input and ideas, a sense of community, and a re-ignited sense of shared purpose that this work was worth doing in the face of challenges.

While writing this conclusion chapter, I received a phone call from Vanessa. In this conversation, she mentioned a person she met in passing who had multiple risk criteria for CPS. Still, care providers brushed off this individual when they broached the subject and it was Vanessa who ultimately empowered this individual with the needed information. This story serves two points. First, it shows that in her own right (and long before our orbits ever intersected), Vanessa advocates for others with or at risk of CPS. Second, it illustrated that although this province has been a paragon for genetic cancer research discoveries for decades, it is not any easier for someone to navigate the implications of cancer genetics to their health in the current healthcare system. In the same conversation, Vanessa and I were brainstorming ideas for all the shared work that she, Mike, Dr. Etchegary, I and the others who will join us, have left to do as part of collective efforts to improve hereditary cancer care in this province. As I reconcile within myself whatever tensions may exist in nursing, as a nurse, I find meaning and resilience in the moments of standing with and for those we serve.

Thus, I offer these dissertation findings as a form of advocacy and as insights into the '*patterns and possibilities*' for how the local nursing discipline could work with and for those they serve to contribute to genomics-informed care in NL. While the title of the qualitative manuscript was rather long and unwieldy, it highlighted an essential truth gleaned from this research: it was not that patients said that a nurse absolutely had to be the one to provide their CPS care. Rather, nurses were among those they considered to fit the bill of "anybody who could clarify or *humanize* their experiences". This can be interpreted in two ways: it doesn't *have* to be nurses who step up to the plate to advocate for and with patients in the genomics era of health. Yet, on the other hand, *it can be nurses*. As technology continues to transform our world, healthcare, as we know it, will surely evolve drastically. Through these evolving contexts, perhaps in leveraging mutual aid and tapping into our power of advocacy- a force that cannot be automated or replaced, we will also protect and preserve our deeper sense of purpose as nurses.

I look forward to working in partnership with others who share this conviction.

Figure 5.1

Concluding Image for the Dissertation from KT Project

(R-L) Vanessa Francis, lived experience expert, and I on this research project. Image by Alex Spracklin Photography.

"Aiming for a good life, with and for others, in just institutions"

- Paul Ricoeur, Little Ethics' (as cited in Ekman, 2022, p.1).

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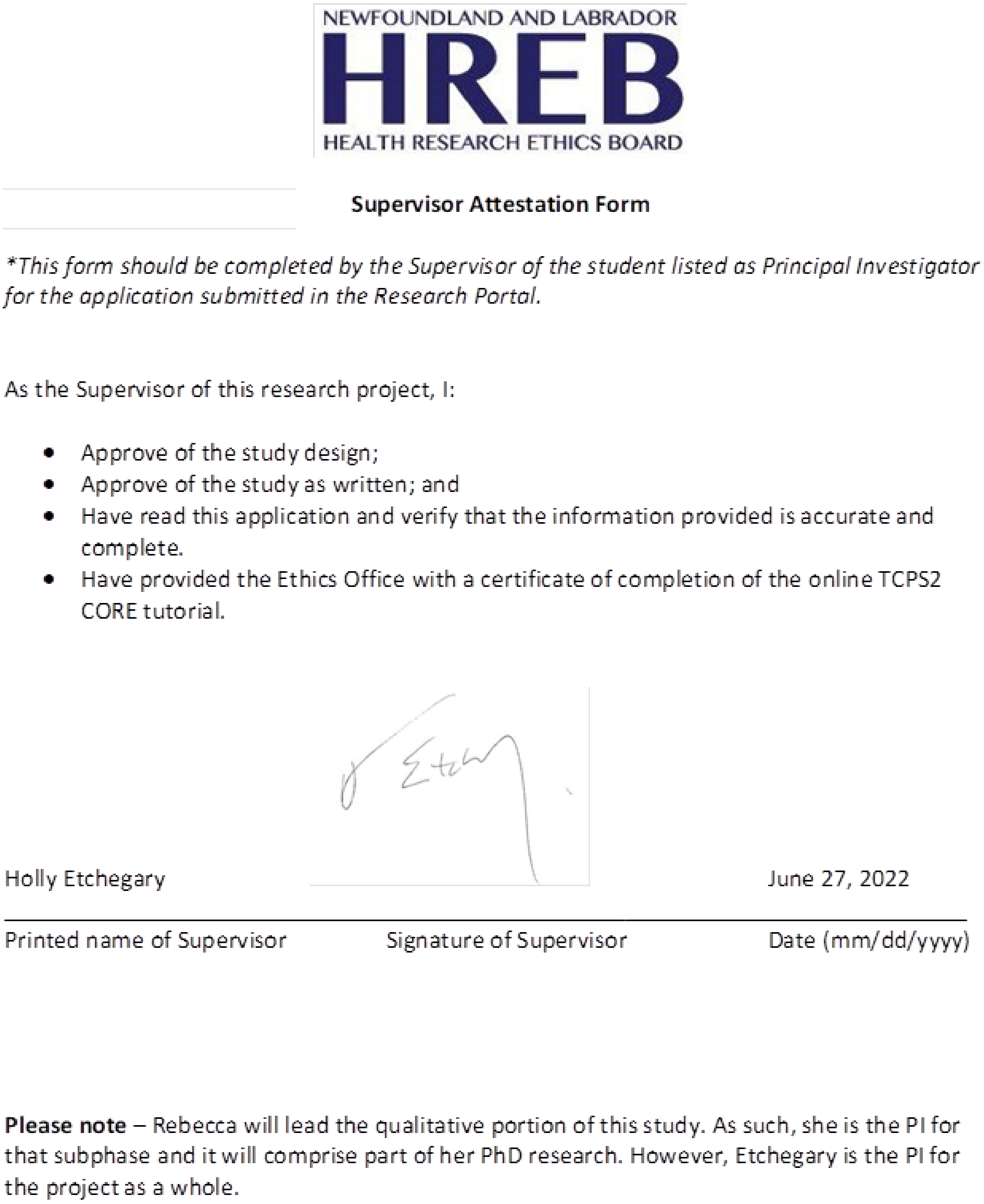
Appendix A: Supervisor Attestation Form (Qualitative)



NEWFOUNDLAND AND LABRADOR
HREB
HEALTH RESEARCH ETHICS BOARD

Supervisor Attestation Form

**This form should be completed by the Supervisor of the student listed as Principal Investigator for the application submitted in the Research Portal.*

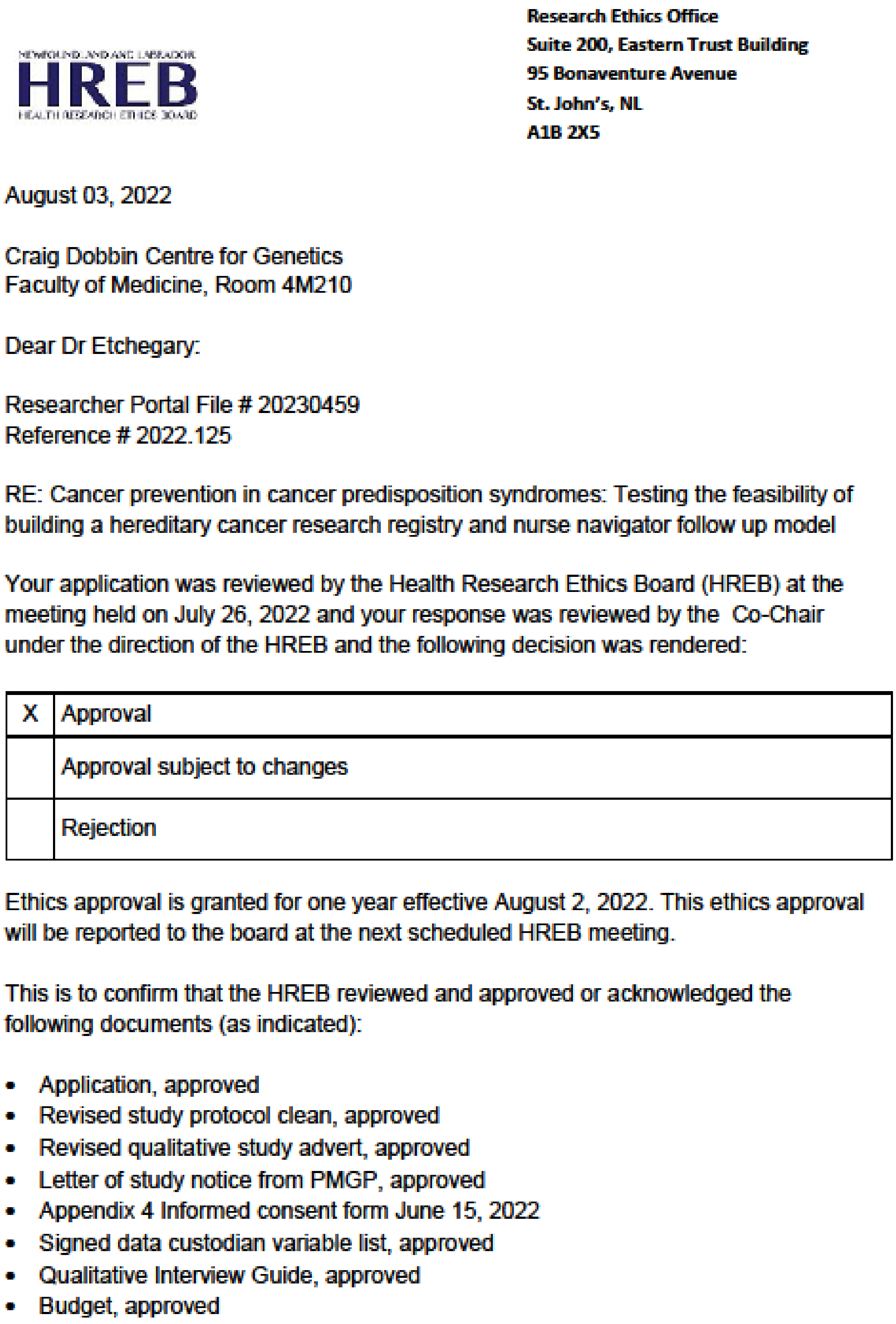
As the Supervisor of this research project, I:

- Approve of the study design;
- Approve of the study as written; and
- Have read this application and verify that the information provided is accurate and complete.
- Have provided the Ethics Office with a certificate of completion of the online TCPS2 CORE tutorial.

Holly Etchegary		June 27, 2022
Printed name of Supervisor	Signature of Supervisor	Date (mm/dd/yyyy)

Please note – Rebecca will lead the qualitative portion of this study. As such, she is the PI for that subphase and it will comprise part of her PhD research. However, Etchegary is the PI for the project as a whole.

Appendix B: Ethics Approval (Qualitative Stream)



NEWFOUNDLAND AND LABRADOR
HREB
HEALTH RESEARCH ETHICS BOARD

Research Ethics Office
Suite 200, Eastern Trust Building
95 Bonaventure Avenue
St. John's, NL
A1B 2X5

August 03, 2022

Craig Dobbin Centre for Genetics
Faculty of Medicine, Room 4M210

Dear Dr Etchegary:

Researcher Portal File # 20230459
Reference # 2022.125

RE: Cancer prevention in cancer predisposition syndromes: Testing the feasibility of building a hereditary cancer research registry and nurse navigator follow up model

Your application was reviewed by the Health Research Ethics Board (HREB) at the meeting held on July 26, 2022 and your response was reviewed by the Co-Chair under the direction of the HREB and the following decision was rendered:

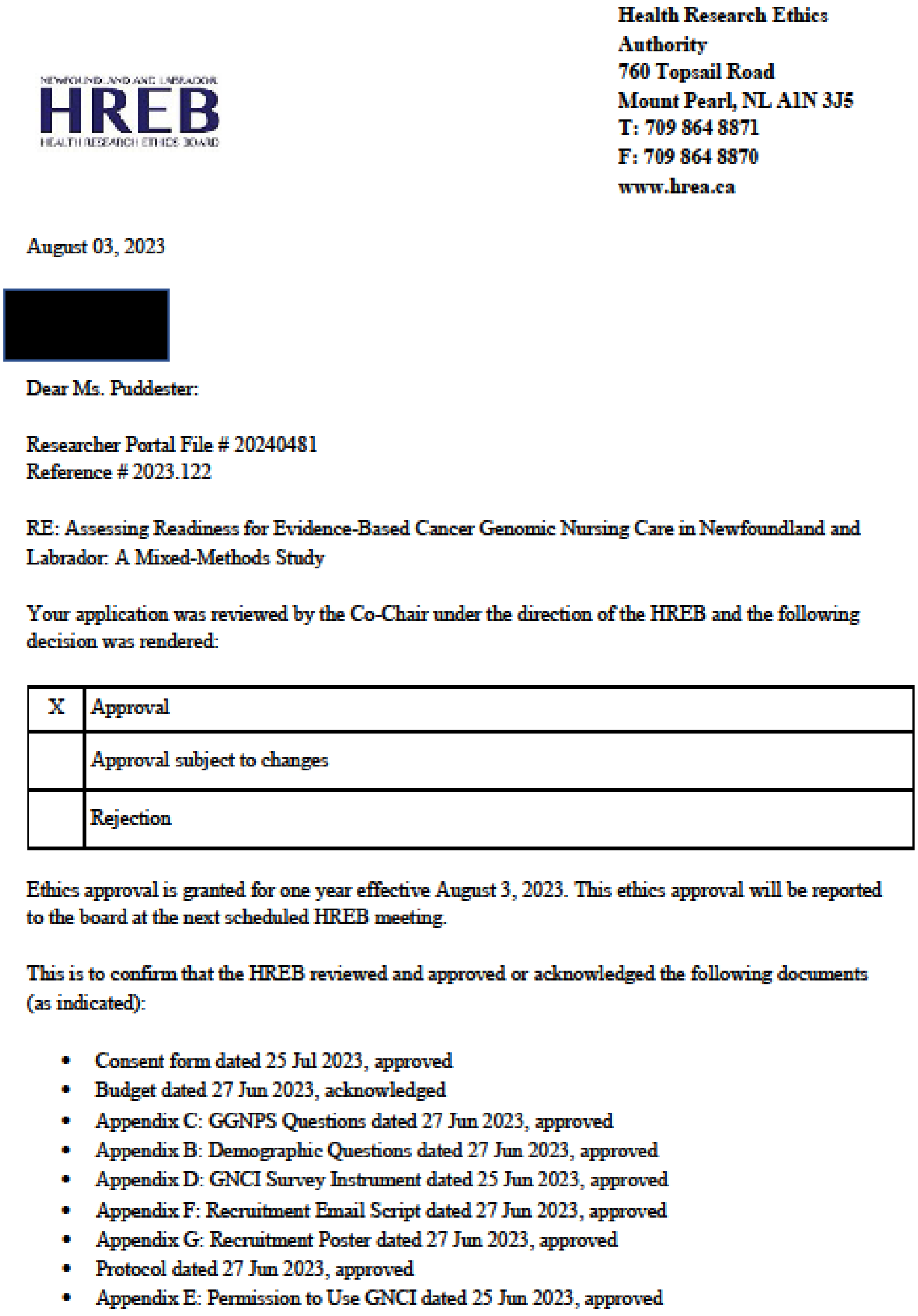
X	Approval
	Approval subject to changes
	Rejection

Ethics approval is granted for one year effective August 2, 2022. This ethics approval will be reported to the board at the next scheduled HREB meeting.

This is to confirm that the HREB reviewed and approved or acknowledged the following documents (as indicated):

- Application, approved
- Revised study protocol clean, approved
- Revised qualitative study advert, approved
- Letter of study notice from PMGP, approved
- Appendix 4 Informed consent form June 15, 2022
- Signed data custodian variable list, approved
- Qualitative Interview Guide, approved
- Budget, approved

Appendix C: Ethics Approval (Quantitative Stream)



NEWFOUNDLAND AND LABRADOR
HREB
HEALTH RESEARCH ETHICS BOARD

Health Research Ethics Authority
760 Topsail Road
Mount Pearl, NL A1N 3J5
T: 709 864 8871
F: 709 864 8870
www.hrea.ca

August 03, 2023

[redacted]

Dear Ms. Puddester:

Researcher Portal File # 20240481
Reference # 2023.122

RE: Assessing Readiness for Evidence-Based Cancer Genomic Nursing Care in Newfoundland and Labrador: A Mixed-Methods Study

Your application was reviewed by the Co-Chair under the direction of the HREB and the following decision was rendered:

X	Approval
	Approval subject to changes
	Rejection

Ethics approval is granted for one year effective August 3, 2023. This ethics approval will be reported to the board at the next scheduled HREB meeting.

This is to confirm that the HREB reviewed and approved or acknowledged the following documents (as indicated):

- Consent form dated 25 Jul 2023, approved
- Budget dated 27 Jun 2023, acknowledged
- Appendix C: GGNPS Questions dated 27 Jun 2023, approved
- Appendix B: Demographic Questions dated 27 Jun 2023, approved
- Appendix D: GNCI Survey Instrument dated 25 Jun 2023, approved
- Appendix F: Recruitment Email Script dated 27 Jun 2023, approved
- Appendix G: Recruitment Poster dated 27 Jun 2023, approved
- Protocol dated 27 Jun 2023, approved
- Appendix E: Permission to Use GNCI dated 25 Jun 2023, approved

Appendix D: Recruitment Poster (Qualitative Stream)

Are you at risk of developing or have a hereditary cancer?

Consider joining a research study titled: **Cancer prevention in cancer predisposition syndromes: Testing the feasibility of building a hereditary cancer research registry and nurse navigator follow up model.**

Lead researcher: Dr. Holly Etchegary, Faculty of Medicine, Memorial University

What is this study about?

This study aims to better understand the experiences of patients with hereditary cancer syndromes and what kind of support they may need to help manage their cancer risks. A better understanding of these patients' needs can help create supports and services for patients with hereditary cancers.

Who Can Participate?

- Tested positive for any hereditary cancer gene (e.g., Lynch syndrome, BRCA 1 or 2, PALB2, CHEK2, CDH1, RAD51C, MEN1, VHL, etc.)
- 18 years and older
- Able to participate in a 45-60 minute interview over the phone or video conferencing

For more information, please contact by [insert date here]:

Rebecca Puddester; [redacted]
Email: rjp823@mun.ca

Or Nadine Burry; [redacted]
Email: tanya.burry@med.mun.ca

If you have questions regarding your rights as a research participant please contact the Health Research Ethics Authority at (709) 777-6974 or info@hrea.ca

Memorial University | Faculty of Medicine

300 Prince Philip Dr.
St. John's, NL A1B 3V6

Appendix E: Consent Form (Qualitative)

TITLE: Cancer prevention in cancer predisposition syndromes: Testing the feasibility of building a hereditary cancer registry and nurse navigator follow up model.

RESEARCHER(S): Study leads Dr. Holly Etchegary and Dr. Lesa Dawson

Co-investigators: Vanessa Francis, Dr. Zhiwei Gao, Dr. April Pike, Rebecca Puddester, Dr. Sevtap Savas, Dr. Melanie Seal, Mike Warren, Dr. Kathy Watkins, Dr. Michael Woods, Dr. Jane Green

Collaborators: Dr. Susan Avery, Fiona Curtis, Dr. Don MacDonald, Andree MacMillan, Dr. Jerry McGrath, Jennifer MacLean. Research staff: Tanya Nadine Burry and Sepideh Rajeziesfahani

Phone Number: Dr. Holly Etchegary 709-864-6605; email holly.etchegary@med.mun.ca

SPONSOR/FUNDER: Canadian Institutes of Health Research, Project Grant # 179835

You have been invited to take part in a research study. Taking part in this study is voluntary. You may choose to take part or you may choose not to take part in this study. You also may change your mind at any time. This consent form has important information to help you make your choice. It may use words that you do not understand. Please ask the researcher(s)/study staff to explain anything that you do not understand. It is important that you have as much information as you need and that all your questions are answered. Please take as much time as you need to think about your decision to participate or not, and ask questions about anything that is not clear. You may find it helpful to discuss it with your friends and family. The researcher/study staff will tell you about the study timelines for making your decision.

1. Why am I being asked to join this study?

You are being invited to join this study because you have a hereditary condition that increases your risk of developing cancer. Hereditary cancer syndromes (HCS) affect hundreds of people in NL. Previous work of our team has shown that patients in NL with HCS can have difficulty getting and keeping track of the screening and appointments recommended for patients living with inherited cancer risk. No research has been done to date where these patients identified their needs and preferences for help with managing this risk. Also, very little research has included patients with cancer genes other than *BRCA* and Lynch Syndrome. This study will help researchers, doctors, and policy makers better understand the experience of living with hereditary cancer risks. Researchers hope this can help inform healthcare services for families affected by inherited cancers. Specifically, information gathered from this study will help researchers develop a nurse navigator model of support for these patients (to be tested in another project). This is someone who has regular contact with patients, helping them make screening or other testing appointments, and providing information and support when needed.

2. How many people will take part in this study?

We will enroll up to 40 patients from Newfoundland and Labrador (NL). We hope to speak to patients with any hereditary cancer syndrome genes including Lynch syndrome, *BRCA1*/2, *ATM*, *CHEK2*, *PALB2*, *RAD51C*, *CDH1*, *MEN1, Von Hippel-Lindau* or any other inherited cancer gene.

3. How long will I be in the study?

You will be expected to take part in one interview over the next month lasting up to an hour. The interview can be done in person, over the phone or a video platform, whatever is your preference.

4. What will happen if I take part in this study?

If you agree to take part in this study, the following procedures will take place:

- Interviews: You will be asked to participate in one interview. During this interview, you will speak with members of the research team, April Pike or Rebecca Puddester. Each interview with either April or Rebecca will be about one hour and will take place in person, over the phone or a video platform, whatever is your preference. You will be asked to discuss your experiences of living with a hereditary cancer syndrome. We wish to better understand your experiences with the healthcare system and living with inherited cancer risks. In particular, we want to explore patients' opinions on whether a nurse navigator could be a helpful support person for coordinating the screening and appointments needed to manage inherited risk.
- Audio recording used: You will be audio/video recorded during the interview, whether by phone or video platform. The audio recording will be transcribed (written down) after the interview and will be analyzed by the research team. Your name or any other identifying information will not be included during the recording, except your voice. The audio recording will be destroyed after it has been transcribed and checked for accuracy.
- Video recording used**:** If you prefer, the interview will be video recorded, rather than done over the telephone. Webex is Memorial University's approved video meeting platform and will be used. As noted above, audio recordings of these interviews (without your face on camera, just your voice) will be transcribed.

5. Are there risks to taking part in this study?

During the interview, you may become uncomfortable or experience some anxiety, emotional and/or psychological distress due to the nature of the questions about living with the risk of hereditary cancer. You can skip questions, take a break or stop answering at any time. As well, the following resources are available for you to contact for psychological support:

Bridge the gApp

If you are finding it difficult to cope or feel anxious, Bridge the gApp offers many services to support you and your wellbeing. bridgethegapp.ca

Provincial CHANNAL Warm Line

You can also reach out and speak with a trained mental health peer supporter.

- 1-855-753-2560 9:00am to 12:00am daily.

Note that, due to large call volumes, you may be prompted to leave a voice mail. CHANNAL will return your call with a blocked phone number to protect your privacy. The Warm Line is a Mental Health Support Service.

Mental Health Crisis Line

If you are experiencing a mental health crisis, you can call the Mental Health Crisis Line 24 hours a day.

- 1-888-737-4668

Other telephone resources include:

Doorways (709) 752-4903; Mobile Crisis Response Team (709) 437-4668; or Adult Central Intake (709) 752-8888

You can also visit any hospital emergency room.

Audio Recording:

There is a potential risk of loss of your confidentiality because even though your name will not be part of the audio recording or the transcription, your voice may still be identifiable as your voice. If anyone mentions identifiers (e.g., your name), during the recording, this may identify you.

Inconvenience of time:

There is an inconvenience of time. Each interview can last up to one hour. We will make every effort to schedule interviews at a time convenient for you.

Confidentiality risk (for all studies):

Despite protections being in place, there is a risk of unintentional release of information. Researchers will make every attempt to protect your privacy.

6. What are the possible benefits of participating in this study?

There may not be direct benefit to you from taking part in this study. We hope that the information learned from this study can be used in the future to benefit other people with hereditary cancers.

If I decide to take part in this study, can I stop later?

It is your choice to take part in this study, participation is voluntary. You can change your mind at any time during the research study. The study team may ask why you are withdrawing for reporting purposes, but you do not need to give a reason to withdraw from the study if you do not want to. Withdrawal from the study will not have any effect on the care you or your family receives related to the hereditary cancer syndrome in your family. Interview honorariums will only be provided following completion of the interview. If you decide to leave the study, simply inform your researcher.

- You may withdraw from this study up to the point of data analysis. This means that the researcher/study staff will no longer use your data for research and all data collected about you will be destroyed. Any data that has already been analyzed cannot be destroyed or removed from the study. This is because we have to preserve the study's scientific integrity. However, your data will not be used in future research.

7. What are my rights when participating in a research study?

You have the right to receive all information that could help you make a decision about participating in this study, in a timely manner. You also have the right to ask questions about this study at any time and to have them answered to your satisfaction.

Your rights to privacy are legally protected by federal and provincial laws that require safeguards to ensure that your privacy is respected.

Signing this form gives us your consent to be in this study. It tells us that you understand the information about the research study. When you sign this form you do not give up any of your legal rights against the study doctor, sponsor or involved institutions for compensation, nor does this form relieve the study doctor, sponsor or their agents of their legal and professional responsibilities.

You have the right to be informed of the results of this study once the entire study is complete. You will be given the option at the end of the interview to provide an email address so we can send you a study summary and information on any public presentations we will do at the end of the study. You can also contact the lead researcher Dr. Holly Etchegary at any time for study updates or to request an end of study summary. Holly can be reached at 864-6605 or by email at holly.etchegary@med.mun.ca

You will be given a copy of this signed and dated consent form prior to participating in this study.

8. What about my privacy and confidentiality?

Protecting your privacy is an important part of this study. If you decide to participate in this study, the researchers/study staff will collect and use information from your interview. They will only collect and use the information they need for this study, including:

- Gender, age, cancer mutation, cancer history, marital status, education level, ethnicity
- information from study interviews

The personal health information or personal information collected about you will have your directly identifiable information removed (i.e., name) and replaced with a code or with a "study number". There will be a master list linking the code numbers to names. The researcher is responsible for keeping it separate from the interview transcripts and demographic information such as gender and age. This link will not be available to the Funding agency.

Study information collected during the study will kept at this site and stored in a secure, locked place that only the study staff will be able to access. After the study closes, study information will kept as long as required by law, which could be 5 years or more. This information will be stored on a password protected computer in the locked office of the lead researcher Dr. Holly Etchegary in the Faculty of Medicine, Craig Dobbin Centre for Genetics, 4th floor, Room 4M210, St. John's, NL. Any paper/hard copy information such as consent forms will be stored in a locked filing cabinet in Dr. Etchegary's office in the Faculty of Medicine. Dr. Holly Etchegary is the person responsible for keeping it secure.

When the results of this study are published or presented at scientific meetings, your name and other personal information will not be used in the publication.

All information that identifies you will be kept confidential, and to the extent permitted by applicable laws, will not be disclosed or made publicly available, except as described in this consent document. Every effort to protect your privacy will be made. Even though the risk of identifying you from the study data is very small, it can never be completely eliminated. If there is a breach of your privacy resulting from your participation in this study you will be notified during the study and for up to five years after the study closes while study information is kept.

Communication via e-mail is not absolutely secure. We do not recommend that you communicate sensitive personal information via e-mail.

9. Who will see my personal information?

Representatives from the following organizations may come to look at the study records and your personal health information under the supervision of the study staff to check that the information collected for the study is correct and to make sure the study followed the required laws and guidelines:

- Representatives of the Health Research Ethics Board

Your access to records

You have the right to see the information that has been collected about you for this study. If you wish to do so, please contact your researcher.

10. Declaration of financial interest, if applicable

There are no conflicts of interest to declare related to this study.

13. Compensation

To acknowledge the value of participants' time, a $25 honorarium is available to all participants at the end of the interview. You may choose to receive or decline this honorarium.

14. What about questions or problems?

If you have any questions about taking part in this study, you can meet with the principal investigator who is in charge of the study. That person is:

Dr. Holly Etchegary; Phone Number: 709-864-6605,

Email: holly.etchegary@med.mun.ca

Or you can talk to someone who is not involved with the study at all, but can advise you on your rights as a participant in a research study. This person can be reached through:

Ethics Office at 709-777-6974

Email at info@hrea.ca

Appendix F: Interview Guide (Qualitative)

1. **Please tell me about your experience with discovering the family's history of inherited cancer.**

Questions:	**Prompts:**
Can you tell me how you came to discover your family's risk for hereditary cancer?	How did you react when you were told you had a cancer predisposition syndrome (name *BRCA* or Lynch, etc. as appropriate)? Can you tell me how you felt when you began to realize the implications of this risk for yourself?
Can you tell me about what happened next when you realized you were at risk for hereditary cancer?	Were you referred to provincial medical genetics for counseling and testing? How long were you waiting to speak to a counselor? How long did it take to get your test results? Tell me how you felt while waiting to get your test result?
Did you have any nurses involved in your care as it related to your experiences with genetic healthcare/testing?	If so, did you find them to be helpful or unhelpful? If yes, what was it about those interactions that were most helpful? Did you feel they had sufficient knowledge to address your concerns? If not, do you that feel that having a nurse (with an understanding of genetics) involved in your care would have made a difference in some of the challenges you experienced? (if applicable)

2. **Can we talk about what having hereditary cancer means for your health (and healthcare)?**

Questions:	**Prompts:**
What recommendations were given to you after being diagnosed with hereditary cancer?	Was any kind of screening recommended? Were appointments made with specialists? Who talked with you about these? Was genetic testing of other relatives recommended?

3. **I would like to talk about how you manage your cancer risk.**

Questions:	Prompts:
What kinds of things have you been doing to manage your cancer risk?	Do you do any regular screening like colonoscopy, endoscopy, urological screening, pelvic exam, endometrial biopsy, transvaginal ultrasound, preventative surgery? [modify as per what the risk management guidelines are for men/women; specific cancer syndrome]. What is challenging about inherited cancer risk management for you? Are you dealing with any other health conditions that might affect how you manage your inherited cancer risk?
Did you feel you had enough information to make an informed decision about how you have decided to manage your cancer risk?	Was there any information you wished you would have had earlier? Or information you felt you didn't get at all? How did you get your information, from whom?

4. Healthcare needs

Questions:	Prompts:
Have there been any barriers to receiving the screening that has been recommended to you? Have there been facilitators – things that helped you do the things recommended to you to manage your risks?	Is there anything you feel you need in relation to managing your risks due to hereditary cancer, but have difficulty accessing? Have you ever received conflicting information about recommended screening?

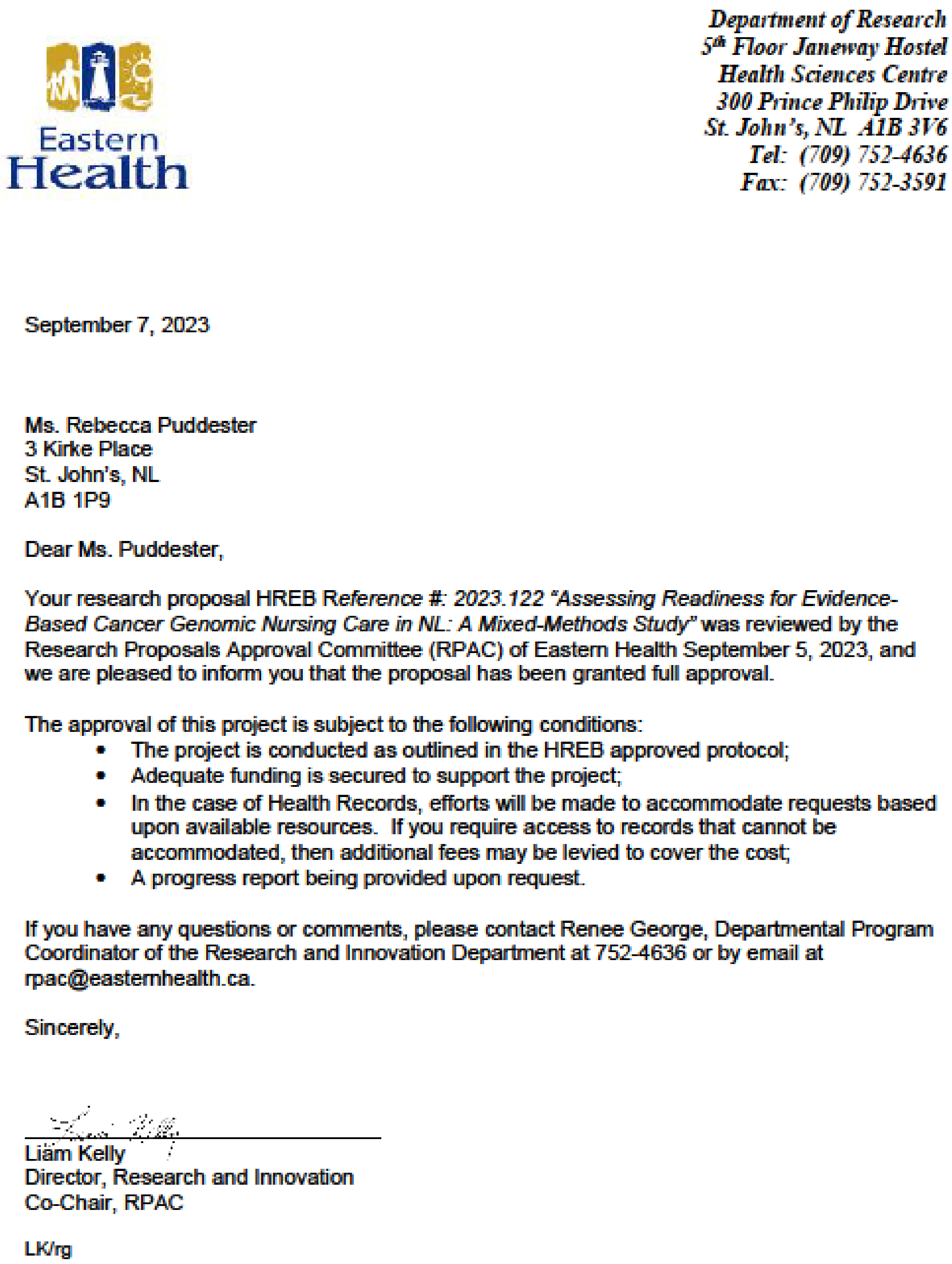
5. Nursing navigation models

We are wondering what you think about having a nurse act as a navigator for patients in an ongoing way. This would include regular contact with patients, reminding them about screening or other testing appointments, assisting with coordination of screening, helping to provide information of any kind (e.g., social support, community supports, help with genetic testing of children/other family members, etc.) as patients ask for it. We have patient navigators in our cancer care program in NL, but not specifically for people who have a mutation for a cancer gene. It is only when a cancer is diagnosed that patients can work with a navigator. Our hope is to create a model like this for individuals at high risk for cancer because of a hereditary cancer syndrome, but we would like to have patient input to help inform how that might look.

Questions:	**Prompts:**
Do you think you would like to have access to a nurse navigator as we've described?	What do you think the advantages of this would be? Would you be okay with a nurse serving in that role? Any other healthcare provider come to mind? Do any immediate concerns come to mind about a nurse navigator? At what point in your health care do you think access to a patient navigator would be important? Would there be an advantage in accessing a navigator before receiving genetic testing results?
What kinds of elements do you think a nurse navigator model should have?	Screening reminders? Coordination of screening? Help with emotional/psychosocial support? Regular contact even if there is no upcoming screening? Available at certain times to take phone calls? Assistance with communicating risk within the family? Advice on genetic testing for children or other relatives such as brothers or sisters? Please tell us anything that comes to mind.
Would you be comfortable with a nurse navigator having access to your medical records and communicating with your doctors?	What is concerning about this? If anything? Would you expect to give informed consent to take part in this kind of model before anything started? How/should your family doctor be involved?
Do you have any suggestions for us about how this model should look?	Who should be responsible for it? How should high risk individuals be informed about it? Should there be online elements (e.g., a patient portal where screening tests and reminders are automatically set up?) Any thoughts that come to mind are welcome.

Please feel free to share any other thoughts you have on managing inherited cancer risk.
Is there something you would like to share or raise that we have not discussed in our interview?

Appendix G: Institutional Approval



Eastern Health

Department of Research
5th Floor Janeway Hostel
Health Sciences Centre
300 Prince Philip Drive
St. John's, NL A1B 3V6
Tel: (709) 752-4636
Fax: (709) 752-3591

September 7, 2023

Ms. Rebecca Puddester
3 Kirke Place
St. John's, NL
A1B 1P9

Dear Ms. Puddester,

Your research proposal HREB R*eference #: 2023.122 "Assessing Readiness for Evidence-Based Cancer Genomic Nursing Care in NL: A Mixed-Methods Study"* was reviewed by the Research Proposals Approval Committee (RPAC) of Eastern Health September 5, 2023, and we are pleased to inform you that the proposal has been granted full approval.

The approval of this project is subject to the following conditions:

- The project is conducted as outlined in the HREB approved protocol;
- Adequate funding is secured to support the project;
- In the case of Health Records, efforts will be made to accommodate requests based upon available resources. If you require access to records that cannot be accommodated, then additional fees may be levied to cover the cost;
- A progress report being provided upon request.

If you have any questions or comments, please contact Renee George, Departmental Program Coordinator of the Research and Innovation Department at 752-4636 or by email at rpac@easternhealth.ca.

Sincerely,

Liam Kelly
Director, Research and Innovation
Co-Chair, RPAC

LK/rg

Appendix H: E-mail Script (Quantitative Stream)

Email Subject line: Invitation to participate in a study about oncology nursing and genomics

Study Title: Assessing Readiness for Evidence-Based Cancer Genomic Nursing Care in Newfoundland and Labrador: A Mixed-Methods Study

Dear NL Oncology Nurse,

You are invited to join a study designed to learn about oncology nurses' knowledge, attitudes and practices related to nursing and genomics. All nurses working in cancer care in NL are invited to join this confidential study. If you volunteer, you will be asked to answer demographic questions and a survey about genomics and nursing practice. The survey should take about 20-25 minutes to complete.

There are **no foreseeable risks or benefits** to you from participating in this study. All responses are anonymous. At the end of the study, participants have the option to follow a link following survey completion to enter their e-mail addresses in a link separate from the survey in a draw to win a $100 gift card. You may choose to enter or not enter this e-mail information.

To participate in this study, follow the link

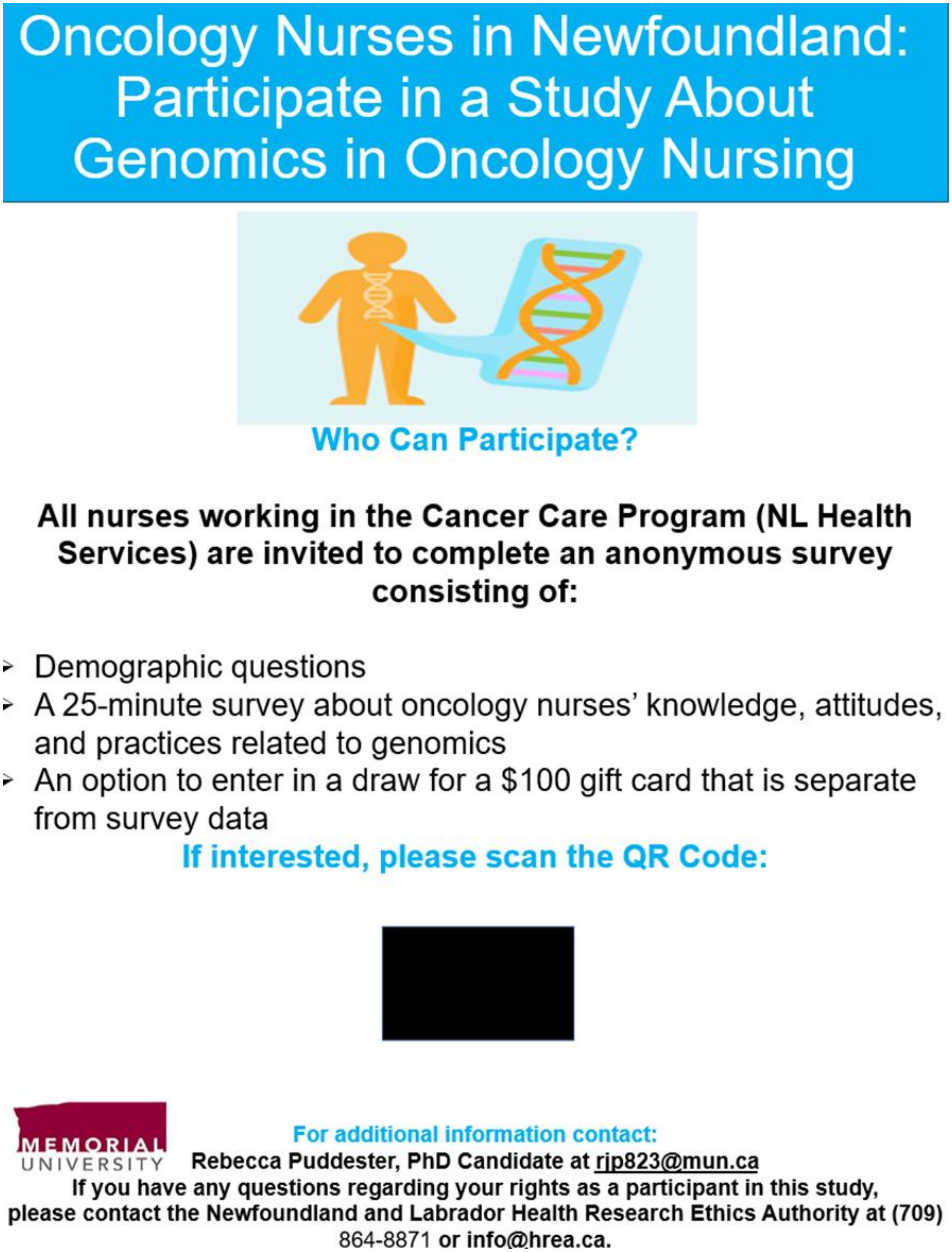
[redacted]

If you have additional questions, please email Rebecca Puddester, PhD (c) at the Faculty of Nursing Memorial University at rjp823@mun.ca

Researcher Information: The study is being conducted by Rebecca Puddester, PhD Candidate, Faculty of Nursing, Memorial University.

If you have any questions regarding your rights as a participant in this study, please contact the Newfoundland and Labrador Health Research Ethics Authority at (709) 864-8871 or info@hrea.ca.

Appendix I: Recruitment Poster (Quantitative Stream)



Oncology Nurses in Newfoundland: Participate in a Study About Genomics in Oncology Nursing

Who Can Participate?

All nurses working in the Cancer Care Program (NL Health Services) are invited to complete an anonymous survey consisting of:

- Demographic questions
- A 25-minute survey about oncology nurses' knowledge, attitudes, and practices related to genomics
- An option to enter in a draw for a $100 gift card that is separate from survey data

If interested, please scan the QR Code:

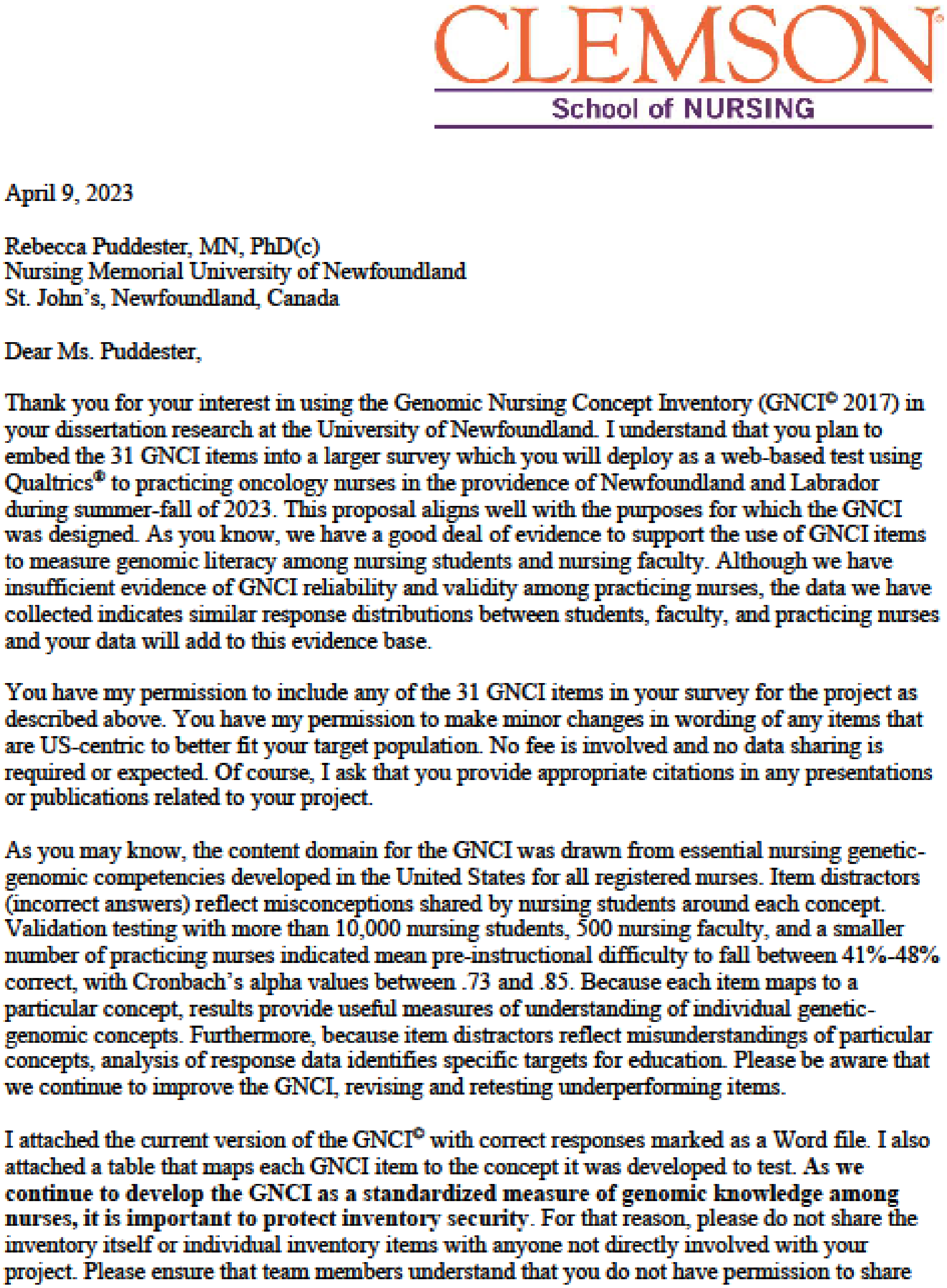
[redacted]

For additional information contact:

Rebecca Puddester, PhD Candidate at rjp823@mun.ca

If you have any questions regarding your rights as a participant in this study, please contact the Newfoundland and Labrador Health Research Ethics Authority at (709) 864-8871 or info@hrea.ca.

Appendix J: Permission to use the GNCI©



CLEMSON
School of NURSING

April 9, 2023

Rebecca Puddester, MN, PhD(c)
Nursing Memorial University of Newfoundland
St. John's, Newfoundland, Canada

Dear Ms. Puddester,

Thank you for your interest in using the Genomic Nursing Concept Inventory (GNCI© 2017) in your dissertation research at the University of Newfoundland. I understand that you plan to embed the 31 GNCI items into a larger survey which you will deploy as a web-based test using Qualtrics® to practicing oncology nurses in the providence of Newfoundland and Labrador during summer-fall of 2023. This proposal aligns well with the purposes for which the GNCI was designed. As you know, we have a good deal of evidence to support the use of GNCI items to measure genomic literacy among nursing students and nursing faculty. Although we have insufficient evidence of GNCI reliability and validity among practicing nurses, the data we have collected indicates similar response distributions between students, faculty, and practicing nurses and your data will add to this evidence base.

You have my permission to include any of the 31 GNCI items in your survey for the project as described above. You have my permission to make minor changes in wording of any items that are US-centric to better fit your target population. No fee is involved and no data sharing is required or expected. Of course, I ask that you provide appropriate citations in any presentations or publications related to your project.

As you may know, the content domain for the GNCI was drawn from essential nursing genetic-genomic competencies developed in the United States for all registered nurses. Item distractors (incorrect answers) reflect misconceptions shared by nursing students around each concept. Validation testing with more than 10,000 nursing students, 500 nursing faculty, and a smaller number of practicing nurses indicated mean pre-instructional difficulty to fall between 41%-48% correct, with Cronbach's alpha values between .73 and .85. Because each item maps to a particular concept, results provide useful measures of understanding of individual genetic-genomic concepts. Furthermore, because item distractors reflect misunderstandings of particular concepts, analysis of response data identifies specific targets for education. Please be aware that we continue to improve the GNCI, revising and retesting underperforming items.

I attached the current version of the GNCI© with correct responses marked as a Word file. I also attached a table that maps each GNCI item to the concept it was developed to test. As we **continue to develop the GNCI as a standardized measure of genomic knowledge among nurses, it is important to protect inventory security**. For that reason, please do not share the inventory itself or individual inventory items with anyone not directly involved with your project. Please ensure that team members understand that you do not have permission to share

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the inventory or individual inventory items beyond the conduct of this particular project. Please control test distribution to prevent participants from acquiring the inventory. Finally, **please do not reproduce inventory items in publications or presentations without my express permission.**

Best wishes with your study! I am honored that you wish to use the GNCI and invite you to contact me if I can provide any further information.

Sincerely,

Linda Ward

Linda D. Ward, PhD, CNE, FNP-C
Associate Professor
Clemson University School of Nursing
411 Edwards Hall
Clemson, SC 29634
(864) 656-5528
ldward@clemson.edu
attached: GNCI version 2017 with answers
GNCI item-concept table

Appendix K: Qualtrics Consent (Quantitative Stream)

Assessing Readiness for Evidence-Based Cancer Genomic Nursing Care in Newfoundland and Labrador: A Mixed-Methods Study

Principal Investigator: Rebecca Puddester MN, PhD(c). RN Faculty of Nursing, Memorial University Email: rjp823@mun.ca

Supervisor: Joy Maddigan, PhD RN Faculty of Nursing, Memorial University Email: jmaddigan@mun.ca

Dear NL Oncology Nurse,

You are invited to complete a survey that will evaluate your knowledge, attitudes, and practices with genomics. Genetics refers to the study of single genes, one at a time, where as genomics is the study of the entire genome of the organism and the interaction of the genome with the environment. Oncology nurses have an opportunity to shape patient care when using genetic information in their practice. Globally and in Newfoundland and Labrador (NL), we need to gather more information about what oncology nurses know about genomics and their readiness to integrate genomics in their practice: Before choosing to participate, please consider that:

You have been invited to complete this survey because you are an oncology nurse in NL.

Questions in the survey relate to your current knowledge, opinions about implications of genetic and genomics-informed health care for preventing and treating diseases such as cancer.

Demographic questions will also be collected such as age, years in oncology nursing practice and practice area. This is to compare responses among different groups.

The survey will take about 20-25 minutes to complete. Your participation in this survey is completely voluntary and you can choose to skip any questions that you do not wish to answer. If you are not sure about an answer, please make your best guess. Several of the questions relate to your attitudes about genetics and genomics for which there are no right or wrong answers.

There are no risks, penalties, or costs to your participation. There are no direct benefits to your participation other than contributing to research. **After the survey, you will have the option to enroll in a draw for a $100 gift card.** The link for the gift card draw is not linked to your survey responses. Only the primary researcher, Rebecca Puddester, will have access to the e-mail addresses for the gift card draw, and this data will be immediately destroyed following the draw.

This survey is administered using Qualtrics online survey platform and is stored securely and not shared. The survey is anonymous meaning that there is no way for the researcher to determine who submitted the survey, thus the results of the survey cannot be returned directly to you.

The overall study findings will be used to inform the development of educational modules to be made available to nurses in the NL Cancer Care Program. This also means that once you have submitted your responses there is no way for your questionnaires to be removed.

If you have any questions about taking part in this study, you can contact the primary investigator who is in charge of the study. That person is: Rebecca Puddester MN RN; PhD (C) (Tel: [redacted] E-mail: rjp823@mun.ca). Alternatively, you can talk to someone who is not involved with the study at all but can advise you on your rights as a participant in a research study. This person can be reached through the Ethics Office (Tel: (709) 864-8871 / Email: info@hrea.ca).

Do you consent to participating in this study?

By selecting “Yes” and click "Next," you are consenting to be in this study. It tells us you understand the information about the research study. When you select “yes” and consent to be a participant in this study, you are not giving up your legal rights. Researchers or agencies involved in this research study still have their legal and professional liabilities.