UNIT 5 - GENOMICS NURSING RESEARCH

Precision Healthcare: Genomics-Informed Nursing by Andrea Gretchev

Please visit the web version of Precision Healthcare: Genomics-Informed Nursing (https://ecampusontario.pressbooks.pub/personalizedhealthnursing/) to access the complete book, interactive activities and ancillary resources.

Unit 5 Contents

Attribution

5.1 UNIT OVERVIEW

Learning Objectives

- Identify key genomic approaches utilized in nursing research and their relevance to patient care.
- Explore barriers to achieving diversity in genomics research and strategies for equitable and inclusive participation.
- Outline the ethical, legal, and social challenges associated with genomic research, with a focus on informed consent, privacy, and inclusion of diverse populations.
- Explain how strategic plans like CIHR's *Sequencing our Future* shape research priorities and funding decisions.
- Describe practical examples of knowledge translation (KT) and knowledge mobilization (KM) outputs.

Outline

Topics covered in this chapter include:

- Genomic research in nursing
- Human genetic research in Canada and Internationally
- Research priorities and funding
- Knowledge translation and mobilization
- Scholarly Posters

Competencies Nurses will Develop in this Chapter

ANA (2023):

Identification:

- Identifies ethical, ethnic or ancestral, cultural, religious, legal, fiscal, and societal issues related to genomic information and technologies.
- Recognizes issues that undermine the rights of all clients for autonomous, informed genomic-related decision-making and voluntary action.

Provision of education, care, and support:

- Advocates for autonomous, informed genomic-related decision-making.
- Evaluates the impact and effectiveness of genomic interventions on clients' outcomes.

NHS (2023):

Advocate for the rights of all individuals to make informed decisions and act voluntarily:

- ensuring that the consent process is person centred; and
- promoting and supporting equitable access to genomic services.

Apply knowledge, understanding and context of genomic testing and information to underpin care and support for individuals and families prior to, during and following decision-making:

- incorporating awareness of the ethical, legal and social issues related to testing, recording, sharing and storage of genomic information and data.

Examine your own competency of practice on a regular basis:

- recognizing areas where professional development related to genomics would be beneficial; and
- maintaining awareness of clinical developments in genomics that are likely to be of most relevance to your area of practice, seeking further information on a case-by-case basis.

Obtain and communicate reliable, current information about genomics, for self, patients, families and colleagues:

- using information technologies and other information sources effectively to do so; and
- applying critical appraisal skills to assess the quality of information accessed.

Key terminology

Candidate gene analysis

The term candidate gene refers to a gene that is believed to be related to a particular trait, such as a disease or a physical attribute. Because of its genomic location or its known function, the gene is suspected to play a role in that trait, thus making it a candidate for additional study. Candidate Gene. The more you know about a trait the better job you can do selecting a candidate gene for further study. For instance, if you're studying the genetics of body size, genes that control bone growth, lipid processing, and insulin growth factors are all excellent candidate genes.

Genome-wide association study (GWAS)

A genome-wide association study is an approach that involves rapidly scanning markers across the complete sets of DNA, or genomes, of many people to find genetic variations associated with a particular disease. Once new genetic associations are identified, researchers can use the information to develop better strategies to detect, treat and prevent the disease. Such studies are particularly useful in finding genetic variations that contribute to common, complex diseases, such as asthma, cancer, diabetes, heart disease and mental illnesses (NGHRI, 2020, para. 1).

Next-Generation DNA Sequencing (NGS)

DNA sequencing establishes the order of the bases that make up DNA. Next-generation DNA sequencing (abbreviated NGS) refers to the use of technologies for sequencing DNA that became available shortly after the completion of the Human Genome Project (which relied on the first-generation method of Sanger sequencing). Faster and cheaper than their predecessors, NGS technologies can sequence an entire human genome in a single day and for less than 1,000.

Symptom science

A field of research focused on understanding the biological and behavioral mechanisms underlying symptoms experienced by patients, such as pain, fatigue, and cognitive impairment.

Translational research

Often described as "bench to bedside," because it involves taking discoveries from basic science (the bench) and applying them to clinical practice (the bedside).

Omics

The branches of science known informally as omics are various disciplines in biology whose names end in the suffix *-omics*, such as genomics (https://en.wikipedia.org/wiki/Genomics), proteomics (https://en.wikipedia.org/wiki/Proteomics), metabolomics (https://en.wikipedia.org/wiki/Metabolomics), metagenomics (https://en.wikipedia.org/wiki/Metagenomics), phenomics (https://en.wikipedia.org/wiki/Phenomics) and transcriptomics (https://en.wikipedia.org/wiki/Transcriptomics). Omics aims at the collective characterization and quantification of pools of biological molecules that translate into the structure, function, and dynamics of an organism or organisms. The related suffix **-ome** is used to address the objects of study of such fields, such as the genome (https://en.wikipedia.org/wiki/Genome), proteome (https://en.wikipedia.org/wiki/Proteome) or metabolome respectively ("Omics", 2024, para. 1).

Attribution & References

Except where otherwise noted, this page is adapted from:

- Talking Glossary of Genomic and Genetic Terms, Courtesy of: National Human Genome Research institute (NGHRI), Public Domain with attribution.
- Symptom science and translational research definitions written by Andrea Gretchev

References

American Nurses Association (ANA). (2023). *Essentials of genomic nursing: Competencies and outcome indicators* (3rd ed.). https://www.nursingworld.org/nurses-books/ana-books/ebook-essentials-of-genomic-nursing-competencies-/

National Health Service (NHS). (2023). *The 2023 genomic competency framework for UK nurses*.

https://www.genomicseducation.hee.nhs.uk/wp-content/uploads/2023/12/2023-Genomic-Competency-Framework-for-UK-Nurses.pdf

National Human Genome Research Institute (NHGRI). (2020, August 17). *Genome-Wide Association Studies Fact Sheet*. Genome.gov. https://www.genome.gov/about-genomics/fact-sheets/Genome-Wide-Association-Studies-Fact-Sheet

Omics. (2024, November 23). In *Wikipedia*. https://en.wikipedia.org/wiki/Omics

5.2 GENOMIC RESEARCH IN NURSING

One of the best aspects of a nursing career is the diverse range of roles nurses can pursue, from direct patient care to leadership positions and specialized areas such as research. This variety allows nurses to continually grow and adapt in their careers, contributing to advancements in healthcare and improving patient outcomes in numerous ways. Advanced practice nurses such as clinical nurse specialists, nurse educators, and nurse practitioners typically have a research component to their role.

Role of Nurses

Nurses play a key role in genomic research by bridging the gap between complex scientific data and patient care. There are many roles for nurses in research. They can contribute by participating in research studies, collecting and managing genetic data, and ensuring ethical standards are upheld. There are many opportunities for nurses to be part of research or simply to refer patients who may be eligible for particular research projects.

Nurses – Research Roles

Type of Research	Role	Description
Bench Scientist	Laboratory Researcher	Conducts lab-based research on biological and physiological processes.
Symptom Science	Symptom Scientist	Studies patient symptoms to improve management and quality of life.
Qualitative Research	Qualitative Researcher	Explores patient experiences and behaviors using interviews, focus groups, and ethnography.
Quantitative Research	Quantitative Researcher	Uses statistical methods to test hypotheses and measure outcomes in clinical trials and surveys.
Mixed Methods Research	Mixed Methods Researcher	Combines qualitative and quantitative approaches for a comprehensive understanding of research questions.
Clinical Research	Clinical Research Nurse	Manages and coordinates clinical trials, ensuring ethical conduct and patient recruitment.
Health Services Research	Health Services Researcher	Studies the organization, delivery, and financing of healthcare services to improve systems and policies.
Implementation Science	Implementation Scientist	Focuses on translating research findings into clinical practice.
Bioinformatics	Bioinformatics Nurse	Analyzes biological data using computational tools to understand complex biological systems.
Genomics	Genomics Nurse	Applies genomic information in clinical care, including genetic testing and counseling.

There is some excellent work being done in genomics research by nurse scholars that has impacts on clinical practice, policy, the nursing workforce, healthcare system transformation, leadership, and education. The following article provides an introductory overview of genomic approaches in nursing research. It reviews essential concepts in genetics and genomics, provides an overview of the research process, and highlights nursing studies that have used genomic technologies. The authors emphasize the potential of genomics to advance nursing research and encourage nurses to incorporate genomics into their research practice. Nurse researchers can utilize diverse methodologies and measurements, consider biological plausibility studies, case studies, patient surveys, qualitative and quantitative research. Table 1 of the article also highlights commonly utilized approaches to genomic analysis such as the more traditional **candidate gene analysis**, which can be effective for investigating specific hypotheses, and a **genome-wide association study (GWAS),** which has become increasingly favored due to advancements like **Next-Generation Sequencing (NGS)** technologies. NGS allows for high-throughput sequencing of the entire genome, transcriptome, and epigenome, offering a more detailed understanding of the intricate interplay between genetic factors and disease.

In order for nurses at the point-of-care to base their practices in current evidence, they must be research consumers. Nurses learn how to critically appraise research and ensure they are accessing quality, peer

reviewed, and current literature. It is critical to stay abreast of new evidence in this rapidly evolving area in order to build and maintain genomic literacy.

Read

Bueser, T., Skinner, A., Bolton Saghdaoui, L., & Moorley, C. (2022). Genomic research: The landscape for nursing. *Journal of Advanced Nursing*, *78*(9), e99–e100. https://doi.org/10.1111/jan.15396

Question for reflection

1. What steps can you take as a future nurse to stay informed about the latest developments in genomics and their implications for nursing?
2. What is the difference between a GWAS and an EWAS (hint: you may need to consult an external source to answer this)?

The Current State of the Science

Thomas et al. (2023) recently conducted a scoping review examining the progress made over the last decade in nursing and midwifery genomics, demonstrating significant growth in the number of publications on the subject. In order for the advances that have been made in genomic technology to truly benefit patients, nurses need to focus on conducting research that generates clinically relevant evidence. The authors emphasize the need for future research to move away from descriptive studies to interventional studies and implementation research.

Symptom Science

Symptom science is a field of research focused on understanding the biological and behavioral mechanisms

underlying symptoms experienced by patients, such as pain, fatigue, and cognitive impairment. This research aims to identify the causes of these symptoms, develop effective interventions, and improve patient outcomes. Symptom science is closely related to **translational research**, often described as "bench to bedside," because it involves taking discoveries from basic science (the bench) and applying them to clinical practice (the bedside). By translating findings from laboratory studies into practical treatments and interventions, symptom science helps bridge the gap between research and patient care, ensuring that new knowledge directly benefits patients. By engaging in symptom science, nurses can develop and implement evidence-based interventions that improve patient outcomes and quality of life. This field also empowers nurses to advocate for patients, ensuring that symptom management strategies are personalized and effective, ultimately enhancing the overall healthcare experience. To read more about advancing symptom science in a precision health context, see Hickey et al. (2019) in the optional readings list at the end of this unit.

Omics

This article explores the use of "**omics**" measures in nursing science research to better understand the biological determinants of health. The authors provide excellent examples of nursing genomics research into health outcomes such as chronic lower back pain and variation in pain outcomes, irritable bowel syndrome, the use of the microbiome in maternal health research, and how metabolomics, the study of chemicals involved in biological function, is used to analyze the impact of dietary interventions on multiple sclerosis. As in the other articles, the authors note the gap between "omics" and clinical practice and recommend integrating "omics" content into core PhD and healthcare provider training. They emphasize the need to educate a larger group of nursing scientists who are skilled in designing experiments, capturing data, and analyzing data in combination with endophenotypic and phenotypic data.

Examples of research nurse scholars and scientists are involved in:

Read

Ferranti, E. P., Grossmann, R., Starkweather, A., & Heitkemper, M. (2017). Biological determinants of health: Genes, microbes, and metabolism exemplars of nursing science. *Nursing outlook*, *65*(5), 506–514. https://doi.org/10.1016/j.outlook.2017.03.013

Question for reflection:

1. How do you envision incorporating knowledge of genomics, the microbiome, and metabolomics into your future nursing practice?

Attribution & References

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Thomas, J., Keels, J., Calzone, K. A., Badzek, L., Dewell, S., Patch, C., Tonkin, E. T., & Dwyer, A. A. (2023, October 27). Current state of genomics in nursing: A scoping review of healthcare provider oriented (clinical and educational) outcomes (2012-2022). *Genes*, *14*(11), 2013. https://doi.org/10.3390/genes14112013

5.3 HUMAN GENETIC RESEARCH IN CANADA AND INTERNATIONALLY

Ethical Practice for Human Genetic Research in Canada

While we will delve into the ethical, legal, and social aspects of genomics in a later unit, it is crucial to address the ethical considerations that arise when discussing genomic research. These include special considerations related to privacy and confidentiality of genomic data, complexities related to informed consent and the unspecified future use of specimens (broad consent), underrepresentation in genomic data, equity and access to genomic services, genetic discrimination, return of findings including incidental findings, cultural considerations, ethical oversight, emerging treatments that pose ethical concerns, and genetic testing in children. It is beyond the scope of this course to delve into all of these ethical issues. A brief review of select issues will be provided as well as information on where to seek guidance to resolve ethical concerns. These concerns are not specific to research in Canada. Globally, genomics research experiences the same ethical issues. However, this chapter will focus on policy that guides human genetics research in Canada.

The Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans (TCPS2 – 2022) is the policy that governs research involving humans conducted by Canadian researchers under the three federal funding agencies: the Canadian Institutes of Health Research (CIHR), the Natural Sciences and Engineering Research Council of Canada (NSERC), and the Social Sciences and Humanities Research Council of Canada (SSHRC). As a condition of funding, researchers must adhere to this policy. It is based upon the Belmont Report (https://www.hhs.gov/ohrp/regulations-and-policy/belmont-report/index.html) and the Nuremberg Code (https://research.unc.edu/human-research-ethics/resources/ccm3_019064/). It should also be noted that TCPS2 (2022) uses the term "genetics." For the purposes of this chapter, this should be considered synonymous with "genomics."

There are learning modules (https://tcps2core.ca/welcome) that all Canadian researchers complete in order to conduct research in Canada and a certificate of completion can be downloaded.

Many of the chapters in the TCPS2 (2022) are specifically applicable to ethical issues arising from genomics research, such as consent, privacy and confidentiality, and storage and use of human biological materials. Additionally, chapter 13 is dedicated to human genetic research considerations.

Read

Briefly Review: Chapter 13: Human Genetic Research in the TCPS2 (https://ethics.gc.ca/eng/tcps2-eptc2_2022_chapter13-chapitre13.html#a).

This chapter focuses on the ethical conduct of human genetic research. It addresses the application of core principles, management of information revealed through genetic research, genetic counselling, and considerations for research involving families, communities, and groups. The chapter emphasizes the importance of privacy, consent, and the potential social impacts of genetic research.

Questions for reflection:

1. How can nurses ensure that patients fully understand the implications of consenting to genetic research, especially regarding privacy and potential future use of their genetic information?
2. What strategies can nurses employ to address the ethical challenges that arise when genetic research findings have implications for a patient's family members or community?
3. As nurses enter a healthcare landscape increasingly reliant on data, how can they advocate for responsible and ethical use of data, especially concerning patient privacy and confidentiality in the context of precision health?

Diversity in Genomic Research (NHGRI, 2023)

The code embedded within the human genome is complex, and genomics research has only scratched the surface of determining everything there is to know about what makes us all different at the DNA level. Historically, the people who have provided their DNA for genomics research have been overwhelmingly of European ancestry, which creates gaps in knowledge about the genomes from people in the rest of the world. Scientists are now expanding their data collection to better understand how genomics can be used to improve the health and wellbeing of all people.

Based on work completed before and after the Human Genome Project, researchers found that the genome

sequences of human populations have changed significantly over 250,000 years of our species' expansion and migration across the Earth. Even with the high degree of similarity between any two human genomes, enough differences exist that it is not appropriate to use a single, or even a few, genomes to represent the world's populations. This highlights that the original human genome reference sequence, produced by the Human Genome Project and based on just a handful of research participants, was just the starting point for human genomics.

To address this limitation, efforts are underway to create human reference genome sequences that better represent diverse populations. NHGRI funds the Human Pangenome Reference Program (https://www.genome.gov/Funded-Programs-Projects/Human-Genome-Reference-Program), which is generating a collection of reference genome sequences that better represent human diversity.

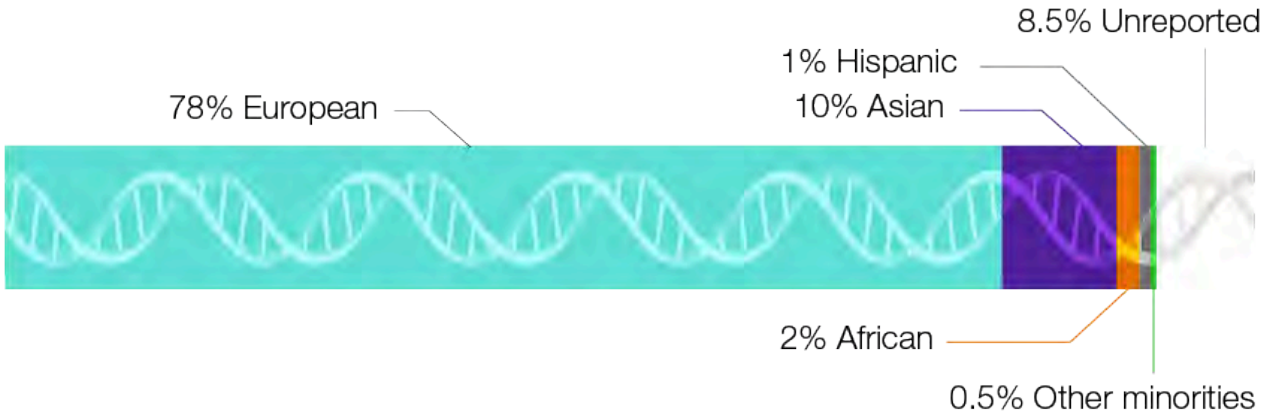

Figure 5.1 The percentage of ancestry populations included in large-scale genomic studies is overwhelmingly European(78%). 10% Asian, 1% Hispanic, 8.5% unreported, 2% African and 0.5% other minorities. **Source:** courtesy: National Human Genome Research Institute, Public Domain with Attribution

How does studying diverse human genomes improve health outcomes?

Every human has some baseline genetic risk of developing a given disease. Extensive research has been performed to both understand and learn how to respond to these risks. In some cases, the same variant consistently causes a disease (e.g., Huntington's disease and cystic fibrosis), but this might not be the case for more complex diseases (e.g., coronary artery disease, obesity, cancer and Alzheimer's disease).

By including populations that reflect the full diversity of human populations in genomic studies, researchers can identify genomic variants associated with various health outcomes at the individual and population levels. This way, researchers can better define a person's risk of developing a specific disease (https://www.genome.gov/Health/Genomics-and-Medicine/Polygenic-risk-scores) and design a clinical management strategy that is tailored to the individual. In addition, they can pursue genomic medicine strategies that benefit specific populations.

Why has enhancing diversity in genomics research been a difficult task?

Increasing the representation of diverse participants in genomics research requires an investment of both resources and time to intentionally establish trusting and respectful long-term relationships between communities and researchers. To ensure that genomics research is both equitable and inclusive, it is crucial for the genomics research workforce to reflect a similar diversity as the communities that the research is intended to serve.

In the past, both inaccessible and insufficient communication left some research participants unclear about the benefits of their participation and how their data would be used after the studies concluded. To overcome this, researchers must seek to understand people's reasons for not participating in genomic studies and to communicate with participants in a more accessible manner. This can take additional time, effort and resources, which may discourage some researchers from including these important, diverse populations in their studies. However, such exclusion can lead to notable gaps in scientific understanding and potentially reenforce existing disparities in genomics research.

Tracking Resource

The GWAS Diversity Monitor (https://gwasdiversitymonitor.com/) (Mills, 2020) is an interactive dashboard that tracks the diversity of participants in all published Genome Wide Association Studies (GWAS).

What are some genomics research projects that are enhancing the diversity?

Genomics researchers have initiated dozens of research projects to enhance the representation of research participants in genomics research. These studies are addressing a variety of research topics, including the effects of genomic diversity on disease risk, how to tailor genomic medicine for underrepresented populations, the impact of genomics research on diverse and the history of the human population.

NIH's *All of Us (https://allofus.nih.gov/)* Research Program is working to build a diverse health resource by collecting genome-related data and other information from about 1 million people. The Global Alliance for Genomics and Health (https://www.ga4gh.org/about-us/) (GA4GH) is developing a framework for storing, analyzing and sharing genomic data among international researchers. The Human Cell Atlas (https://www.humancellatlas.org/) aims to be a resource that includes in-depth information about all cell types found in people across the world.

How is NHGRI helping to improve diversity in genomics research?

NHGRI is dedicated to increasing diversity of the genomics workforce (https://www.genome.gov/about-nhgri/leadership-initiatives/diversity-in-genomics-workforce). In addition, NHGRI supports projects that work to increase the diversity of people participating in genomics research, including:

- The 1,000 Genomes Project (https://www.internationalgenome.org/) (2002 – 2015)
  The most extensive public catalog of human variation and genomic data, with over 2,000 genomic samples from 26 populations across the North and South America, Africa, Asia and Europe.
- Human Heredity and Health in Africa (H3Africa) (https://h3africa.org/) (2012 – 2022)
  The largest pan-African genomic research consortium that investigates the genomics of disease in Africa. The project also aims to build a sustainable African genomics research enterprise. This project is a collaborative effort that also involves the NIH Common Fund, the Wellcome Trust and the African Academy of Sciences.
- Polygenic Risk Score (PRS) Diversity Consortium (https://www.genome.gov/Funded-Programs-Projects/Polygenic-Risk-Score-Diversity-Consortium) (2021 – 2027)
  The consortium uses insights from genomic diversity to predict health and disease risk across diverse populations using a PRS approach.
- Implementing Genomics in Practice (IGNITE) Network (https://www.genome.gov/Funded-Programs-Projects/Implementing-Genomics-in-Practice-IGNITE) (2018 – 2022)
  This network assesses approaches for real-world applications of genomic medicine in diverse clinical settings.
- Electronic Medical Records and Genomics (eMERGE) Network (https://www.genome.gov/Funded-Programs-Projects/Electronic-Medical-Records-and-Genomics-Network-eMERGE) (2020 – 2025)
  This network establishes protocols and methodologies for improved genomic risk assessments for diverse populations and to integrate their use in clinical care.

Unethical Research Conduct Consequences

Historical abuses of research ethics have led to a lack of trust in scientific research and medical systems. This has led to the development of stricter policies guiding research ethics to protect participants and researchers. Ethical guidelines draw particular attention to the protection of vulnerable subjects because history has taught us that these populations are most easily exploited and have the most to lose. We will explore scientific racism more in the unit on ethical, legal and social implications of genomics. However, it seems fitting to include mention of this research history here.

HeLa Cells: A Lasting Contribution to Biomedical Research

Figure 5.2 Source: National Institutes of Health Office of Science Policy, PDM

In 1951, Henrietta Lacks, a 31-year-old African-American woman, went to Baltimore's Johns Hopkins Hospital to be treated for cervical cancer. Some of her cancer cells began being used in research due to their unique ability to continuously grow and divide in the laboratory. These so-called "immortal" cells were later named "HeLa" after the first two letters of **He**nrietta **La**cks first and last name.

Since Ms. Lacks' untimely death in 1952, HeLa cells have been a vital tool in biomedical research, leading to an increased understanding of the fundamentals of human health and disease. Some of the research involving HeLa cells also served as the underpinning of several Nobel Prize winning discoveries.

While Henrietta Lacks' story has been known in the research community for some time, it raised further awareness after the publication of the best-selling book *The Immortal Life of Henrietta Lacks* (https://search.worldcat.org/title/The-immortal-life-of-Henrietta-Lacks/oclc/326529053) (Skloot, 2010).

To honor Ms. Lacks' and her family's continued support of biomedical research, NIH analyzed and evaluated the scientific literature involving HeLa cells and found over 110,000 publications that cited the use of HeLa cells between 1953 to 2018. This analysis further highlights the persistent impact of HeLa cells in science and medicine, proving that they have been a consistent, essential tool that has allowed researchers to expand the knowledge base in fields such as cancer biology, infectious disease, and many others.

This website aims to act as a transparent, accessible resource to the general public, scientific researchers, and the Lacks' family that is in keeping with the spirit of the historic 2013 NIH-Lacks Family Agreement. NIH remains grateful to Henrietta Lacks and her family for the contributions of HeLa cells to science and medicine, and for her family's continued support of biomedical research.

Source: HeLa Cells: A Lasting Contribution to Biomedical Research courtesy of the NIH Office of Science Policy.

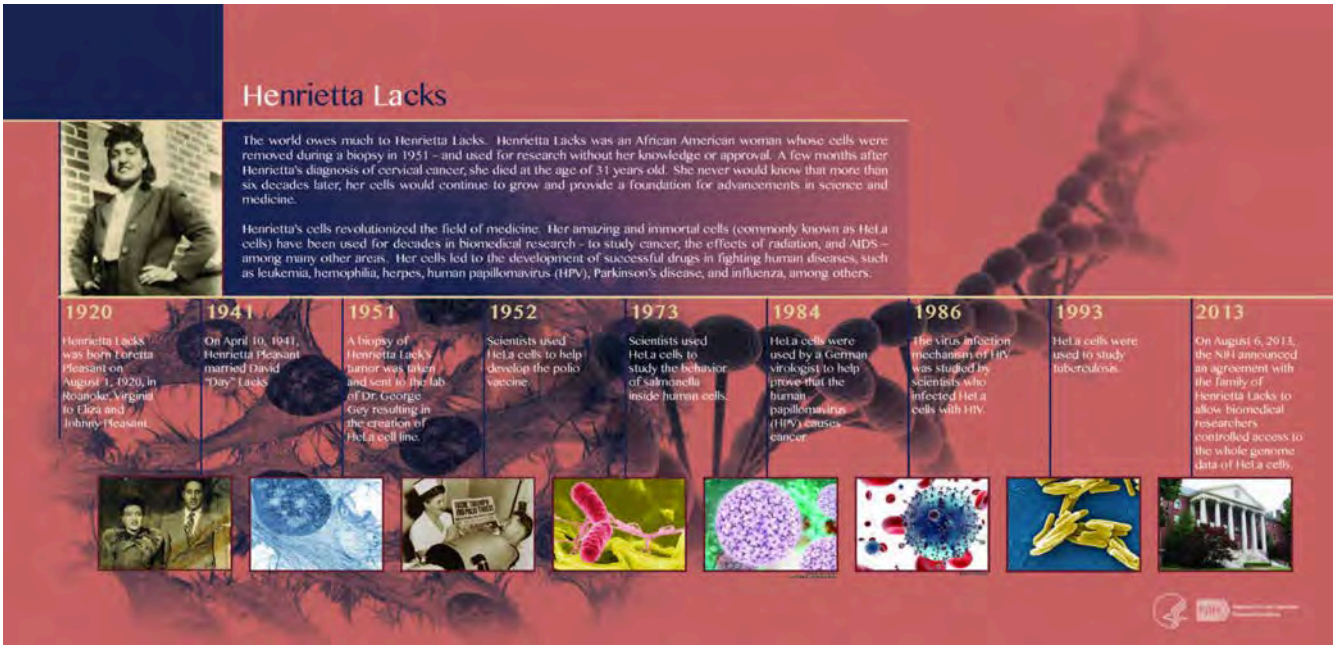

Figure 5.3 HeLa timeline. Courtesy: National Human Genome Research Institute. Figure 5.3 image description. **Source:** Image by Darryl Leja, NHGRI. PDM

Concept in Action: Henrietta Lacks

Use the bars on the bottom of the interactive slide show to navigate and watch the following videos, or use the text links below to access the videos on YouTube.

Concept in Action: Henrietta Lacks (text version)

- Watch Henrietta Lacks: The 'immortal' cells that changed the world – BBC REEL (8 mins) on YouTube (https://youtu.be/pgB1lqGp8BE)
- Watch Henrietta Lacks' family settles with biotech company over cancer cells (2 min) on YouTube (https://youtu.be/AENPGhVWBvE)

Source: Created by Andrea Gretchev, CC BY-NC 4.0 except where otherwise noted.

Here in Canada, the Nuu-chah-nulth case brought attention to the ethical concerns surrounding the use of stored biological samples, especially those collected from indigenous communities. The Nuu-chah-nulth tribe donated blood samples for research into the genetic causes of rheumatoid arthritis but later learned their samples were used for unrelated research without their consent, which they considered an example of exploitation. The tribe's blood samples were collected by geneticist Ryk Ward at the University of British Columbia, who took the samples with him when he left the university, continuing his research at the

University of Utah and later at the University of Oxford. Ward shared data from the samples with collaborators and published half a dozen articles based on his research. In response to the case, the University of British Columbia and the University of Utah implemented new policies requiring researchers to obtain consent for any new research conducted on stored samples.

Special Considerations for Genomics Research with Indigenous Populations

The article below focuses on consultations with First Nations communities in northern British Columbia regarding the establishment of a First Nations biobank for use in genomic health research. Some key ethical considerations that emerged in the consultations were: the need to rebuild trust in research among First Nations communities, the need to incorporate cultural safety and traditional knowledge into all stages of the biobank's development and implementation, the importance of ensuring First Nations ownership and control of the biobank and all research undertaken using its materials, and the need for comprehensive and culturally sensitive consent processes.

Read

Caron, N. R., Adam, W., Anderson, K., Boswell, B. T., Chongo, M., Deineko, V., Dick, A., Hall, S. E., Hatcher, J. T., Howard, P., Hunt, M., Linn, K., & O'Neill, A. (2023). Partnering with First Nations in Northern British Columbia Canada to reduce inequity in access to genomic research. *International Journal of Environmental Research and Public Health, 20*(10), 5783-. https://doi.org/10.3390/ijerph20105783

OPEN ACCESS ONLINE

Other important work being done to protect the rights and interests of Indigenous Peoples include the Silent Genomes Project (https://www.bcchr.ca/silent-genomes-project)is a collaborative effort involving various partners, including the First Nations, Inuit, and Métis communities, and is led by experts in the field of genomics and Indigenous health. It is aimed at reducing health care disparities, and improving diagnostic success and health outcomes for Indigenous children in Canada with genetic diseases. Some key aspects of the project include addressing health inequities, Indigenous governance, creating an Indigenous biobank and variant library, and providing culturally safe genomic testing.

Concept in Action: First Nations Principles of OCAP™

Research with Indigenous Peoples should follow the principles of OCAP® which "asserts that First Nations alone have control over data collection processes in their communities, and that they own and control how this information can be stored, interpreted, used, or shared" (First Nations Information Governance Centre, 2024, para. 2).

Watch Understanding the First Nations Principles of OCAP™ : Our Road Map to Information Governance (6 min) on YouTube (https://youtu.be/y32aUFVfCM0) to learn about these principles.

Image Description

5.3 HeLa Timeline: Henrietta Lacks

The world owes much to Henrietta Lacks. Henrietta Lacks was an African American woman whose cells were removed during a biopsy in 1951 – and used for research without her knowledge or approval. A few months after Henrietta's diagnosis of cervical cancer, she died at the age of 31 years old. She would never know that more than six decades later, her cells would continue to grow and provide a foundation for advancements in science and medicine.

Henrietta's cells revolutionized the field of medicine. Her amazing and immortal cells (commonly known as HeLa cells) have been used for decades in biomedical research – to study cancer, the effects of radiation, and AIDS – among many other areas. Her cells led to the development of successful drugs in fighting human diseases, such as leukemia, hemophilia, herpes, human papillomavirus (HPV), Parkinson's disease, and influenza, among others.

HeLa Timeline

- 1920: Henrietta Lacks was born Loretta Pleasant on August 1, 1920 in Roanoke, Virginia to Eliza and Johnny Pleasant.
- 1941: On April 10, 1941, Henrietta Pleasant married David "Day" Lacks.
- 1951: A biopsy of Henrietta Lacks' tumour was taken and sent to the lab of Dr. George Gey resulting in the creation of the HeLa cell line.
- 1952: Scientists used HeLa cells to help develop the polio vaccine.
- 1973: Scientists used HeLa cells to study the behavior of samonella inside human cells.

- 1984: HeLa cells were used by a German virologist to help prove that the human papillomavirus (HPV) causes cancer.
- 1986: The virus infection mechanism of HIV w2as studied by scientists who infected HeLa cells with HIV.
- 1993: HeLa cells were used to study tuberculosis.
- 2013: On August 6, 2013, the NIH announced an agreement with the family of Henrietta Lacks to allow biomedical researchers controlled access to the whole genome data of HeLa cells. [Back to Fig. 5.3]

Attribution & References

- Diversity in Genomic Research section is adapted from Diversity in Genomic Research, Courtesy: National Human Genome Research Institute, Public Domain with Attribution
- HeLa Cells: A Lasting Contribution to Biomedical Research is reused from the NIH Office of Science Policy, Public Domain, with additional content added in (videos)

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First Nations Information Governance Centre. (2024). *The First Nations principles of OCAP®*. https://fnigc.ca/ocap-training/

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Skloot, R. (2010). The immortal life of Henrietta Lacks. Crown Publishers. https://search.worldcat.org/title/The-immortal-life-of-Henrietta-Lacks/oclc/326529053

5.4 RESEARCH PRIORITIES AND FUNDING

Research Funding in Canada

The Canadian Institutes of Health Research (CIHR) is Canada's primary federal agency responsible for funding health research. Within it, there are 13 virtual institutes, each focusing on specific areas of health research, such as cancer, genetics, and indigenous health (CIHR, 2021; CIHR, 2022). These institutes bring together researchers, health professionals, and policymakers to collaborate on improving the health of Canadians through innovative research and knowledge translation (CIHR, 2022).

Another important source of research funding for genomics come from Genome Canada [PDF] (https://genomecanada.ca/wp-content/uploads/2023/02/Genome-Canada-Corporate-Plan-2023-24-EN-Accessible-Version.pdf). The organization supports genomics research impact areas of health, climate and environment, and food and agriculture.

Setting Research Priorities

Although there are many excellent research questions, funding research is competitive. How can a researcher know what topics have the best chance of being funded? Competitions for research grants are established through the various funding agencies and organizations, each with specific mandates and priorities. Major funding bodies like the Canadian Institutes of Health Research (CIHR), the Natural Sciences and Engineering Research Council (NSERC), and the Social Sciences and Humanities Research Council (SSHRC) regularly announce calls for proposals. These calls outline the eligibility criteria, application guidelines, and evaluation processes for different types of research projects.

The CIHR has an Institute of Genetics (CIHR-IG) (https://cihr-irsc.gc.ca/e/13147.html) which facilitates research on human and model genomes, encompassing all areas of genetics related to health and disease. This includes translating research findings into health policies and practices, as well as addressing the societal impacts of genetic discoveries. The Institute's strategic plan titled *Sequencing our Future (2022-2027) [PDF]* (https://cihr-irsc.gc.ca/e/documents/ig-strat-plan-2022-2027-en.pdf). This document describes the Institute's strategic priorities for genomic research. Funding for research is often prioritized based on these types of documents which outline key research priorities and goals to guide funding decisions to areas that promise the most significant impacts.

Priorities for nursing genomics research are determined through a combination of national health priorities, emerging scientific evidence, and the needs identified by healthcare professionals and patients.

Additionally, professional organizations and academic institutions play a significant role in setting research agendas and securing funding to advance the field of nursing genomics.

Attribution & References

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Canadian Institutes of Health Research (CIHR). (2021). *CIHR Strategic Plan 2021-2031*. https://cihr-irsc.gc.ca/e/52331.html

Canadian Institutes of Health Research (CIHR). (2022). *Annual Report 2021-22*. https://cihr-irsc.gc.ca/e/53108.html

5.5 KNOWLEDGE TRANSLATION AND MOBILIZATION

Knowledge Translation and Mobilization

"**Knowledge translation (KT)** is defined as a dynamic and iterative process that includes synthesis, dissemination, exchange and ethically-sound application of knowledge to improve the health of Canadians, provide more effective health services and products, and strengthen the health care system" (CHIR, 2010, para. 4).

Download the Government of Canada *Knowledge Translation Planner (https://www.canada.ca/en/health-canada/corporate/about-health-canada/reports-publications/grants-contributions/knowledge-transfer-planner.html#section2.4).*

"**Knowledge mobilization (KM)** is an umbrella term encompassing a wide range of activities relating to the production and use of research results, including knowledge synthesis, dissemination, transfer, exchange, and co-creation or co-production by researchers and knowledge users" (SSHRC, 2023, para 4.).

Examples of KM outputs include, but are not limited to:

- Toolkits
- Websites
- Policy briefs
- Journal articles
- Videos

While both concepts aim to bridge the gap between research and practice, knowledge translation is more focused on the application of research in specific fields like healthcare, whereas knowledge mobilization emphasizes a collaborative and inclusive approach across various disciplines.
Research Impact Canada (RIC) has created an excellent online learning resource, KMb101: Introduction to Knowledge Mobilization (https://rise.articulate.com/share/qV54-kftJACqH_QXUcaMODQ3W9qDw3-Y#/). This is a free short course containing 8 modules and numerous useful resources. Have a brief look and file this away for when you next need to plan for knowledge mobilization. RIC also has an excellent learning module on Infographic Design for Knowledge Mobilization (https://rise.articulate.com/share/sUG_m7CTkldQ6phbVlUqV45evvihETL4#/).

Examples of Knowledge Translation and Mobilization in Nursing and Genomics

Examples of knowledge mobilization in genomics for nurses include these excellent resources which will enhance your practice. Bookmark or save these for future reference. It is not required to read these in detail now though readers may wish to explore them in brief.

Online Resources

1. The Genetics and Genomics Toolkit for Canadian Nurses: (https://genomicstoolkit.my.canva.site/) A resource developed to enhance the genomic literacy of Canadian nurses, providing essential knowledge and tools for integrating genomics into nursing practice.
2. Canadian Nurses and Genomics: (https://www.nursingandgenomics.com/) An initiative aimed at supporting Canadian nurses in developing genomic literacy and integrating genomics into their practice to improve patient care and health outcomes.
3. Linkage: (https://linkage.trubox.ca/) an online knowledge engagement hub which includes educational content about foundational genomic concepts in the context of nursing practice. There are opportunities for nurses to learn about how health, genes, and nursing care are connected.
4. Oncology Nursing Society Genetics and Genomics: (https://www.ons.org/taxonomy/term/876) A professional organization dedicated to advancing excellence in oncology nursing through education, research, and advocacy, supporting over 35,000 members in providing high-quality cancer care.
5. Many of the NGHRI resources for nurses in genomics are the result of knowledge mobilization from nurse scientists. The Talking Glossary (https://www.genome.gov/genetics-glossary) that definitions are drawn from at the start of each unit in this book is one example. The Method for Introducing a New Competency (MINC) (https://www.genome.gov/minc) is a toolkit for healthcare professional that is the output of an implementation study.

Policies Examples

These have already been introduced but the competency documents for nurses working with genomics are examples of evidence-based policies developed by nurses.

- NHS. (2023). Genomic competency framework for UK nurses [PDF] (https://www.genomicseducation.hee.nhs.uk/wp-content/uploads/2023/12/2023-Genomic-Competency-Framework-for-UK-Nurses.pdf).
- ANA. (2023). Essentials of genomic nursing: Competencies and outcome indicators (https://www.nursingworld.org/nurses-books/ana-books/ebook-essentials-of-genomic-nursing-competencies-/) (3rd ed.).

Journal Articles Featuring Nursing Genomics Knowledge Mobilization

This paper discusses how the new regulatory model for nurse practitioners in Canada can be optimized to better integrate genomics into healthcare, highlighting the potential benefits and challenges of this integration:

> Acorn, M., Chiu, P., Limoges, J., & **Gretchev, A**. (2024). Optimizing the new model of nurse practitioner regulation in Canada to support the integration of genomics. *Canadian Journal of Nursing Leadership, 37*(2), 49-56. https://doi.org/10.12927/cjnl.2024.27468

This case study explores the collaborative efforts across Canada to advance nursing practices through genomics, emphasizing the importance of nationwide cooperation and shared learning to enhance healthcare outcomes:

> Chiu, P., **Gretchev, A.**, Limoges, J., Puddester, R., Carlsson, L., Pike, A., Leslie, K., Dordunoo, D. (2024). Fostering pan-Canadian collaboration to advance new nursing practice: A case study from the genomics experience. *Canadian Journal of Nursing Leadership, 37*(2), 41.48. https://doi.org/10.12927/cjnl.2024.27470

Genomics Medicine Professional Organizations

There are several organizations that foster collaboration, networking, research, policy development, and professional development for nurses working in genomics. Many of these organizations are interdisciplinary. Many of these organizations offer discounts or free membership for students. These are another excellent avenue for knowledge translation and mobilization.

Nursing Specific

- ISONG (https://www.isong.org/memberbenefits) – International Society of Nurses in Genetics
- G2NA (https://g2na.org/index.php?option=com_content&view=article&layout=edit&id=47) – Global Genomics Nursing Alliance
- CANO (https://www.cano-acio.ca/page/member_benefits) – Canadian Association of Nurses in Oncology – has a special interest group for genomics oncology nursing

Interdisciplinary

- DOHAD – (https://dohadsoc.org/join-us/) International and local chapters
- ACMG (https://www.acmg.net/ACMG/Membership/ACMG_Membership.aspx?hkey=1827209f-0d45-4c96-8771-267c281ac3a1&WebsiteKey=6e814a8c-3077-4552-ba39-f7fcacff42d6) – American College of Medical Genetics
- ASHG (https://www.ashg.org/membership/member-benefits/) – American Society of Human

Genetics

Attribution & References

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Canadian Institutes of Health Research (CHIR). (2010). *About us: Knowledge translation*. https://www.cihr-irsc.gc.ca/e/29418.html

Social Sciences and Humanities Research Council (SSHRC). (2023, November 24). *Guidelines for effective knowledge mobilization*. https://www.sshrc-crsh.gc.ca/funding-financement/policies-politiques/knowledge_mobilisation-mobilisation_des_connaissances-eng.aspx#a1

5.6 SCHOLARLY POSTERS

Scholarly Posters

Scholarly posters are an effective form of knowledge translation, serving as a visual and concise medium to communicate research findings to a broad audience. They distill complex information into accessible formats, using visuals like charts, graphs, and images to enhance understanding and retention. At academic conferences, posters facilitate direct interaction between researchers and attendees, fostering discussions that can clarify and expand on the presented data. This interactive element not only aids in the dissemination of new knowledge but also encourages feedback and collaboration, further advancing the research field. By making research more approachable and engaging, scholarly posters play a crucial role in bridging the gap between scientific discovery and practical application.

Assignment – Scholarly Poster

See the Blackboard course shell for resources to develop a scholarly poster.

Scholarly poster assignment guidelines and due dates can be located in Blackboard under Assessments.

Attribution

5.7 UNIT SUMMARY AND REVIEW

Key Takeaways

Nurses are integral to advancing genomic research, leveraging their expertise to bridge the gap between complex scientific discoveries and patient care. Through roles in direct research, data management, and ethical oversight, nurses contribute significantly to the genomics field. Studies using genomic approaches, such as genome-wide association studies (GWAS), have highlighted the potential of genomics to improve clinical practice, policy, education, and healthcare systems. However, the integration of genomic findings into clinical settings remains an ongoing challenge.

Diversity and ethical considerations are critical considerations in genomics research. Historically, studies have overrepresented populations of European ancestry, creating knowledge gaps about diverse genomic variants and their impact on health outcomes. Efforts such as the Human Pangenome Reference Program and initiatives like the NIH's *All of Us* Research Program aim to address these disparities by including underrepresented populations. Additionally, ethical frameworks, such as Canada's Tri-Council Policy Statement (TCPS), guide the conduct of human genetic research, emphasizing informed consent, privacy, and inclusivity. Addressing diversity challenges requires building trust with communities and ensuring equitable participation, which enhances the global applicability and fairness of genomic medicine.

Research funding in Canada is primarily supported by organizations such as the Canadian Institutes of Health Research (CIHR) and Genome Canada. CIHR operates through 13 virtual institutes focused on areas such as genetics, cancer, and Indigenous health, promoting collaboration among researchers, health professionals, and policymakers. Genome Canada funds genomics research in areas like health, climate, and food systems. Setting research priorities involves understanding the mandates and strategic plans of funding agencies. Nursing genomics research priorities are informed by national health needs, emerging evidence, and input from professional and academic organizations.

Knowledge translation (KT) and knowledge mobilization (KM) are integral to bridging research and

practice. KT emphasizes the application of research findings to specific fields, while KM promotes collaborative and inclusive approaches across disciplines. Examples include resources like the *Genetics and Genomics Toolkit for Canadian Nurses* and initiatives supporting genomic literacy among nurses. Policymakers, educators, and researchers play a critical role in developing tools, policies, and competencies to advance nursing genomics. Collaborative efforts among organizations and professionals are vital for integrating genomics into healthcare and nursing practice.

Resources

See **Appendix A** for a list of online resources for genomics.

Additional Optional Readings:

Guidelines for Genomic Data

World Health Organization. (2024, November 20). *Guidance for human genome data collection, access, use and sharing*. https://www.who.int/publications/i/item/9789240102149

Diversity in Genomics Research

1. Fatumo, S., Chikowore, T., Choudhury, A., Ayub, M., Martin, A. R., & Kuchenbaecker, K. (2022). A roadmap to increase diversity in genomic studies. *Nature Medicine, 28*, 243–250. https://doi.org/10.1038/s41591-021-01672-4
2. Koch, L. Global genomic diversity for All of Us. (2024). *Nature Reviews Genetics,25*, 303. https://doi.org/10.1038/s41576-024-00727-9

Developing Nursing Research Priorities

This is an older article which is in the process of being updated. However, this paper provides an excellent example of priority areas of nursing genomics research mapped to the National Institute of Nursing Research Strategic Plan.

1. Genomic Nursing State of the Science Advisory Panel, Calzone, K. A., Jenkins, J., Bakos, A. D., Cashion, A. K., Donaldson, N., Feero, W. G., Feetham, S., Grady, P. A., Hinshaw, A. S., Knebel, A. R.,

Robinson, N., Ropka, M. E., Seibert, D., Stevens, K. R., Tully, L. A., & Webb, J. A. (2013). A blueprint for genomic nursing science. *Journal of Nursing Scholarship*, *45*(1), 96–104. https://doi.org/10.1111/jnu.12007

2. Lee, H., Gill, J., Barr, T., Yun, S., & Kim, H. (2017). Primer in genetics and genomics, Article 2-advancing nursing research with genomic approaches. *Biological Research for Nursing*, *19*(2), 229–239. https://doi.org/10.1177/1099800416689822

Health Disparities

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2. Limoges, J., Chiu, P., Dordunoo, D., Puddester, R., Pike, A., Wonsiak, T., Zakher, B., Carlsson, L., & Mussell, J. K. (2024). Nursing strategies to address health disparities in genomics-informed care: a scoping review. *JBI Evidence Synthesis*, *22*(11), 2267–2312. https://doi.org/10.11124/JBIES-24-00009

Public Trust and Ethics Review

1. Samuel, G.N., & Farsides, B. (2018). Public trust and 'ethics review' as a commodity: the case of Genomics England Limited and the UK's 100,000 genomes project. *Medicine, Health Care and Philosophy,* 21, 159–168. https://doi.org/10.1007/s11019-017-9810-1
2. Milne, R., Morley, K. I., Almarri, M. A., Anwer, S., Atutornu, J., Baranova, E. E., Bevan, P., Cerezo, M., Cong, Y., Costa, A., Critchley, C., Fernow, J., Goodhand, P., Hasan, Q., Hibino, A., Houeland, G., Howard, H. C., Hussain, S. Z., Malmgren, C. I., ... Middleton, A. (2021). Demonstrating trustworthiness when collecting and sharing genomic data: public views across 22 countries. *Genome Medicine, 13*(1), 92–92. https://doi.org/10.1186/s13073-021-00903-0

Symptom Science

1. Hickey, K. T., Bakken, S., Byrne, M. W., Bailey, D. E., Demiris, G., Docherty, S. L., Dorsey, S. G., Guthrie, B. J., Heitkemper, M. M., Jacelon, C. S., Kelechi, T. J., Moore, S. M., Redeker, N. S., Renn, C. L., Resnick, B., Starkweather, A., Thompson, H., Ward, T. M., McCloskey, D. J., Austin, J. K., & Grady, P. A. (2019). Precision health: Advancing symptom and self-management science. *Nursing Outlook, 67*(4), 462-475. https://doi.org/10.1016/j.outlook.2019.01.003.

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1. Chiu, P., Limoges, J., Pike, A., Calzone, K., Tonkin, E., Puddester, R., Gretchev, A., Dewell, S., Newton, L., & Leslie, K. (2024). Integrating genomics into Canadian oncology nursing policy: Insights from a comparative policy analysis. *Journal of Advanced Nursing, 0*(0), 1–22. https://doi.org/10.1111/jan.16099
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Attribution & References

Key takeaways generated using ChatGPT. Prompt: “summarize this text in a few sentences, ignoring images, captions, citations and web references.” The output was then edited by Andrea Gretchev.

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