UNIT 1 - INTRODUCTION TO PRECISION HEALTHCARE AND THE ROLE OF THE NURSE

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 1 Contents

Attribution

1.1 UNIT OVERVIEW

Learning Objectives

- Define common genomic terms.
- Establish the relevance of genomics for nursing and the need for nurses to become literate in this emerging knowledge form.
- Describe nurses' distinct and overlapping contributions to the interdisciplinary team delivering genomics-informed care.
- Identify policies to guide genomics-informed nursing practice.
- Explore professional organizations for nurses interested in genomics.

Outline

Topics covered in this chapter include:

- Genetics and genomics introduction
- The contributions of nursing professionals in genomics healthcare

Competencies Nurses will Develop in this Chapter

ANA (2023):

Nursing assessment: Applying/integrating genomic knowledge:

- Demonstrates an understanding of the relationship of genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.

Provision of education, care, and support:

- Advocates for autonomous, informed genomic-related decision-making.
- Provides genomic health care in collaboration with interdisciplinary professionals and when possible clients and their families.

NHS (2023):

Examine your own competency of practice on a regular basis:

- recognizing areas where professional development related to genomics would be beneficial;
- 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; and
- based on an understanding of the boundaries of your professional role in delivering genomic healthcare, including referral, provision, or follow-up of genomic services.

Provide ongoing nursing care and support to patients, caregivers, families and communities with genomic healthcare needs:

- working in partnership with family members, multidisciplinary teams, and other agencies in the management of conditions.

Key terminology

Family history

A family health history is a record of the diseases and health conditions of an individual and that person's biological family members, both living and deceased. A family history can help determine whether someone has an increased genetic risk of having or developing certain diseases, disorders or conditions. It is often recorded by drawing a pedigree (a family tree) that illustrates the relationships among individuals.

Gene

The gene is considered the basic unit of inheritance. Genes are passed from parents to offspring and contain the information needed to specify physical and biological traits. Most genes code for specific proteins, or segments of proteins, which have differing functions within the body. Humans have approximately 20,000 protein-coding genes.

Genetics

Genetics is the branch of biology concerned with the study of inheritance, including the interplay of genes, DNA variation and their interactions with environmental factors.

Genome

The genome is the entire set of DNA instructions found in a cell. In humans, the genome consists of 23 pairs of chromosomes located in the cell's nucleus, as well as a small chromosome in the cell's mitochondria. A genome contains all the information needed for an individual to develop and function.

Genomics

Genomics is a field of biology focused on studying all the DNA of an organism — that is, its genome. Such work includes identifying and characterizing all the genes and functional elements in an organism's genome as well as how they interact.

Human Genome Project

The Human Genome Project was a large international, collaborative effort that mapped and sequenced the human genome for the first time. Conducted from 1990 to 2003, the project was historic in its scope and scale as well as its groundbreaking approach for the free release of genomic data well ahead of publication, leading to a new ethos for data sharing in biomedical research.

Inherited

Inherited, as related to genetics, refers to a trait or variants encoded in DNA and passed from parent to offspring during reproduction. Inheritance is determined by the rules of Mendelian genetics.

Precision medicine (also referred to as precision healthcare/ medicine or personalized medicine)

Precision medicine (generally considered analogous to personalized medicine or individualized medicine) is an innovative approach that uses information about an individual's genomic, environmental and lifestyle information to guide decisions related to their medical management. The goal of precision medicine is to provide more a precise approach for the prevention, diagnosis and treatment of disease.

Protein

Proteins are large, complex molecules that play many important roles in the body. They are critical to most of the work done by cells and are required for the structure, function and regulation of the body's tissues and organs. A protein is made up of one or more long, folded chains of amino acids (each called a polypeptide), whose sequences are determined by the DNA sequence of the protein-encoding gene.

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.

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). (n.d.). *Talking glossary of genetic and genomic terms*. www.genome.gov

1.2 GENETICS AND GENOMICS INTRODUCTION

Understanding Genomics

Genetics and Genomics – What's the Difference?

Genetics and genomics both play roles in health and disease. **Genetics** refers to the study of genes and the way that certain traits or conditions are passed down from one generation to another. **Genomics** describes the study of all of a person's genes (the **genome**).

Genetics is a term that refers to the study of genes and their roles in inheritance – in other words, the way that certain traits or conditions are passed down from one generation to another. Genetics involves scientific studies of genes and their effects. **Genes** (units of heredity) carry the instructions for making **proteins**, which direct the activities of cells and functions of the body. Examples of genetic or **inherited** disorders include cystic fibrosis (See: Learning About Cystic Fibrosis (https://www.genome.gov/10001213/learning-about-cystic-fibrosis/)), Huntington's disease (Learning About Huntington's Disease (https://www.genome.gov/10001215/learning-about-huntingtons-disease/)), and phenylketonuria (PKU) (Learning About Phenylketonuria (https://www.genome.gov/25020037/learning-about-phenylketonuria/)).

Genomics is a more recent term that describes the study of a person's genes (the genome), including interactions of those genes with each other and the person's environment. Genomics includes the scientific study of complex diseases such as heart disease, asthma, diabetes, and cancer because these diseases are typically caused more by a combination of genetic and environmental factors than by individual genes. Genomics offers new possibilities for therapies and treatments for some complex diseases and new diagnostic methods.

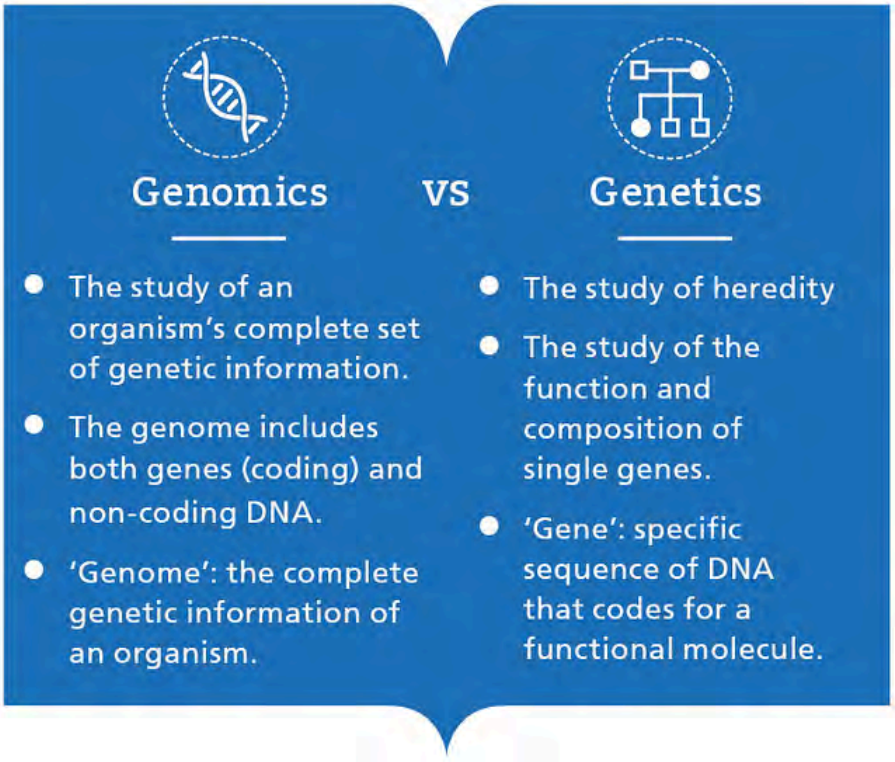

Figure 1.1. Genomics vs. Genetics: Genomics is the study of an organism's complete set of genetic information. The genome includes both genes (coding) and non-coding DNA. 'Genome': the complete genetic information of an organism. Genetics is the study of heredity, the function and composition of single genes. 'Gene': specific sequence of DNA that codes for a functional molecule. **Source:** What is Genomics? by Genomics Education Programme (GEP), CC BY-NC 4.0

Personalized medicine (or healthcare) is an emerging practice that uses an individual's genetic profile to guide decisions made regarding disease prevention, diagnosis, and treatment. Knowing a patient's genetic profile can help doctors select the proper medication or therapy and administer it using the appropriate dose or regimen. Personalized medicine is being advanced through data from the Human Genome Project.

Personalized medicine is a fantastic opportunity to take a "one size fits all" approach to diagnostics, drug therapy, and prevention and turn it into an individualized approach. We all are similar, of course, but we are also different. The idea that medicine would be applied in a fashion that ignores those differences can't be any more correct than going to the shoe store and buying any old pair of shoes without checking the size. Genomics plays a significant role in the emergence of personalized medicine as it gives us a window in a particular molecular way into those differences between us and allows the opportunity for making individual predictions about disease risk that can help somebody choose a prevention plan that is right for them. It also allows the possibility, in some instances, of picking the right drug at the right dose for the right person instead of the "one size fits all" approach to drug therapy. And ultimately, it will be hard to see how any kind of medicine will not be affected by this as we learn more and more about the individual and as many of us find

our complete genomes being sequenced and placed into our medical records to empower that kind of personalized approach. Lots of work to do here, but it may be the biggest revolution in medicine in a very long time.

Precision medicine is a newer term with a similar meaning to the often analogously used "personalized medicine or healthcare." The shift away from the word personalized was in response to concerns over misinterpretation of the word "personalized" to imply that each individual could have uniquely tailored care and treatment. Precision medicine/healthcare takes advantage of large data sets of individuals such as their genome or their entire electronic health record to tailor their healthcare to their unique attributes. "Precision medicine consists of identifying which approaches/treatment will be effective for which patients according to the group to which they belong based on their biological characteristics. In this sense, it is more stratified medicine than personalized medicine" (Delpierre & Lefèvre, 2023). It is common sense that no two individuals are the same, so they should not get the same healthcare. Precision healthcare embodies that simple idea.

Personalized Medicine (text version)
Watch the video Personalized Medicine (5 mins) at BC Campus Media (https://media.bccampus.ca/media/0_78tjylkd)

Pause the video at 4:25 to answer the following question:

True or false? How drugs interact with your unique genetic makeup is a part of personalized medicine.

Check your answer in footnote[1]

Activity source: Concepts of Biology – 1st Canadian Edition, CC BY 4.0

The Human Genome Project

The Human Genome Project (HGP) is one of the most remarkable scientific feats in history. The project was a voyage of biological discovery led by an international group of researchers looking to comprehensively study all of the DNA (known as a genome) of a select set of organisms. Launched in October 1990 and completed in April 2003, the Human Genome Project's signature accomplishment – generating the first sequence of the human genome – provided fundamental information about the human blueprint, which has since accelerated the study of human biology and improved the practice of medicine.

1. True

Checkout this Human Genome Project timeline of events (https://www.genome.gov/human-genome-project/timeline) from the NHGRI

Visit NHGRI (https://www.genome.gov/about-genomics/educational-resources/fact-sheets/human-genome-project) to learn more about the Human Genome Project.

Figure 1.2. Francis Collins, M.D., Ph.D., announces the successful completion of the Human Genome Project. **Source:** Photo by Ernie Branson, NIH, PDM.

Facts About the Human Genome

- Humans have approximately 20,000 genes
- DNA contains 6 billion nucleotides, which amounts to 3 billion base pairs.
- There is a 99.9% similarity in the DNA sequences of humans.
- A human gene may have 1-3 letters that differ from person to person.
- Humans have 23 pairs of chromosomes

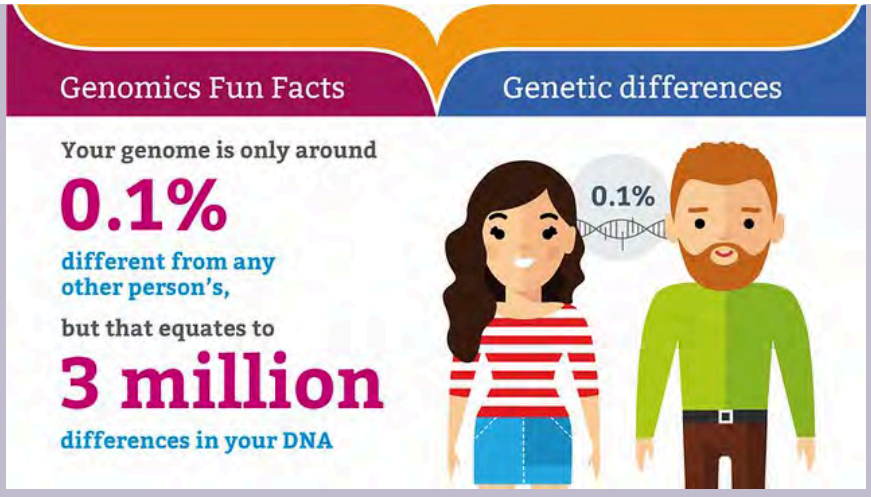

Figure 1.3. Source: Genomics Fun Facts: Genetic Differences by Genomics Education Programme (GEP), CC BY-NC 4.0

Watch History – Making Sense of the Human Genome (4 mins) on YouTube (https://youtu.be/iHuUkb-Mvz8)

Genomics Applications

Why Are Genetics and Genomics Important to Health?

Genetics and genomics both play roles in health and disease. Genetics helps individuals and families learn about how conditions such as sickle cell anemia and cystic fibrosis are inherited in families, what screening and testing options are available, and, for some genetic conditions, what treatments are available.

Genomics is helping researchers discover why some people get sick from certain infections, environmental factors, and behaviours while others do not. For example, some people exercise their whole lives, eat a healthy diet, have regular medical checkups, and die of a heart attack at age 40. Some people smoke, never exercise, eat unhealthy foods and live to be 100. Genomics may hold the key to understanding these differences.

Apart from accidents (such as falls, motor vehicle accidents or poisoning), genomic factors play a role in nine of the ten leading causes of death in the United States (for example, heart disease, cancer and diabetes. See: Leading Causes of Death (http://www.cdc.gov/nchs/FASTATS/lcod.htm)). All human beings are 99.9 percent identical in their genetic makeup. Differences in the remaining 0.1 percent hold important clues about the causes of diseases. Gaining a better understanding of the interactions between genes and the environment using genomics is helping researchers find better ways to improve health and prevent disease,

such as modifying diet and exercise plans to prevent or delay the onset of type 2 diabetes in people who carry genetic predispositions to developing this disease.

Why Are Genetics and Genomics Important to the Health of Families?

Understanding more about diseases caused by a single gene (using genetics) and complex diseases caused by multiple genes and environmental factors (using genomics) can lead to earlier diagnoses, interventions, and targeted treatments. A person's health is influenced by their family history and shared environmental factors. This makes family history an important, personalized tool that can help identify many of the causative factors for conditions with a genetic component. The **family history** can serve as the cornerstone for learning about genetic and genomic conditions in a family and for developing individualized approaches to disease prevention, intervention, and treatment.

Concept in Action

Watch My Genomics Journey: three perspectives (5 mins) on Vimeo (https://vimeo.com/888710908)

15 ways genomics influences our world:

1. DNA sequencing – Advances in genomics are reducing the cost of genome sequencing by a million-fold.
2. Human genomic variation – Genomics is helping us understand what makes each of us different and what makes us the same.
3. Cancer genomics – Genomics transforms how we study, diagnose and treat cancer.
4. Human origins and ancestry – Genomics is illuminating human and family origins at a

previously impossible level.
5. Agriculture – Genomics is empowering farmers to improve the food supply.
6. Genomes at work – We are learning how our genomes serve as blueprints for life.
7. Rare genetic diseases – Genomics is ending diagnostic odysseys for patients with rare diseases.
8. Pharmacogenomics – Genomics is helping us choose the right medication at the right dose for each patient.
9. Noninvasive prenatal genetic testing – Genomics is revolutionizing health assessments before birth.
10. Enhanced forensics – Genomics establishes more robust methods for DNA-based forensic analyses.
11. Microbes and microbiomes – Genomics is advancing the study of individual and communities of microbes.
12. Direct-to-consumer genomic testing – Genomics is helping you access information about your genome from your home.
13. The natural world – Genomics helps us understand evolution and protect our biological ecosystems.
14. Genome editing – Genomics alters a genome with unparalleled efficiency and precision.
15. Social context – Genomics fosters an appreciation of what our DNA means for our health, identities and culture.

To view this source and for more details on these applications, visit NHGRI. (https://www.genome.gov/dna-day/15-ways)

Genomic Literacy

What exactly is meant by "genomic literacy?"

Watch the short animation Canadian Nursing and Genomics: Whiteboard Video 1 (1 minute) on YouTube (https://youtu.be/4yYfMNE39JE)

Evidence Informing the Definition of Genomic Literacy

Boerwinkel et al. (2017) conducted a Delphi study to achieve expert consensus on the essential knowledge required for informed decision-making in genomics. Genomic literacy is necessary not only for healthcare professionals delivering genomic services but also for patients receiving them. The authors classified this knowledge into three categories:

> Conceptual knowledge: knowledge of genetic concepts;
> Sociocultural knowledge: knowledge of how applications of genetic technologies are used in societal activities and in what ways they influence human lives;
> Epistemic knowledge: knowledge of the meaning of genetic information. This concerns the knowledge needed to interpret genetic information from different sources and how to use these in argument and decision-making. This knowledge includes Nature of Science aspects such as the certainty and uncertainty of genetic information and how genetic concepts have evolved (Boerwinkel et al., 2017, p.1106).

How Can Genomic Literacy Assist Nurses in Addressing the Social, Ethical, and Equity Issues Associated with Nursing?

Watch this short animation: Canadian Nursing and Genomics Whiteboard Video 4 (2 mins) from YouTube (https://youtu.be/IFo8IgltQDY)

Attribution & References

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- Personalized Medicine, Courtesy of: National Human Genome Research Institute, Public Domain with attribution
- Precision Medicine, Courtesy of: National Human Genome Research Institute, Public Domain with attribution
- 15 Ways Genomics Influences Our World, Courtesy of: National Human Genome Research Institute, Public Domain with attribution
- H5P/video from 10.3 Genomics and Proteomics In *Concepts of Biology – 1st Canadian Edition* by Charles Molnar and Jane Gair, CC BY 4.0

Adaptations include combining sources, minor grammatical changes, and addition of quotes, videos and commentary to improve student understanding.

References

Boerwinkel, D.J., Yarden, A. & Waarlo, A.J. (2017). Reaching a consensus on the definition of genetic literacy that is required from a twenty-first-century citizen. *Science & Education, 26*, 1087–1114. https://doi.org/10.1007/s11191-017-9934-y

Delpierre, C., & Lefèvre, T. (2023). Precision and personalized medicine: What their current definition says and silences about the model of health they promote. Implication for the development of personalized health. *Frontiers in Sociology*, *8*, 1112159. https://doi.org/10.3389/fsoc.2023.1112159

1.3 THE CONTRIBUTIONS OF NURSING PROFESSIONALS IN GENOMICS HEALTHCARE

There is a wide variety of ways that healthcare professionals can contribute to providing genomic services. As technologies advance and genomics becomes increasingly integrated into routine practices, new roles will emerge.

Visit the NHS Genomics Education Programme website (from the United Kingdom) – Careers in Genomics (https://www.genomicseducation.hee.nhs.uk/careers/), to learn more about some of the interdisciplinary team members that nurses will work with.

To learn more about different practice contexts and medical specialties, visit the Genomics Education Programme website's section on Genomics in Healthcare.

Nurses' Contributions to Genomics

Watch this short video on the relevance of genomics to nursing practice. This video briefly overviews some critical takeaways nurses will gain from reading this book.

Watch Nursing in the Genomic Era (3 mins) on YouTube (https://youtu.be/LaHSMJGUco8)

Why should nurses understand basic genomic principles?

Genomics plays a role in every aspect of nursing, which includes providing one-on-one care, treating entire populations, teaching the next generation of nurses, or making discoveries in the lab.

Genomic principles form the foundation for disease pathophysiology

- Genomics underlies all diseases
- Genomics contributes to our understanding of the mechanisms of disease

Genomics principles influence clinical actions

- Genomics knowledge supports clinical practice and potential interventions (e.g., newborn screening tests).

- Obtaining genomic information about a patient can change recommendations for prevention and screening (e.g., recommendations for annual MRI with elevated breast cancer risk).
- Genomics affects pharmacokinetics and pharmacodynamics, affecting drug selection and dose (e.g., antidepressants, anticoagulants).

Understanding genomic principles facilitates communication with patients and family members and supports genomics-informed care

- Patients often need help understanding how genomics influences their health and the health of their family members.
- Effective patient and family education requires foundational knowledge of genomics.
- Nurses can help patients understand how genomics affects disease risk.
- Ethical nursing care includes understanding the ethical considerations of genomic healthcare (e.g., right not to know).

Figure 1.4 Nursing in the Genomic Era. Nurses communicate by explaining processes and tests, listening for clinical clues, advising and signposting. Nurses are hands on: Taking samples, administering drugs, managing infection control. Nurses collaborate: Leading and guiding others, escalating concerns, representing patients at meetings. **Source:** Nursing in the Genomic Era by Genomics Education Programme, CC BY 2.0.

In What Settings Can Genomics be Used?

Genomics is used in all practice settings across all aspects of nursing care, including educating patients and families, administering medications, advocating for health promotion and disease prevention and interpreting family histories.

While all nurses will encounter genomics at some point in their practice, there are particular areas where genomics is highly relevant, such as oncology, pediatrics, and maternity. As new genomics applications are developed, these areas are likely to increase.

Rare disease

Nurses who care for patients with rare diseases support these families and help them through the health and social care system. Some nurses specialize in a particular rare condition, such as:

- Familial hypercholesterolemia: These roles usually involve identifying and supporting affected families. You can read more about this work here: North East and Cumbria [PDF]. (http://www.ahsn-nenc.org.uk/wp-content/uploads/2018/07/The-role-of-the-FH-Nurse-.pdf)
- Monogenic diabetes: Specialist genetic diabetes nurses (https://www.genomicseducation.hee.nhs.uk/about-us/training-a-network-of-genetic-diabetes-nurses/) support patients with this rare form of diabetes and their families and increase recognition and diagnosis of monogenic diabetes among healthcare professionals. More information can be found on the Diabetes Genes website. (https://www.diabetesgenes.org/training/genetic-diabetes-nurses/)

Advanced practice settings

Nurses working as experts in a particular clinical area, such as oncology, cardiology or pediatrics, will increasingly be trained to manage patients with inherited genetic conditions and targeted treatments. Keeping abreast of rapid developments in genomics is a vital part of the role of many specialisms. Here is an example of the work that a clinical nurse specialist with expertise in genomics may do [PDF] (https://www.thinkkidneys.nhs.uk/kquip/wp-content/uploads/sites/5/2017/12/FamilyHistoryOfGeneticRenalDiseases.pdf), with a special focus on renal medicine.

As new initiatives are developed, it is often appropriate for advanced practice and specialist nurses to become involved with service development to help meet patient needs and address patient demands, aiming to improve patient outcomes. Advanced practice and specialist nurses are uniquely situated to provide these services.

Research

Although advances in genomics have been rapid and impressive, we still have a long way to go. Ongoing research is vital to ensure that all patients can benefit. There are many opportunities to be involved with research projects as a nurse, and you may choose to specialize in this area. The work may include developing hypotheses, recruiting patients to research projects, training other staff members, collecting and analyzing data, and caring for and communicating with those involved. Nursing research in genomics will be explored further in a subsequent unit.

How Can Nurses Apply Genomics to Practice?

- Record and interpret a comprehensive family history to identify risk for heritable genetic conditions.
- Understand the genomic basis for specific health conditions and the associated prognosis.
- Recognize newborn risk for morbidity and mortality based on identified genetic conditions.
- Identify risk for asymptomatic individuals with high risk for hereditary cancers.
- Advocate for interventions based on individual genetic needs.
- Facilitate screening and follow-up for individuals with known genetic conditions.
- Encourage genetic testing for at-risk family members.
- Obtain and verify informed consent.
- Ensure understanding of all risks, benefits, and limitations of genetic testing and research.
- Use current credible information to answer questions about genomic information or services and assess for understanding.
- Identify at-risk populations.
- Form empathetic relationships to support individuals and families at risk for or affected by genetic conditions.
- Educate individuals, families, and communities about the role of genomics in medication response.
- Discuss and educate about the function of the new genomics-based vaccine.

How Are Nurses Prepared and Positioned to Provide Genomics-Informed Care?

Nurses practice from a holistic viewpoint. Nurses consider patients, families and communities within their contexts.

- Genomics must be considered in context, including patients' genomic information, clinical presentation and the environments in which they live.
- Nurses consider ethical, legal and social issues for individuals and families.

Nurses have a central position in the healthcare team and interact the most with patients.

- Nurses are central to interdisciplinary communication and coordination.
- Nurses translate and relay the concerns of patients and family members to the healthcare team.

Nurses have an optimal skill set for applying genomics in all practice settings and roles. Nurses apply genomics principles at the bedside, in the community, in policy and research.

- Nurses are skilled at obtaining individual and family histories to provide critical information for genomics-informed care.
- Nurses obtain and analyze data from physical assessments, individual and family histories, laboratory tests and other sources.
- Nurses plan and implement interventions central to genomics-informed care.
- Nurses champion patient education and translate complex topics into lay terms.
- Nurses are ethical practitioners prepared to advocate for patient needs.

Concept in Action

This podcast (https://www.genomicseducation.hee.nhs.uk/education/podcasts/genomics-conversation-genomics-in-nursing-with-dr-christine-patch/)(approx 5min) features Dr Christine Patch, Clinical Lead for Genetic Counselling at Genomics England and the new Chair of the Global Genomics Nursing Alliance, also known as 'G2NA'. Christine talks about the influence of genomics on nursing practice and how nurses need to be aware of its potential in patient care. She also talks about the work of the G2NA and other international collaborations to raise the genomics profile. She discusses the need for genomics to become an established part of nursing training.

Source: Health Education England's Genomics Education Programme (GEP)

Read

Dewell, S., Benzies, K., & Ginn, C. (2020). Precision health and nursing: Seeing the familiar in the foreign. *The Canadian Journal of Nursing Research*, *52*(3), 199–208. https://doi.org/10.1177/0844562120945159

(https://georgian.primo.exlibrisgroup.com/permalink/01OCLS_GEORG/21p491/cdi_proquest_miscellaneous_2440663648) 

Fu, M. R., Kurnat-Thoma, E., Starkweather, A., Henderson, W. A., Cashion, A. K., Williams, J. K., Katapodi, M. C., Reuter-Rice, K., Hickey, K. T., Barcelona de Mendoza, V., Calzone, K., Conley, Y. P., Anderson, C. M., Lyon, D. E., Weaver, M. T., Shiao, P. K., Constantino, E. E., Wung, S., Hammer, M. J., Voss, J. G., Coleman, B. (2020). Precision health: A nursing perspective, International Journal of Nursing Sciences, 7(1), 5-12. https://doi.org/10.1016/j.ijnss.2019.12.008.

(https://doi.org/10.1016/j.ijnss.2019.12.008)

Genomics-Informed Nursing Policies

Nurses typically rely on policy to guide practice. Examples of nursing policies include professional or practice standards, scope of practice, competencies, practice guidelines, and educational frameworks. Currently, in Canada, there are no policies to guide genomics-informed nursing (Puddester et al., 2023). The United States and the UK have modelled the way about interdisciplinary genomics integration. These countries have developed evidence-based competencies for nurses delivering genomic services. One aspect of preparing the health system for genomics integration is developing policies for healthcare professionals, including nurses, to delineate roles and responsibilities and foster the development of genomic literacy.

Briefly review the following documents. As you progress through each chapter in this book, consider the competencies you are beginning to develop. The unit overview sections will list the competencies developed in each unit.

- NHS. (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).
- 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.).

Note: the ANA website will request some personal information, including an email address, to distribute the competency document.

Genomics Organizations for Nurses and Other Healthcare Professionals

The following organizations offer member benefits, including continuing professional development, practice resources, conferences, journals, scholarships, grants, and more. These organizations also offer opportunities to provide policy input or for nurses to get involved in governance.

Most of them offer **student discounts** for membership and conference attendance.

- The International Society of Nurses in Genetics (ISONG (https://www.isong.org/))
- Global Genomics Nursing Alliance (G2NA (https://g2na.org/))
- The Canadian College of Medical Genetics (CCMG (https://cihr-irsc.gc.ca/e/13147.html))
- The American College of Medical Genetics (ACMG (https://www.acmg.net/))
- The American Society of Human Genetics (ASHG (https://www.ashg.org/))
- Oncology Nursing Society (ONS (https://www.ons.org/)) – not specific to genomics but has excellent education resources on cancer genetics.

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Puddester, R., Limoges, J., Dewell, S., Maddigan, J., Carlsson, L., & Pike, A. (2023). The Canadian landscape of genetics and genomics in nursing: A policy document analysis. *The Canadian Journal of Nursing Research*, *55*(4), 494–509. https://doi.org/10.1177/08445621231159164

1.4 UNIT SUMMARY AND REVIEW

Key Takeaways

This chapter introduced genomics as a field of study with a related yet distinct focus from genetics. Some key terminology was provided that will be utilized throughout the book. The completion of the Human Genome Project in 2003 provided fundamental information about the human blueprint and was a launching point for the acceleration of genomic sciences. As the use of genomic technologies becomes more commonplace in healthcare settings, nurses need genomic literacy to be able to provide safe and equitable care. While all nurses will encounter genomics at some point in their practice, certain roles require more specialized knowledge. It is also likely that as genomics is advanced in healthcare system, new roles for nurses will be created to meet workforce demands. The Canadian healthcare system has more work to do to be ready for the complete adoption of genomics. One area where nurses can advocate for change relates to developing policy infrastructure to guide genomics-informed nursing practice, education, and research. Finally, nurses are encouraged to get involved in genomics organizations in order to have a voice in how genomics is integrated into the nursing profession and the healthcare system to benefit patient care.

Additional Optional Readings:

Although this is an older article, it has an excellent summary table. Review p.171, Table 1 – Standards in Genetics and Genomics for General Nursing Practice.

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Discover how nurses and nursing leaders across Canada are working to accelerate the integration of genomics in practice, education, and research.

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