

Precision Healthcare

Genomics-Informed Nursing

Created by Andrea Gretchev, RN, MN, CCNE

PRECISION HEALTHCARE: GENOMICS-INFORMED NURSING

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ACCESSING AND USING PERSONALIZED HEALTH IN NURSING: GENOMICS-INFORMED PRACTICE

Welcome to Personalized Health in Nursing: Genomics-Informed Practice

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- Unit 9
- Unit 10
- Unit 11
- Unit 12
- Unit 13

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- All images in this guide are described fully in the text, alt-tag or in an image description section for complex images.
- Information is not conveyed by colour alone.
- Pressbooks has built in features such as the ability to change font size.

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This statement was last updated on November 30, 2024

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I am so grateful for the work that Jen Booth has done to ensure this book appears professional and adheres to best practices in copyright and accessibility. Georgian College is fortunate to have someone with her knowledge and expertise.

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Source Authors

This book is primarily a curation of open access materials on genetics and genomics. I would like to express my sincere gratitude to the original source authors for creating this important and timely content and for making it openly accessible.

Book Cover

- Cover created by Andrea Gretchev, using Canva (<https://www.canva.com/>)

I am grateful to Georgian College, Dr. Sara Lankshear, Associate Dean – Nursing programs, and the faculty in the Honours Bachelor of Science – Nursing (HBSN) Program for having the foresight to include genomics in their undergraduate nursing curriculum. Their innovation in curriculum development and the creation of open access resources is leading the way for increasing nursing genomic literacy in Canada and globally.

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This OER was first published on **January 10, 2024**

This OER, *Personalized Health in Nursing: Genomics-Informed Practice*, is a collection of resources adapted by Andrea Gretchev to meet the needs of students enrolled in the Georgian College Honours Bachelor of Science – Nursing (HBSN) Program, NURS 4001: Precision Healthcare: Genomics-Informed Nursing course. In most sections of this OER, updates have been made to the existing content to improve usability and accessibility, incorporate interactive elements and improve the overall student experience. This collection reuses content from the following key resources:

- *Talking Glossary of Genomic and Genetic Terms* (<https://www.genome.gov/genetics-glossary>), Courtesy of: National Human Genome Research Institute (NHGRI), Public Domain with attribution (<https://www.genome.gov/about-nhgri/Policies-Guidance/Copyright>).
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INTRODUCTION

About the Book

Precision Healthcare: Genomics-Informed Nursing was developed for Georgian College as course material for NURS 4001.

Genomics is changing the healthcare landscape and providing opportunities to personalize patient care. With the widespread integration of genomics, nurses will increasingly need to incorporate genomic knowledge into their practice to provide safe, equitable, timely, and accessible care. This book will provide nursing professionals with the foundational genomic knowledge to navigate this rapidly evolving field.

Readers will explore genomics integration in personalized healthcare and how it relates to nursing practice. Genomic literacy is vital to understanding how genetic variations and environmental and lifestyle factors contribute to disease susceptibility and progression. Nurses with a strong foundation in genomics will be better equipped to assess genetic risk factors, interpret genetic and genomic data, and communicate with patients about their genomic health.

The chapters in this book will allow readers to explore the many factors that influence gene expression and lead to disease development, such as obesity, cancer, diabetes, cardiovascular disease and mental health disorders. Nurses will gain insight into modifiable and non-modifiable risk factors to develop evidence-based interventions that promote health and improve patient outcomes. By applying genomics-informed practices, nurses can advocate for personalized healthcare strategies that meet the needs of individual patients and populations. Nurses will also consider their role as part of an interdisciplinary team delivering genomic services and future nursing practice initiatives.

Navigation

The content of this book is divided into thirteen units. Students are expected to work through the materials in a unit for each week of study. This course was designed for an asynchronous course. The 28-hours of course time is to be used reviewing materials. Additional time will be spent on reviewing and completing assignments in further depth. Units 7 and 11 provide time to apply learning. The intent of these two weeks is to give students time to work on discussion posts, case studies, and



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scholarly posters. Additional case studies and learning activities are provided for optional independent practice.

Each chapter begins with an overview of the content covered and learning outcomes for the unit. Canada has not developed genomic competencies for nurses. Therefore, the NHS competencies from the UK and the ANA competencies from the US will be used. It should be noted that the competencies are meant to be demonstrated in practice. Each chapter aims to provide foundational theoretical knowledge that nurses need to be able to demonstrate these competencies in practice. However, this course does not include a practical component, so the competencies should be interpreted with this in mind. The competency documents should be consulted for clinical performance indicators.

The National Institutes of Health (NIH), National Human Genome Research Institute (NHGRI) talking glossary, and other sources provide definitions of key terminology in the unit. For pronunciation of terminology and audiovisual resources to enhance understanding of the term, visit the NHGRI talking glossary website. Select terms are listed at the start of each chapter and are highlighted as they appear in the body of the text. The subsequent sections of each unit contain the course content, learning activities, external resources, additional required reading, and related media. Key takeaways and additional optional readings are in the final chapter of each unit.

Overview

Unit 1: Introduction to Personalized Healthcare and the Role of the Nurse

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

Unit 2: Molecular Genetics Review

- DNA structure and function
- The genome and the cell cycle
- Cancer and the cell cycle
- The cellular basis of inheritance
- Patterns of inheritance

Unit 3: The Exposome

- Nature vs nurture

- Epigenetics
- Developmental origins of health and disease
- The exposome
- Adverse early childhood experiences
- Epigenetics in practice

Unit 4: Genetic Disorders

- Gene variants
- Genetic disorders
- Single gene disorders
- Polygenic disorders
- Chromosomal disorders
- Mitochondrial disorders

Unit 5: Genomics Nursing Research

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

Unit 6: Assessing Genetic Risk

- Family history
- Constructing a pedigree chart
- Pedigree analysis and modes of inheritance
- Calculating probabilities using pedigree charts
- Polygenic risk scores

Unit 7: Application of Theory in Practice Part 1

- There is no new reading material for this unit.
- Some case studies and exercises are presented for optional additional independent review and practice.
- Students are given time to complete the discussion post group assignment and begin work on the case study assignment.

Unit 8: Genetic Testing

- Genetic testing overview
- Types of Genetic tests
- Interpreting genetic test results

Unit 9: Pharmacogenomics

- Pharmacogenomics overview
- Genomic variation in drug response
- Personalized drug therapy
- Limitations of pharmacogenomic testing

Unit 10: Ethical, Legal, and Social Issues Arising from Genomics

- Genetic discrimination
- Eugenics and scientific racism
- Use of population descriptors in genomics
- Nursing implications

Unit 11: Application of Theory in Practice Part 2

- There is no new reading material for this unit.
- Some case studies and exercises are presented for optional additional independent review and practice.
- Students are given time to complete the discussion post group assignment and complete work on the case study assignment.

Unit 12: Special Topics in Genomics

- Genomics and global health
- Cancer genomics
- Genomics application by specialty

Unit 13: The Future of Genomics and Nursing

- Gene editing
- Other genomic technologies

- Health system readiness for the genomic era

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FOR EDUCATORS

About Precision Healthcare: Genomics-Informed Nursing

This book introduces nurses and nursing students to the applications of genomics in practice and research. It is designed to be engaging, promote self-directed learning, and remain accessible, with content presented in a clear and reader-friendly format. The text is tailored for a single-semester, 28-hour online asynchronous undergraduate course. However, it can also benefit practicing nurses seeking to enhance their genomic literacy.

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Supplementary course materials for instructors are available upon request and with confirmation of educator status. These include a sample syllabus, quiz question bank, case studies with instructor keys, assignment guidelines for a scholarly poster project, and a comprehensive reading list.

Journal articles have been assigned as required reading throughout the book, in addition to textbook content, for the purposes of the 28-hour course.

At the end of each unit there is a list of recommended additional readings and resources. For in-person courses, educators may elect to present the textbook contents, or reduce some of the journal articles. For a 40-hour course, see the optional additional readings. The citations below are recommended first-choice additions from the additional optional readings for expanded course content.

Chapter 1

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.

Kerber, A. S., & Ledbetter, N. J. (2017). Standards of practice: Applying genetics and genomics resources to oncology. *Clinical Journal of Oncology Nursing*, 21(2), 169–173. <https://doi.org/10.1188/17.CJON.169-173>

Chapter 3

Focus on definitions and distinctions between terms:

Harden, K.P. (2023). Genetic determinism, essentialism and reductionism: semantic clarity for contested science. *Nature Reviews Genetics*, 24, 197–204. <https://doi.org/10.1038/s41576-022-00537-x>

Chapter 5

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>

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. Precision health: Advancing symptom and self-management science. (2019). *Nursing Outlook*, 67(4), 462-475. <https://doi.org/10.1016/j.outlook.2019.01.003>

Chapter 8

Miller, D. T., Lee, K., Abul-Husn, N. S., Amendola, L. M., Brothers, K., Chung, W. K., Gollob, M. H., Gordon, A. S., Harrison, S. M., Hershberger, R. E., Klein, T. E., Richards, C. S., Stewart, D. R., Martin, C. L., & ACMG Secondary Findings Working Group. Electronic address: documents@acmg.net (2023). ACMG SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*, 25(8), 100866. <https://doi.org/10.1016/j.gim.2023.100866>

Chapter 10

The full article that the news brief is based on is available here:

Fernando, A., Kondrup, E., Cheung, K., Uberoi, D., & Joly, Y. (2024). Still using genetic data? A comparative review of Canadian life insurance application forms before and after the GNDA. *FACETS*, 9, 1-10. <https://doi.org/10.1139/facets-2023-0101>

Thomas, G. M. & Katz Rothman, B. (2016). Keeping the backdoor of eugenics ajar: Disability and future prenatal screening. *AMA Journal of Ethics*, 18(4), 406-415. <https://journalofethics.ama-assn.org/article/keeping-backdoor-eugenics-ajar-disability-and-future-prenatal-screening/2016-04>

Peer Review and Feedback

The book has undergone expert review by professionals in nursing and genomics. Feedback is welcomed from

healthcare professionals with genomic expertise. Suggestions for additional activities, examples, video content, emerging research, or revisions can be submitted to our contact addresses below. See the acknowledgement section for a list of peer reviewers.

Contact

Please contact us with feedback, suggestions or to request supplemental materials at:

- andrea.gretchev [at] gmail.com
- oer [at] georgiancollege.ca

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