UNIT 10 - ETHICAL, LEGAL, AND SOCIAL ISSUES ARISING FROM GENOMICS

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 10 Contents

Attribution

10.1 UNIT OVERVIEW

Learning Objectives

- Critically assess some of the ethical, legal, and social implications of genomics.
- Evaluate the historical context and origins of eugenics and scientific racism and relate this to contemporary healthcare practices and policies.
- Distinguish between race, ethnicity, and genetic ancestry as applied in biomedical contexts.
- Examine how historical misuse of population descriptors has contributed to health disparities and reinforced inequities in healthcare and genomic research.

Outline

Topics covered in this chapter include:

- Genetic discrimination
- Eugenics
- Scientific racism
- Population descriptors
- Nursing implications

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.
- Demonstrates in practice the importance of tailoring genomic information and services that are responsive to the unique attributes of every person.

NHS (2023):

Demonstrate effective communication in tailoring genomic information and services to the individual:

- recognizing factors (such as ethnicity, culture, religion, ethical values, developmental stage or language) that may influence the individual's ability to use information and services.

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

- understanding the importance of delivering genomic information and counselling fairly, accurately and without coercion or personal bias, to facilitate decision-making and manage expectations;
- recognizing that your values and the values of others may influence the care and support provided during decision-making, and that choices and actions may change over time;
- ensuring that the consent process is person centered; 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; and
- incorporating awareness of the potential physical, emotional, psychological and social consequences of genomic information for individuals, family members and communities.

Key terminology

Ethnicity

Refers to a group of people with shared language, religion, customs, beliefs, heritage and history, even though such attributes are not always confined to a single ethnic group. Ethnicity may also refer to groups that are considered indigenous to an area. Ethnicity is not a biological characteristic (NHGRI, 2024).

Eugenics

Eugenics is a discredited belief that selective breeding for certain inherited human traits can improve the "fitness" of future generations. For eugenicists, "fitness" corresponded to a narrow view of humanity and society that developed directly from the ideologies and practices of racism, colonialism, ableism and imperialism.

Genetic ancestry

Genetic ancestry refers to information about the people that an individual is biologically descended from, including their genetic relationships. Genetic information can be combined with historical information to infer where an individual's distant ancestors lived.

Genealogical ancestry

An individuals family origins and ancestral history established through records, family trees, and other forms of documentation tracing lineage and relationships (NHGRI, 2024, para. 10).

Population descriptors

Population descriptors are ways of describing or distinguishing people from each other based on perceived or actual differences. They capture the various ways in which people can differ from one another. A wide variety of population descriptors are used to describe groups of people in research, healthcare or society. Examples of population descriptors include race, ethnicity, genealogical ancestry, genetic ancestry, Indigenous, primary language spoken, nationality, geographic origin, sex at birth, gender identity, disability status and age. Each population descriptor captures a different aspect of a group or individual. One population descriptor is not enough to fully describe or distinguish any

individual or group. Depending on the situation, some population descriptors may be more relevant than others (NHGRI, 2024).

Race

Race is a social construct used to group people. Race was constructed as a hierarchal human-grouping system, generating racial classifications to identify, distinguish and marginalize some groups across nations, regions and the world. Race divides human populations into groups often based on physical appearance, social factors and cultural backgrounds.

Reference populations

To determine genetic ancestry, researchers compare DNA variants in an individual to the frequency of those DNA variants in reference populations – groups of people from around the world who have provided samples of their DNA (NHGRI, 2024. para. 13).

Scientific racism

Scientific racism is the practice of using data derived from pseudoscience to support racial biases and other forms of discrimination. Leading scientists across scientific institutions in the 19th and early 20th centuries were proponents of such ideologies. By the mid-20th century, data derived from pseudo-science were widely disproven. However, evidence shows that the practice of scientific racism persists in science and research.

Attribution & References

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- Talking Glossary of Genomic and Genetic Terms, Courtesy of: National Human Genome Research institute (NGHRI), Public Domain with attribution.

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National Human Genome Research Institute (NHGRI). (2024, October 24). *Explainer: Use of population descriptors in genomics.* https://www.genome.gov/about-genomics/policy-issues/population-descriptors-in-genomics

10.2 GENETIC DISCRIMINATION

Ethical, Legal and Social issues

There are a multitude of ethical, legal, and social issues arising from genomic-informed healthcare and research. An entire textbook could be devoted to exploring these ethical issues. While some of these topics have been covered in previous units, as noted in the brackets beside each topic, this course cannot encompass all of them. This chapter intends to build on nursing students' previous studies of legal and ethical issues in healthcare. Some of the ethical issues in genomics include:

- Returning genomic results (5.3, 8.4)
- Predictive testing of minors (5.3, 8.2, 12.4)
- Informed consent (5.3)
- Privacy and confidentiality (8.3)
- Individual vs relational autonomy and the right to know (12.3)
- Genetic discrimination (3.3)
- Data sharing and security (5.3, 13.2)
- Biobanking (5.3)
- Concerns about assistive reproductive technologies (8.3, 12.4, 13.2)
- Gene editing (13.2)
- Eugenics and scientific racism (10.3)
- Population descriptors (10.4)

This unit will specifically focus on genetic discrimination, the dark history of genetics and genomics (eugenics), ongoing scientific racism, and the preferred use of population descriptors. The final section will review implications for nursing, particularly in respect to the nursing Code of Ethics, professional and practice standards, and competencies. The lessons from this unit also apply to other healthcare areas beyond genomics and are rooted in the bioethical principles of autonomy, beneficence, non-maleficence, and justice that nurses learn about in their ethics courses. The Tri-council Policy Statement (TCPS2) was reviewed in chapter 5.3. Recall that chapter 13 of the TCPS2 provides guidance on conducting human genetic research and examines ethical issues in genetic research including biobanking, privacy and confidentiality, vulnerable populations (e.g. predictive testing of children), consent, returning genomic test results (e.g. incidental and secondary findings), and gene transfer.

Genetic Discrimination

Chapter 3.3 briefly explored how everyday discrimination happens through genetic discourse. The assigned reading for that chapter reviewed the problematic concepts of genetic determinism, essentialism, and reductionism which, when misunderstood or misused, can exacerbate health disparities and perpetuate prejudice. These concepts underlie scientific racism and eugenics due to the misbelief that certain people are advantaged over others due to their 'genetic endowment' or 'good genes'.

In certain cases, genetic information may be misused in ways that impact access to employment, goods, and services. In chapter 8.2, it was mentioned that the possibility of genetic discrimination in employment or insurance is a concern of genetic testing. An example of institutional genetic discrimination is when employers or insurance companies treat people differently based on genetic information that shows a predisposition to an inherited disorder. There are many other forms of individual genetic discrimination. For example, knowledge about carrier status for sickle cell trait is used to select or reject partners for marriage and childbearing. This can make those who carry the trait feel less-than, particularly in cultures where marriage is linked to social status. Fear of discrimination is a common concern among people considering genetic testing" (MedlinePlus, n.d.).

In 2017, parliament passed the Genetic Non-Discrimination Act (GNDA) into law to prevent institutional discrimination based on genetic test results by employers or insurance agencies. Under the GNDA, companies cannot require genetic testing or deny services based on a refusal to obtain testing (Canadian Civil Liberties Association, 2018). Furthermore, a person's genetic test results cannot be collected, used, or disclosed without written consent (Canadian Civil Liberties Association, 2018).

The protections afforded by the act protect individuals being denied goods and services, such as obtaining life or health insurance or entering into contracts, such as employment, based on genetic test results. The complete legislation can be viewed here (https://laws-lois.justice.gc.ca/eng/acts/g-2.5/page-1.html). Nurses should be familiar with the legislation, as patients pursuing genetic testing will likely have questions about what happens to their genetic data, who can access it, and how it can be used. Alaire et al. (2021) found that many Canadian women lacked knowledge or were misinformed about the scope and content of the GNDA, with an average rate of 50.94% correct responses to their questionnaire. Dalpé et al. (2021) analyzed online discussion posts about the GNDA and found that in the forum about fears around the GNDA, 69% of the posts related to avoiding genetic testing.

Several countries have their own form of protection against genetic discrimination and Canada's GNDA is one of the strongest, as it is enforced under the criminal code. Penalties for violation can include fines of up to $1 million dollars or five years imprisonment (Genetic Non-discrimination Act, 2017). However, there is some validity to concerns as there is evidence that some insurance companies have attempted to find loopholes in the legislation (Fernando et al., 2024). The GNDA is not all encompassing and, as genomic sciences advances, the limited frame used by the GNDA to define genetic testing will need to be amended to be more inclusive of other forms of genetic data.

Read

A news brief on a McGill study (https://www.mcgill.ca/newsroom/channels/news/mcgill-study-finds-some-canadians-may-still-be-risk-genetic-discrimination-despite-new-federal-law-355104) that exposes the ongoing risks of discrimination that are not protected by the GNDA

Attribution & References

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10.3 EUGENICS AND SCIENTIFIC RACISM

Trigger Warning:

This chapter contains material that may be upsetting or distressing. Contents may include discussion of traumatic experiences related to mental or physical disability, genocide, slavery, and racism.

Eugenics and Scientific Racism

Genetics has an ugly past – Eugenics. Eugenics is an inaccurate theory linked to historical and present-day forms of discrimination, racism, ableism and colonialism. It has persisted in policies and beliefs worldwide, including in Canada. Have we moved on, or are we just finding different ways to manifest the same thinking?

Hard Language

In the 21st century, we are accustomed to using vocabulary that is sensitive, respectful, and person-centered regarding physical and mental illnesses and challenges. The language used in the 19th and most of the 20th centuries was far more direct and judgmental. These terms enabled 19th and 20th-century reformers to objectify the individuals they were referring to. Understanding that relationship between language and reform is critical to understanding the attraction and authority of movements like eugenics. The language used in the original source material has been adapted to be more respectful.

Source: *Canadian History: Post-Confederation*, CC BY 4.0

The Big Picture:

- Eugenics is the scientifically inaccurate theory that humans can be improved through selective breeding of populations.
- Eugenicists believed in a prejudiced and incorrect understanding of Mendelian genetics that claimed abstract human qualities (e.g., intelligence and social behaviours) were inherited in a simple fashion. Similarly, they believed complex diseases and disorders were solely the outcome of genetic inheritance.
- The implementation of eugenics practices has caused widespread harm, particularly to populations that are being marginalized.
- Eugenics is not a fringe movement. Starting in the late 1800s, leaders and intellectuals worldwide

perpetuated eugenic beliefs and policies based on common racist and xenophobic attitudes. Many of these beliefs and policies still exist.
- The genomics communities continue to work to scientifically debunk eugenic myths and combat modern-day manifestations of eugenics and scientific racism, particularly as they affect Black, Indigenous, and people of colour, people with disabilities, and LGBTQAI2+ individuals.

Visit the NHGRI website – Eugenics: Its Origin and Development (1883-Present) (https://www.genome.gov/about-genomics/educational-resources/timelines/eugenics). Scroll down to the timeline and use the navigation arrows to learn about some of the more predominant historical examples of eugenics.

What are eugenics and scientific racism?

Eugenics is the scientifically erroneous and immoral theory of "racial improvement" and "planned breeding," which gained popularity during the early 20th century. Eugenicists worldwide believed that they could perfect human beings and eliminate so-called social ills through genetics and heredity. They thought the use of methods such as involuntary sterilization, segregation and social exclusion would rid society of individuals deemed by them to be unfit.

Eugenics can be separated into two equally erroneous categories. Negative eugenics is described above (NIH, 2022). Positive eugenics is the encouragement of reproduction among genetically advantaged people (Thomas et al., 2016). It should be noted that the characteristics that were deemed to be more or less desirable were not determined by genes at all. These were entirely social constructs. Associating these beliefs with science distorts the evidence and undermines the credibility and validity of the findings.

Scientific racism is a practice that appropriates the methods and legitimacy of science to argue for the superiority of one race over another. Like eugenics, scientific racism grew out of:

- the misappropriation of revolutionary advances in medicine, anatomy and statistics during the 18th and 19th centuries.
- Charles Darwin's theory of evolution through the mechanism of natural selection.
- Gregor Mendel's laws of inheritance.

Eugenic theories and scientific racism drew support from contemporary xenophobia, antisemitism, sexism, colonialism and imperialism, as well as justifications for slavery.

How did eugenics begin?

Listen:

Genetics Unzipped is a great podcast that covers a vast array of topics in genomics. Listen to Living with the Eugenic Past: Michele Goodwin (17:36) (https://geneticsunzipped.com/blog/2022/11/27/adelphi-genetics-michele-goodwin?rq=living%20with%20the%20eugenic). This is one of several talks on the subject from the conference titled the same. Michele Goodwin is a Professor of Law at the Centre for Biotechnology, Global Health Policy Director at the University of California Irvine, and a senior lecturer at Harvard Medical School.

“Her talk focused on how the long shadow of eugenics and white supremacy persists into the present day and remains embedded in contemporary political frameworks, and why this pernicious ideology is taking so long to die” (The Genetics Society, 2022).

Galton defined eugenics as "the study of agencies under social control that may improve or impair the racial qualities of future generations either physically or mentally." Galton claimed that health, disease, and social and intellectual characteristics were based upon heredity and the concept of race.

During the 1870s and 1880s, discussions of "human improvement" and the ideology of scientific racism became increasingly common. So-called experts determined individuals and groups to be superior or inferior. They believed biological and behavioural characteristics were fixed and unchangeable, placing individuals, populations and nations inside that hierarchy.

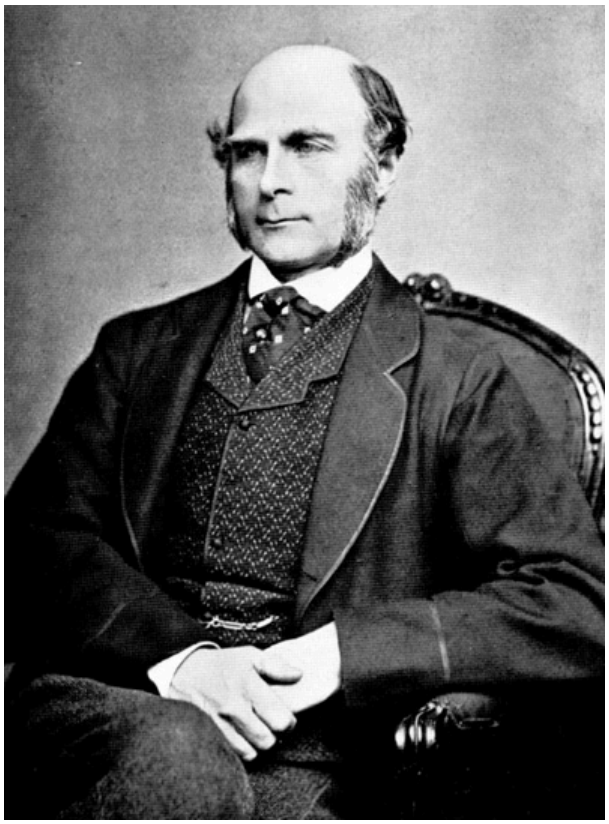

Figure 10.1 Sir Francis Galton (1822-1911) was a largely self-trained British social scientist, a half-cousin of Charles Darwin, and the figure most readily associated with Eugenics. It is Galton who is credited with coining the dichotomy of nurture vs. nature. **Source:** Francis Galton, 1850s, PDM

What did eugenics look like across the globe?

By the 1920s, eugenics had become a global movement. There was popular, elite and governmental support for eugenics in Germany, the United States, Great Britain, Italy, Mexico, Canada and other countries. Statisticians, economists, anthropologists, sociologists, social reformers, geneticists, public health officials and members of the general public supported eugenics through various academic and popular literature.

The most well-known application of eugenics occurred in Nazi Germany in the lead-up to World War II and the Holocaust. Nazis in Germany, Austria and other occupied territories euthanized at least 70,000 adults and 5,200 children. They implemented a campaign of forced sterilization that claimed at least 400,000 victims (https://www.ushmm.org/learn/timeline-of-events/1939-1941/euthanasia-killings-continue). This culminated in the near destruction of the Jewish people, as well as an effort to eliminate other marginalized ethnic minorities, such as the Sinti and Roma, individuals with disabilities and LGBTQ+ people.

Slavery and its legacies, fears of "miscegenation, (https://www.britannica.com/topic/miscegenation)" and eugenics were deeply connected in the early 20th century. Prominent American eugenicists expounded on their concerns of "race suicide," or the increasingly differential birthrates between immigrants and non-Nordic races compared to native-born Nordic whites. Eugenicists used these concerns to promote discriminatory policies like anti-immigration and sterilization.

American eugenicists from a variety of disciplines declared specific individuals unfit or anti-social, which resulted in the involuntary sterilization of at least 60,000 people through 30 states' laws by the 1970s.

These eugenicists disproportionately targeted non-white people, particularly those with lower socio-economic status, and people with disabilities during the entirety of the 20th century. Eugenicists were also crucial to the enactment of discriminatory immigration legislation that was passed in 1924 (the Johnson-Reed Act), which completely excluded immigrants from Asia.

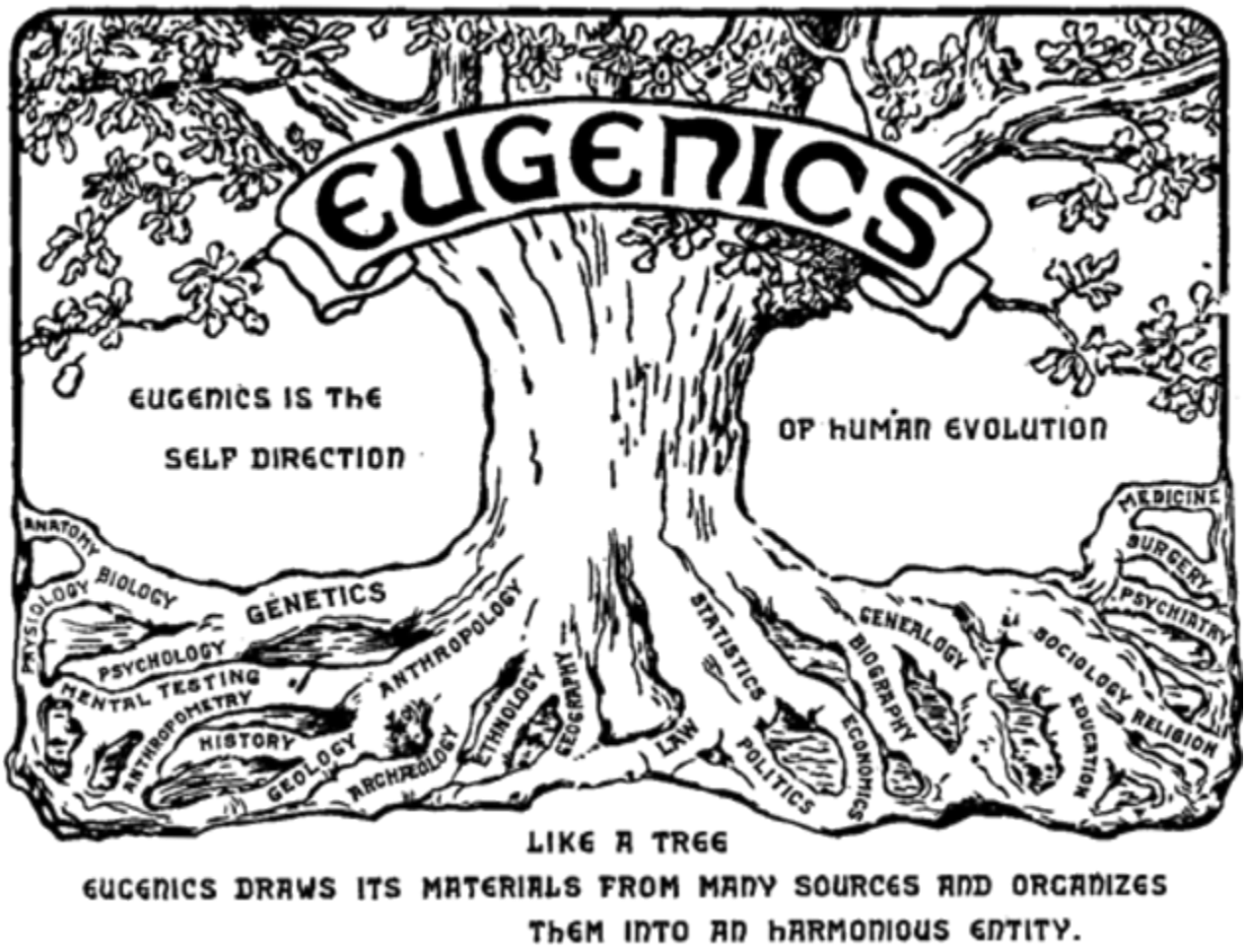

Figure 10.2 Eugenics Congress logo shows a tree, with the label "Eugenics" across the trunk just before the branches extend out. Label stats that "Eugenics is the self direction of human evolution" and the roots of the tree are labeled with many different areas of study, including biology, genetics, statistics, sociology, etc.. **Source**: Image in the PDM scanned from Harry H. Laughlin, *The Second International Exhibition of Eugenics held September 22 to October 22, 1921, in connection with the Second International Congress of Eugenics in the American Museum of Natural History, New York* (Baltimore: William & Wilkins Co., 1923).

Figure 10.3 American Eugenics Society photograph of Eugenics Building, Fitter Families Contest, Kansas Free Fair. Staff of Fitter Families Contest, Kansas Free Fair. **Source:** Photo by unknown, courtesy of American Philosophical Society Library (https://diglib.amphilsoc.org/islandora/object/graphics%3A1660), PDM.

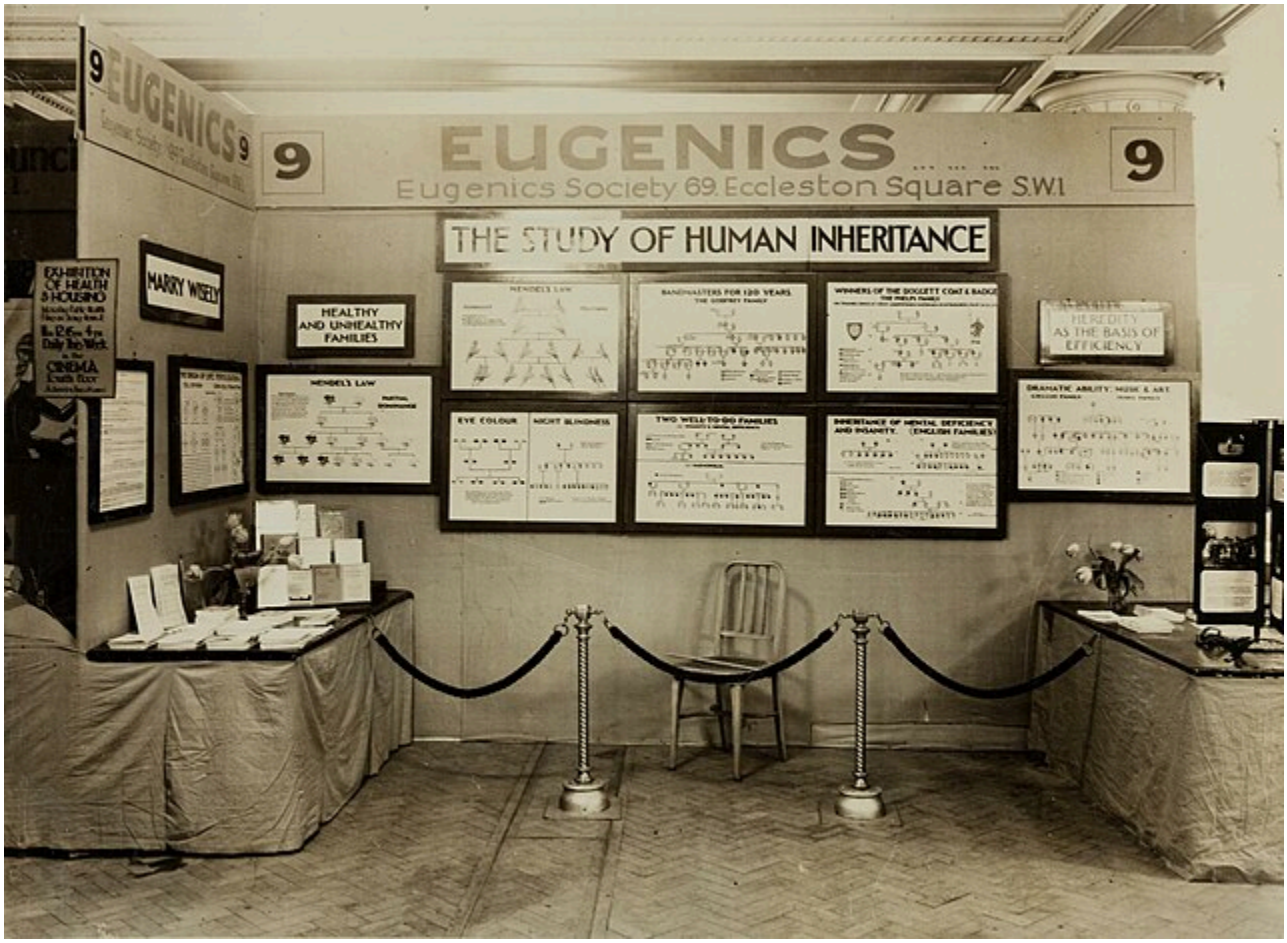

Figure 10.4 Eugenics Society Exhibit. Image from the Wellcome Library (1930s). **Source:** Photo by Wellcome Library, CC BY 4.0.

Was there eugenics in Canada?

The Eugenic Archive has a wealth of historical information in an accessible format. Check out this interactive timeline of Eugenics in Canada (https://www.eugenicsarchive.ca/timeline)

The eugenicist strategy has been one of *selective breeding*, but that term does not do it justice. "Selective breeding" invariably involves *selecting in*: that is, encouraging people who were deemed to possess more desirable qualities to have a significant number of children. However, the eugenics message in Canada was more about *selecting out*: to find ways to deter the reproduction of what they regarded as fated populations doomed by their genes to imperil themselves, successive generations, and the nation.

Canada sterilized proportionately fewer people than the United States — the total number is believed to

be slightly more than 3,000 – but record keeping was inconsistent and there is little doubt that an accurate total is unknowable (Hanson and Kind, 2013).

Figure 10.5 A residential institution for people with mental health challenges at Orillia, ON, ca. 1910.
Source: The Asylum, Orillia, ON, about 1910 © McCord Museum (MP-0000.724.13), CC BY-ND.

Listen:

Judy Lytton is a eugenics survivor and currently is on the Board for the Living Archives in Eugenics in Western Canada. Hear her story (https://www.eugenicsarchive.ca/our-stories?id=531cdbd7132156674b0001f1).

Figure 10.6 The Provincial institution for people with mental health challenges in New Westminster, BC, shortly after it was opened in 1878. It would subsequently become known as the Provincial Hospital for the Insane and, from 1950, as Woodlands School. **Source:** Provincial Asylum for the Insane, c. 1878 by S.J. Thompson, Public Domain

Concept in Action

The eugenics movement in Western Canada and the application of sterilization are discussed by historian of institutionalization Megan Davies (York University).

Watch Dr. Megan J. Davies Question 9 – Eugenics (5 mins) on YouTube (https://youtu.be/1rzLfGfcwMg)

Do eugenics and scientific racism still exist?

While eugenics movements especially flourished during the three decades before the end of World War II, eugenics practices such as involuntary sterilization, forced institutionalization, social ostracization, and stigma were common in many parts of Canada until at least the 1970s and, in some instances, have continued into the present in various forms. Historical injustices also lead people to fear and mistrust government bodies.

Watch Kyle Lillo (https://www.eugenicsarchive.ca/our-stories?id=544724c7d4cf080000000003) describe his concerns about having children. He fears that due to his disability, the government will apprehend his children and deem him unfit to parent.

Compulsory Sterilization in Canada

Compulsory sterilization in Canada is an ongoing practice that has a documented history in the provinces of Alberta, Saskatchewan, and British Columbia. Sixty Indigenous women in Saskatchewan sued the provincial government, claiming they had been forced to accept sterilization before seeing their newborn babies (Moran, 2016). In June 2021, the Standing Committee on Human Rights in Canada found that compulsory sterilization is ongoing in Canada, and its extent has been underestimated (Government of Canada, 2022). A bill was introduced to Parliament in 2024 to end the practice (Ryckewaert, 2024).

Over 9,000 Indigenous People in British Columbia were surveyed and interviewed in an inquiry into racism and discrimination in BC's healthcare system. The In Plain Sight Report (https://engage.gov.bc.ca/app/uploads/sites/613/2020/11/In-Plain-Sight-Full-Report-2020.pdf) summarized the findings, which included examples of forced sterilization of Indigenous women in Canada.

With the completion of the Human Genome Project (HGP) and, more recently, advances in genomic screening technologies, there is some concern about whether generating an increasing amount of genomic information in the prenatal setting would lead to new societal pressures to terminate pregnancies where the fetus is at heightened risk for genetic disorders, such as Down Syndrome and spina bifida.

The possible genomic-based screening of embryos for behavioural, psychosocial and intellectual traits would be reminiscent of the history of eugenics in its attempt to eliminate specific individuals.

In fact, some geneticists view both genomic screening and genetic counselling as an extension of eugenics.

Human enhancement through consensual gene editing has also been described as the new eugenics. Despite its basis in science and

Optional Video

Watch the Eugenics Crusade on AppleTV or Prime Video for a

deeper dive into Eugenic history.

Honeycutt, C., De La Uz, R., & Ferrari, M. (October 16, 2018). The eugenics crusade: What's wrong with perfect? (https://www.pbs.org/wgbh/americanexperience/films/eugenics-crusade/) PBS. USA: 42nd Parallel Films.

being consensual, it has underpinnings of the historical examples of eugenics – the pursuit of a superior being at the expense of those considered inferior.

How can nurses address eugenics and scientific racism?

Nurses can commit to interrogating the legacies of eugenics and scientific racism to further develop ethical and equitable uses of genomics. Understanding the history of eugenics, including forced sterilizations and discriminatory practices, helps healthcare providers recognize the deep-seated mistrust some patients may have toward medical institutions. This awareness can guide providers in delivering trauma-informed care. By integrating these practices, healthcare providers can create a safer, more supportive environment for all patients, particularly those who have experienced trauma. This history emphasizes the potential for misuse of genetic information and reinforces the need for nurses to remain vigilant against discrimination and stigmatization based on genetic traits. Nurses can integrate this knowledge and awareness into practice to ensure genomic services are provided ethically, equitably, and inclusively.

It is also crucial that nurses critically evaluate medical literature to identify scientific racism, and challenge unscientific medical practices such as the misrepresentation of race as a biological variable rather than a social construct.

Read:

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Questions for Reflection

1. What can nurses do to avoid making assumptions about their patients based on skin colour or perceived racial background?
2. How can nurses identify and address the impacts of racism, including frequency, intensity, and duration of racist encounters in their patients' lives when assessing their health and planning their care?
3. Why do the authors propose using the term *racism* instead of *race* as a variable in health research and practice and how would this help quantify the social determinants of health that are related to racism?

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10.4 USE OF POPULATION DESCRIPTORS IN GENOMICS

Population Descriptors

Appropriate use of **population descriptors** is a critical scientific issue that is important for advancing genomic research and improving healthcare across human populations. Given the ethical, legal and social implications of their historical and current use, thoughtful use by researchers and other interested parties is essential.

The inaccurate belief that human populations are biologically distinct has contributed to harm, such as justifying eugenics through the practice of scientific racism, and marginalizing groups. In turn, misapplication of concepts of population groups has contributed to health disparities, alienated marginalized groups from research participation, and led to harmful stereotypes that have reinforced inequities.

More work is needed to educate researchers, clinicians, policymakers, and the public on the distinctions between race, ethnicity, and genetic ancestry and advance the use of population descriptors in genomics and biomedical research.

The National Academies of Sciences, Engineering and Medicine (NASEM) assessed the methods, benefits and challenges in a review of the use of population descriptors in genomics research. The NASEM Report (https://www.nationalacademies.org/our-work/use-of-race-ethnicity-and-ancestry-as-population-descriptors-in-genomics-research) includes 13 recommendations to transform how population descriptors are used in human genetics and genomics research.

Types of population descriptors

Population descriptors are ways of describing or distinguishing people from each other based on perceived or actual differences. They capture the various ways in which people can differ from one another.

A wide variety of population descriptors describe groups of people in research, healthcare or society. Examples of population descriptors include race, ethnicity, skin colour, genealogical ancestry, genetic ancestry, Indigenous, primary language spoken, nationality, geographic origin, sex at birth, gender identity, disability status and age. Each population descriptor captures a different aspect of a group or individual. One population descriptor cannot fully describe or distinguish any individual or group. Depending on the situation, some population descriptors may be more relevant than others.

People commonly use population descriptors and their corresponding categories or numerical scales to

describe themselves and others. For example, we use categories like female, male or intersex when referring to the biological sex assigned at birth (Phenex Toolkit, 2024-a). We use numerical values like months and years when referring to age. We also use categories like newborn, adolescent or older adult.

Researchers in genomics and healthcare also use population descriptors and corresponding categories to describe who is participating in a research study, what groups are being compared as part of the study and to whom their study findings may apply. These are collectively referred to as demographics. Researchers can obtain information about population descriptors in many ways, for instance, by asking participants how they identify, looking in an electronic medical record, using data from a prior research study that was shared, or searching public records. Researchers may also assign a population descriptor to an individual or group using a specific analytical approach, such as using statistics to look at the frequency of DNA variants across the genome.

The definitions, measurements, uses, and interpretations of population descriptors have varied over time across users and worldwide. Human rights movements or social and political action can bring about such changes. In addition, new scientific discoveries or knowledge, such as in the fields of genomics, archaeology or social science, can lead to changes. New scientific discoveries and well-established facts present an opportunity to improve our understanding of human genetic variation and our knowledge of what types of differences between or across groups may be important for health. For example, the first modern humans lived somewhere in Africa approximately 300,000 years ago (Hublin et al., 2017), and physical barriers to the migration of humans, such as oceans and mountains, led to geographical differences in the frequency of genetic variants we see within and between populations (Rosenberg, 2011).

While we're often searching for differences, we must remember that human beings are far more similar than they are different. When identifying groups that differ genetically, researchers have found that **most of the variation occurs *within* groups of people rather than between them**. This means that nearly all differences are not specific to a group. Instead, they are sometimes found at different frequencies between groups.

Understanding genetic ancestry, race, and ethnicity

Concept in Action

Watch **this video** from the Canadian Nursing and Genomics Steering Committee on the intersection of genomics, social determinants of health, race and racism for an description and illustration of how these concepts impact nursing practice.

There is not one agreed-upon definition for these terms. The descriptions below highlight key differences across them.

Genetic ancestry

Genetic ancestry refers to the biological relationships between individuals resulting from inheriting common ancestors' DNA. These common ancestors are tied to their geographical origins from many centuries ago when long-distance travel was extremely difficult. Parents do not pass down all their DNA to their children. Therefore, **genealogical ancestry** and genetic ancestry can be different. Genetic ancestry is based on a statistical measure of genetic similarity across individuals.

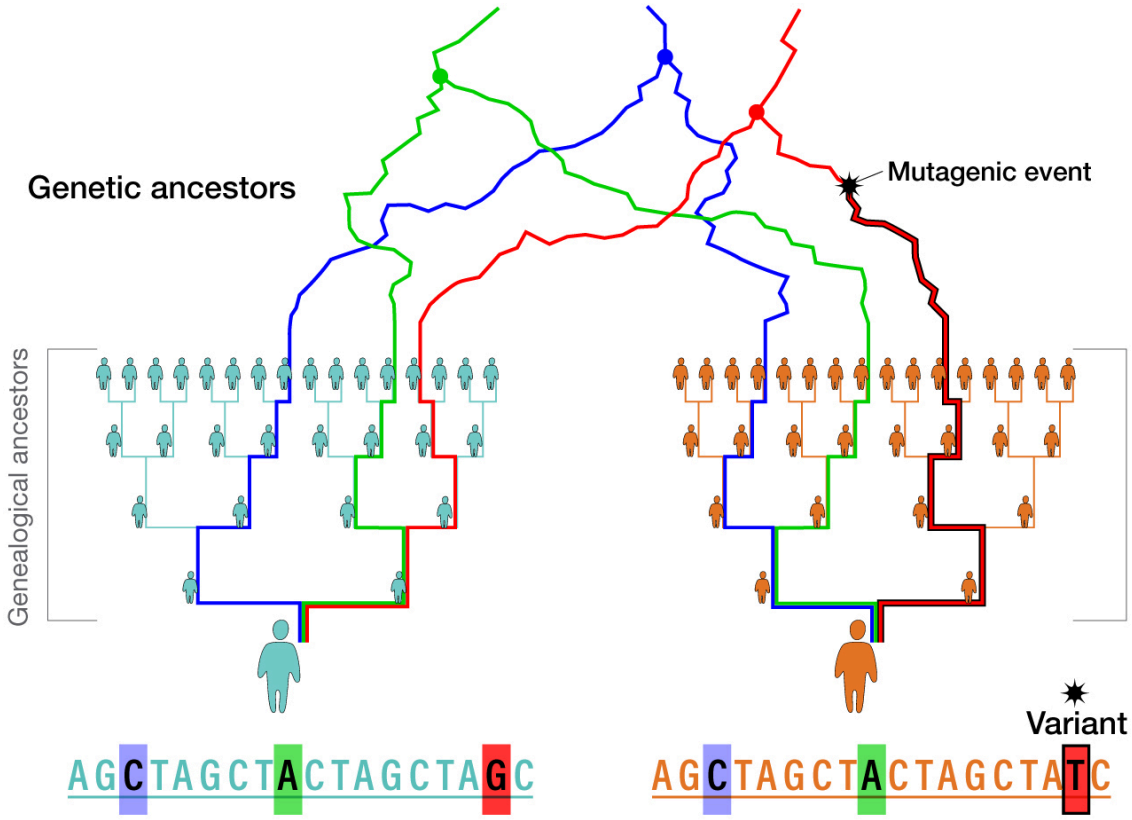


Figure 10.7 Genetic ancestry refers to the bits of DNA a person or group has inherited over time from prior generations. **Source:** National Human Genome Research Institute, PDM with attribution.

Figure 10.8 Race is a population descriptor with multiple definitions. Race has been used to group humans into a hierarchical system that identifies, distinguishes and marginalizes some groups across nations, regions and the world. **Source:** National Human Genome Research Institute, PDM with attribution.

Race

People created the concept of race. **Race** typically divides human populations into groups based on perceived physical appearance (such as skin colour), social factors and cultural backgrounds (NHGRI, 2022). Race has been used to inappropriately group people into a hierarchical system to "establish and justify systems of power, privilege, disenfranchisement and oppression" (National Museum of African American History and Culture, n.d.-b, para. 1).

Ethnicity

Ethnicity refers to a group of people with shared language, religion, customs, beliefs, heritage and history, even though such attributes are not always confined to a single ethnic group. Ethnicity may also refer to groups that are considered indigenous to an area. Ethnicity is not a biological characteristic.

Figure 10.9 Ethnicity refers to a group of people with a shared language, religion, customs, beliefs and history. **Source:** National Human Genome Research Institute, PDM with attribution.

How well can researchers determine genetic ancestry?

Methods for estimating genetic ancestry are evolving. To determine an individual's genetic ancestry, researchers compare DNA variants in that individual to the frequency of those DNA variants in groups of people from around the world who have provided samples of their DNA. These groups of people form what is referred to as **reference populations.** Genetic ancestry is estimated using statistical techniques and is typically based on some measure of genetic similarity. An individual with a collection of genetic variants that appear in the highest frequency within a reference population is estimated to have ancestors from that reference population. Individuals may have a collection of genetic variants that appear in more than one reference populations, which indicates they likely have ancestors from more than one group. Some have argued that instead of thinking about genetic ancestry as broad groups or categories, genetic ancestry should be considered a continuum.

Currently, genomic researchers do not have DNA samples from many groups of people around the world, which means genetic ancestries for some geographical locations cannot be estimated accurately. In addition, as mentioned above, scientists use reference datasets to calculate ancestries. Genetic ancestry estimations can differ from one analysis to another due to differences in the frequencies of genetic variants in the datasets used. Furthermore, when someone is estimated to have ancestors from more than one group, researchers sometimes lump individuals together into a single group to simplify analyses. Therefore, determining genetic ancestry is a statistical estimate based on available data and is inconsistent across studies. More recently, companies offering ancestry-related services directly to consumers have combined genetic ancestry information with family history information.

Read

Cerdeña, J. P., Grubbs, V. & Non, A.L. (2022). Genomic supremacy: the harm of conflating genetic ancestry and race. *Human Genomics, 16*(18), 1-5. https://doi.org/10.1186/s40246-022-00391-2

OPEN ACCESS ONLINE

A closer look: Genetic ancestry and identification

Regardless of the outcome of a genetic ancestry test, people will choose how they want to be identified by others. DNA, social factors, personal or familial preferences, or lived experiences may inform these choices.

How do I identify?

Imagine your friend received an ancestry test for the holidays and was surprised by some results. After taking the test, your friend had a primary care appointment with a new doctor. When completing the required forms, they answered questions about their race and ethnicity differently than in the past based on the latest information provided in their ancestry test. While their physical body and health status did not change, their social identity did change. People vary in their response to ancestry testing. For some, the outcome may lead to an identity change. For others, they may maintain their original identity (Roth & Ivemark, 2018).

But how meaningful is this change for healthcare decision-making? An estimate of genetic ancestry (not race) can be informative for some conditions. For example, some heritable cancers are more common in certain groups than others. Should self-identified race or ethnicity change a doctor's decision about medical treatment? The answer can depend on a variety of factors.

How am I identified?

As another example, the U.S. government has changed the reporting of race and ethnicity over time, with categories being renamed, merged, removed or expanded (United States Census Bureau, 2015). Was this change due to new information about how people differ genetically or biologically? No. The change was made to reflect better perceptions of growing diversity across groups in the country, better reflect how different people identify themselves and improve the quality of available demographic data. The way people self-identify can change in their lifetime or across generations, along with the questions and forms intended to capture this information.

Are population descriptors social constructs?

A social construct is an idea or collection of ideas created, agreed upon, accepted, or acknowledged by groups of people in a society. Social constructs offer ways to organize, explain or make sense of the world. Many population descriptors are social constructs. A socially constructed population descriptor can change and be used and defined differently in different parts of the world (National Museum of African American History and Culture, n.d.-a).

Race and ethnicity are social constructs. There is no clear or consistent way to place people into racial or ethnic groups using biology or innate characteristics. For example, people of similar skin colour or hair texture have been defined as being of different races. Skin colour variation has arisen from people adapting to varying levels of sun exposure (Jablonski & Chaplin, 2010), and people of similar skin colour may have very little in common genetically.

For centuries, definitions of race and ethnicity were overly simplistic, unscientific, unethical and regularly used to support colonialism, slavery, imperialism, scientific racism and eugenics. Race has been used to group people into a hierarchical system that identifies, distinguishes and marginalizes some groups across nations, regions and the world. Race also has been used to "establish and justify systems of power, privilege, disenfranchisement and oppression" (National Museum of African American History and Culture, n.d.-b).

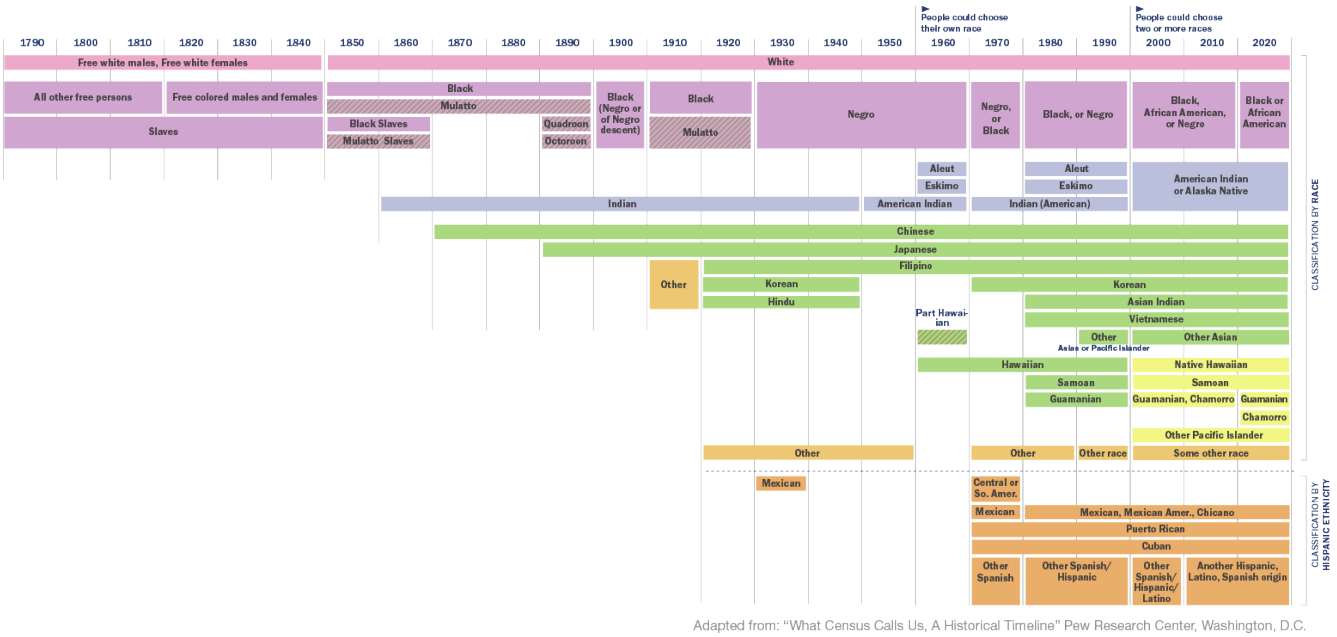
The U.S. federal government notes that the race and ethnicity categories established by the Office of Budget and Management (OMB) are sociopolitical constructs and do not attempt to define race and ethnicity biologically or genetically. Furthermore, these categories reflect a social definition of race and ethnicity recognized in the U.S. and do not conform to biological, anthropological or genetic criteria (United States Office of Management and Budget, 1997).

While genetic ancestry involves analyzing DNA variants and is tied to biology, as described above, the availability of reference populations can influence the ancestral group(s) a person is categorized into. Scientists must decide what level of resolution they will use to group people and what terms they will use to label these genetic ancestry groups. The categories that result when calculating genetic ancestry are sometimes aligned with social constructs of race and ethnicity. Broad genetic ancestry categories are sometimes labelled using the

continents from where people are believed to have origins or roots, such as African, European, and Asian. In this way, descriptors for race, ethnicity and genetic ancestry are often intertwined and misused. Because of these factors, some have described genetic ancestry as socially constructed, too (Dauda et al., 2023).

How are population descriptors used?

The categories used to describe racial and ethnic groups around the world vary. Different ways of categorizing race and ethnicity arise from various historical and modern-day experiences. Some of these experiences include colonization, forced and voluntary migration, racial or ethnic stratification and governance systems. For example, Australia's census form (Australian Bureau of Statistics, 2021) in 2021 asked residents to indicate whether they are "of Aboriginal or Torres Strait Islander origin" and to indicate their "ancestry" using no more than two categories. Some of the ancestry categories included "English," "Italian," "Chinese," "Maori," and "Australian South Sea Islander." There is no mention of "race" on the 2021 form, but it did appear in prior years (Australian Bureau of Statistics, 2011). In some countries, language spoken, origins, or religion matter more than race or ethnicity. France and Italy are examples of countries that do not include race or ethnicity in their census. Some have argued that including race and ethnicity on censuses is essential for understanding and addressing inequities or racism.



Adapted from: "What Census Calls Us, A Historical Timeline" Pew Research Center, Washington, D.C.

Figure 10.10 Racial categories changed over time due to shifts in scientific, political and social thinking about race and ethnicity. According to Pew Research Center (2020), "this graphic displays the different race, ethnicity and origin categories used in the U.S. decennial census, from the first one in 1790 to the latest count in 2020. The category names often changed from one decade to the next, in a reflection of current politics, science and public attitudes. For example, "colored" became "black," with "Negro" and "African American" added later. The term "Negro" was dropped for the 2020 census. Through 1950, census-takers commonly determined the race of the people they counted. From 1960 on, Americans could choose their own race. Starting in 2000, Americans could include themselves in more than one racial category. Before that, many multiracial people were counted in only one racial category" (para. 1). Visit the Pew Research Centre version for an expanded copy in PDF. **Source:** Adapted from "What Census Calls Us, A Historical Timeline" Pew Research Center, Washington, D. C. by National Human Genome Research Institute, used with permission.

When the U.S. government established racial categories around 1790, they were tied to colonialism and flawed science. They were used in population surveys for taxation, government representation, counting enslaved persons and maintaining power (Diamond, 2020). The names and number of categories changed over time due to shifts in scientific, political and social thinking about race and ethnicity (United States Census Bureau, 2015).

The major categories used in the U.S. 2020 Census (U.S. Census Bureau, 2020) included Hispanic, Latino or Spanish for ethnicity, and White, Black or African American, American Indian or Alaskan Native and Asian or Pacific Islander.

In addition to their use in the census, race and ethnicity have been used to measure racial and ethnic health disparities and track progress in reducing inequalities. Race and ethnicity are also commonly used as a **proxy** (Proxy, n.d.). These uses may be helpful for research and public health, especially when other data are unavailable.

Why should we be intentional about how population descriptors are used in genomics research and healthcare?

Advances in genomic medicine greatly amplify the urgency of ensuring the field exemplifies scientific and social accuracy in all our work. Simply stated, the design of some genomic research studies has exacerbated scientific flaws due to how data are being analyzed, interpreted, reported and aligned across data sets. In no small part, this is because of how we *misuse* population descriptors.

Race and ethnicity are not valid or reliable proxies for genetic ancestry. In addition, genetic ancestry is a poor proxy for the geographic area where someone is from, where they currently live or things that may be part of their surrounding environment. Relying on race, ethnicity or genetic ancestry as a proxy for something not measured in research often hides underlying biological, environmental or social factors that may contribute to health and disease. In healthcare, race and ethnicity have been improperly treated as biological or innate characteristics.

In society, there are tangible and measurable impacts of one's racial or ethnic identity on health, wellness and status in the United States, whether self-identified or assigned by someone else. Thus, race and ethnicity may help examine social or political issues, document racial/ethnic health disparities, explore the impact of racial bias in health service delivery (Smedley et al., 2003) and monitor diversity, equity and inclusion efforts (https://diversity.nih.gov/about-us/population-underrepresented) within the biomedical workforce. Directly measuring and analyzing social determinants of health (SDOH), such as racism, violence, access to nutritious food or safe water, or exposure to trees and nature, would improve the rigour and usefulness of research. A growing collection of SDOH measures is available in a toolkit for researchers (PhenX Toolkit, 2024-b).

In all types of research, when using population descriptors, researchers should be clear and transparent about which population descriptor(s) they are using, how they are measured and why they were chosen. Researchers should have a reasonable hypothesis for why specific descriptors may or may not be important to their research questions. Research should use labels and categories that accurately reflect what is being measured. Researchers should carefully consider whether race, ethnicity or genetic ancestry directly causes the health differences across individuals or groups. If proxies are used in research because data of interest are unavailable or cannot be collected, then the challenges and limitations of doing so should be acknowledged.

A closer look: Measuring "race" in heart disease research

Imagine three different studies that look at the severity of heart disease among people living in three different regions of the United States. "Race" is one of several variables analyzed in each study:

- The first study measures race by asking participants to select a category that best describes their race by

checking a box on a form.

- The second study measures race by asking each participant for a saliva sample and using genetic analysis to group study participants into different races.
- The third study uses the birth certificates of participants and their parents to assign a race to each participant.

All three studies use similar labels — Black, White, Native American, Hispanic, Asian and Other — when reporting their findings of heart disease across groups. After completing their analysis, all three studies conclude that "race" is a key factor in the severity of heart disease.

In this scenario, the same population descriptor and group labels are used in each study, but their measurements are different and range from self-reporting to DNA analysis to using vital records. In the second study, race and genetic ancestry appear to be merged as if they are similar or equal. We don't know from this scenario why each study is including race as a variable. The reasons may be varied.

Figure 10.11 In this scenario, three different approaches are used to measure "race," including self-reported race, DNA analysis and vital records. **Source:** National Human Genome Research Institute, PDM with attribution.

Suppose studies are unclear or inconsistent in the labels, definitions, measurements or justifications used for population descriptors in research. Our ability to advance science and improve health outcomes is compromised. For example, when research approaches are not specified, it is hard to repeat a study to confirm its accuracy or to see if the same outcome occurs in a different population or part of the world. Furthermore, broad categories for genetic ancestry can obscure DNA variation relevant to understanding certain health conditions (Rotimi & Jorde, 2010).

Poor use of population descriptors can also cause harm to communities. Findings from such studies are more likely to be misinterpreted and misused. For example, readers may believe there is something biological about race when a study uses DNA analysis to analyze "race" differences. Over the last seven decades, various population descriptors have been used in genomic research studies and have varied (Ganguly, 2021).

Using population descriptors in genomic and biomedical research is a critical scientific issue with varied ethical, legal and social implications (ELSI). NHGRI will continue focusing on this issue to promote the ethical, responsible and scientifically rigorous advancement of genomic science, genomic medicine and ELSI research. NHGRI is also focusing on this issue to:

- Recognize that people have been and continue to be harmed by the misuse of race in genomic research and the misinterpretation of research findings.
- Avoid repeating mistakes of the past, which have caused immediate and long-lasting harm to minoritized and disenfranchised groups here in the U.S. and worldwide.
- Earn the public's trust by ensuring that researchers thoughtfully consider whether, when and how to use population descriptors and ensure they are used ethically.
- Build and maintain trust in science among those we hope will participate in genomic research.
- Ensure a more complete understanding of the diversity across people who participate in research.
- Ensure that all populations benefit from advances in genomic and biomedical research.
- Improve health equity and eliminate disparities in genomic medicine.

Looking forward

Understanding the true role of genomics in health and wellness will require careful attention to the full spectrum of potential contributing factors, including genomic, biological or clinical traits; components of the natural, built or social environment in which people live; and more significant systemic or structural issues. Clarity and specificity around population descriptors used in genomic research can improve the scientific integrity of research while also showing respect for the people represented in genomic research.

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10.5 NURSING IMPLICATIONS

Implications for Nursing Practice

Each province and territory in Canada has a nursing regulatory body that sets out the professional and practice standards and entry-level competencies. These are based on the Canadian Nursing Associations (CNA) *Framework for the Practice of Registered Nurses in Canada* (2015). Nurses practicing in genomics apply these standards to ethical issues as they arise. These standards and competencies promote the provision of safe, ethical, compassionate, competent, and evidence-informed nursing care. Nurses must consider issues around consent, privacy, and confidentiality when working with clients and families receiving genomic services. Trauma-informed practices are a vital component of genomic nursing care, particularly given historical harms related to genetics and genomics. Establishing a therapeutic nurse-client relationship can help build trust and promote informed decision-making.

The CNA Code of Ethics (the Code) provides a framework to guide nurses in navigating ethical dilemmas and decision-making that align with professional values and principles. In addition, the Code promotes equity and addresses social determinants of health by encouraging nurses to challenge systems of oppression, discrimination, and inequity in healthcare. The Code Recognizes the importance of culturally safe care, particularly in addressing the health needs of Indigenous peoples and other communities that experience disproportionate burdens. Thus, the Code can serve as a resource for nurses to address ethical issues arising from genomics-informed nursing. The previous version of the Code (CNA, 2008) included mention of genetic endowment as a health determinant (Puddester et al., 2023). Additionally, the document noted that advances in genetics and genomics impact healthcare system transformation in Canada (Puddester et al., 2023). The absence of any mention of genetics or genomics in the current (CNA, 2017) version of the Code is concerning, given the mainstreaming of genetics and genomics in medicine.

The following article explores how the American Nurses Association (ANA) Code of Ethics can be applied to genomics nursing, emphasizing the ethical responsibilities of nurses working in this rapidly evolving area. The article explains how the core principles of the ANA code – including respect for persons, beneficence, justice, and the nurse's duty to advocate for patients – are applicable to genetic/genomic nursing. Additionally, it highlights the importance of informed consent, ensuring patients understand complex genetic information, and the need to protect patient privacy and confidentiality.

Although the ANA Code of Ethics only applies to American nurses, there are many similarities to the CNA Code of ethics. While both the CNA and ANA Codes of Ethics align in their commitment to patient care, advocacy, and professional integrity, the CNA Code reflects Canada's focus on global health, cultural safety, and reconciliation with Indigenous communities. In contrast, the ANA Code addresses issues relevant

to the U.S., such as privatized healthcare and disaster response. These distinctions highlight how each code adapts ethical nursing principles to meet the specific needs of their populations and healthcare systems.

The article below focuses on the nurse's role in patient education, advocacy, and addressing health disparities related to access to genetic services. While the specific context is American, the ethical considerations and the role of the nurse outlined in this article can be readily related to the CNA code of ethics and to the practice of nursing in Canada.

Read

Tluczek, A., Twal, M. E., Beamer, L. C., Burton, C. W., Darmofal, L., Kracun, M., Zanni, K. L., & Turner, M. (2019). How American nurses association code of ethics informs genetic/genomic nursing. *Nursing ethics*, *26*(5), 1505-1517. https://doi.org/10.1177/0969733018767248

Questions for Reflection

While reading the above article, consider the following questions:

1. How can nurses ensure truly informed consent when patients may have limited health literacy? Consider the challenges nurses face in conveying complicated genetic concepts, such as DNA, variants, and inheritance patterns.
2. How can nurses balance the patient's right to know with the right not-to-know, particularly in cases where genetic findings may have no clear treatment?
3. In what ways can nurses advocate for equitable access to genetic/genomic services and address potential health disparities? Given that genetic technologies and information are not always equally available or accessible to all populations, what steps can nurses take to ensure that all patients receive appropriate genetic services?
4. What strategies can nurses use to protect patient privacy and confidentiality in the age of electronic health records and increased genetic testing?

5. How does having knowledge and awareness of the eugenic past contribute to trauma-informed care and ensuring a safe practice environment?

Position Statements

A position statement is a strategic tool used to define and communicate an organization's perspective on a topic, provide guidance, and advocate for change. It is essential for influencing practice, shaping policy, and fostering awareness and alignment on critical issues.

The International Society of Nurses in Genetics (ISONG) have issued policy statements that align with nurses responsibilities related to ethical issues arising from genomics. Note that these are also founded on nursing guidance from the United States, therefore, should be read with a critical view to their relevance in the Canadian context.

Briefly review the following documents:

- Access to Genomic Healthcare: The Role of the Nurse [PDF] (https://www.isong.org/resources/Documents/PS_Access%20to%20Genomic%20Healthcare_November%202021.pdf)
- Privacy and Confidentiality of Genetic Information: The Role of the Nurse [PDF] (https://www.isong.org/resources/Documents/ISONG%20Position%20Statement_Privacy%20and%20Confidentiality%20of%20Genetic%20Information%20approved%20May%201%202018.pdf)
- Informed Decision-Making and Consent Related to Genetic Testing (Clinical and Research): The Role of Nursing [PDF] (https://www.isong.org/resources/Documents/Informed%20decision%20making%20position%20statement%20approved%20Dec%202018.pdf)

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10.6 UNIT SUMMARY AND REVIEW

Key Takeaways

Genomic healthcare and research raise numerous ethical, legal, and social issues, such as privacy and confidentiality, informed consent, genetic discrimination, and the implications of gene editing and biobanking. Addressing these concerns requires careful consideration of justice, equity, and access to ensure ethical and fair practices. Protections like Canada's Genetic Non-Discrimination Act (GNDA) aim to safeguard against misuse of genetic data.The history of eugenics highlights its scientifically flawed and discriminatory roots, which have caused widespread harm through practices like forced sterilization, segregation, and social exclusion. Eugenics gained global traction in the 20th century, driven by racist, ableist, and colonial ideologies, influencing policies in countries such as Nazi Germany, the United States, Canada, and beyond. While the most infamous applications include Nazi racial policies and sterilizations targeting marginalized groups, eugenics also shaped immigration and public health policies in the U.S. and Canada, with Indigenous populations disproportionately affected. Modern concerns center on the potential for new genomic technologies to revive eugenic-like practices, emphasizing the need for ethical vigilance in genomics research and healthcare

Population descriptors, such as race, ethnicity, skin colour, and genetic ancestry, are essential yet complex tools in genomics and healthcare research. Historically, their misuse has justified eugenics through the practice of scientific racism, and marginalized groups, contributing to health disparities and social inequities. The National Academies of Sciences, Engineering, and Medicine (NASEM) has issued recommendations to transform how these descriptors are applied in research, emphasizing the importance of understanding their distinctions and implications.

Descriptors like race and ethnicity are social constructs, often conflated with genetic ancestry, a biological measure based on inherited DNA. While genetic ancestry provides insights into human migration and variation, most genetic differences occur within populations rather than between them. Misinterpretation of these terms can reinforce stereotypes and inequities. Effective use of

population descriptors requires researchers and healthcare providers to consider their relevance and limitations, acknowledging their social, historical, and scientific contexts. Greater education and precision in their application are needed to ensure ethical and equitable practices in research and healthcare.

The Canadian Nurses Association (CNA) Code of Ethics provides a foundational framework to guide nurses in navigating ethical challenges, including those related to equity, culturally safe care, and social determinants of health. The integration of principles from both the CNA and American Nurses Association (ANA) Codes of Ethics emphasizes the ethical responsibilities of nurses in informed consent, patient advocacy, education, and addressing disparities in access to genetic services, which are critical to ensuring equitable and patient-centered care in the genomic era.

Resource

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Additional Optional Readings

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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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