



## CHAPTER 17

# Genetics and Identity: Ethical Considerations in Practice, Policy, and Research

**Alice Virani and Drew B. A. Clark**

*“During the past two decades, the field of human genetics has undergone significant change. The sequencing of the human genome has fueled understanding of the relationship between genetic variation and human health. Demand is such that clinical nurses and physicians working in a variety of clinical disciplines are now required to integrate genetics into routine care.”* (White et al., 2020, p. 1149)

### **The Field of Genetics and Genomics**

**THE HUMAN GENOME PROJECT**, an international effort to map the entire human genome physically and functionally, was completed in 2003. The completion of the Human Genome Project spurred on significant growth and development in the field of genetics and genomics. Initially, the field of genetic testing focused on determining risk based on family history and offering genetic testing for single gene disorders inherited in a predictable pattern, where testing was

available. Now genetic technology enables testing, genome sequencing, and screening for more than just single gene disorders; it also offers opportunities to understand the underpinnings of complex conditions, to determine whether a patient will respond favourably or unfavourably to certain pharmaceuticals (pharmacogenomics), to target cancer therapy, and to identify predispositions to future conditions. Preimplantation genetic diagnosis and prenatal testing, for example, allow prospective parents to predict and select risk of certain genetic conditions in embryos prior to implantation, or in developing fetuses during pregnancy.

Genetic testing and screening have evolved from being offered only in specialized tertiary or university hospitals to being readily available and used in primary care, cardiology, cancer care, and midwifery, among other places. Innovations in genetic and genomic technology have led to a significant decrease in cost and corresponding increase in accessibility to genetic testing and services. Over the last twenty years—the emergence of direct-to-consumer testing—has changed the landscape of genetic testing (Allyse et al., 2018). At-home test kits can now be accessed to obtain information about an individual's geographic ancestry, as well as their predisposition for certain conditions in the future.

Development of genetic and genomic technology has led to a concomitant recognition of the need for legislation and guidance about acceptable use of such technology. For example, there have been calls for international moratoriums on gene cloning and use of human germline editing (i.e., technology that changes heritable DNA in sperm, eggs, or embryos) to make genetically modified humans (Lander et al, 2019). Some countries with more widespread access to genetic and genomic technologies have put legislation in place to prevent forms of genetic discrimination in employment and life insurance, providing protection for individuals seeking genetic or pre-disposition testing for personal health and life planning (*Genetic Information Nondiscrimination Act*, 2008; Office of the Privacy Commissioner of Canada, 2017). In addition, professional bodies have offered guidance around key areas of ethical complexity including genetic testing in the pediatric context and the return of incidental findings, which we discuss later in this

chapter (Boycott et al., 2015; Committee on Bioethics, 2001; Miller et al., 2021).

Genetic and genomic research has also resulted in problematic practices, particularly for groups marginalized in Canadian and United States (US) health systems (e.g., Indigenous and racialized people). For example, there are multiple incidences of Indigenous groups being subject to genetic research to which they did not consent (Begay et al., 2019). Mistrust of genetic research is also apparent in research recruitment disparities among Black Americans. Research that fails to account for the immense genetic diversity within marginalized communities may lead to further health disparities (Scherr et al., 2019).

Clinical applications of genetic and genomic technologies hold potential to relieve suffering and promote well-being. However, they also raise ethical concerns in relation to marginalized populations. Of significant concern is the potential for eugenic applications which target certain genes based on subjective views of the burden of living with conditions related to particular genetic expressions. Thus, further advancements in genetic research carry implications not only for the human genome, but also for the value of human diversity within our societies.

In this chapter, we begin with an overview of genetic and genomic tools now used in a broad range of health care settings. These include genetic testing for single gene disorders, genetic screening, whole exome and whole genome sequencing, direct-to-consumer testing, prenatal screening, and preimplantation genetic diagnosis. We discuss complexity in analysis and interpretation of genetic findings. Lastly, we highlight ethical issues relevant to nurses and advanced practice nurse leaders related to informed consent, variants of uncertain significance, incidental findings, continuation and termination of pregnancy, and access to genetic technologies.

In the section *Counselling Individuals and Families*, we focus on decision making surrounding genetic and genomic technologies for individuals and families. We highlight the relevance of genetics and genomics throughout the human life cycle through an exploration of prenatal genetics, newborn screening and sequencing, inherited conditions with pediatric and adult onset, and multifactorial disorders which emerge in adulthood. We identify

how approaches to genetic counselling for people facing difficult decisions can effectively be grounded in feminist, relational ethics. Finally, we discuss ethical challenges in counselling related to working with families, newborn screening, genomic sequencing, prenatal and preimplantation genetic technologies, direct-to-consumer genetic testing, and genetic enhancement.

In the section *Genetics, Identity, and Society*, we consider the intersections of genetics and identity; for example, how human identity relates to genetics and how access to genetic information can shift identity. We consider the role of dominant cultural values in shaping biases concerning “favourable” and “unfavourable” traits and how such assignments can lead to practices, policies, and research that are arguably eugenic in nature. Here, we explore challenging ethical questions on how we should proceed as our technological capabilities continue to expand.

Finally, in the *Practical Challenges and Opportunities for Nurses* section, we include an overview of the role of advanced practice nurses in supporting individuals and families making decisions related to genetic testing. Areas of focus include education surrounding genetic and genomic technologies, communication with patients and families, decisional support grounded in relational ethics, culturally safer practices, and policy. Throughout the chapter, we provide several case examples to highlight common ethical challenges related to genetics and identity in diverse Canadian health care practice settings.

## **Genetic and Genomic Testing and Screening**

Genetic testing initially focused on identifying a specific, known, disease-causing mutation in a single gene, such as one of the known mutations in the *BRCA1/2* genes associated with hereditary breast and ovarian cancer. As technology has evolved and costs of testing and analysis have decreased, further options have become available (Guzauskas et al., 2020; National Human Genome Research Institute, 2021). For example, an individual with a personal history of inherited cardiomyopathy can now access gene panel testing, which looks for mutations in multiple genes associated with this condition. Additionally, over the last decade, more patients have

been offered whole genome sequencing or whole exome sequencing (that is, the part of the genome usually correlated with genetic conditions).<sup>1</sup> A child with an undiagnosed developmental difference (e.g., significant cognitive delays and physical differences) can, for instance, have either their whole genome or exome sequenced to determine if there is a genetic basis for their phenotype (Elliott et al., 2019; Ontario Health [Quality], 2020).

There are also a number of technologies that may be used during the prenatal period. One is genetic screening, which determines, for example, whether a pregnant person may have an increased chance of carrying a fetus with a chromosomal difference such as Down syndrome or Trisomy 18. Screening tests are followed by diagnostic tests, such as amniocentesis or chorionic villus sampling. These diagnostic tests can pinpoint single gene conditions such as cystic fibrosis, alpha and beta thalassemia, and autosomal dominant polycystic kidney disease. Preimplantation genetic diagnosis is a tool that enables genetic profiling of an embryo during in vitro fertilization (IVF) prior to implantation of the embryo. Genome editing—involving targeted genetic modification of a cell's genome through cutting, inserting, or otherwise altering DNA in specific somatic (non-heritable) cells in an individual with a specific genetic condition—offers potential treatment promise for those with genetic conditions. More controversially, the ability to perform germline editing (on sperm, eggs, or embryos) is now more technologically feasible. This process—which affects all cells in an organism—ensures that genetic conditions are no longer passed down through generations. However, this technology is not available clinically due to an international moratorium based on ethical and safety concerns. These concerns relate to the moral status of the embryos and the potential for permanent modification on the germline to affect future generations, as well as off-target implications whereby germline editing results in unplanned, harmful changes (Greely, 2019; Hildt, 2016; Ormond et al., 2017).

## Ethical Challenges in Practice

### Informed Consent

One of the foremost ethical considerations in genetic and genomic testing relates to ensuring adequate, informed consent. Traditional, individualistic models of consent may not be sufficient in genetic counselling. This is a uniquely complex area of health care: because genes are familial in nature, testing may have implications and ripple effects not just for an individual patient, but also for their close and more distant relatives, as well as their wider community. In the Ethics in Practice example that follows, we describe some of the concerns that are evident in relation to genetic testing.

#### **ETHICS IN PRACTICE 17-1**

### *Informed Consent*

Abigail is a nurse practitioner (NP) in a remote community with a large Indigenous population. Abigail was born in the community and has served as the primary health care provider there for over a decade. Bill, one of Abigail's Indigenous patients, recently came to her with a letter he received from a cousin whom he has not seen in years. The letter states that his cousin, who now lives in a large city, has been found to have long QT syndrome. This condition is prevalent in certain Indigenous communities and causes an abnormal QT heart rhythm and may result in seizures, fainting, or even sudden death. The letter states that Bill is at risk and should get tested. Bill is distressed and very anxious upon receiving this information. He states that he is concerned about what might be done with his DNA if he goes ahead with testing. However, he is also concerned about his duty to share this information with his own siblings and children, whom Abigail also sees as part of her practice.

In the pediatric setting—or in any setting where testing may benefit someone who does not possess the capacity to consent—issues related to autonomy and choice can become even more complex. In general, there is recognition that a child or youth should only be offered genetic testing that will benefit them in their childhood. For adult-onset disorders, professional guidelines maintain that, in general, a minor's self-determination should be respected by delaying testing until the minor can make a fully

autonomous choice as an adult (Committee on Bioethics, 2001). However, this has been challenged as being overly paternalistic in the context of minor youth who have the capacity to make such decisions, particularly when the genetic information may have immediate impacts in terms of life planning and relieving anxiety about the unknown. Additionally, while this approach generally stands up to ethical scrutiny, it is complicated when incidental findings revealed through pediatric genome sequencing may reveal not only the child's risk for an adult-onset cancer, but also their parent's risk. Revealing adult-onset conditions during childhood may take away a child's right to decide, but the child also has a vested interest in their parent's well-being which would necessitate disclosure of the incidental finding. Knowledge needed for parents to take preventative measures (such as, screening or prophylactic surgery) may be identified through such testing; however, disclosing this information is at odds with delaying revealing a child's risk for an adult-onset disorder until the child has the ability to make an informed choice about whether they want to receive the information (see Chapter 12 for more information about children and informed consent).

### **Variants of Uncertain Significance and Incidental Findings**

Another ethical issue that arises in genetic counselling concerns *variants of uncertain significance*. These occur when a mutation in the gene of interest being tested is revealed, but it is unclear whether this mutation is benign or disease-causing. Sensitive disclosure of such findings is essential to avoid unnecessary anxiety and also ensure follow-up is possible if this variant is reclassified in the future as either benign or disease causing. Discussions regarding whether the health care provider or the patient is responsible for initiating future follow-up to determine whether a variant is reclassified as benign or disease-causing are complex, and involve ethical tensions related to duties of health care professionals, the length of the therapeutic relationship, and potential for disclosure to result in harm or benefit.

Incidental findings are those known to have clinical significance that are uncovered unintentionally in the course of testing. Such findings are not related to the initial reason for the test and have

been the subject of many ethical debates, including the “right to know” versus the “right not to know” (Christenhusz et al., 2013; Ells & Thombs, 2014). Some have argued that there is an ethical duty to disclose findings that are clinically actionable, as many people may wish to be informed; for example, in situations where individuals may decide to access further screening, or pursue prophylactic surgeries to reduce the risk of developing a health condition (Green et al., 2013; McGuire et al., 2013). Others have argued that mandatory disclosure fails to recognize that a significant portion of individuals at risk for genetic conditions prefer not to know their risk, as knowledge of this risk may cause unwanted distress and anxiety (Burke et al., 2013). Determining whether an individual would want to know about their incidental findings as part of the informed consent process may decrease the potential for unforeseen moral dilemmas on the part of clinicians (Cox & Starzomski, 2004). However, unforeseen incidental findings are sometimes unavoidable. As noted above, particularly challenging ethical issues may arise in the context of pediatric testing, wherein the incidental finding may only have relevance for the patient decades down the line, but may be immediately salient for the parents (and thus impactful, too, for the child).

### **Prenatal Screening and Testing**

Genetic screening and diagnostic tests available during the prenatal period may lead to challenging ethical decisions related to continuation or termination of pregnancy. Preimplantation genetic diagnosis exacerbates already heated ethical debates around in vitro fertilization regarding the burden on the individual undergoing fertility treatments, the inequities in access to this technology, and the creation of multiple embryos, many of which will ultimately be used for other purposes (e.g., donated for research) (Dondorp & de Wert, 2019). Human germline editing, while not currently available, has raised similar ethical questions, in addition to bigger concerns related to the acceptability of permanently altering the human genome (such that changes are inherited through generations). Additionally, the safety of such technology and potential for off-target effects (whereby there are unintended changes in other areas of the genome with unknown consequences) has also raised con-

cern (Ormond et al., 2017). Each of these prenatal technologies raises ethical questions related to justice; they underline, for example, issues of ableist stigmatization of genetic difference, discrimination, and eugenic messaging. Issues of equity also arise related to access and costs of these technologies.

### **Direct-to-Consumer Testing**

Another ethical complexity that has become increasingly apparent over the last decade involves access to genetic technologies provided by direct-to-consumer genetic testing companies. Home testing kits are available for a nominal fee, usually in the range of one to two hundred dollars. Individuals interested in learning more about their genetic histories can send blood or saliva samples to these companies for analysis. While some of the results may be low stakes or curiosity-driven, such as ancestry or risk of male pattern baldness, implications of other test results may be more profound in terms of their clinical significance (e.g., risk for developing Alzheimer's disease). A multitude of questions related to the ethical rationale for offering these tests has arisen, including how to ensure proper informed consent, accuracy of results, protection of personal data, and equitable access to testing. There are also implications for health care systems that are burdened with helping patients make sense of the test results (Caulfield & McGuire, 2012; Hawkins & Ho, 2012; Middleton et al., 2017).

In this section, we have highlighted the evolution of genetic and genomic technology over the last several decades, along with the simultaneous emergence of ethical considerations in relation to the use and application of these technologies. While questions related to discrimination, ableism, privacy, consent, and the familial implications of genetic and genomic testing in an individualistic health care model are not new, the increased availability of such technology has meant that the public, and accordingly health care systems more broadly, increasingly have to grapple with these complex ethical issues. As such, it behooves new and practicing health care providers to have awareness of these issues.

## Counselling Individuals and Families

To illustrate the ethical complexity of genetic and genomic technologies, it is helpful to consider how genetic conditions manifest throughout the lifecycle and how relevant technologies are applied in clinical practice. Preconception or prenatal genetic offerings provide an opportunity to assess risk for certain conditions that manifest during pregnancy due to family history, gamete age, or generalized chance for a condition in a given population. It is more common for genetic testing to be offered following initial screening tests (e.g., non-invasive chromosome screening) or ultrasounds that suggest certain genetic conditions. In these circumstances, further and more invasive testing (e.g., amniocentesis) may be offered to either confirm or rule out a condition. This ensures that the locally available range of reproductive choices, including termination of pregnancy, can be offered during the prenatal period. In the pediatric genetic context, counselling surrounding testing is complicated by the potential for genetic information about a parent being revealed that may have immediate relevance for the health of the parent, and, therefore, the well-being of the child and the entire family.

Genetic testing and genomic sequencing in the adult context also generate discussion and debate. With the increased availability and awareness of genetic tests, practitioners outside specialized genetic centres have become more familiar with obtaining family histories and offering (and interpreting) genetic tests. For example, practitioners in the fields of neurology, cardiology, and oncology have developed specialized clinics to identify and care for individuals at risk for certain conditions (Lynce & Isaacs, 2016; Musunuru et al., 2020; Rexach et al., 2019). This care often includes regular screening tests, prophylactic risk-reducing surgeries and implantation devices, and behavioural modification. While the ability to act on genetic findings to modify risks of clinical manifestation of certain aspects of disease has improved, complexities related to predictive and confirmatory testing remain significant.

## Genetic Counselling and Relational Ethics

### Interpretation of Findings

Genetic and genomic testing mechanisms—and the ensuing results—are complex and require skilled analysis and interpretation. Sound genetic counselling practices are necessary to ensure patients are adequately supported and can provide fully informed consent before undergoing testing. Genetic counselling entails an explanation of complex inheritance patterns, including autosomal dominantly inherited conditions (in which first-degree relatives of an individual with a genetic condition have a 50% chance of inheriting that condition), autosomal recessive conditions (in which there is a 25% chance of having a child with the same condition), X-linked conditions (which usually manifest more severely in males), and multifactorial conditions such as, cardiovascular disease or cancer, which are caused by many genetic and environmental factors (Canadian Association of Genetic Counsellors, n.d.; National Human Genome Research Institute, n.d.; Resta et al., 2006).

Counselling must be tailored according to the specific type of genetic test and expected result. For example, single gene, *fully penetrant* disorders are conditions that have a clear inheritance pattern and will definitely lead to predictable clinical manifestations (e.g., hemochromatosis). The term *variable expressivity* is used when the same genetic profile can have different manifestations, even within a single family (e.g., Marfan syndrome). *Reduced penetrance* is present when some individuals with a genetic mutation will not manifest a condition while others will (e.g., familial cancers). Highly complex multifactorial inheritance involves several genetic and environmental factors that can lead to a condition (e.g., heart disease and diabetes). Finally, interpretation of test results involves explanation of variants of uncertain significance and incidental findings.

### Relational Ethics

Genetic counselling is strongly rooted in feminist relational ethics approaches (i.e., ethics of care) (Jamal et al., 2020; Ryan et al., 2015). In contrast to predominantly individualistic health care models,

trained genetic counsellors embrace the interconnectedness and interdependence of individuals. The context in which patients live, as well as their relationships with others, help shape the ways in which counselling and health services are provided. Using relational ethics supports practitioners to respectfully attend to the complex interplay of familial, social, and cultural relationships, as well as power dynamics, learning styles, and cultural ways of knowing (Hauskeller, 2020; Noddings, 2012). In addition to its grounding in relational ethics, genetic counselling is non-directive in nature, meaning practitioners aim to create environments in which empathic listening supports patients to feel understood, to have their personal values clarified and validated, and to reflect and make choices that are in keeping with their beliefs and circumstances.

## **Ethical Challenges in Practice**

### **Familial Risk**

One complicating factor in providing genetic counselling relates to the familial nature of genetic conditions. For example, when counselling an individual about their familial risk for polycystic kidney disease, a practitioner needs to consider the familial and cultural context of decision making. This includes addressing concerns related to genetic discrimination, as well as contending with ethical tensions surrounding the right not to know one's genetic information. When patients choose not to know, practitioners may experience moral distress as they attempt to balance their duty to protect patient privacy and confidentiality with their perceived and sometimes real duty to warn at-risk relatives of their genetic predisposition. However, as many common conditions—such as heart disease, diabetes, and cancers—are multifactorial in terms of genetic and environmental risk factors, it must be taken into account that risks may be over- or under-estimated. In the Ethics in Practice example that follows we describe challenges that occur in genetic counselling when respecting the divergent wishes of multiple family members.

**ETHICS IN PRACTICE 17-2***Respecting Competing Wishes in a Family*

Emiliano is an advanced practice nurse in a neurology clinic. They have been following a family for several years after the grandfather died of Huntington's disease. Huntington's disease is a dominantly inherited adult-onset condition, which means first-degree relatives have a 50% chance of inheriting the condition. Part of follow-up care involves a neurological exam every six months for each at-risk family member. During a visit to the clinic, Angela, the granddaughter of Emiliano's original patient, mentions that she was recently married and wishes to get pregnant. She states that she would like to undergo predictive testing for Huntington's disease to aid in her reproductive planning. However, Angela's mother Lucia is adamant that she does not want to know her own status and would not consider testing. As a daughter of Emiliano's original patient, Lucia has a 50% chance of having inherited the condition. By supporting Angela's request for testing, Emiliano may inadvertently reveal her mother's Huntington's disease status. How should they proceed?

Relational ethics approaches can be particularly helpful in navigating complex family situations and balancing the needs of various parties whose genetic information is inextricably connected. Practitioners must be skilled at navigating familial and psychosocial implications of genetic knowledge and technologies, such as the potential for family conflict and genetic discrimination. For example, applying individualistic models of consent may be challenging when there are familial and community ramifications to genetic and genomic testing. Practitioners can encourage patients to consider the broad range of repercussions of testing on family members, including implications for genetic status as well as mechanisms to balance harms and benefits. This is done by assisting patients in sharing information with their family members via family counselling sessions or informational letters. The sessions or letters provide relevant information about the condition, inheritance, and potential for testing, which family members can then use to inform their personal decision making.

Use of newborn screening and sequencing technologies has also stimulated ethical debate related to consent, utility, and potential for discrimination. Newborn screening is available and offered

under public health schemes in many countries. Use of these screening programs helps practitioners identify rare metabolic, endocrine, blood, and other disorders in the first few days of life, so that effective treatment may be provided. While such testing in and of itself is widely heralded for improving and lengthening lives, questions have arisen related to appropriate consent on the part of parents, both for the testing itself and for the use of leftover samples for population health planning and research purposes. Utility is also questioned in terms of whether the testing yields significant, actionable health-related information. This is particularly important in relation to the potential for discrimination based on health conditions or predispositions identified through testing. In the Ethics in Practice example that follows we highlight the importance of cultural safety and humility in providing care related to genetics in the prenatal setting.

### **ETHICS IN PRACTICE 17-3**

#### *Navigating Cultural Safety in Prenatal Testing*

Chioma is a nurse practitioner working in a clinic that provides primary care to new immigrant patients. One of Chioma's patients, Fatima, recently went for a prenatal ultrasound. During the ultrasound she was informed that there were findings suggesting Down syndrome. Fatima was offered an amniocentesis test to confirm the diagnosis and have the option for termination of the pregnancy. She returns to Chioma for a follow-up clinic visit very upset. Fatima did not understand that the ultrasound could reveal this information. She now feels pressure to do the amniocentesis test even though she would never consider termination, as it is against her religious and cultural views. Fatima asks for Chioma's guidance on how to proceed.

The use of genomic sequencing in the neonatal period is increasingly available, yet still expensive and not widespread. While such testing may identify the underlying genetic cause of a newborn's congenital differences and suggest appropriate treatments, it can also flag variants of uncertain significance, incidental findings, and predispositions to adult-onset conditions (Johnston et al., 2018). This presents a challenge for practitioners in determining how

much genomic sequence information should be shared with parents, especially when such information may impact their child (e.g., a variant of uncertain significance that might lead to childhood onset diabetes) or even themselves (e.g., a genetic variant that leads to risk of sudden cardiac death).

### **Prenatal and Preimplantation Genetic Technologies**

Prenatal and preimplantation genetic technologies have led to some of the most heated ethical debates regarding appropriate use of genetic technology. Such technologies have expanded reproductive choice and freedom, but also led to an increase in selective termination and a significant decrease in the number of children born with conditions such as Down syndrome and spina bifida. Meanwhile, disability communities have voiced eugenics concerns regarding “genetic genocide,” arguing that offering prenatal genetic screening and testing sends harmful messages to prospective parents about certain traits being inherently problematic, when they need not limit prospects of living a worthwhile life (Parens & Asch, 2003). These critiques extend to societal implications, as wide-scale use and acceptance of prenatal tests sends discriminatory messaging about disability rather than using resources to create more inclusive communities that address the needs of genetically diverse people.

These ethical tensions challenge practitioners to recognize and put aside their own beliefs and preconceived notions about what constitutes a life worth living. Practitioners are bound by their duty to ensure that patients are empowered to make choices that best reflect each individual’s values and context. Practitioners should also be prepared to address ethical concerns related to equity and justice for disability communities that are raised by these technologies.

### **Accessibility of Services**

Direct-to-consumer tests, while considered by some as democratizing genetic testing by expanding access, have also impacted health care providers in settings ranging from primary care to specialized centres. As the availability and popularity of these tests rise, the health care system may face increasing pressure to manage the fallout of such testing, as people turn to the publicly funded health care

system for assistance in interpreting and responding to their results. This may lead to further diagnostic testing—such as, medical imaging or monitoring—which is particularly problematic when the predictive value of direct-to-consumer tests is poor, and people may receive unnecessary follow-up testing in an already overburdened public health care system.

Finally, while genetic technologies that can bestow a perceived enhancement or advantage (e.g., height, intelligence) are not currently clinically available, advancements in gene editing have made such services theoretically possible. Use of so-called “enhancement” technologies raises similar ethical concerns as pre-natal testing: there is a potential for creating an increasingly disparate society. If only specific segments of the population were able to access such technologies, the creation of genetically privileged and marginalized classes could widen existing community and global inequities. As a society, as health professionals, and as individual practitioners, we must consider the ethics of genetic “enhancement,” and how we will respond to future requests to apply this technology in practice.

In this section, we have addressed ways in which genetic conditions affect people across the lifespan. We have also considered the expanded role of genetics in various health care settings. Relational ethics and non-directive counselling have been identified as central to the field of genetic counselling. Ethical challenges are not uncommon for those who help patients and families navigate complex issues related to privacy, confidentiality, consent, culture, diversity, and use of resources.

## **Identity, Genetics, and Society**

The concept of identity concerns who we are, our unique characteristics, how we think about ourselves in relation to one another, and what connects us with others. If we conceptualize identity as socially constructed, we understand it as a phenomenon that emerges through interactions between the individual and others within society. In other words, identity is grounded in how we see ourselves in relation to others. Each of us likely has many facets to our identity; these may include race, Indigeneity, culture, gender,

sexuality, ability, and health. Many identity traits are willingly assumed by an individual; however, identity labels may also be assigned by others without consent of the individual. Some aspects of identity may shift over time in response to acquired information, individual and collective experiences, personal development, and shifting societal norms.

An individual can hold multiple, intersecting cultural and social identities. A person may be Indigenous (e.g., Anishinaabe, Métis), be religious (e.g., Jewish, Catholic), identify with a settler-state (e.g., Canada, United States), be part of a Queer community, be part of a Crip community, or even be a dedicated fan of a particular hockey team. Some identities are accompanied by strong genetic links, while others are not; for example, some families share strong genetic connections, while other families' connections are primarily social (e.g., adoptive families). One well-recognized form of genetic identity is having a genetic lineage that connects a person to family members and to culture. Another form of genetic identity is connection to others who share a common genetic expression (e.g., Down syndrome). These identities may also intersect; for example, sickle cell disease is an inherited blood disorder most common among people of African, Middle Eastern, and Indian descent. The commonality between all of these identities is that they are relational: we are either aligned with, or differentiated from, others in society based on individual traits, many of which have genetic links.

Some aspects of identity, once developed, may remain fairly constant across the lifespan. However, there are also events that can shift a person's identity surrounding family, culture, and health. Of relevance for us in this chapter are experiences related to genetic and genomic information which can affect a person's identity. For example, genetic testing can reveal unexpected parentage or predisposition to disease. Discovering a genetic predisposition to a medical condition can have positive and negative aspects. It could lead to identification as an unwell person or, alternatively, to welcome connections with others with the same genetic condition (Zeiler, 2009). Responses can be complex. For example, a person receiving a negative test result for Huntington's disease may feel both relief with regard to their own health and guilt if other family members have tested positive.

Since identity and its development occur in the context of society, it is important to consider the power of societal norms in shaping identity. A human society can be understood as a large social group of people who live in community and share a set of cultural norms. Identity traits that are considered normative (i.e., perceived as “normal”) vary across cultures and subcultures. However, within a given society, dominant views exist of certain traits as normative and non-normative; by extension, certain traits are therefore considered desirable or undesirable; for example, consider normative expectations surrounding race, gender, sexuality, and ability across various societies. Prevailing dominant cultural values can generate biases concerning favourable and unfavourable traits. An individual’s personal traits, whether self-identified or assigned by others, can determine their social location within a given community. For example, a Deaf or Hard-of-Hearing (DHH) person may be considered disabled by others in some spaces, but not within Deaf communities.

Certain groups may be subject to subtle or overt forms of discrimination or eugenics based on dominant cultural norms (e.g., conscious or unconscious bias against race, religion, sexuality, gender, or ability). Such groups may be deprived of liberties or resources necessary to thrive and reproduce. They may also be subjected to policies and services intended to prevent the existence of future members of the group (e.g., Down syndrome, DHH). Societal norms have the power to shape individual decisions, policy, and resource availability.

### **Ethical Implications**

While some may consider genetic data to be purely objective, it is important to understand the value-laden nature of such information and the technologies used to acquire it (Newell, 2000). In this section, we consider ethical implications of how genetic information is obtained and applied in relation to individual identity, cultural identities, and prenatal decision making. We also examine how bias, expressed through genetic and genomic technologies, iteratively shapes how certain lives are valued in relation to others in ways that may alter the scope of human diversity.

## Individual Identities

As noted in the section on Genetic and Genomic Testing, the ways in which people acquire genetic information is ethically relevant. Some may exercise autonomy in actively choosing to learn this information through, for example, seeking genetic testing for hereditary diseases like breast and ovarian cancer. Others may inadvertently discover identity-shifting genetic information through a direct-to-consumer test or a family member's disclosure. In the case of disclosure by a family member, the individual may not have the opportunity to determine whether they want to know this information, but may benefit from opportunities to seek medical intervention (e.g., screening, prophylactic surgery). Regardless, knowledge of a single gene disorder or predisposition to a condition can change how a person views themselves and how they are treated by family members, health care providers, and society at large.

Furthermore, assumptions made by health care providers on the basis of someone's perceived identity can determine whether they have access to health care services informed by genetic research. For example, multiracial people may not be offered screening for cystic fibrosis based on an assumption that they are not White and, therefore, have low risk of having a child with this condition. In another example, Black Canadians with sickle cell disease presenting in emergency departments with pain crises (i.e., vaso-occlusive crises) have reported encountering medical racism; they are racially profiled as drug-seeking, and denied timely access to necessary care (Favaro et al., 2021; Sickle Cell Awareness Group of Ontario, 2020). Ultimately, genetic information can significantly influence a person's identity, their relationships with others, and their treatment in society (e.g., facing unjust discrimination or stigma, gaining access to social support).

## Cultural Identities

On both individual and cultural levels, genetic information may be either highly valued or considered to have minimal utility. For example, within many Ashkenazi Jewish communities, genetic testing is considered integral to the health of their communities and future generations. Tay-Sachs disease is a life-limiting condition

that historically has disproportionately affected people of Ashkenazi Jewish descent; however, the disease has been largely eradicated within this population. Initiatives that have contributed to this include confidential marriage matching programs and pre-marriage genetic testing (e.g., *Dor Yeshorim*) within Orthodox Jewish communities. A connection between Jewish identity and genetic testing is demonstrated through the religious and cultural obligation to protect one's health (Lipinsky, 2021). This is enacted through genetic testing to prevent multiple heritable conditions, thereby strengthening the genetic future of the community (Lipinsky, 2021).

Another example of genetic information being used to support specific populations has emerged in the form of human rights reparations. The right to identity has been established as a fundamental human right within the United Nations Convention on the Rights of the Child (United Nations, 1989). This right has been argued to include the right to one's genetic identity. During the genocidal military dictatorship in Argentina from 1976–1983, thousands of adults targeted as political dissidents, or sympathizers, were killed or disappeared and approximately 500 babies born in captivity were given to people connected to security forces to be raised in what was considered a morally superior culture (Penchaszadeh, 2015). In 1983, a program was established to use genetic information both to identify human remains and to reunite abducted children with their grandparents.

In Canada, the period of mass child apprehension known as the "Sixties Scoop" resulted in thousands of Indigenous children being taken from their families and placed in adoptive families across Canada and the US, spanning the 1960s into the 1980s (First Nations Studies Program, 2009). Today, survivors continue to reunite with family members and communities with the assistance of direct-to-consumer genetic testing, social media networking, and generational knowledge of Indigenous communities (Martens, 2018). For many, these reconnections are immensely important to identity and well-being, but they do not erase the pain of being estranged from family, community, culture, and the land (MacDonald, 2019). In both the Argentinian and Canadian

examples, genetic information has been important in upholding the rights of stolen children to their cultural identities.

While genetic information can help re-establish cultural identities, human cultures and cultural identities existed long before genetic testing. Genetic information can be useful in establishing connection for some groups, but it does not necessarily function as a sole basis of cultural identity and may even, in some circumstances, threaten strongly held identities. For example, Native American tribes and First Nations have resisted attempts to reduce identity to genetics, instead identifying concepts such as kinship and citizenship as holding cultural relevance for Indigenous identities (TallBear, 2013).

A specific area of ethical concern involves acts of *biocolonialism*, a term that can be used to describe harms that genetic and genomic technologies can cause when used to exploit knowledge and resources from Indigenous peoples (TallBear, 2013). In one such incident, a researcher took samples from the Nuu-Chah-Nulth First Nations (British Columbia, Canada), purportedly for arthritis research, but kept samples for many years, moved them internationally, and conducted highly sensitive research on unrelated topics without the consent of the Nuu-Chah-Nulth people (Wiwchar, 2013). A similar story unfolded in the Havasupai Tribe (Arizona, US) when DNA collected for diabetes research was used without individual or community consent for research on schizophrenia and geographic origins, the latter leading to conclusions that did not align with the tribe's cultural beliefs and claims to traditional lands (Blakemore, 2018; Greenberg, 2020). In 2002, this led the Navajo Nation to issue a moratorium on genetic research. This topic is discussed in further detail in Chapter 2.

Unethical research conduct has perpetuated mistrust of Western medical researchers, contributing to underrepresentation of Indigenous people in genetic and genomic research on various diseases. This has led to efforts to understand Indigenous perspectives on genomic data and support Indigenous people to benefit from this research without exploitation (Morgan et al., 2018). Research ethics guidelines have been created for genomic research with Indigenous communities, specifically addressing the needs for cultural competency, collaboration, capacity building, community

control of data and biological samples, transparency, trust, consent, and accountability (Canadian Institutes of Health Research, Natural Sciences and Engineering Research Council of Canada, & Social Sciences and Humanities Research Council, 2018; Claw et al., 2018; Taniguchi et al., 2012).

### **Prenatal Decisions**

As noted in the previous section, one of the most ethically provocative areas of genetics involves prenatal interventions. Many decisions centre on whether to access screening for genetic differences and how to act on information obtained through any resulting diagnostic testing. In advance of pregnancy, people may think through what available prenatal screening they plan to access and how they will act on findings. For some, continuation of a pregnancy is the only acceptable option, while for others, termination may be preferred under some circumstances (e.g., Trisomy 18, Down syndrome). Other people may only consider these choices when faced with the results of screening. In the realm of preimplantation genetic diagnosis, the timeline is moved back: these choices are not made about an already implanted embryo, but rather about which embryos will be given the opportunity to develop.

In terms of identity and prenatal decision making, a key question is: what does it mean to a person to be a “good (prospective) parent”? There is no simple answer to this question. Each person must answer for themselves the value-laden question of what prospective parents owe their potential future children and what constitutes a life worth living. Some people may highly value genetic diversity or believe that selecting children with specific genetic traits is inconsistent with their faith (e.g., “playing God”). Others may wish to give their potential future children every possible advantage through genetic and genomic technologies (Savulescu, 2001), believing it would be wrong to bring a child into existence with a preventable genetic condition. Finally, some parents who have a child with a genetic condition may wish to have another child via IVF with preimplantation diagnosis who shares DNA with their sibling, but not their genetic condition (i.e., saviour sibling), in hopes that the subsequent child’s stem cells can be used to treat the older sibling’s condition (Glover, 2006).

People's values, which are connected to their identities, guide them as they make decisions about prenatal screening, continuation and termination of pregnancy, and preimplantation genetic diagnosis. However, individuals' values may come into conflict and their identities may be challenged as they make these decisions. People's partners and other influential figures in their lives (e.g., parents, friends, religious community members) may hold different values, adding complexity to decision-making processes. Ultimately, choices about prenatal interventions can influence both how a person sees themselves and how others view them.

### **Genetic Diversity and Health Policy**

Underlying many decisions about genetic and genomic technologies are understandings of how genetic conditions affect people's lives. These ideas can differ greatly and may be impacted by personal experiences and societal norms. For example, genetic diversity is separated into genetic norms as well as disabling genetic disorders on a societal level. What it means to be disabled is fundamentally based on socially constructed differences: these differences may be framed as undesirable at best, constitutive of a life not worth living at worst. If a difference judged to be disabling has a genetic link, screening may be developed. Once screening is available, a person may decide to terminate a pregnancy or not transfer an embryo based on available genetic information.

A key ethical tension that arises here is the eugenic application of genetic and genomic technologies to eliminate certain differences. While many people living with disabilities report good quality of life, perceptions of quality of life on the part of non-disabled people are often much more negative (Childress, 2003; Goering, 2008). The absence of disabled voices from discourses and policy development on the use of genetic and genomic technologies to eliminate disability is ethically problematic (Boardman et al., 2018). In recent years, the eugenic application of genetic screening has become evident, perhaps most clearly in the precipitous drop in births of infants with Down syndrome, with the termination rate for pregnancies with a positive screening result exceeding 90% in some countries (Will, 2018).

Ideas about what constitutes disability or socially unacceptable traits can shift over time. For example, some sexual orientations have been pathologized in the past, but are now considered a natural part of human diversity. Work is ongoing to depathologize certain genders, efforts that are reflected in human rights protections within Canada, notably through the recent addition of gender identity and gender expression as protected categories within the *Canadian Human Rights Act* (Bill C-16, 2017). However, given this history, it is understandable that people from marginalized groups may be wary of researchers seeking to pinpoint genetic causes of difference (e.g., gender identity), as there is potential for this information to not only shift how people identify themselves and are identified by others, but also for it to be used to attempt to eradicate segments of the population (Rajkovic et al., 2021). Another concern is the risk of creating a genetically privileged class with access to genetic screening, prophylactic measures, and pharmacogenetic treatments, alongside a genetically marginalized class without access to such technologies.

Finally, ableism not only manifests within the social construction of disability, but also in the (re)production of environments that are inhospitable to some, thereby reinforcing the idea of disability by limiting inclusion in society for many with genetic and other differences. From a disability ethics perspective, it can be said that it is society that is disabling (i.e., by creating and perpetuating norms of exclusion and oppression), rather than genetics or physiology (Newell, 2000). The ethical imperative to uphold the dignity and worth of all people requires us to consider how genetic and genomic technologies are applied on a societal level and the harms that may be caused by fostering bias against those identified as disabled. It calls into question why some forms of diversity are considered a disability or undesirable within certain cultures, as well as the implications of eliminating genetic differences based on potentially temporary socially constructed ideas of which lives are most worth living. To uphold the ideals of justice, equity, diversity, inclusion, and cultural safety, we must consider whether more resources should be allocated to creating societies in which people with genetic differences are better supported, to counteract eugenic elimination of such differences. As we seek to find the best pathways

forward as individuals, families, clinicians, scientists, and policy-makers, one thing is clear—we must ensure those with genetic differences are centred among the stakeholders involved in these deliberations (Conti, 2017). Ultimately, our use of genetic and genomic technologies, along with our treatment of people with genetic and other differences, reflects and shapes who we are as a society—our collective societal identity. For more discussion on disability please see Chapter 14.

We have explored the connection between genetics and identity on both individual and cultural levels. Genetic information can be used in beneficial ways; for example, to reduce the occurrence of genetic disease in a population and to restore genetic identity following human rights violations. However, genetic and genomic technologies can also be used in problematic ways—as evidenced through biocolonialism, unethical research practices, and eugenic policies—raising questions about the ethical use and limitations of these technologies. We have highlighted many ethical challenges in integrating genetic and genomic technologies within our health care systems in this chapter. Next, we focus on the role advanced practice nurse leaders can play in supporting patients, families, and interprofessional practice related to genetic and genomic technologies. We focus specifically on the areas of education, communication, relational ethics, cultural safety, and policy.

## **Practical Challenges and Opportunities for Nurses**

### **Education**

Nurses can access continuing professional education to increase their knowledge of genetic and genomic technology applications within health care. This includes understanding the impacts of having—and of being at risk for—genetic conditions, and how it affects patients and families across their lifespan (Sloand et al., 2018). Nurses can better support patients and families when they are familiar with the history and application of technologies that may be offered or accessed, including genetic testing, genome sequencing, prenatal screening, preimplantation genetic diagnosis, and, in the

future, gene editing (Lea et al., 2011). Knowledge of the availability, utility, and predictive value of certain tests, particularly those offered through direct-to-consumer genetic testing companies, may be helpful in supporting patients and families who have questions about testing and health risks. Advanced practice nurse leaders can make important contributions to the field, whether through self-study, continuing professional education, educating the public, or designing nursing courses about genetics and genomics.

### **Communication**

The ability to communicate about complex genetic information at individual, family, and collective levels is essential for supporting decision making about genetic and genomic technologies (Medendorp et al., 2021; Snyder et al., 2009). Nurses are well-positioned to provide this support, drawing on education and experience in caring for patients and families, particularly those with complex relationships (Williamson & LeBlanc, 2008). Clear, tailored, and nondirective counselling can support informed consent about pursuing or declining testing or intervention. Building interprofessional relationships with genetics specialists and other health care providers in genomic medicine is key, as nurses increasingly need to have genomic acumen to support patients and families from their position as trusted health professionals (Montgomery, 2017). The ability of nurses to respond to patient questions (e.g., around informing family members, or whether and how to follow up on results of direct-to-consumer tests) and facilitate family and patient information sessions can reduce demand on genetic counsellors and geneticists, making care more accessible overall (Yoes & Thomas, 2020). Expanding the scope of nursing practice in this manner may also have important policy implications in terms of who can provide genetic services.

### **Relational Ethics**

Relational ethics provides a strong foundation for engagement among nurses, genetic counsellors, geneticists, patients, and families. Drawing on relational ethics approaches, nurses are well-prepared to work with patients and families facing difficult

decisions, such as whether to pursue predictive testing or the continuation or termination of pregnancy (Evans et al., 2004). Similarly, working with patients and families to clarify their values and perspectives is essential for supporting sound decisions that may have profound impacts on identity. For example, nurses should be prepared to explore people's ideas of what "a life worth living" means to them and how that shapes their decisions in relation to genetic and genomic technologies (Janvier, 2011; Janvier & Watkins, 2013). In addition, expertise or access to supports in areas such as grief counselling, conflict resolution, nondirective counselling, and anticipatory guidance will allow nurses to support patients and families through these complex and values-based decisions.

### **Cultural Safety**

Cultural safety is defined by the BC First Nations Health Authority (n.d.) as "an outcome based on respectful engagement that recognizes and strives to address power imbalances inherent in the health care system. It results in an environment free of racism and discrimination, where people feel safe when receiving health care" (p. 5). Broadly speaking, nurses can support cultural safety through learning about the histories (e.g., since time immemorial, colonization), cultures, ways of knowing, and health care experiences (e.g., medical racism) of the communities with which they work, through promoting mutual trust through open communication, and through providing individualized care that accounts for physical, psychosocial, and spiritual needs (Greenwood, 2019; Morgan et al., 2021). Nurses should be aware that based on their histories, identities, and lived experiences, people have diverse relationships with genetic and genomic technologies. Nurses should, therefore, avoid making assumptions about what decisions people will make based on stereotypes about cultural identities (Ward et al., 2016). In the context of genetic counselling, attention should be paid to historical context, trust, language, and beliefs, as well as individual and cultural values in relation to genetic differences, dis/ability, and relevance of genetic and genomic technologies to decision making (Morgan et al., 2018; Shen et al.,

2018). Further information about cultural safety and research in Indigenous communities can be found in Chapter 2 and Chapter 5.

## Policy

Nurses, alone or in collaboration with interprofessional colleagues, can play a key role in shaping institutional and governmental policy related to genetic and genomic technologies (Limoges & Carlsson, 2020). Examples of policy issues of relevance based on current literature include establishing roles and genetic and genomic competencies for nurses; resource allocation for patients and families (e.g., equitable access to testing, dealing with demands for unnecessary testing); limiting testing that leads to eugenic outcomes; and data privacy (Bottorff et al., 2005; Jamal, 2015; Taniguchi et al., 2012). In the future, issues such as germline editing and genetic enhancement may need to be addressed through policy review and development. Nurses can play an important role in ensuring relevant groups (e.g., the disability community) are represented in policy development processes, working toward a health care system that allows people to benefit from genetic technologies without reinforcing ableism or promoting eugenic practices.

## Conclusion

In this chapter, we have reviewed genetic and genomic tools currently in use for testing, screening, and diagnosis, and how application of these technologies raises ethical issues related to informed consent, privacy, medical decision making, and resource allocation. We presented relational ethics and nondirective counselling as approaches applied in genetic counselling to support individuals and families making complex decisions. In discussing genetics in relation to identity and society, we delved into unresolved ethical concerns about how to balance beneficial application of genetic technologies with harms related to biocolonialism and eugenic practices. Finally, we suggest that advanced practice nurse leaders can make contributions within the area of genetics and genomics in five key areas. Through education, communication, relational ethics, cultural safety, and policy work, nurses can engage

in interprofessional practice to support individuals, families, and communities around their use of genetic and genomic technologies. As the field of genomics continues to evolve, so too do fundamental societal questions, thereby stretching us to consider not only personal values, but also our collective societal values. Advanced practice nurse leaders have a key role to play in these conversations, and in contributing to shaping the future of genetic and genomic technologies within our health care systems.

### **QUESTIONS FOR REFLECTION**

---

1. *Would you want to be informed by a family member if you were at risk for a genetic condition? Would you want to take a test for a genetic condition for which there is treatment? Would you want to take a genetic test for a genetic condition for which there is no treatment? What values inform your thoughts about notification and testing?*
2. *What skills do you have as a nurse that you could apply in counselling individuals and families about issues related to human genetics?*
3. *How does your identity influence your perspectives on genetic and genomic technologies? What perspectives or biases might you hold that could affect your ability to provide culturally safer care?*

## Endnotes

---

- 1 The exome makes up only 1.5% of the whole human genome; however, all protein coding genes are found in the exome. As most genetic disorders are correlated with changes in the protein coding genes, exome sequencing is often used when trying to determine if there is a discoverable genetic cause of a condition. Exome sequencing and analysis is cheaper and quicker than whole genome sequencing.

## References

---

- Allyse, M. A., Robinson, D. H., Ferber, M. J., & Sharp, R. R. (2018). Direct-to-consumer testing 2.0: Emerging models of direct-to-consumer genetic testing. *Mayo Clinic Proceedings*, 93(1), 113–120. <https://doi.org/10.1016/j.mayocp.2017.11.001>
- Begay, R. L., Garrison, N. A., Sage, F., Bauer, M., Knoki-Wilson, U., Begay, D. H., Becenti-Pigman, B., & Claw, K. G. (2019). Weaving the Strands of Life (Iná Bit'ool): History of genetic research involving Navajo People. *Human Biology*, 91(3), 189–208. <https://doi.org/10.13110/humanbiology.91.3.04>
- Bill C-16, An act to amend the Canadian Human Rights Act and the Criminal Code*, 42nd Parliament, 1st session. (2017). <https://www.parl.ca/DocumentViewer/en/42-1/bill/c-16/royal-assent>
- Blakemore, E. (2018, August 31). *Why the Navajo Nation banned genetic research*. History. A&E Television Networks. <https://www.history.com/news/why-the-navajo-nation-banned-genetic-research>
- Boardman, F. K., Young, P. J., & Griffiths, F. E. (2018). Impairment experiences, identity and attitudes towards genetic screening: The views of people with spinal muscular atrophy. *Journal of Genetic Counseling*, 27(1), 69–84. <https://doi.org/10.1007/s10897-017-0122-7>
- Bottorff, J. L., McCullum, M., Balneaves, L. G., Esplen, M. J., Carroll, J., Kelly, M., & Kieffer, S. (2005). Establishing roles in genetic nursing: Interviews with Canadian nurses. *The Canadian Journal of Nursing Research*, 37(4), 96–115.
- Boycott, K., Hartley, T., Adam, S., Bernier, F., Chong, K., Fernandez, B. A., Friedman, J. M., Geraghty, M. T., Hume, S., Knoppers, B. M., Laberge, A. M., Majewski, J., Mendoza-Londono, R., Meyn, M. S., Michaud, J. L., Nelson, T. N., Richer, J., Sadikovic, B., Skidmore, D. L., ... Stockley, T. Canadian College of Medical Geneticists (2015). The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position statement of the Canadian College of Medical Geneticists. *Journal of Medical Genetics*, 52(7), 431–437. <https://doi.org/10.1136/jmedgenet-2015-103144>
- Burke, W., Antommaria, A. H. M., Bennett, R., Botkin, J., Clayton, E. W., Henderson, G. E., Holm, I. A., Jarvik, G. P., Khoury, M. J., Knoppers, B. M., Press, N. A., Ross, L. F., Rothstein, M. A., Saal, H., Uhlmann, W. R., Wilfond, B., Wolf, S. M., & Zimmern, R. (2013). Recommendations for returning genomic incidental findings? We need to talk! *Genetics in Medicine*, 15(11), 854–859. <https://doi.org/10.1038/gim.2013.113>

- Canadian Association of Genetic Counsellors. (n.d.). *What is a genetic counsellor?*  
<https://www.cagc-accg.ca/index.php?page=353&id>
- Canadian Institutes of Health Research, Natural Sciences and Engineering Research Council of Canada, & Social Sciences and Humanities Research Council. (2018). *Tri-council policy statement: Ethical conduct for research involving humans – TCPS 2*. [https://ethics.gc.ca/eng/policy-politique\\_tcps2-epts2\\_2018.html](https://ethics.gc.ca/eng/policy-politique_tcps2-epts2_2018.html)
- Caulfield, T., & McGuire, A. L. (2012). Direct-to-consumer genetic testing: perceptions, problems, and policy responses. *Annual Review of Medicine*, 63, 23–33.  
<https://doi.org/10.1146/annurev-med-062110-123753>
- Childress, K. D. (2003). Genetics, disability, and ethics: Could applied technologies lead to a new eugenics? *The Journal of Women and Religion*, 20, 157–178.
- Christenhusz, G. M., Devriendt, K., & Dierickx, K. (2013). To tell or not to tell? A systematic review of ethical reflections on incidental findings arising in genetics contexts. *European Journal of Human Genetics*, 21(3), 248–255.  
<https://doi.org/10.1038/ejhg.2012.130>
- Claw, K.G., Anderson, M.Z., Begay, R.L., Tsosie, K. S., Fox, K., Summer internship for Indigenous peoples in Genomics (SING) Consortium, & Garrison, N A. (2018). A framework for enhancing ethical genomic research with Indigenous communities. *Nature Communications*, 9, Article 2957.  
<https://doi.org/10.1038/s41467-018-05188-3>
- Committee on Bioethics. (2001). Ethical issues with genetic testing in pediatrics. *Pediatrics*, 107(6), 1451–1455. <https://doi.org/10.1542/peds.107.6.1451>
- Conti, A. (2017). Drawing the line: Disability, genetic intervention and bioethics. *Laws*, 6(3), Article 9. <https://doi.org/10.3390/laws6030009>
- Cox, S., & Starzomski, R. (2004). Genes and geneticization? The social construction of autosomal dominant polycystic kidney disease. *New Genetics and Society*, 23(2), 137–166. <https://doi.org/10.1080/1463677042000237071>
- Dondorp, W., & de Wert, G. (2019). Refining the ethics of preimplantation genetic diagnosis: A plea for contextualized proportionality. *Bioethics*, 33(2), 294–301.  
<https://doi.org/10.1111/bioe.12534>
- Elliott, A. M., du Souich, C., Lehman, A., Guella, I., Evans, D. M., Candido, T., Tooman, L., Armstrong, L., Clarke, L., Gibson, W., Gill, H., Lavoie, P. M., Lewis, S., McKinnon, M. L., Nikkel, S. M., Patel, M., Solimano, A., Synnes, A., Ting, J., van Allen, M., ... Osiovich, H. (2019). RAPIDOMICS: Rapid genome-wide sequencing in a neonatal intensive care unit—successes and challenges. *European Journal of Pediatrics*, 178(8), 1207–1218.  
<https://doi.org/10.1007/s00431-019-03399-4>
- Ells, C., & Thombs, B. D. (2014). The ethics of how to manage incidental findings. *Canadian Medical Association Journal*, 186(9), 655–656.  
<https://doi.org/10.1503/cmaj.140136>
- Evans, M., Bergum, V., Bamforth, S., & MacPhail, S. (2004). Relational ethics and genetic counseling. *Nursing Ethics*, 11(5), 459–471.  
<https://doi.org/10.1191/0969733004ne7240a>

- Favaro, A., St. Philip, E., & Ho, S. (2021, June 19). *Racially profiled sickle cell disease patients say they face poor care*. CTV. <https://www.ctvnews.ca/health/racially-profiled-sickle-cell-disease-patients-say-they-face-poor-care-1.5477996>
- First Nations Health Authority. (n.d.). *Creating a climate for change*. <https://www.fnha.ca/Documents/FNHA-Creating-a-Climate-For-Change-Cultural-Humility-Resource-Booklet.pdf>
- First Nations Studies Program. (2009). *Sixties Scoop*. [https://indigenousfoundations.arts.ubc.ca/sixties\\_scoop/](https://indigenousfoundations.arts.ubc.ca/sixties_scoop/)
- Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233 (2008), <https://www.govinfo.gov/app/details/PLAW-110publ233>
- Glover, J. (2006). *Choosing children: Genes, disability, and design*. Clarendon Press. <https://doi.org/10.1093/acprof:oso/9780199290925.001.0001>
- Goering, S. (2008). 'You say you're happy, but ...': Contested quality of life judgments in bioethics and disability studies. *Journal of Bioethical Inquiry*, 5(2), 125–135. <https://doi.org/10.1007/s11673-007-9076-z>
- Greely, H. T. (2019). CRISPR'd babies: Human germline genome editing in the 'He Jiankui affair.' *Journal of Law and the Biosciences*, 6(1), 111–183. <https://doi.org/10.1093/jlb/lso10>
- Green, R. C., Berg, J. S., Grody, W. W., Kalia, S. S., Korf, B. R., Martin, C. L., McGuire, A. L., Nussbaum, R. L., O'Daniel, J. M., Ormond, K. E., Rehm, H. L., Watson, M. S., Williams, M. S., Biesecker, L. G., & American College of Medical Genetics and Genomics (2013). ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genetics in Medicine*, 15(7), 565–574. <https://doi.org/10.1038/gim.2013.73>
- Greenberg, A. (2020, September 11). *An Indigenous bioethicist on CRISPR and decolonizing DNA*. Nova. <https://www.pbs.org/wgbh/nova/article/bioethics-crispr-indigenous-genome/>
- Greenwood M. (2019). Modelling change and cultural safety: A case study in northern British Columbia health system transformation. *Healthcare Management Forum*, 32(1), 11–14. <https://doi.org/10.1177/0840470418807948>
- Guzauskas, G. F., Garbett, S., Zhou, Z., Spencer, S. J., Smith, H. S., Hao, J., Hassen, D., Snyder, S. R., Graves, J. A., Peterson, J. F., Williams, M. S., & Veenstra, D. L. (2020). Cost-effectiveness of population-wide genomic screening for hereditary breast and ovarian cancer in the United States. *JAMA Network Open*, 3(10), Article e2022874. <https://doi.org/10.1001/jamanetworkopen.2020.22874>
- Hauskeller, C. (2020). Care ethics and care contexts: Contributions from feminist philosophy. *East Asian Science, Technology and Society*, 14(1), 153–161. <https://doi.org/10.1215/18752160-8234663>
- Hawkins, A. K., & Ho, A. (2012). Genetic counseling and the ethical issues around direct to consumer genetic testing. *Journal of Genetic Counseling*, 21(3), 367–373. <https://doi.org/10.1007/s10897-012-9488-8>
- Hildt, E. (2016). Human germline interventions—think first. *Frontiers in Genetics*, 7, 81.

- Jamal, L. (2015). Ethical and policy issues in clinical genetics and genomics. In C. E. Kasper, T. A. Schneidereith, & F. R. Lashley (Eds.), *Lashley's essentials of clinical genetics in nursing practice* (2nd ed.). Springer.
- Jamal, L., Schupmann, W., & Berkman, B. E. (2020). An ethical framework for genetic counseling in the genomic era. *Journal of Genetic Counseling*, 29(5), 718–727. <https://doi.org/10.1002/jgc4.1207>
- Janvier, A. (2011). Pepperoni pizza and sex. *Current Problems in Pediatric and Adolescent Health Care*, 41(4), 106–108. <https://doi.org/10.1016/j.cppeds.2010.11.002>
- Janvier, A., & Watkins, A. (2013). Medical interventions for children with trisomy 13 and trisomy 18: What is the value of a short disabled life? *Acta Paediatrica*, 102(12), 1112–1117. <https://doi.org/10.1111/apa.12424>
- Johnston, J., Lantos, J. D., Goldenberg, A., Chen, F., Parens, E., Koenig, B. A., & the members of NSIGHT Ethics and Policy Advisory Board. (2018). Sequencing newborns: A call for nuanced use of genomic technologies. *Hastings Center Report*, 48, S2–S6. <https://doi.org/10.1002/hast.874>
- Lander, E. S., Baylis, F., Zhang, F., Charpentier, E., Berg, P., Bourgain, C., Friedrich, B., Joung, J. K., Li, J., Liu, D., Naldini, L., Nie, J. B., Qiu, R., Schoene-Seifert, B., Shao, F., Terry, S., Wei, W., & Winnacker, E. L. (2019). Adopt a moratorium on heritable genome editing. *Nature*, 567(7747), 165–168. <https://doi.org/10.1038/d41586-019-00726-5>
- Lea, D. H., Skirton, H., Read, C. Y., & Williams, J. K. (2011). Implications for educating the next generation of nurses on genetics and genomics in the 21st century. *Journal of Nursing Scholarship*, 43(1), 3–12. <https://doi.org/10.1111/j.1547-5069.2010.01373.x>
- Limoges, J., & Carlsson, L. (2020, October 26). *Nurses posed to play key role in advancing precision care through genetics and genomics*. Canadian Nurse. <https://canadian-nurse.com/en/articles/issues/2020/october-2020/nurses-poised-to-play-key-role-in-advancing-precision-care-through-genetics-and-genomics>
- Lipinsky, Y. (2021). *Clash of Titans: An ethical framework towards limitations in religious accommodation within healthcare* [Doctoral dissertation, Duquesne University]. Duquesne Scholarship Collection. <https://dsc.duq.edu/etd/1983>
- Lynce, F., & Isaacs, C. (2016). How far do we go with genetic evaluation? Gene, panel, and tumor testing. *American Society of Clinical Oncology Educational Book*, 36, e72–e78. [https://doi.org/10.1200/EDBK\\_160391](https://doi.org/10.1200/EDBK_160391)
- MacDonald, S. (2019, Winter). Finding lost roots: Emotional challenges and obstacles of tracing ancestry using DNA testing. *Insights Magazine*, 18–23. [https://bcacc.ca/wp-content/uploads/2022/10/Insights\\_2019\\_Winter.pdf](https://bcacc.ca/wp-content/uploads/2022/10/Insights_2019_Winter.pdf)
- Martens, K. (2018, October 10). *'I really felt my mother's presence': '60s Scoop adoptee comes home*. APTN News. <https://www.aptnnews.ca/national-news/i-really-felt-my-mothers-presence-60s-scoop-adoptee-come-home/>

- McGuire, A. L., Joffe, S., Koenig, B. A., Biesecker, B. B., McCullough, L. B., Blumenthal-Barby, J. S., Caulfield, T., Terry, S. F., & Green, R. C. (2013). Point-counterpoint. Ethics and genomic incidental findings. *Science*, *340*(6136), 1047–1048. <https://doi.org/10.1126/science.1240156>
- Medendorp, N. M., van den Heuvel, L. M., Han, P. K. J., Hillen, M. A., & Smets, E. M. A. (2021). Communication skills training for healthcare professionals in providing genetic counseling: A scoping literature review. *Patient Education and Counseling*, *104*(1), 20–32. <https://doi.org/10.1016/j.pec.2020.07.018>
- Middleton, A., Mendes, Á., Benjamin, C. M., & Howard, H. C. (2017). Direct-to-consumer genetic testing: Where and how does genetic counseling fit? *Personalized Medicine*, *14*(3), 249–257. <https://doi.org/10.2217/pme-2017-0001>
- Miller, D. T., Lee, K., Chung, W. K., Gordon, A. S., Herman, G. E., Klein, T. E., Stewart, D. R., Amendola, L. M., Adelman, K., Bale, S. J., Gollob, M. H., Harrison, S. M., Hershberger, R. E., McKelvey, K., Richards, C. S., Vlangos, C. N., Watson, M. S., Martin, C. L., & the members of ACMG Secondary Findings Working Group (2021). ACMG SF v3.0 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*, *23*(8), 1381–1390. <https://doi.org/10.1038/s41436-021-01172-3>
- Montgomery, S. (2017, October 11). Genetics in the clinical setting. *American Nurse*. <https://www.myamericannurse.com/genetics-clinical-setting/>
- Morgan, J., Coe, R. R., Lesueur, R., Kenny, R., Price, R., Makela, N., & Birch, P. H. (2018). Indigenous Peoples and genomics: Starting a conversation. *Journal of Genetic Counseling*, *28*, 407–418. <https://doi.org/10.1002/jgc4.1073>
- Morgan, J., Jameson, K., Joolae, S., & Ho, A. (2020). Calls to action: Truth, reconciliation, and Indigenous rights for supportive decision-making in healthcare. *International Journal of Indigenous Health*, *16*(1), 243–260. <https://jps.library.utoronto.ca/index.php/ijih/article/view/33201>
- Musunuru, K., Hershberger, R. E., Day, S. M., Klinedinst, N. J., Landstrom, A. P., Parikh, V. N., Prakash, S., Semsarian, C., Sturm, A. C., & American Heart Association Council on Genomic and Precision Medicine; Council on Arteriosclerosis, Thrombosis and Vascular Biology; Council on Cardiovascular and Stroke Nursing; and Council on Clinical Cardiology. (2020). Genetic testing for inherited cardiovascular diseases: A scientific statement from the American Heart Association. *Circulation: Genomic and Precision Medicine*, *13*(4), Article e000067. <https://doi.org/10.1161/HCG.0000000000000067>
- National Human Genome Research Institute. (n.d.). *Genetic counseling*. <https://www.genome.gov/genetics-glossary/Genetic-Counseling>
- National Human Genome Research Institute. (2021, November 1). *The cost of sequencing a human genome*. <https://www.genome.gov/about-genomics/factsheets/Sequencing-Human-Genome-cost>
- Newell, C. (2000). Biomedicine, genetics and disability: Reflections on nursing and a philosophy of holism. *Nursing Ethics*, *7*(3), 227–236. <https://doi.org/10.1177/096973300000700305>

- Noddings, N. (2012). The language of care ethics. *Knowledge Quest*, 40(5), 52–56.
- Office of the Privacy Commissioner of Canada. (2017, December). *Direct-to-consumer genetic testing and privacy*. [https://www.priv.gc.ca/en/privacy-topics/health-genetic-and-other-body-information/02\\_05\\_d\\_69\\_gen/](https://www.priv.gc.ca/en/privacy-topics/health-genetic-and-other-body-information/02_05_d_69_gen/)
- Ontario Health (Quality). (2020). Genome-wide sequencing for unexplained developmental disabilities or multiple congenital abnormalities: A health technology assessment. *Ontario Health Technology Assessment Series*, 20, 1–178.
- Ormond, K. E., Mortlock, D. P., Scholes, D. T., Bombard, Y., Brody, L. C., Faucett, W. A., Garrison, N. A., Hercher, L., Isasi, R., Middleton, A., Musunuru, K., Shriner, D., Virani, A., & Young, C. E. (2017). Human germline genome editing. *American Journal of Human Genetics*, 101(2), 167–176. <https://doi.org/10.1016/j.ajhg.2017.06.012>
- Parens, E., & Asch, A. (2003). Disability rights critique of prenatal genetic testing: Reflections and recommendations. *Mental Retardation and Developmental Disabilities Research Reviews*, 9(1), 40–47. <https://doi.org/10.1002/mrdd.10056>
- Penchaszadeh, V. B. (2015). Ethical, legal and social issues in restoring genetic identity after forced disappearance and suppression of identity in Argentina. *Journal of Community Genetics*, 6(3), 207–213. <https://doi.org/10.1007/s12687-015-0219-3>
- Rajkovic, A., Cirino, A. L., Berro, T., Koeller, D. R., & Zayhowski, K. (2021). Transgender and gender-diverse (TGD) individuals' perspectives on research seeking genetic variants associated with TGD identities: A qualitative study. *Journal of Community Genetics*, 13, 31–48. <https://doi.org/10.1007/s12687-021-00554-z>
- Resta, R., Biesecker, B. B., Bennett, R. L., Blum, S., Hahn, S. E., Strecker, M. N., & Williams, J. L. (2006). A new definition of genetic counseling: National Society of Genetic Counselors' task force report. *Journal of Genetic Counseling*, 15(2), 77–83. <https://doi.org/10.1007/s10897-005-9014-3>
- Rexach, J., Lee, H., Martinez-Agosto, J. A., Németh, A. H., & Fogel, B. L. (2019). Clinical application of next-generation sequencing to the practice of neurology. *The Lancet Neurology*, 18(5), 492–503. [https://doi.org/10.1016/S1474-4422\(19\)30033-X](https://doi.org/10.1016/S1474-4422(19)30033-X)
- Ryan, J., Virani, A., & Austin, J. C. (2015). Ethical issues associated with genetic counseling in the context of adolescent psychiatry. *Applied & Translational Genomics*, 5, 23–29. <https://doi.org/10.1016/j.atg.2015.06.001>
- Savulescu, J. (2001). Procreative beneficence: Why we should select the best children. *Bioethics*, 15(5-6), 413–426. <https://doi.org/10.1111/1467-8519.00251>
- Scherr, C. L., Ramesh, S., Marshall-Fricker, C., & Perera, M. A. (2019). A review of African Americans' beliefs and attitudes about genomic studies: Opportunities for message design. *Frontiers in Genetics*, 10, Article 548. <https://doi.org/10.3389/fgene.2019.00548>

- Shen, J. J., Carmichael, J., & Vásquez Santos, L. (2018). Identifying and addressing genetic counseling challenges among Indigenous people of Oaxaca—One center's experience with two immigrant farmworker families in the Central Valley of California. *Journal of Genetic Counseling*, 27(4), 996–1004. <https://doi.org/10.1007/s10897-018-0221-0>
- Sickle Cell Awareness Group of Ontario. (2020, June 12). *Black health matters*. <https://sicklecellanemia.ca/black-health-matters>
- Sloand, E., Bourguet, A. N., Engle-Pratt, W., & Bodurtha, J. (2018). Striving for precision: Enhancing genetic competency in primary care nurse practitioner students. *Journal of Nursing Education*, 57(11), 690–693. <https://doi.org/10.3928/01484834-20181022-12>
- Snyder, C. L., Lynch, J. F., & Lynch, H. T. (2009). Genetic counseling and the advanced practice oncology nursing role in a hereditary cancer prevention clinic: Hereditary breast cancer focus (part I). *The Breast Journal*, 15, S2–S10. <https://doi.org/10.1111/j.1524-4741.2009.00802.x>
- TallBear, K. (2013). *Native American DNA: Tribal belonging and the false promise of genetic science*. University of Minnesota Press.
- Taniguchi, N. K., Tualii, M., & Maddock, J. (2012). A comparative analysis of Indigenous research guidelines to inform genomic research in Indigenous communities. *The International Indigenous Policy Journal*, 3(1), 1–20. <https://doi.org/10.18584/iipj.2012.3.1.6>
- United Nations. (1989). *Convention on the rights of the child*. <http://www.ohchr.org/en/professionalinterest/pages/crc.aspx>
- Ward, C., Branch, C., & Fridkin, A. (2016). What is Indigenous cultural safety—And why should I care about it? *Visions; BC's Mental Health and Substance Use Journal*, 11(4), 29–32.
- White, S., Jacobs, C., & Phillips, J. (2020). Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care. *Genetics in Medicine*, 22(7), 1149–1155. <https://doi.org/10.1038/s41436-020-0785-6>
- Will, G. F. (2018, March 14). The real Down syndrome problem: Accepting genocide. *The Washington Post*. [https://www.washingtonpost.com/opinions/whats-the-real-down-syndrome-problem-the-genocide/2018/03/14/3c4f8ab8-26ee-11e8-b79d-f3d931db7f68\\_story.html](https://www.washingtonpost.com/opinions/whats-the-real-down-syndrome-problem-the-genocide/2018/03/14/3c4f8ab8-26ee-11e8-b79d-f3d931db7f68_story.html)
- Williamson, L., & LeBlanc, D. B. (2008). A genetic services practice model: Advanced practice nurse and genetic counselor team. *Newborn and Infant Nursing Reviews*, 8(1), 30–35. <https://doi.org/10.1053/j.nainr.2007.12.011>
- Wiwchar, D. (2013, July 22). Genetic researcher uses Nuu-chah-nulth blood for unapproved studies in genetic anthropology. *Ha-Shilth-Sa*. <https://hashilthsa.com/news/2013-07-22/genetic-researcher-uses-nuu-chah-nulth-blood-unapproved-studies-genetic>

- Yoes, M.-V., & Thomas, L. (2020). Hereditary cancer genetic risk assessment, testing, and counseling: A nurse practitioner–led program in a community setting. *The Journal for Nurse Practitioners*, 16(9), 660–665. <https://doi.org/10.1016/j.nurpra.2020.07.006>
- Zeiler, K. (2009). Symposium on genetics, identity and ethics. *New Genetics and Society*, 28(2), 153–156. <https://doi.org/10.1080/14636770902901561>