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ORIGINAL PAPER

The Ethical, Legal, and Social Implications of Genomics and Disability: Findings from a Scoping Review and Their Human Rights Implications

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Abstract

Objectives Genomic advancements affect people with disabilities. This paper presents the findings of a scoping literature review on the ethical, legal, and social implications (ELSI) of genomic technologies for people with disability. The human rights implications of the ELSI findings are then discussed briefly with reference to the United Nations Convention on the Rights of Persons with Disabilities (UNCRPD).

Methods A systematic search of the ELSI literature was conducted. Via a process of abstract screening and full-text review, 288 sources of evidence were included in the review. Data extraction involved identifying the ELSI discussed in each source, which were thematically analysed to generate ELSI themes and to identify relevant linkages to the UNCRPD.

Results Ten ELSI themes were identified as having relevant UNCRPD linkages including reproductive autonomy, issues related to cost and access, the downside of knowing about one's genetic makeup, lagging legislation in light of the rapid advancement of genomic technologies, genetic discrimination, the stigmatisation and devaluation of people with disabilities, the potential resurgence of eugenics and the medical model of disability, and the involvement of people with disabilities in conversations about genomic technologies. These themes have relevant and direct linkages to several UNCRPD rights including equality, non-discrimination, diversity, accessibility, full participation, identity, and freedom of expression.

Conclusions The review findings highlight that there is scope for the development of a charter on human rights specific to genomic technologies in the context of disability, which could guide ethical and socially appropriate developments in the field of genomic technologies in future.

Keywords Genomics · Disability · Human Rights · Ethical · Legal · Social · Implications · UNCRPD

With the emergence of biological discoveries in genetics, there is now a wealth of knowledge available regarding genetic variation in humans, and the diversity of human experience linked to this variation. Genetic variation refers to differences in genetic structure between individual people, and even populations of people (National Human Genome Research Institute, 2019). Most genetic variations have no effect on an individual person. However, some variations can lead to the development of a condition, disorder, or disease. Such variations can occur (1) in a single gene, e.g., cystic fibrosis is caused by variation in the CFTR gene (Farrell et al., 2017), (2) across multiple genes (polygenic), e.g., multiple sclerosis (Shams et al, 2023), or (3) at the chromosome level, e.g., 95% of people born with Down syndrome have three copies of chromosome 21 rather than two (Antonarakis et al., 2020).

People who have conditions, disorders, or diseases due to genetic variation (i.e., genetic conditions) may experience disability (United Nations, 2006). The International Classification of Functioning, Disability and Health Framework (ICF) (World Health Organisation; WHO, 2001) views disability as the interaction between health (or genetic) conditions (e.g., diseases, disorders, injuries) and the context in which a person lives. Contextual factors can be either personal (e.g., gender, age, education) or external (e.g., geographic location, community attitudes, social structures).

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This interaction between health (or genetic) conditions and the person's context can result in impairment of one's bodily structures and functions, limitations in one's ability to undertake activities, and restrictions around one's participation in social and economic life. The ICF therefore acknowledges that both biological (including genetic) factors and external factors influence the extent of a person's disability.

Scientific and medical developments around genetics and genomics are advancing rapidly, with many technologies focusing on the identification of genetic variations (through testing) that play a role in the diagnosis of genetic conditions that lead to disability. Genetic testing refers to the analysis of a single gene for genetic variations, while genomic testing involves analysing multiple genes simultaneously (Sandesh et al., 2020). Genomic testing techniques include panel testing (where a panel of genes is examined) and whole genome sequencing (where the genetic code of an entire genome is sequenced/analysed). The third type is cytogenetic testing which explicitly assesses for chromosomal anomalies (Sandesh et al., 2020), for example, chromosomal microarray to check for the deletion or insertion of genetic material.

Genetic, genomic, and cytogenetic testing technologies are used across the lifespan (Bilkey et al., 2019) to identify genetic conditions (referred to as genomic testing from here on). There are three testing technologies available prior to birth. Reproductive genetic testing can be used by people to understand the likelihood of passing a genetic condition onto their future children based on whether they are carriers of a genetic variant or not. Preimplantation genetic diagnosis involves testing cells extracted from embryos created through in vitro fertilisation for genetic conditions and is typically offered to prospective parents with a known family history of genetic conditions. Prenatal screening involves screening a foetus in utero for genetic conditions; it involves screening techniques such as non-invasive prenatal testing, which test cell-free foetal genetic material present in maternal blood to determine the probability of the presence of genetic conditions. Screening is then followed up with specific prenatal testing (e.g., amniocentesis—the testing of amniotic fluid for the presence of genetic conditions) to facilitate diagnosis if a high likelihood of the presence of a genetic condition is identified.

Three genomic testing technologies available post-birth are newborn screening, diagnostic testing, and predictive testing. Newborn screening involves testing the blood of newborn babies up to 3 days old and can be undertaken as a population-wide program. Newborn screening focuses on detecting the likelihood that the newborn baby may have childhood onset genetic conditions for which evidence-based treatments are available and if applied early in the child's life, will result in significant health benefits for the child (e.g., phenylketonuria). Like prenatal screening, diagnostic testing is carried out to facilitate diagnosis when newborn screening indicates a high likelihood of the presence of a genetic condition, but it should be noted that diagnostic testing can be used at any stage of life. Diagnostic testing tests the blood or saliva of a person to identify the genetic cause of a person's symptoms or to confirm a clinical diagnosis. Lastly, predictive testing is used to screen for genetic conditions in asymptomatic people with a known family history of a genetic condition to determine the likelihood that the person will develop the condition in future.

Advancements in genomic technologies have led to the development and research of novel therapeutic approaches such as gene therapy and gene editing. These methods are gaining attention in the scientific literature and hold significant promise for the treatment of various conditions through correcting the functioning of genes. Gene editing "alters the genome at a specific location to correct or alter the genetic sequence" (Delhove et al., 2020, p.20). This can be done by adding or removing genes or replacing abnormally functioning genes in the genetic sequence with correct copies. Gene editing is not currently viable or legal for human use in reproductive cells (where genetic changes could be passed from generation to generation); however, gene editing is being cautiously used with animals to investigate potential treatments for human genetic conditions (Li et al., 2020). Gene therapies are being developed using gene replacement techniques in the somatic (non-reproductive) cells of humans. These therapies typically insert correct copies of abnormally functioning genes in the cells of the target organ/tissue (Delhove et al., 2020). The abnormally functioning genes are not removed—the correct copies act to properly express whatever the abnormally functioning genes do not. Gene therapies are not widely available; however, there are some promising therapies in development for genetic conditions such as spinal muscular atrophy (Rao et al., 2018).

Innovations in genomic technologies such as those described above, raise issues beyond the science. In 1990, the USA created the ongoing ethical, legal, and social implications (ELSI) Research Program (National Human Genome Research Institute, 2023), which funds research activities that specifically address the ELSI of genomic research and technologies for individuals, families, and the broader community. McEwan et al. (2014) stated that research funded through this program has contributed to the development of genomic research guidelines to ensure ethical research practice (e.g., simplified informed consent forms to improve the accessibility of the language to the wider population) and supported the creation of legislation to protect people from discrimination as a result of genomic testing. Parker et al. (2019) observed that other scientific disciplines (e.g., neuroscience) have adopted the ELSI terminology and promote the undertaking of research that investigates the implications associated with emerging findings and technologies from their discipline. The ELSI term is broad, with no specific

definitions provided in the literature regarding what constitutes an ethical, legal, or social implication.

Even so, the ELSI of genomic technologies for people with disability has received considerable attention in the literature, with studies documenting the perspectives of people with disabilities and relevant stakeholders (parents, families, health professionals, etc.). Some of the ELSI discussed by people with disabilities and their families include reproductive autonomy (one's choice and control over reproduction, including choice and control around if, and when one chooses to have children), devaluation (people with disabilities feeling a sense of devaluation due to the availability of genomic technologies that can prevent people with disabilities from being born), and the potential impact on human diversity if the use of genomic technologies results in fewer people with genetic conditions (and disability) being born (see for example, Barter et al., 2017; Boardman & Hale, 2018; Olesen et al., 2017). It should be noted that conceptual or theoretical papers commenting on the ELSI of genomic technologies for people with disability are also plentiful in the literature (e.g., Bunnik et al., 2020; Scully, 2008). These are referred to as commentaries from here on.

Some of the documented ELSI of genomic technologies for people with disability have roots in the concept of human rights. Donnelly (2013) stated that human rights are "literally the rights that one has simply because one is a human being" (p. 10) and notes that these rights are afforded to all regardless of one's personal circumstance. The Universal Declaration of Human Rights was established by the United Nations in 1948 in response to the atrocities that occurred during World War II (United Nations, 1948), and paved the way for countries around the world to adopt human rights charters and treaties into legislation to ensure and uphold the rights of their people. The United Nations has also developed human rights charters specific to certain populations in the community, one being people with disabilities. In 2006, the United Nations opened the Convention on the Rights of Persons with Disabilities (UNCRPD; United Nations, 2006) for signature. The UNCRPD adopts eight guiding principles, which are specified in Article 3: (1) respect for the dignity, independence, and autonomy, (2) non-discrimination, (3) full inclusion and participation in society, (4) respect for difference and acceptance of disability as part of human diversity, (5) equality of opportunity, (6) accessibility, (7) equality between men and women, and (8) respect for the capabilities of children with disabilities and respect to preserve their identities. These eight principles are elaborated upon across 50 articles in the UNCRPD, in relation to aspects of society including health, education, and employment to name a few.

There is limited literature that specifically discusses the UNCRPD and the ELSI of genomic technologies for people with disability. MacKellar (2021) stated that the increasing acceptance of the use of human germline editing for the purpose of bringing children into the world with or without specific genetic traits is incompatible with the underlying concept of equality, value, and worth of all human beings expressed in Article 1 of the UNCRPD. Also, authors such as de Paor (2016) and Tiller and Delatycki (2021) discussed the significance of genetic discrimination—"the differential treatment of asymptomatic individuals or their relatives on the basis of their actual or presumed genetic characteristics" (Otlowski et al., 2012, p. 434)—with reference to Article 25 of the UNCRPD. Article 25 focuses on a person's right to quality health care without discrimination because of disability (or future disability in the case of genetic discrimination), including access to health and life insurance.

A small body of research literature is emerging that specifically discusses the human rights implications of the ELSI of genomic technologies for people with disabilities, but this literature is limited in depth and breadth. To contribute further to this body of evidence, the aim of this paper is twofold. First, this paper will report the findings of a scoping review that identified and described the common ELSI discussed in relation to genomic technologies for people with disabilities. Second, the human rights implications of the commonly identified ELSI will be briefly discussed with reference to the UNCRPD. It should be noted that this discussion of human rights implications is not meant to be exhaustive—the intention of this discussion was to highlight which human rights may have direct relevance to the identified ELSI, with recommendations for research and policy initiatives highlighted to facilitate more in-depth exploration in future. A discussion of the human rights implications of the ELSI of genomic technologies for people with disabilities would be incomplete without a deep understanding of the ELSI themselves, hence why a scoping review was conducted as the first point of call.

Method

The objective of the review was to scope the current evidence base regarding the ELSI of genomic technologies for people with disabilities to (1) summarise the extent, range, and nature of the available sources of evidence and (2) identify the range of ELSI reported in the sources. The review protocol was registered via Open Science Framework (Registration Number: osf.io/8ehqg) and was informed by the JBI Manual of Evidence Synthesis (Peters et al., 2020) and the PRISMA checklist for scoping reviews (PRISMA-ScR; Tricco et al., 2018).

Eligibility Criteria

The review focused on genetic conditions that result in people experiencing disability. Definitions of disability vary in the literature and for the purposes of this review, the ICF definition of disability (World Health Organisation, 2001) was used (see Introduction). The populations of interest to the review were (a) people with disability and their parents, carers, families, and/or guardians; (b) disability advocates; (c) medical and health professionals; (d) genomic researchers and educators, and (e) genetic counsellors, with no limits placed on the populations of interest regarding age, gender, or cultural background.

Given that the relevant body of literature contains a mix of commentaries and works that report the findings of research studies, both were included in the review. No limits were stipulated regarding the methodology of published research studies. Only sources of evidence written in English (or an English translation) were included in the review. Also, no limits were placed regarding source context, for example, if the source was specifically related to a particular country, geographic location (e.g., metropolitan), or setting (e.g., community).

Sources of evidence—including journal articles (accepted and online advanced publication), book chapters, conference papers, and theses/dissertations—published between 2000 and 2020 were included in the review. This timeframe coincides with the completion of the Human Genome Project and the implementation of genomic technologies such as testing (National Human Genome Research Institute, 2022). Genomic technologies have progressed dramatically since the completion of the Human Genome Project, meaning that much of the earlier literature discussing ELSI may be uninformative for the purposes of this review.

Information Sources and Search Strategy

Six electronic database platforms were consulted (EBSCO, Ovid, Proquest, PsycInfo, Scopus and Web of Science) and were chosen to cover topic areas of relevance to the review (e.g., disability, medical, social, legal). A list of search terms using Boolean operators was used across all platforms. The search terms reflected were (1) keywords related to disability (e.g., disability, impairment), (2) keywords related to stakeholders (e.g., person, parent, clinician, researcher), (3) keywords related to genomic technologies (e.g., genomic, genetic), and (4) keywords related to the review objectives (e.g., ethical, legal, social, implications, views). Searches were limited to the title, abstract/summary, and identified keywords or subject headings of the evidence source. Searches were undertaken by the first author with the assistance of an experienced librarian from the University of Queensland. More information regarding the database platforms searched and the search strategies used can be found in the supplementary file that accompanies this paper.

Selection of Sources of Evidence

Figure 1 depicts the PRISMA diagram that summarises the statistics related to source selection and full-text review. The total number of search results across the six platforms was 7176. Duplicate entries were then removed, leaving 4565 results. These were then imported into Covidence for screening. A checklist was developed that outlined the eligibility criteria as yes/no questions. To assess the checklist's inter-rater reliability, the first and third authors screened the titles and abstracts of 167 search results using the checklist. Percentage of agreement regarding inclusion was high (93%). Conflicting inclusion ratings were resolved by the second author. Given the high level of agreement, all remaining search results were screened for selection by the first author. A total of 537 sources of evidence were selected for full-text review. English full-text documents could not be obtained for 46 sources, leaving 491 documents available for full-text review.

Full-Text Review and Data Extraction

Full-text review and data extraction were undertaken by the first and fourth authors. Each full-text document was read by one reviewer with a decision made regarding inclusion using the previously discussed eligibility criteria to guide decision-making. If the reviewer was unsure, the other reviewer read the full-text document also, and a collaborative decision was made regarding inclusion. The first and fourth authors reviewed 268 and 223 full-text documents respectively. A total of 288 sources of evidence were included in the review. A common reason for exclusion was that the source presented content that was irrelevant to the review, e.g., issues that hinder the patient/genetic counsellor relationship, or sources that discussed a genomic technology with no discussion of ELSI. Another common reason for exclusion was that the source presented philosophical discussion about ethical issues related to genomic technologies, which did not align with the objective of the review.

Using an electronic spreadsheet, the first and fourth authors extracted the following data from the included sources: (a) citation information (author, year, country), (b) work presented (research study, commentary), (c) disability population of interest (e.g., Down syndrome), (d) genomic technology of interest (e.g., gene therapy), (e) research methodology for studies (participants group, methodological approach), and (f) the ELSI identified a short description of the ELSI discussed within the source.

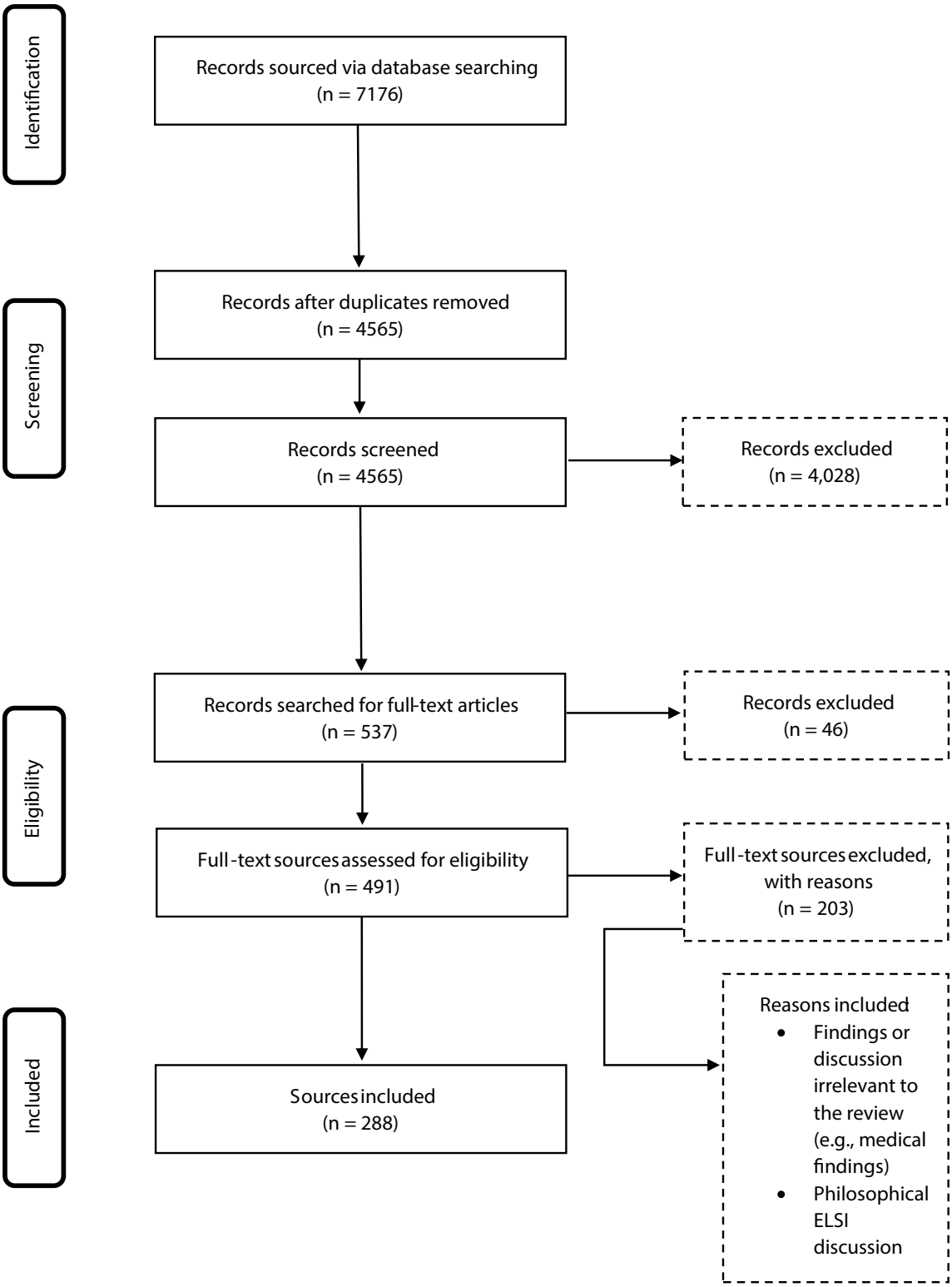

Fig. 1 PRISMA diagram

Data Synthesis and the Approach to Data Analysis

Data synthesis was completed by the first author. Frequency counts were tallied to synthesise the categorical descriptive data related to the included sources of evidence (e.g., disability population of interest). The qualitative data (the short ELSI descriptions) were synthesised using Braun and Clarke's (2022) thematic analysis method to identify ELSI themes. This method involves data familiarisation, generating codes from the data, generating initial themes, and reviewing them; and lastly, naming and defining the final set of themes. A total of 14 ELSI themes were identified via thematic analysis.

Next, the first three authors continued using the thematic analysis approach to identify direct, noticeable, and relevant interlinkages between the 14 identified ELSI themes and the eight guiding principles and 50 articles of the UNCRPD. Each author spent several hours familiarising themselves with the content of the UNCRPD and the ELSI themes separately and individually identified direct, noticeable, and relevant interlinkages between the ELSI themes and UNCRPD. They then came together to discuss their individual coding, and this discussion was concluded when consensus was gained between the three authors regarding which UNCRPD guiding principles and articles aligned with the identified ELSI themes.

When it came to understanding the interlinkages between the UNCRPD and the ELSI themes uncovered, the authors' ascribed to Braun and Clarke's (2022) view on reflexive thematic analysis—that interlinkages are not waiting to be discovered in the data or that other researchers would expose the same interlinkages. Rather, the authors acknowledge that each member of the authorship team brought their own perspectives as social science researchers to the data analysis, and the interlinkages generated are the product of the authorship team's collective reading and construction of knowledge.

Results

Only ten of the 14 ELSI themes had a direct, relevant, and noticeable linkage to one or more of the guiding principles and articles of the UNCRPD. The Results section to follow will only focus on these ten themes due to space limitations. Briefly, the four themes found to have no direct linkage to the UNCRPD were (1) the definition of a "serious" condition (the idea that only genetic conditions deemed to be "serious" should be the focus of genomic technologies), (2) "wrongful life" lawsuits (legalities around seeking financial compensation from medical and health professionals when a child is born with a genetic condition when prenatal screening and testing indicated no or a low chance of the child being born with a genetic condition), (3) informed consent (that the currently used informed consent processes for genomic testing procedures may be inadequate or incomplete), and (4) termination of pregnancy (the specific legalities around termination of pregnancy following prenatal screening and testing).

Descriptive Analysis of the Included Sources of Evidence

Country of origin for each source of evidence was defined by the country affiliation of the lead author, with 82% of the authors affiliated with Western countries led by the USA, UK, Canada, and Australia. Collectively, European authors made up the next largest group (11%). Regarding source type, journal articles feature most (90%), then theses/dissertations (6%), and book chapters (4%). Regarding genomic technologies, testing approaches were discussed most (92%), with prenatal screening/testing the focus of 39% of sources followed by multiple testing approaches (25%). Gene editing and gene therapy were discussed in 5% of the sources, with 3% discussing genomic research. In 70% of the sources, disability was discussed generally without reference to a specific disability population, and when a specific population was discussed, intellectual disability (7%), Down syndrome (6%), and deafness (5%) featured most. Only 26% of the sources presented study findings, with the other 74% providing commentary. Most of the studies used qualitative methodologies (63%), followed by quantitative (21%), and mixed methodologies (16%). Lastly, 32% of the studies sampled parents and 19% sampled multiple stakeholders (e.g., people with disabilities and parents). Only 11% of the studies sampled only people with disabilities.

Thematic Analysis

The ten themes identified as having a direct link to guiding principles and articles of the UNCRPD are briefly described below, with reference to relevant literature sourced from the scoping review. Table 1 provides extracts from the UNCRPD to demonstrate the linkage between the ELSI theme and the UNCRPD.

Reproductive Autonomy

Article 10 of the UNCRPD recognises every human being's inherent right to life, which has relevance when understanding reproductive autonomy. Reproductive autonomy was discussed in the literature taking into consideration the increased availability of genomic testing technologies, and therefore more information being available to people about their own genetic makeup and the makeup of their prospective or actual children. This information was reported

Table 1 Identified ELSI and their alignment with the articles of the UNCRPD (2006)

ELSI	Identified connections to the UNCRPD
Reproductive autonomy Information from genomic technologies is seen as beneficial when it comes to exercising one's right to make reproductive choices	• Article 10: Right to life—every human being has the inherent right to life, and measures are needed to ensure its effective enjoyment by persons with disabilities on an equal basis with others
Stigmatisation The availability of genomic technologies that prevent the birth of people with genetic conditions perpetuates negative attitudes towards disability	• Article 8: Awareness raising—(a) to raise awareness throughout society, including at the family level, regarding persons with disabilities, and to foster respect for the rights and dignity of persons with disabilities; (b) to combat stereotypes, prejudices, and harmful practices relating to persons with disabilities in all areas of life; (c) to promote awareness of the capabilities and contributions of persons with disabilities
Devaluation The availability of genomic technologies that prevent people being born with disability can make existing people with disability feel devalued	• Preamble: (m) Recognising the valued existing and potential contributions made by persons with disabilities to the overall wellbeing and diversity of their communities and that the promotion of the full enjoyment by persons with disabilities of their human rights and fundamental freedoms and of full participation by persons with disabilities will result in their enhanced sense of belonging and in significant advances in the human, social, and economic development of society
Eugenics The increasing use of genomic technologies may constitute eugenic practice as it results in less people with genetic conditions being born	• Article 8: Awareness raising—(b) to combat stereotypes, prejudices, and harmful practices relating to persons with disabilities
Medical model of disability The medical model of disability will again become the dominant view of disability as a result of increased use of genomic technologies—that disability is a medical problem that can be cured or prevented with medical intervention	• Article 8: Awareness raising—(a) to raise awareness throughout society, including at the family level, regarding persons with disabilities, and to foster respect for the rights and dignity of persons with disabilities; (b) to combat stereotypes, prejudices, and harmful practices relating to persons with disabilities in all areas of life; (c) to promote awareness of the capabilities and contributions of persons with disabilities
The downside of knowing Knowing relevant genetic information can have a negative impact on people	• General Principle: (h) Respect for the right of children with disabilities to preserve their identities
Cost and access The high costs associated with genomic technologies and associated issues regarding access have implications not only for individuals, but society also	• Article 9: Accessibility—to enable persons with disabilities to live independently and participate fully in all aspects of life. States Parties will take appropriate measures to ensure all people with disabilities access, on an equal basis with others
Lagging legislation and policy Genomic technologies are advancing at such a rate that legislation and policy is not keeping up	• Preamble: (o) Persons with disabilities should have the opportunity to be actively involved in decision-making processes about policies and programmes, including those directly concerning them • Preamble: (v) Recognising the importance of accessibility…to information and communication, in enabling persons with disabilities to fully enjoy all human rights and fundamental freedoms • Article 4: General Obligations: (1c) To take into account the protection and promotion of the human rights of persons with disabilities in all policies and programmes • General Principle: (c) Full and effective participation and inclusion in society • Article 4: General Obligations – (3) In the development and implementation of legislation and policies to implement the present convention, and in other decision-making processes concerning issues relating to persons with disabilities, States Parties shall closely consult with and actively involve persons with disabilities, including children with disabilities, through their representative organisations • Article 21: Freedom of expression and opinion, and access to information

Table 1 (continued)

ELSI	Identified connections to the UNCRPD
Genetic discrimination Unaffected people may experience discrimination (e.g., the denial of health or life insurance) based on genomic testing results that indicate the person will or may develop a genetic condition in future	• Preamble: (h) discrimination against any person on the basis of disability is a violation of inherent dignity and worth of the human person • Article 5: Equality and non-discrimination—State Parties shall prohibit all discrimination on the basis of disability and guarantee to persons with disabilities equal and effective legal protection against discrimination on all grounds • Article 25: Health—(e) prohibit discrimination against persons with disabilities in the provision of health insurance and life insurance
Including people with disability in conversations People with disabilities have had little opportunity to express their views in relation to genomic technologies, when research indicates that they would like to	• Preamble: (o) persons with disabilities should have the opportunity to be actively involved in decision-making processes about policies and programmes, including those directly concerning them • Article 21: Freedom of expression and opinion, and access to information—(a) providing information intended for the general public to persons with disabilities in accessible formats and technologies appropriate to different kinds of disabilities in a timely manner and without additional cost

Direct quotations provided from the UNCRPD

as being a key benefit of genomic testing as it supports informed reproductive decision-making. Research found that the availability of genetic information from genomic testing was viewed favourably by some people with genetic conditions that lead to disability (e.g., Boardman & Hale, 2018; Nahar et al., 2013) as it allowed them to make an informed decision around whether to have children who may be born with the same genetic condition they have. Also, Metcalfe (2018) and Siegel and Milunski (2004) discussed that information from genomic testing has implications not only for the person tested, but also their family members, including that people may choose to not have children if a genetic condition is known to run in families. But some authors (e.g., Benston, 2016; Kemper et al., 2019) commented that clear legal boundaries regarding reproductive autonomy are needed in light of the availability of genomic testing prenatally and pre-conception as theoretically, genomic technologies could be used to ensure that a child is born with a desired genetic condition.

Stigmatisation

Article 8 of the UNCRPD (awareness-raising) advocates for measures to be adopted in society that positively promote people with disability (e.g., respect for their rights and their dignity and the contributions they make to society). Regarding genomic testing technologies, several authors commented that the use of genomic testing to prevent the birth of people with disabilities reinforces negative stereotypes and societal bias against people with disabilities and perceptions of disability being a burden (e.g., Asch, 2003; Doxzen & Halpren, 2020). This can influence reproductive decision-making, as studies have found that some prospective or expectant parents held the view that preventing a child being born with a genetic condition prevents that child from experiencing disability stigma that is already present in society (e.g., Boardman & Hale, 2018; Bryant et al., 2011). Alternatively, some authors (Malek, 2010; v. Hammerstein et al., 2019) stated that reproductive decisions following genomic testing do not necessarily reflect negative societal attitudes; rather, they reflect reproductive autonomy and an individual's choice to not raise children with disabilities. Stigma around decision-making following genomic testing was also identified in the literature, with studies finding that some mothers experienced negative attitudes from family members and health professionals for choosing to continue their pregnancy when prenatal screening/testing indicated the presence of a genetic condition (e.g., Guon et al., 2014; Hickerton et al., 2012).

Devaluation

Section (m) of the UNCRPD Preamble recognises the valued existence of people with disabilities to society, including enhanced community diversity that comes with people with disability being included in society on an equal basis with others. The devaluation theme encompassed the view that the availability of genomic technologies that can prevent people with disability being born in the first place can make existing people with disabilities feel devalued by society. This is discussed as the expressivist argument by several authors (Hofmann, 2017; Peterson, 2012), and this perspective has been supported through research that specifically includes people with disabilities (e.g., Barter et al., 2017; Boardman, 2014). However, some authors (e.g., Shakespeare, 2005a, b) commented that this consideration

should not limit reproductive autonomy, especially if society continues to focus on positively supporting and including people with disabilities in society.

Eugenics

Article 8 of the UNCRPD discusses the need to raise awareness of disability in society to combat harmful practices relating to people with disability. Some authors argued that the use of genomic technologies that can eliminate or cure inherited disability risks a return of eugenic beliefs, where disability is seen as an undesirable heritable trait (e.g., Çaha, 2014; Lord, 2014), and in the past resulted in harmful practices towards people with disabilities (e.g., programs of sterilisation to prevent disability in future generations). Studies have found that maternal decisions to pursue prenatal testing are impacted by their personal belief that they could not cope with raising children with disabilities (Carroll et al., 2000; Kibel & Vanstone, 2017). Based on such findings, other authors such as Bruni et al. (2012) and Lemke and Rüppel (2019) argued that reproductive decisions based on genomic testing are usually framed within a personal and family context, and that the coercive eugenic practices of the past are not present in current programs related to genomic testing and other genomic technologies.

Medical Model of Disability

Some authors expressed concern that the widespread use and promotion of genomic technologies could return the medical model as the dominant driver of disability and social policy (e.g., Miller & Levine, 2013). As summarised by Haegele and Hodge (2016), the medical model views disability as a medical issue (an impairment of bodily structures and/or functions), and emphasises the treatment and/or cure of these disabilities through medical intervention. In contrast, Haegele and Hodge (2016) stated that the more contemporary social model of disability emphasises that disability is actually a social construct—that society imposes disability onto people with impairments as a result of how society is organised (e.g., inaccessible environments that people with physical impairments may not be able to access). Research by Nagle et al. (2008) showed that a medicalised view of disability held by health professionals influenced the type of advice provided to patients following genomic testing, for example, a greater emphasis on treatment options (as opposed to intervention options) available when testing indicated the presence of a genetic condition. Several authors stated that information provided to people about genetic conditions, disability, and quality of life following genomic testing needs to be neutral, non-directive, and reflective of more than just the medical facts related to the condition (e.g., de Montgolfier, 2018; Perez Gomez, 2020). This highlights the role that health professionals can play when it comes to raising awareness of disability in the community (Article 8 of the UNCRPD), again to promote the rights and dignity of people with disability, and their contributions to society.

The Downside of Knowing

A stated benefit of genomic technologies is the ability to diagnose genetic conditions which allows people to access genetic and other information (e.g., possible interventions) to assist them across their lifespan. Though, research showed that this information could also have negative effects. These include hindering parental joy around having the baby and their initial bonding following newborn screening (Boardman et al., 2020; Riley & Wheeler, 2017) and the emotional impact of being informed that their child's condition is permanent or that no treatments or interventions are available for the child's diagnosed condition (Tremblay et al., 2019). General Principle (h) of the UNCRPD has relevance here, which affirms the right of children with disabilities to preserve their identities. The findings above describe instances when the child's disability became the core part of the child's identity for the parents due to genomic testing, which led to emotional impacts.

Cost and Access

Access to all aspects of life, on an equal basis with others is discussed in Article 9 (accessibility) of the UNCRPD. However, commentary in the literature about the cost of, and access to genomic technologies has focused on (1) inequity of access based on the financial resources of individuals and governments (Çaha, 2014; Kumar, 2008), (2) the possibility that funding to improve the accessibility of genomics technologies could actually compete with the allocation of funding to resource supports for people with disabilities (Bunnik et al., 2020; Doble et al., 2020), and (3) that low-cost direct-to-consumer genomic testing options could provide genetic information to people without appropriate professional support to allow the consumer to fully understand that information (Bailey et al., 2014).

Lagging Legislation and Policy

Several authors commented that legislation and policy is not keeping up with rapid advances in genomic technologies (e.g., Lord, 2014; Mannion, 2006), especially the current advancements in gene editing (e.g., Doxzen & Halpern, 2020; Segers & Mertes, 2020). As a result, some authors remarked that there is the need for continued debate and reviews of legislation and policies to take into consideration these advances, with some advocating that such reviews should consider and incorporate guidance from human

rights charters like the UNCRPD (e.g., Cunningham, 2019; Doxzen & Halpern, 2020), and should include the views of people with disabilities (Ward et al., 2002). The participation of people with disabilities in supporting the development of up-to-date legislation/policies regarding genomic technologies is a right stipulated in several sections of the UNCRPD including the right to accessible information [Preamble (v)]. This would allow people with disabilities to take part in relevant legislative debates and reviews with more ease. It would also enable them to exercise their right to full participation and inclusion in society, especially around the development and implementation of legislation and policy [Preamble (o), General Principles (c), and (i) General Obligation (c)], and the right to freedom of expression and opinion (Article 21).

Genetic Discrimination

The UNCRPD states in the Preamble (h) and Article 5 that people with disability have the right to equality and to live a life free of discrimination. Genetic discrimination—which refers to unaffected or asymptomatic people experiencing discrimination because of their actual or presumed genetic makeup—was discussed in the literature, mainly in the context of people being denied health insurance or life insurance based on the result of genomic testing that indicated the person will or may develop a genetic condition (and potentially disability) in future (e.g., Huntington's disease). Countries such as the USA, Germany, and Sweden have specific legislation for genetic discrimination that is separate from disability discrimination legislation, while other countries (e.g., Australia, Ireland, Netherlands) have amended existing disability discrimination legislation to include genetic discrimination (de Paor, 2016; Karpin, 2016). Via a systematic review, Wauters and Van Hoyweghan (2016) found little evidence of the actual occurrence of genetic discrimination and recommends further research be done that aims to distinguish genetic discrimination from disability-specific discrimination.

Including People with Disability in the Conversation

The UNCRPD advocates for people with disabilities to be actively involved in decision-making processes about policies and programs of direct relevance to them [Preamble (o)], and the right to express their opinions (Article 21). Authors such as Scully (2008) and McKee et al. (2013) commented that people with disabilities have had little opportunity to express their views on genomic technologies, with one study (Ward et al., 2002) finding that people with disabilities want to take part in discussions and want to inform policy regarding reproductive autonomy and the use of prenatal screening. Without meaningful collaboration between genomic researchers and people with disabilities, Miller and Levine (2013) stated that people with disabilities could lose confidence in genomic technologies. There is also the possibility that people with intellectual and cognitive disabilities may be assumed as lacking in capacity to participate in such discussions/debates; however, research has demonstrated that their contribution is viable when accommodations are made such as relevant information being presented to them in accessible formats (e.g., Alderson, 2001; Barter et al., 2017).

Discussion

Using scoping literature review methodology and thematic analysis, the first aim of this paper was to identify and understand the documented ELSI of genomic technologies for people with disabilities. Using thematic analysis again, the second aim was to understand the human rights implications of the ELSI uncovered by identifying a set of direct, noticeable, and relevant interlinkages between the ELSI and the guiding principles and articles of the UNCRPD. There are eight guiding principles (in Article 3) and 50 articles in the UNCRPD and as indicated in Table 1; not all were relevant to this discussion of human rights and the ELSI of genomic technologies for people with disability. In reverse, though, it is important to note that most of the ELSI themes identified via the scoping review relate to several of the guiding principles and articles of the UNCRPD. Ten ELSI themes with relevant linkages to the UNCRPD were agreed upon by the first three authors, with the identified ELSI themes deemed to be related to rights such as equality, non-discrimination, diversity, accessibility, full participation, identity, and freedom of expression. What follows is a brief (and by no means exhaustive) discussion of the interlinkages between the uncovered ELSI themes and the UNCRPD, with a focus on recommendations for future research and policy to guide further discussion and analysis of the ELSI of genomic technologies for people with disabilities and their human rights (as stipulated by the UNCRPD).

Implications for Human Rights in the Context of the UNCRPD

Article 10 of the UNCRPD reaffirms that all human beings have an inherent right to life, and that people with disabilities have the right to its effective enjoyment on an equal basis with others. A link between the ELSI of reproductive autonomy and Article 10 was flagged in the Results section; however, unpacking and understanding the link between Article 10, reproductive autonomy and genomic technologies requires careful thought for several reasons. Selective termination of pregnancy is commonly discussed

in conjunction with prenatal screening, disability, and reproductive autonomy; however the UNCRPD is silent on the issue of termination of pregnancy. Petersen (2015) documented that the draft version of the UNCRPD included language around disability not being used as a justification for the termination of life; however, this language was removed from the final version of the UNCRPD in preference of the simple statement that currently forms Article 10. Considering genomic technologies like preimplantation genetic diagnosis when embryos created through IVF are screened for genetic conditions, understanding reproductive autonomy through the lens of the UNCRPD is even more difficult as Petersen (2015) noted that the UNCRPD makes no reference to persons unborn, or when life begins (e.g., at birth, at conception). In a similar vein, Wohbring and Diep (2016) stated that it is unclear if the UNCRPD could be used to question the use of gene editing technologies. Given this, the reproductive autonomy ELSI theme may not be as relevant when it comes to understanding the ELSI of genomic technologies for people with disabilities from a human rights perspective, as the UNCRPD is more focused on ensuring that existing people with disabilities live a life of quality equal to others, as opposed to those not born yet.

A common thread through many of the ELSI themes is ignorance or outdated views about disability, for example, that the promotion and application of genomic technologies that have the potential to eliminate inherited disability promote negative attitudes towards existing people with disabilities and devalues their contribution to society, and may result in the resurgence of outdated eugenic thinking and viewing disability as a medical issue that requires diagnosis, treatment, or remediation (i.e., the medical model of disability). Findings from some of the research studies reviewed highlighted the impact that outdated views on disability had on people's decisions following genomic testing, e.g., reproductive decisions to prevent a person being born with a genetic condition to avoid experiencing disability stigma (Boardman & Hale, 2018; Bryant et al., 2011), and an extreme interpretation of the medical model could lead to a view that a cure or treatment is needed to fix disability, and if these are not available, a person will inevitably have a poor quality of life (Faragher, 2019). The perseverance and reemergence of outdated thinking about disability could give rise to concerns about the goal of removing certain types of disability from the population using genomic technologies (Reinders et al., 2019).

To safeguard against outdated views impacting the way genomic technologies are used in society, the UNCRPD has a significant role to play. The UNCRPD is an important internationally agreed upon framework that affirms the rights of people with disabilities across all aspects of life, and if legislation, policies, and practices related to the development and application of genomic technologies take into consideration the human rights of people with disabilities, this is a positive step forward in protecting people with disabilities from the potentially negative consequences of genomic technologies in the context of disability. While human rights charters specific to genomic technologies are available, for example, the United Nations Universal Declaration on the Human Genome and Human Rights (UDHGHR, 1997) and the Council of Europe Convention on Human Rights and Biomedicine (1997), these charters do not specifically address disability. Of interest, the UNCRPD does not specifically address genomic technologies. Given this, bringing together the concepts outlined in both the UNCRPD and the human rights charters focused on genomic technologies to create a unified set of human rights related to genomics and people with disabilities may be an appropriate direction for future research and policy. In line with this, authors such as Lord (2013), de Paor and Blanck (2016), and Wolbring and Diep (2016) advocated for the use of the UNCRPD as a framework to inform the development of best practice guidelines and legislation to guide the ethical, legal, and socially appropriate use of genomic technologies in the context of people with disabilities.

Article 8 of the UNCRPD advocates that States Parties need to continually raise awareness of disability in society to combat stereotypes and prejudice and promote the capabilities and contributions of people with disabilities. Awareness raising will also play a crucial role in protecting people with disabilities from the potentially negative consequences of genomic technologies. This is plausible because studies are available that demonstrate that raising awareness of people with disabilities can positively change people's attitudes towards disability (Lu et al., 2018), and by enhancing positive attitudes in the community, this may have a flow-on effect around how people with disabilities are viewed in light of the presence of genomic technologies that have the potential to remove inherited disability from society. Of interest, Shakespeare (2005a, b)—who is a prominent disability advocate—commented that if there is continued disability awareness and the positive inclusion of people with disabilities in society to ensure people with disabilities can live a life of quality, how genomic technologies are used by people to exercise reproductive autonomy (i.e., choice around raising a child with disabilities) should not impacted.

A theme of interest was that genomic advancements are largely seen as outpacing legislation and policy. And this may always be the case as it is impossible to legislate for advances yet to be discovered (take gene editing for example). Authors such as Miller and Levine (2013) emphasised that including people with disability in genomics research and genomics policy development further safeguards the human rights of the very people who are likely to be most affected by the wide use of genomic technologies. The

authors take this view also. Involving people with disability as partners in research, at a minimum on advisory panels, offers the possibility of safeguarding against potential inadvertent or detrimental impacts of research developments prior to protections by legislation and policy. The lack of genomics research (including ELSI-focused research) including people with disabilities on research teams or in other forms of co-design and co-production was noted in the literature included in the scoping review. This is interesting given government health bodies such as the National Health and Medical Research Council (NHMRC, 2018) in Australia advocated for consumer involvement and have even made it mandatory when it comes to health guideline development.

The biopsychosocial model (Engel, 1977, 1981) considers biological, psychological, and social factors and their complex interactions when it comes to understanding a person's health, illness, and how to best support that person through the delivery of health care. This model has been discussed extensively in the literature, even in the context of disability. For example, the World Health Organisation (2002) highlights one of the key advantages of the biopsychosocial model of disability is that it acknowledges that although some aspects of disability are inherent to an individual, disability is a more complex phenomenon that also includes important environmental and social interactions. In the context of genomics, genetic information gained from genomic testing is not determinative in providing predictions or prognosis of the impact of a genetic condition on the person's functioning or their (or their family's) quality of life. There are many factors outside of a person's genetic makeup that impacts on functioning and quality of life, for example, socioeconomic status (McMahon et al., 2022) and accessible environments (Roos et al., 2022). Given this, the biopsychosocial model may also be useful when it comes to considering the human rights implications of genomic technologies for people with disabilities as the UNCRPD adopts a biopsychosocial approach to defining disability and understanding human rights in a disability context (Kazou, 2017).

Ignoring the biological or medical basis of disability risks missing opportunities for making improvements to one's quality of life utilising biological interventions such as gene therapy, but ignoring societal interactions that impact upon disability due to the availability of genomic technologies may also lead to the possibilities of social stigma, discrimination, and devaluation of those with disability from a genetic cause. Therefore, using a framework of disability that considers the complexity and richness of the disability phenomenon (i.e., the biopsychosocial model) could lead to guiding principles that can underpin a response to genomic technologies that affirms and upholds the rights of people with disabilities. Some of the themes identified via the scoping review demonstrate the value of the biopsychosocial model in the interplay between the genetic basis of some disabilities and the impact of the social context on the person. For example, the identified ELSI theme relating to cost of, and access to, genomic technologies expose considerations around maintaining public funding for societal supports for people with disabilities against the potential desire to reduce the economic cost of funding these supports by preventing inherited disability.

Limitations and Future Directions

Regarding the scoping review specifically, a limitation of the review was that the search methodology employed has likely missed some key sources of evidence. The body of evidence that discusses the ELSI of genomic technologies is large to begin with, and the field of genomics contains complicated terminology which is sometimes not applied consistently across the literature. For example, prenatal testing, prenatal screening, and prenatal diagnosis all refer to genomic testing carried out on expectant mothers to ascertain if their unborn child may have a genetic condition. While it would have been preferable to use the name of each technology in the literature search strategy, when tested, it resulted in an unmanageable number of search results (over 10,000 results) hence why general terms (genetic, genomic) were favoured. Also, a search strategy to identify grey literature was not included. This was considered but abandoned due to the large sample of search results available for selection and subsequently full-text review from the electronic databases.

The findings of this scoping review should be viewed in the context of ELSI literature published post-2000, and in the context of an era of genomic research and practice informed by the findings of the Human Genome Project (National Human Genome Research Institute, 2022). However, when engaging with the ELSI research literature published pre-2000, many of the same ELSI identified via this scoping review are also discussed and debated in this literature. This is an interesting direction for future research—to map and analyse the ELSI of genomic technologies for people with disabilities across time to see if increased genomic knowledge, the development of new genomic technologies such as gene editing, and the passage of time have changed the rhetoric around the identified ELSI, and what these changes means in the context of human rights also.

This work has been drawn from a vast body of literature around the ELSI of genomic technologies that is rapidly increasing. To this point, considerably fewer studies have been undertaken into the ELSI of genomic technologies in the context of disability and even fewer with a specific consideration of human rights. The ELSI themes uncovered and the identified interlinkages between these themes and the rights outlined in the UNCRPD are the authors' analytical view of the research in the area. This paper, therefore, aims to

open the field up for further work, debate, and discussion and is not meant to be an exhaustive discussion on the ELSI of genomic technologies in the context of disability and human rights. This work also imposed a view of human rights from the perspective of the UNCRPD. This was a deliberate choice to account for the relevance of this convention to almost all countries in the world (United Nations Treaty Collection, 2023) and its recognition of the diverse community of people with disabilities and that they make a valued existing and potential contribution to the overall community—a view that is contradictory to the disability prevention rhetoric that may be implied by the availability of genomic technologies.

It should be acknowledged that most of the literature reviewed contained studies conducted in high-income countries (e.g., the USA, Australia) or commentaries written by authors from these countries discussing the ELSI of genomic technologies for people with disabilities within the context of their country. Further work is needed to understand the ELSI of genomic technologies in low- and middle-income countries. It is not just further research that is needed; researchers in those countries need support to have their work published for study by the international community. This is critical as the proportion of people with disabilities is greater in low- and middle-income countries, with 80% of people with disabilities living in developing countries (United Nations Department of Economic and Social Affairs, n.d.), and access to genomic technologies in these countries may be limited and available only to some.

There is great promise in genomics advances to improve the quality of life of people with disabilities and society in general. Through a lens of human rights, potential harm can be mitigated. While the focus here has been on the impact on people with disabilities and those around them, there are wider implications for the advancement of genomics technologies. There is scope for a charter of human rights specific to people with disabilities in the context of genomic technologies and is a recommendation for future work. This charter could form an ethical framework to guide future developments in the field and should include the views of people with disabilities to facilitate the ethical and socially appropriate application of genomic technologies.

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