Disability and genomic technologies – A scoping review of issues and implications

The "We Need to Talk" Project

- A team of multi-disciplinary researchers from The University of Queensland is embarking on a collaborative research project – the 'We Need to Talk' project - focused on identifying, understanding, and addressing the issues and implications of genomic technologies for people with disability.
- The end goal of the project is to combine the views of people with disabilities and their families with the views of health professionals, policy makers and researchers to create a shared dialogue regarding genomic technologies and their implications for people with disability.
- Using co-design methodology, the project includes people with disability, other key stakeholders and the research team working collaboratively to identify practical solutions to key issues, and to create resources to support disability-informed and evidence-based clinical practice in genomics.
- To inform the project moving forward, a *scoping review* of available research literature discussing the issues and implications of genomic technologies for people with disability and their families was undertaken. Such issues and implications are broadly described as the ethical, legal, and social implications (ELSI) of genomic technologies in the literature.

Overview of the Scoping Review

- The review focused on: (1) describing the extent, range, and nature of the current body of evidence discussing the ELSI of genomic technologies for people with disability, and (2) briefly summarising the ELSI reported in the evidence.
- The eligibility (inclusion) criteria for peer-reviewed sources of evidence were:
    - The source discussed ELSI of genomic technologies for people with disability.
    - The source discussed ELSI of genomic technologies from the perspective of people with disability and/or other stakeholders such as family members, disability advocates, health professionals, and genomics educators, researchers, and counsellors.
    - The source was a published, online advance or accepted for publication journal article or conference paper, thesis or dissertation, or book chapter.
    - The source was written in English, or an English translation was available.
    - The source was dated from 2000 to 2020.
    - The source presented findings from a research study, opinion, or literary analysis.
- Relevant databases within the EBSCO, Ovid, ProQuest, PsycInfo, Scopus and Web of Science electronic database platforms were searched for sources of evidence.
- Terms such as 'disability', 'genomic', 'genetic', 'person', 'family', 'carer', 'ethical', 'legal', and 'social' were used to the search the databases and searches were limited to titles, keywords, and abstracts.

Search Results

- Literature searches were completed on November 9 and November 17, 2020. The total number of search results across the six platforms was 8304. After the removal of 2918 duplicate results, 5380 sources of evidence remained in an Endnote library.

- The titles and abstracts of the 5380 sources of evidence were screened against the eligibility criteria using the Covidence platform, with 671 sources chosen for full-text review. Full-text documents for 625 sources were obtained via The University of Queensland Library.
- The sample of full-text documents were read in full and assessed against the eligibility criteria. If the source of evidence was included in the review, relevant data were extracted from the document and catalogued in a spreadsheet.
- Categorical data extracted were synthesised using frequency counts.
- Qualitative data extracted were synthesised via simple coding; each identified ELSI was categorised by genomic technology (e.g., prenatal testing), then the identified ELSI in each category were coded into a refined group of ELSI by merging similar issues/implications where possible.
- A total of 288 sources of evidence were included in the review. Sources were excluded from the review if they presented: (1) evidence or discussion that was deemed outdated, (2) philosophical discussion regarding issues/implications, or (2) content that was deemed irrelevant.

Key Findings

Descriptive Analysis of the Body of Evidence

- 81% of the included sources of evidence were written by authors from Anglo countries (i.e., United States, United Kingdom, Canada, and Australia).
- 26% of the included sources presented findings from a research project. The other 74% presented either opinion and/or literary analysis. Regarding the research studies:
    - Interviews and surveys were the main methods of data collection used.
    - 32% of the studies sampled parents (including prospective parents) and 12% sampled people with disability or a lived experience of disability.
    - For the studies that sampled multiple stakeholders, all included people with a disability and/or parents alongside other stakeholders such as health professionals.
- Most of the included sources were peer-reviewed journal articles (90%).
- Disability was discussed in general terms in most of the included sources (70%). When a specific population was discussed, 7% discussed people with intellectual and developmental disabilities, 6% discussed people with Down syndrome and 5% discussed people with hearing impairments.
- Most of the included sources discussed *genomic testing* approaches (e.g., preimplantation genetic diagnosis prenatal testing, newborn screening, carrier screening, predictive testing and testing for diagnosis).

Ethical Legal and Social Issues of Genomics Identified within the Sources of Evidence

- *Reproductive autonomy:* Information from genomic testing is seen as beneficial and valuable when it comes to exercising one's right to make choices regarding their own reproduction. Such information allows prospective/expectant parents to make choices around whether they wish to parent a child with disability. However, some authors state that clear boundaries around reproductive autonomy are needed as theoretically, genomic technologies could be used to ensure that a child is born with a desired genetic condition.
- *The definition of a 'serious' condition:* The concept of a 'serious' condition is used in legislation and policy around the world to place boundaries around which genetic conditions are subject to the use of genomic technologies, and which genetic conditions selective termination of pregnancy applies to in the later stages of gestation. However, this term is not clearly defined in legislation and policy and

what constitutes a 'serious' condition is open to interpretation. Studies that sought the perspectives of parents identified that genetic conditions that lead to premature death and are associated with significant physical and/or intellectual impairments aligned closely with their perceived notion of what constitutes a 'serious' condition.

- *Stigmatisation:* Some authors argued that the availability of genomic technologies prolongs negative attitudes towards disability. Studies found that many prospective/expectant parents express the view that by preventing children being born with disability, they are preventing them from experiencing disability stigma already present in society. Stigmatisation of reproductive decisions was also discussed, with evidence showing mothers who chose to continue their pregnancy when prenatal testing indicated the presence of a genetic condition experienced negative attitudes from family and health professionals for their decision. Some authors expressed that this sort of stigma may become more prevalent if genomic technologies become free or more affordable to use, and widely available.
- *Devaluation of people with disabilities:* Many authors discussed the expressivist argument – that the availability of genomic technologies that prevent people being born with disability can make existing people with disability feel devalued. Studies that sampled the views of people with disability support the expressivist argument. However, several authors stated that if there is continued societal focus on supporting people with disabilities to live their best life, the expressivist argument should not limit reproductive autonomy.
- *Eugenics:* Some authors state that the increasing use of genomic technologies constitutes eugenic practice because the result is the prevention of people with genetic conditions being born. However, some authors hold the view that reproductive decisions based on information gleaned from genomic testing (such as selective termination of pregnancy following prenatal testing) are rooted in personal and/or family reasons that do not have a basis in eugenics, which is more of a political, cultural, or philosophical position.
- *Medical model of disability:* With increased use/promotion of genomic technologies, some authors stated that the medical model of disability will gain prominence as the dominant view of disability. The medical model conceptualises disability as a medical problem that can be cured or prevented with medical intervention (e.g., medication, surgery, termination of pregnancy). Some authors noted that a medicalised view of disability may influence how health professionals impart information about disability to people undergoing genomic testing. To support reproductive autonomy, these authors advocated that information provided to people about genetic conditions, disability, and quality of life needs to be neutral and non-directive.
- *The downside of knowing:* While the diagnosis of genetic conditions and access to relevant genetic information is a key benefit of genomic technologies, there can be a downside to knowing. Studies have found that diagnosis following newborn screening can hinder parental joy around having a newborn baby and their initial bonding. Also, some authors discuss the wider implications of genetic information; carrier screening, predictive testing and testing for diagnosis results have implications not only for the person undergoing testing, but for members of their immediate family, which can lead to family conflict if family members are ambivalent about knowing.
- *Cost and access:* High costs associated with genomic technologies may lead to unequal access. Some authors stated that disability could become a socioeconomic issue linked with disadvantage if access to genomic technologies is limited to those with financial means. Another issue relates to the availability of low-cost options such as direct-to-consumer testing for carrier screening; some authors caution that this form of testing has questionable reliability/validity, and people should seek support from a genetic counsellor to fully understand any positive results received. Some authors observe that genomic technologies benefits society by preventing disability, subsequently reducing support costs through public health and social service systems. However, these same authors acknowledge

that existing supports for people with disabilities should not diminish in any way due to the availability of genomic technologies.

- *Legislation and policy are lagging:* Some authors noted that genomic technologies are advancing at such a rate that legislation and policy is not keeping up. Also, the legality of gene editing was also discussed; while the technology is not currently legal (or viable) for human use, many authors state that with effective regulation in place to ensure safe and responsible implementation, gene editing should be made available for human use when viable.
- *'Wrongful life' lawsuits:* In some countries (including Australia), 'wrongful life' lawsuits are acceptable within the bounds of the law if health professionals provide an inaccurate interpretation of genomic testing results. Varying opinion was expressed in relation to these lawsuits in the literature. Some argued that 'wrongful life' lawsuits are a natural extension of medical malpractice lawsuits, and governments should not be enacting laws to prevent them. Others noted the potential ramifications of 'wrongful life' lawsuits for health professionals such as the overuse of genomic testing to avoid litigation. Lastly, some authors argued that 'wrongful life' lawsuits perpetuate the societal devaluing of people with disability and their continued stigmatisation.
- *Genetic discrimination:* Genetic discrimination constitutes people experiencing discrimination due to being diagnosed with a genetic condition such as the denial of employment and insurance. Evidence indicates that while fear of genetic discrimination is reported by many people with genetic conditions, there is little evidence of its actual occurrence. Also, evidence shows that many people with genetic conditions are unaware that many countries around the world have laws in place to protect people from genetic discrimination. Lastly, studies have found that people who have a personal experience of genetic discrimination decline genomic testing (out of fear of discrimination) even though testing would be beneficial to their current and future health.
- *Informed consent:* Some authors discuss informed consent when it comes to genomic technologies, with some questioning whether patient consent is compromised if prenatal screening/testing and newborn screening is presented as routine antenatal/postnatal care. Regarding expanded carrier screening programs, health professionals indicated that they weren't sure how to impart accurate information about all possible genetic conditions screened for without overloading the patient with information and potentially compromising their ability to provide consent.
- *Legalities regarding termination of pregnancy:* In some countries, timelines for prenatal testing do not align with timelines for legal termination of pregnancy, be it for personal or medical reasons. Some authors also note significant ambiguity in existing laws regarding termination of pregnancy, e.g., a lack of clarity around what constitutes a 'serious' condition. Given such issues, the benefit of prenatal testing was questioned by some authors if termination of pregnancy is not an available option.
- *Inclusion of people with disability in conversations about genomic technologies:* Many authors outlined that people with disabilities have had little opportunity to express their views in relation to genomic technologies. Studies that sampled people with disability show that many people want to be involved in discussions to inform policy and legislation regarding the development and use of genomic technologies.

Summary and Conclusion

- There is a considerable amount of literature available that discuss the ELSI of genomic technologies for people with disability, with prenatal testing and preimplantation genetic diagnosis receiving more focus compared to other technologies. A total of fourteen issues/implications were identified in this scoping review.

- Reproductive autonomy and stigmatisation appear to be central to the ELSI identified and discussed. Most ELSI are discussed in relation to: (a) how they either uphold or threaten reproductive autonomy or (b) continue to stigmatise or perhaps protect people from negativity because of disability.
- Given that lived experience of disability varies across and within disability diagnostic groups, the generalist way that disability is discussed in the body of evidence fails to account for experiences that may be unique to certain disability groups and individuals.
- The body of evidence is lacking in research studies that directly sample the views of people with intellectual or cognitive disabilities. This is a limitation given that the ELSI of genomic technologies are likely to impact this group of people more than others.
- There were some limitations surrounding the literature search methodology used, e.g., limiting the number of search terms used to allow a more manageable amount of search results could have potentially excluded relevant literature from the review.
- In conclusion, the findings from this scoping review lay a solid foundation to inform the ‘We Need to Talk’ project moving forward. The lack of inclusion of people with disabilities in past research further solidifies the premise of the project – that people with disability need a seat at the table when the ELSI of genomic technologies are being debated, and policy/legislation is being created to ensure the safe, ethical, and socially appropriate implementation of genomic technologies in practice and research.