

# The Genetic Discrimination Observatory: confronting novel issues in genetic discrimination

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Genetic discrimination (GD) is the differential or unfair profiling of an individual on the basis of genetic data. This article summarizes the actions of the Genetic Discrimination Observatory (GDO) in addressing GD and recent developments in GD since late 2020. It shows how GD can take many forms in today's rapidly evolving society.

**Keywords:** genetic discrimination; ethical, legal, and social implications (ELSI); intersex; moratorium; insurance contract; genetic databases

## Context

With rapid technological advancements in today's postgenomic era, GD is a persistent issue associated with the use of genetic and genomic data. Simply stated, GD involves the differential, negative treatment or unfair profiling of an individual relative to the rest of the population on the basis of actual or presumed genetic characteristics that is intended to infringe upon or has the effect of infringing human rights, fundamental freedoms, and human dignity<sup>i</sup>. In recent years, this concept has been expanded to include discrimination based on other high-throughput omics technologies such as polygenic risk score and epigenetic clocks<sup>ii</sup>. (See Box 1, Box 2.)

### Box 1

#### About the GDO

Conceived in early 2018 and launched internationally in 2020, the GDO is an international organization of researchers and stakeholders dedicated to documenting and addressing instances of genetic discrimination around the world<sup>xii</sup>. The GDO upholds the human rights principles expressed by the United Nations Educational, Scientific and Cultural Organization's (UNESCO's) International Bioethics Committee, stated in the Universal Declaration on the Human Genome and Human Rights adopted at UNESCO's 29th General Conference on November 11, 1997<sup>xiii</sup>. The observatory has collaborated with the Global Alliance for Genomics and Health, a policy-oriented and technical standards-setting organization aimed at responsible genomic data sharing within a human rights framework<sup>xiv</sup>.

Following its launch, additional experts from Kazakhstan, South Africa, Colombia, and Ukraine joined the GDO's International Expert Panel. This panel now includes 22 researchers in 20 jurisdictions across six world regions and supports the development of international comparative research projects on GD<sup>xv</sup>. The GDO has also developed tools such as real-time maps documenting various aspects of GD<sup>xvi</sup> and a 'Share Your Story' module to encourage GD victims to report discriminatory events securely and anonymously in North America and the UK<sup>xvii</sup>. This tool serves as the GD barometer, allowing the GDO to keep abreast of new GD trends that have yet to be reported in formal settings. The GDO is currently assessing the logistics and ethical requirements to open 'Share Your Story' to other regions.

The GDO's most recent initiative is a Delphi study to identify essential features of genetic nondiscrimination policies. The Delphi design is used to (i) identify issues where information is not readily available; (ii) explore a full range of alternatives and underlying assumptions, correlating informed judgments; and (iii) educate a respondent group [3.]. It is forward looking and aims to identify areas of consensus and disagreement among recruited experts regarding new technologies and emerging challenges. Our study will solicit views of interdisciplinary experts on GD and genomic research from around the world. The first round of the study has just begun, and we look forward to analyzing the results in late 2021.

The GDO is seeking experts willing to find innovative solutions to address GD, particularly from unrepresented jurisdictions around the world. Representatives of patient groups and vulnerable communities who would like to collaborate with the GDO are invited to reach out to us.

Box 2

**GD in the coronavirus disease 2019 (COVID-19) pandemic context**

Since 2020, the world's attention has been focused on the COVID-19 pandemic. In this context, the use of genetic tests could increase the potential for GD at both the individual and population levels [10.]. According to Milne [10.], if a relationship is demonstrated between genotype and susceptibility/severity of COVID-19, genetic susceptibility screening may contribute to shaping societal responses to COVID-19. In this possible scenario, individuals belonging to a genetically determined high-risk group could then have to contend with more restrictive and potentially discriminatory public health measures, such as continuing to undergo shielding or self-isolation measures for a longer amount of time than the rest of the population [11.]. However, it should be noted that most of the recent research on COVID-19 does not provide evidence of this genetic correlation to warrant such exceptional and controversial measures<sup>xi</sup>. There is also the risk that the absence of sufficient legal, administrative, and technical protection for the large amount of human genetic data collected under broad consent by researchers and public health authorities for surveillance and COVID-19–related research could be misused for discriminatory objectives by third parties [12.].

Today, GD is a global challenge that international organizations, countries, and institutions have attempted to address through public policy, including national laws [1.]. However, these policies are often limited by important shortcomings: (i) a lack of public visibility or awareness about GD and existing informational resources; (ii) restrictive, rigid policy formulation that may become out of step with rapidly paced scientific advancement; and/or (iii) complex administrative procedures that limit the access and timely implementation of protection [2.]. Furthermore, GD is an insidious phenomenon that often happens without the knowledge of the person being discriminated against, thus depriving them of the opportunity to seek redress [1.].

This article provides an overview of recent trends in GD since the GDO's inception in late 2020. These developments point to the pervasive and evolving forms of GD and the need for anticipatory and flexible public policies in response.

**Recent trends in GD**

Public policies are essential to prevention and sanction of occurrences of GD. However, successfully addressing GD through law is an ongoing challenge with which many jurisdictions struggle. In Europe, for example, in February 2021, the Danish Supreme Court delivered a controversial decision in favor of an insurance company, which may impact the protections against GD available in Denmark<sup>xix</sup>. The court ruled that an asymptomatic person referred for genetic screening on the basis of family history should have disclosed to the insurer that they had been referred (but not diagnosed) at the time of taking out the insurance. The decision is particularly disturbing because Section 3a of the Danish Insurance Contracts Act, 2015 (as amended), contains a nondiscrimination provision prohibiting insurance companies from asking for more information or demanding further examinations on inheritable conditions. The presence of a referral was deemed sufficient to render the condition 'current' and thereby legal for an insurer to request information. It is hoped that such a narrow interpretation of a genetic nondiscrimination law will not impact legal developments in neighboring European countries. Genetic nondiscrimination laws are often passed to encourage individuals with family histories of diseases to learn of their potential genetic risk without fear of reprisal. The Danish ruling could therefore have the unfortunate consequence of causing individuals to avoid learning about their genetic predisposition to serious diseases – endangering their very lives.

In July 2020, Canada settled a jurisdictional conflict over its recent federal legislation that aims to prevent GD. The Canadian Genetic Non-Discrimination Act (GNDA) prohibits individuals from being compelled to undergo genetic testing or share the results of their genetic tests when seeking goods and services or entering contracts, including in the context of insurance or employment<sup>iii</sup> [4.]. After a 3-year constitutional battle, the Supreme Court of Canada, by a small majority (five to four), ruled that the GNDA was constitutional under the legislative field of criminal law, which is under the jurisdiction of the federal government<sup>iv</sup>. Thus, Canadians nationwide can now benefit from increased protection from GD. Meanwhile, genetic nondiscrimination policy in the USA is more complex, because the country currently offers no uniform federal protection against GD by life insurance companies. Instead, genetic nondiscrimination policy on this matter is regulated at the level of the individual states. In July 2020, Florida became the first US state to prohibit life insurance companies from using genetic data obtained by predictive genetic tests in underwriting, unless the test was accompanied by a medical diagnosis [5.]. Similarly, states such as Arizona, California, and Vermont have recently enacted genetic nondiscrimination laws in underwriting. Other state laws, however, generally have a more modest reach than Florida's Genetic Information for Insurance Purpose (HB 1189) [5.].

On the other side of the world, the Australian government has not yet adopted legislation to prevent GD by insurers. The Australian insurance industry's peak body, the Financial Services Council, voluntarily introduced a moratorium restricting the use of genetic test results in life insurance underwriting for policies worth up to AU\$500 000 [6.]. In response, the Australian government's Genomic Health Future Mission funded a national project from 2020 to 2023 called 'the Australian Genetics and Life Insurance Moratorium: Monitoring the Effectiveness and Response (A-GLIMMER)' [6.]. This project has recruited researchers, clinicians, patient groups, and policy experts to determine if the moratorium is an adequate and effective long-term regulatory solution in the interests of preventing GD [6.]. These recent legal developments showcase the variety of legal approaches put forward to prevent GD and their inherent limitations.

The European Society of Human Genetics raised the alarm earlier this year about a different facet of GD that is less often highlighted: government use of genetic tests and data to profile specific populations in ways that exacerbate existing social inequities [7.]. For example, in recent years, the Chinese police forces, supported by the Chinese Communist Party, have begun establishing DNA databases of children and members of the general population in Xinjiang province for public safety and surveillance purposes, without their explicit consent [7.].<sup>v</sup> These actions echo similar troubling instances of compulsory genetic profiling around the world, such as the US government conducting DNA profiling on immigrants crossing the Mexican border, or in Kuwait, where a 2015 law (Law No. 78 of 2015, known as 'the DNA Law') was planned to impose the compulsory collection of DNA samples from all citizens, residents, and visitors to the country [7.]. Alongside growing international pressure, Kuwait's Constitutional Court invalidated this controversial law in 2017<sup>vi</sup>. Such state-sponsored brands of GD are often overlooked by traditional genetic nondiscrimination laws, which tend to focus on private actors such as insurers and employers rather than government departments and agencies.

Another type of GD, unnoticed for many years, involves the treatment of intersex individuals. The term ‘intersex’ describes individuals with congenital variations in sex characteristics and/or reproductive anatomy, reflecting underlying variation in genitalia, gonads, hormones, or genetic differences. It is estimated that intersex individuals represent approximately 1.7% of the general population [8.]. Intersex individuals are often victims of GD, in the contexts of both healthcare (e.g., victims of unnecessary sex assignment surgery) and sports [9.]. For example, in September 2020, the Swiss Federal Supreme Court rejected athlete Caster Semenya’s appeal against the Court of Arbitration for Sport to uphold the legality of the Difference of Sex Development Regulations of World Athletics (previously called the International Amateur Athletic Federation)<sup>xviii</sup>. Caster Semenya’s lengthy administrative, legal, and social ordeal to compete as a woman demonstrates the existence of GD against intersex individuals in sports and public media. In Ireland and Canada, studies to document this emerging form of GD are currently ongoing<sup>vii,viii</sup>.

Lastly, genetic nondiscrimination laws adopted over 2 decades ago are unlikely to provide sufficient protection against new types of GD. This is exemplified by the growing use of polygenic risk scores along with health data to inform risk prediction models<sup>ix</sup>. These models, already on the radar of insurers, calculate the future risk of an individual developing a specific disease using genetic, health, and lifestyle data<sup>x</sup>. Individual results from such a prediction model, expressed as a risk level, are unlikely to be covered by most genetic nondiscrimination laws currently in force. Another potential, more distant threat is that of new human genome editing applications resulting in preferred genomic ‘improvements and designs,’ as well as emerging genomic data and artificial intelligence applications<sup>x</sup>, that may lead to new and unforeseen forms of discrimination.

### **Concluding remarks**

GD is one of the main challenges that comes to mind in omics research and personalized healthcare. Although it already has a long history, GD has kept pace with developments in genetics. As new tests, drugs, and treatments have emerged, new ways to discriminate between individuals on the basis of omics data have been identified. Therefore, at a time when omics sciences play such an increasingly important role in our lives, there is a strong need for a dedicated international organization to monitor and advise on emerging developments in GD. The multijurisdictional and decentralized structure of the GDO enables members of the international expert panel to quickly identify new incidents of GD and collaboratively address them through research, public engagement, and the development of robust, forward-looking policy models for the international community. Thus, in the coming years, the GDO will continue to serve the international community by developing surveys, information briefs, web applications, policy models, recommendations, and a multitude of other tools to address the complex challenges raised by GD. Although the integration of omics sciences to mainstream medical practice is becoming a positive reality, omics knowledge should never be hijacked to discriminate and stigmatize individuals.

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### **Declaration of interests**

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### **Author contributions**

Y.J. developed and drafted the manuscript with the assistance of K.H. Y.J., K.H., M.A., Y.B., A.D.P., E.S.D., P.G.M., C.W.L.H., C.-H.H., I.V.H., H.K., A.L., T.M., K.Ó.C., A.E.R.P., A.P.S.N., M.O., M.S.P., R.S., L.S., T.H.V., M.H.Z., and G.D. reviewed and edited the manuscript before approving its submission.

### **Resources**

<sup>i</sup><https://gdo.global/en/gdo-faq>

<sup>ii</sup><https://theconversation.com/new-dna-test-that-reveals-a-childs-true-age-has-promise-but-ethical-pitfalls-126676>

<sup>iii</sup><https://www.canlii.org/en/ca/laws/stat/sc-2017-c-3/latest/sc-2017-c-3.html>

<sup>iv</sup><https://www.scc-csc.ca/case-dossier/cb/2020/38478-eng.aspx>

<sup>v</sup>[www.aspi.org.au/report/genomic-surveillance](http://www.aspi.org.au/report/genomic-surveillance)

<sup>vi</sup>[www.newscientist.com/article/2149830-kuwaits-plans-for-mandatory-dna-database-have-been-cancelled/](http://www.newscientist.com/article/2149830-kuwaits-plans-for-mandatory-dna-database-have-been-cancelled/)

<sup>vii</sup>[www.dcu.ie/intersex](http://www.dcu.ie/intersex)

<sup>viii</sup>[www.sciencedirect.com/science/article/abs/pii/S1701216320304904](http://www.sciencedirect.com/science/article/abs/pii/S1701216320304904)

<sup>ix</sup>[www.swissre.com/reinsurance/life-and-health/l-h-risk-trends/polygenic-risk-scores-better-cancer-predictor-for-insurers.html](http://www.swissre.com/reinsurance/life-and-health/l-h-risk-trends/polygenic-risk-scores-better-cancer-predictor-for-insurers.html)

<sup>x</sup><https://emerj.com/ai-sector-overviews/machine-learning-in-genomics-applications/>

<sup>xi</sup>[www.nature.com/articles/s41586-021-03767-x](http://www.nature.com/articles/s41586-021-03767-x)

<sup>xii</sup><https://gdo.global/en>

<sup>xiii</sup><https://en.unesco.org/themes/ethics-science-and-technology/human-genome-and-human-rights>

<sup>xiv</sup>[www.ga4gh.org](http://www.ga4gh.org)

<sup>xv</sup><https://gdo.global/en/gdo-team>

<sup>xvi</sup><https://gdo.global/en/world-views>

<sup>xvii</sup><https://gdo.global/en/share-your-story-consent>

<sup>xviii</sup>[www.washingtonpost.com/sports/2020/09/08/caster-semenya-gender-ruling/](http://www.washingtonpost.com/sports/2020/09/08/caster-semenya-gender-ruling/)

<sup>xix</sup><https://www.domstol.dk/media/i1bas5lw/14231-2020-anonym.pdf>

## References

1. Joly, Y. et al. (2020) Looking beyond GINA: policy approaches to address genetic discrimination. *Annu. Rev. Genom. Hum. Genet.* 21, 491–507
2. Joly, Y. et al. (2017) Comparative approaches to genetic discrimination: chasing shadows? *Trends Genet.* 33, 299–302
3. Okoli, C. and Pawlowski, S.D. (2004) The Delphi method as a research tool: an example, design considerations and applications. *Inf. Manag.* 42, 15–29
4. Joly, Y. et al. (2021) Erring in law and in fact. *Can. Bar Rev.* 99, 172–192
5. Rothstein, M.A. and Brothers, K.B. (2020) Banning genetic discrimination in life insurance — time to follow Florida’s lead. *N. Engl. J. Med.* 383, 2099–2101
6. Tiller, J. et al. (2021) Monitoring the genetic testing and life insurance moratorium in Australia: a national research project. *Med. J. Aust.* 214, 157–159
7. Forzano, F. et al. (2021) ESHG warns against misuses of genetic tests and biobanks for discrimination purposes. *Eur. J. Hum. Genet.* 29, 894–896
8. Keating, A. (2019) Intersex: beyond the sex binary. In *Spotlight on Current Events: Essays on Contemporary World Issues*, ABC-CLIO
9. Bauer, M. et al. (2020) Intersex human rights. *Int. J. Hum. Rights* 24, 724–749
10. Milne, R. (2020) Societal considerations in host genome testing for COVID-19. *Genet. Med.* 22, 1464–1466
11. McQuillen, M. and Terry, S.F. (2020) Genetic and disability discrimination during COVID-19. *Genet. Test. Mol. Biomarkers* 24, 759–760
12. Hollenstein, M. et al. (2020) Preservation of DNA privacy during the large scale detection of COVID-19. *arXiv* Published online August 1, 2020. <http://arxiv.org/abs/2007.09085>