Global Alliance for Genomics & Health

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Genetic Discrimination: Implications for Data Sharing Projects (GeDI) (Ver. 1.0)

1. Genetic Discrimination

Genetic Discrimination (GD) involves the differential adverse treatment, or unfair profiling, of an individual relative to the rest of the population based on actual or presumed genetic/genomic information and other 'omic' data. Like sexual, ethnic, or disability-based discrimination, genetic discrimination is a source of exclusion with a negative effect on that individual's life. Thus, genetic discrimination can limit the social and professional opportunities of a person and can lead to psychological, social, and economic disadvantage and distress.

2. Is discrimination closer to myth than reality? A look at the existing evidence

Generally speaking, there are too few studies about the incidence and impact of GD. See https://gdo.global/en/gdo-map-life-insurance for a complete listing of published studies on GD in the context of life insurance sorted by geographical location. As seen from this overview, there are large regions of the world where GD incidence is poorly documented. GD may also be occurring in other fields, but it is insufficiently documented to allow for any systematic review or meta-analysis. The studies that we do have, in life insurances and in other fields, also differ significantly in the quality of methodologies that they use. Common methodological problems include positive bias, non-representative samples, leading questions, etc. However, based on these sources, media reports and government statistics (available in the U.S. only), GD incidents - especially in the context of insurance and access to employment - have been documented in North America, Western Europe, Australia and East Asia. Such incidents usually involve the results of predictive tests for serious or life-threatening, monogenic, highly heritable conditions, such as Huntington's disease, sickle cell anemia, and hereditary breast and colon cancers. Some studies indicated that GD occurs more often based on family history than on genetic test results. The frequency and spread of these events are impossible to determine because of the aforementioned problems with existing studies. Concerns over the risk of GD have also been expressed in countries where anti-genetic discrimination policies were already in place or where no clear evidence of GD incidents has yet been reported.

3. Are there existing laws to prevent genetic discrimination?

Following international declarations from leading scientific and legal organizations proscribing GD, many countries worldwide, especially in North America and Western Europe, have adopted non-GD laws (see https://gdo.global/en/gdo-map-approaches for a global overview of existing laws and policies). In low- and middle-income countries with less capacity to perform genetic tests on a wide scale, non-GD laws are rarely a priority. Adopted laws either primarily provide sector-specific protection or contain some imprecisions. The landscape of these laws is complex and different

regulations may even find application within a single country, for example in the U.S and Argentina. Laws also struggle to keep up with the pace of scientific advancement in the 'omic' fields, quickly becoming outdated. Existing laws mainly prohibit the imposition of genetic tests, or use of genetic test results, in the context of personal insurance (ex. health, life, or disability) and in the context of employment. Genetic test results are defined differently, broadly, or more narrowly, from one law/jurisdiction to another. For example, they sometimes include family history of disease but most of the time do not. Work on the international harmonization of those laws has not started. In the absence of genetic-specific laws, traditional data privacy and human rights laws can protect against GD to some extent. However, discrimination is an insidious phenomenon that can happen unbeknownst to the person subject to discrimination. Even when discrimination is known, victims may be reluctant to seek a legal solution to this type of problem for various reasons that could include cost of the proceeding, complexity of the process, uncertainty about the judicial outcome of their case, a lack of awareness of avenues for recourse, etc.

4. **What are the implications of genetic discrimination for the sharing of genetic/genomic and health data?**

The fear of GD has been shown to dissuade individuals from participating in genetic research and from clinically recommended genetic testing. While private insurers, employers and other third parties are generally not interested in raw genetic information held in databases, government agencies in several countries have shown interest in such databases for profiling and forensic purposes. Results of genetic tests validated for clinical use could be of interest to many stakeholders, who may attempt to access and use them for discriminatory purposes unless they are legally prevented from doing so. Thus, stakeholders facilitating the sharing of genomic and health data can be doubly impacted by GD concerns and regulations and in the following manner: 1) recruitment in their projects may be negatively impacted because of existing GD concerns; and 2) they may be obligated to provide general information about the risk of, and protections against, GD in their region in informed consent documents. International data sharing compounds GD issues further as it becomes possible to access the data in multiple countries with various legal and ethical norms. Further, participants may not be aware that their data will be shared with another country with different privacy/discrimination laws or expectations.

5. **What should research participants be told about genetic discrimination?**

The information that should be provided to research participants is context dependent. The scope of data sharing, the incidence of discrimination, existing concerns, available protections, and ethical and cultural norms relevant to research projects should also be considered when deciding if, and how, to discuss GD in informed consent documents. Ultimately, the risk of GD will vary depending on multiple factors including the country where the research is taking place (as well as the country with whom the data is being shared), population group(s) included and/or excluded, topic of the research project, genetic information and other personal data sought and existing ethical, administrative, and legal protections. Any statement about the potential for GD should be kept short, as well as, avoid jargon and overly legalistic or technical language. While the risk of discrimination for most projects is

likely to remain relatively low, there are some genomic research projects in which it may be higher. It can be useful to remind patients/participants that the risk of GD is generally similar to or lower than one's risk of being discriminated against from the results of a medical diagnostic. A possible approach is to convey that the risk of GD, while generally low, remains a part of almost every genetic data sharing enterprise. If existing protections in the project's region make GD highly unlikely, it may not be necessary to include it as a potential risk. Instead, in these cases, the informed consent document may include a statement that the risk of GD is greatly reduced because of existing legal protections such as, including provisions to protect against/mitigate re-identification risks.

6. **A call, to the GA4GH community, for information about genetic discrimination Consent Clauses on genetic discrimination**

The GA4GH Workgroup on GD is interested in learning from the experiences of GA4GH members with GD. If you are involved, as a PI, Co-Applicant or ethics collaborator, in a project that collects genomic data from human participants and that has developed consent forms, or other tools, to address GD, please share them with us at: rews-coordinator@ga4gh.org. We are also interested in collecting data about any discriminatory use of genetic information (or request to obtain such information by individuals outside the health/health research context) that you may have encountered. Following a short validation process and provided with sufficient information, we will de-identify and upload any information provided to an open web application, so that it is available for all interested researchers to consult.

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