Draft WHO principles for human genome data access, use and sharing

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Background

Genomics holds great promise with the potential to improve individual and population health. To realise this potential, there is a need to enable access to, use, and sharing of human genome data within and across differing health and research sectors. This can only be achieved by proactively addressing the ethical, legal, social, religious and cultural issues that can arise in the process, but also acknowledging that there can also be risks if these data are not shared. Such risks must be balanced and mitigated so that human genome data access, use, and sharing can occur in a manner that protects individuals and communities and at the same time, promotes their health.

Efforts to scale up access to, use, and sharing of human genome data must recognise the mistrust that can exist when accessing, using and sharing human genome data from some individuals and communities. This is an ongoing challenge that is in part due to persisting exploitative practices, and capacity and power imbalances between different actors and regions. It is also critical that the lack of diverse data sets and the consequent under-representation of many populations in those data sets be addressed if global equity in genomics is to be achieved.

The integration of genomics into health systems requires an all of data life cycle approach with guidance enabling access to, use, and sharing of human genome data within and across health sectors locally, nationally, and internationally. To support this work and following the WHO's Science Council 2022 Report "Accelerating access to genomic for public health" that recommended the promotion of ethical, legal, and equitable use and responsible sharing of human genome data, WHO has developed this document setting out principles for human genome data access, use and sharing.

Purpose of this document

This document sets out globally applicable and inter-connected principles on the access to, use and sharing of human genome data, to promote human health and wellbeing. This document is to complement local legislation and builds upon the many policies, frameworks, and other guiding documents in this space. These principles aim to:

- Promote social and cultural inclusiveness, equity, and justice in access to, use, and sharing of human genome data.
- Promote trustworthiness within the human genome data life cycle.
- Foster integrity and good stewardship of access to, use, and sharing of human genome data.
- Promote communal and personal benefits arising from human genome data access, use, and sharing.

In addition, this document seeks to strengthen capacity and awareness of individuals from whom, and communities in which, human genome data are collected, so that they can exercise greater control over its access, use and sharing if they so desire.

Data life cycle
Data life cycle refers to the different stages of data use, including collection, generation, storage, use, sharing, access, and subsequent deletion or archiving. The term is intended to reflect that data can be used beyond the lifespan of the individual from whom the data originates and that these principles apply to the use of data throughout the data life cycle and not just the lifespan of the individual.

Scope of this document

These principles apply to human genome data access, use, and sharing in health and research contexts, recognising that implementing these principles requires a comprehensive approach along the entire data life cycle. They apply to all prospective and retrospective collections of human genome data. These principles are intended to complement and inspire national legal and ethical regulations, frameworks and guidelines.

Although the scope of this document applies to human genome data only, these data are typically linked with other health information that is critically important to interpreting human genome data. WHO strongly encourages making other health data available with human genome data when this is necessary and does not impose unacceptable risks.

Human genome data are obtained from biological samples thus these principles can equally apply to them. Biological samples, however, require specific considerations that are outside of the scope of this document. Nevertheless, providing access to, use, and sharing of human biological samples should not be used to avoid the application of these principles.

Pathogen genome data[1] and microbiome data do not fall within the remit of this document.

This document sets out principles with guidance on their application, but it is not an implementation document.

Human genome data
Human genome data are data that are derived directly and/or indirectly from the human genome.
These data include but are not limited to:

- DNA sequence from the nuclear and mitochondrial genomes
- Transcriptome
- Proteome (e.g. by deriving genetic information from protein structure)
- Methylome and other epigenetic modifications.

Context

The application of these principles in practice will depend upon the context in which human genome data are accessed, used, and shared. This will include the individuals and community providing the data, those collecting, accessing, using, or sharing human genome data, the purpose of access, the capacity, the resources, the skills, and the expertise of those accessing, using, and sharing the data, amongst others.

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[1] See WHO guiding principles for pathogen genome data sharing.

Equally, national legal and ethical frameworks, as well as social and cultural beliefs may impact the application of these principles.

Audience

These principles are intended to apply to those involved in governing, generating, storing, accessing, using, sharing, and disposing of human genome data in health and research contexts, and individuals and communities from whom human genome data originates.

Principles for human genome access, use, and sharing

To affirm and value the right of individuals and communities to make decisions

A commitment to value and affirm the rights and interests of individuals with capacity to make decisions on what should be done with their human genome data throughout the life cycle of the data. In addition, a commitment to promote the best interests of individuals who do not have the capacity to make decisions for themselves.

The use of human genome data has implications beyond an individual, and the views of communities should be ascertained on access to, use, and sharing of these data throughout its lifecycle.

Actions:

- Human genome data use, access, and sharing should be in line with the needs, preferences and values of individuals and communities.
- Clear, transparent, accessible, understandable, and ongoing information should be provided on human genome data access, use, and sharing to individuals and communities.
- Communities should be engaged in the governance and decision-making process regarding appropriate access to, use, and sharing of human genome data, including the development of appropriate consent models and processes.
- Consent should be as specific and granular as possible about potential uses of the data and should provide information on who is intended to benefit.
- Consent should be supported by appropriate governance processes and individuals should be informed of such processes.
- Where specific consent is not adopted, consent to broad categories of human genome data use may be permissible provided there are safeguards in place. This includes, at a minimum, governance frameworks on the re-use of the data, independent oversight, and such processes should be informed and supported by community engagement.
- Where possible, interactive and dynamic consent models that facilitate ongoing communication and enable individuals to quickly update their consent preferences are encouraged.
- Any uncertainty on the scope of consent on human genome data access, use, and sharing should be determined by an independent body to consider, amongst others, the context and local customs and ethical norms in which the consent was provided and the adequacy of the consent for current technologies.

- Children, upon reaching the age of maturity, can if desired, provide a new consent or change their preference relating to the use of their human genome data.

Social justice

A commitment to provide access to, use, and sharing of human genome data in ways that (i) promote human health, individual and collective well-being; (ii) address the needs of under-served individuals and communities, and those experiencing greater health burdens; (iii) reduce socioeconomic inequalities and health inequities; and (iv) avoid individual and group discrimination and stigmatisation.

A commitment to strengthening the capacity and enable access to adequate resources, skills, training and capacity-strengthening for researchers, clinicians, policymakers, genetic counsellors, all healthcare staff and individuals and communities involved in human genome data access, use, and sharing.

Actions

- The genomics agenda, specifically on the purpose for human genome data access, use, and sharing, should be locally set according to local health needs and burdens, but with an awareness of existing interconnectedness between the local, national, and international health ecosystems.
- Results should be returned to individuals, if such results are clinically relevant, validated, and legally and ethically permissible. Any return of results must follow an approved policy and be in line with the individuals' consent.
- Policies and procedures to protect individuals and communities from stigmatisation and discrimination that can result from the association between genome data, community membership and health status should be developed in advance.

Genetic discrimination & genetic stigmatisation

Genetic discrimination involves an individual or a group being negatively treated, or harmed, relative to the rest of the population, based on actual or presumed human genome data.[2]

Genetic stigmatisation involves an individual or a group being labelled, devalued, negatively stereotyped, or socially excluded, based on actual or presumed human genome data.

Solidarity

Solidarity denotes a commitment to stand with others and to collectively and equitably share the benefits and burdens of human genome data access, use, and sharing, within and across communities.

Actions:

- Interests and rights of individuals providing human genome data access, use, and sharing should continue to be protected, particularly as efforts to scale up diversity and representation are increased.

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[2] Definition as amended from the Genetic Discrimination Observatory

- Decisions on human genome data access, use, and sharing, should include discussions on the risks, benefits, and commitments to facilitating access to the resulting benefits for individuals and their communities.
- Commercial interests should not be used to justify limiting access to, use, and sharing of human genome data.
- Governance processes should be introduced to clearly identify duties for data users and associated sanctions in case of non-compliance that act as deterrents, in cases of individual and collective data harms.

Equitable access to, and benefit from, human genome data

A commitment to provide equitable access to and use of human genome data and its resulting benefits, recognizing that this requires addressing current capacity and power imbalances between different actors that may exist due to exploitative practices and inequities in health systems.

A special commitment to increase diversity and representation in datasets and decisions on access to, use, and sharing of human genome data.

A special commitment to benefit sharing that includes giving back to individuals and communities whose human genome data are accessed, used, and shared.

<u>Actions:</u>

- Increasing representation of datasets across diverse populations is critical but inclusion alone is insufficient to achieving equity. It must be accompanied with, at a minimum, equitable representation in decisions on human genome data access, use, and sharing and decisions on benefits from such activities.
- Special attention should be paid to the equitable sharing of risks and benefits across and within societies, including affordable access to resulting benefits. This should be considered in advance of access, use, and sharing of human genome data, and where applicable, informed by community engagement.
- Capacity strengthening should be considered as part of any access to, use, and sharing of human genome data.

Benefit sharing

Benefit sharing involves the equitable distribution of benefits from human genome data access, use, and sharing. This can include profit sharing agreements, access to resulting diagnostics and therapies, technology transfer, and capacity strengthening.

Collaboration, cooperation, and partnership

A commitment to promote genuine collaboration, cooperation, and partnership between those involved in all aspects of human genome data access, use, and sharing, acknowledging that to achieve this will require a rebalancing of power between individuals, entities, and regions.

<u>Actions</u>:

- Decisions on human genome access, use, and sharing should be made collaboratively between researchers, individuals, their healthcare providers, and communities, amongst others, depending on the context.
- Policies should clarify that human genome data should be accessed, used, and shared within and across the health sectors to improve human health and wellbeing.
- To promote collaborative decision making and partnership development, efforts should be made to increase health literacy on genomics and human genome data access, use, and sharing amongst all stakeholders, including those contributing human genome data and those involved in decision making.

Transparency

A commitment to provide openly available and easily accessible information on policies and processes that describe human genome data access, use, and sharing, including how the data are to be protected.

Actions:

- Publicly available policies should describe who has responsibility for deciding on access to, use, and sharing of human genome data, the criteria for decision-making, and how human genome data will be protected.
- Systems should be put in place to enable individuals and communities to be informed about the use of their human genome data and the resulting outcomes.
- Outputs, including publications from human genome data access, use, and sharing should be made available to those who provided the data within a reasonable timeframe. Lay summaries of key insights should be openly available for all.

Accountability

A commitment to establish processes preventing human genome data misuse, accompanied by mechanisms that hold individuals accountable for their misuses of human genome data.

Actions:

- Mechanisms should be put in place to guard against the misuse of human genome data, including limiting access, use, and sharing by individuals and entities who cannot or will not adequately protect the data.
- Mechanisms should be put in place to ensure that individuals or institutions responsible for human genome data misuses are held to account.
- Those responsible for safeguarding and securing the data, and that can be held accountable for cases of gross negligence or perpetuate data misuse, should be identified prior to human genome data collection, access, use, and sharing.

Stewardship of human genome data

A commitment to encourage, enable, and sustain ethical, legal, socially and culturally appropriate, and responsible, human genome data access, use, and sharing by committing to (i) develop processes to enable access to, use, and sharing of human genome data (ii) follow the current ethical practices on human genome data (iii) identify and minimize potential harms in human genome data access, use, and sharing, (iv) respect applicable law, including laws on privacy and data protection.

Actions:

- Identify and implement appropriate models that provide equitable access in a manner that best protects individuals and communities across contexts.
- Attention should be given to the environmental impact of data processing, storage, and use.
- Human genome data should be accessed, used, and shared, as applicable with the FAIR and CARE principles.
- There should be appropriate attribution of those who provide the human genome data
- Timely data access should be granted but justified, reasonable and proportionate time delays can be permitted. Where appropriate, community engagement should be ongoing throughout the data life cycle to ensure that data collection and subsequent use is in line with cultural and social considerations.
- Resources required (including financing, infrastructure, and personnel) to sustain the use of human genome data should be considered at the outset of the collection and should be reviewed through the data life cycle.
- Current ethical, legal, privacy, data protection, and security standards and practices should be followed, recognising that they may be informed by standards and practices on health data generally.