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The WHO genomics program of work for equitable implementation of human genomics for global health

The publication of the first draft of the human genome sequence in 2001[1] was followed by the advent of high-throughput methodologies, which together have driven the expansion of genomic tools and technologies. These efforts have enabled the sequencing of millions of genomes and expedited the functional and clinical annotation of the human genome. The resulting discovery of thousands of gene–disease associations[2] and the characterization of human genetic variation at scale[3] has extended the application of genomics to enable diagnosis, prognosis and clinical management of disease, as well as risk prediction and implementation of preventive measures.

Despite the health benefits from these innovations and the potential to improve sectors related to agriculture, livestock and the economy (with indirect effects on health), the availability of genomic technologies remains inconsistent and often limited, particularly in low- and middle-income countries[4,5], in part because of resource shortages and low adoption capabilities within health systems. The lack of representation of genetic data from many underserved communities exacerbates this disparity, resulting in a predominant focus of human genomic studies on European ancestry populations[6,7].

The World Health Organization (WHO) Science Council recognized the potential of genomics alongside global disparities in the adoption and sharing of its benefits, and produced a report[8] outlining a roadmap to accelerate access to genomic technologies and applications across all continents. The report focused on four overarching themes: promotion; implementation; collaboration; and ethical, legal and social issues.

Subsequently, WHO launched a program of work that acknowledges the many efforts in the realm of human genomics[9], and is strategically designed to advocate for investments in the field, promote collaboration, overcome obstacles to implementation, and address inequitable access to genomic services.

Table 1 | The WHO genomics program of work

Topic	Details
Challenges	Poor awareness of the benefits of human genomics for individual and public health
	Lack of alignment and collaboration between human genomic stakeholders
	Limited financial and technological resources, and trained workforce
	Inconsistent equity, ethics and governance frameworks in human genomics
	Limited tools and frameworks to evaluate impact of interventions globally
	Missing guidance to implement human genomics in research and clinical practice
Actions	Communicate benefits and potential of genomics to different audiences
	Advocate for investing in genomics
	Map gaps, opportunities and priorities
	Guide genomic implementation based on good practices and local priorities
	Engage stakeholders across sectors and regions
	Promote a community of practice in genomics
	Support workforce education and training in genomics
	Guide human genome data access, use and sharing
	Guide equitable genomics research and practice
Outcomes	Genomics stakeholders and expertise centers aligned and collaborating to accelerate the implementation of human genomics in research and clinical practice
	Available guidance and technical support to increase human genomic implementation for individual and public health
	Expanded availability of human genomic knowledge, technologies and expertise in research and clinical practice
Impact	Equitable access and use of human genomic knowledge and technologies in research and practice
	Enhanced human genome research and knowledge of the role of genetic variation in health and disease across diverse countries and populations
	Accessible genome-based technologies and interventions for health prevention, promotion, diagnosis and management of diseases in various settings
	Improvement of individual and population health globally

To provide ongoing guidance to the program's actions and to monitor its progress, the WHO Technical Advisory Group on Genomics (TAG-G)[10] was established, consisting of experts from diverse disciplines, practice contexts and regions. Strategic actions prioritized by the WHO TAG-G are expected to advance the utilization of genomic technologies in clinical practice and research, ultimately contributing to the improvement of individual and population health globally (Table 1).

The WHO genomics program advocates for genomics through tailored communication efforts aimed at various audiences, such as governments, decision-makers, funders, the public and other pertinent stakeholders. These endeavors might include explainer videos, posters, leaflets and infographics designed in consultation with the WHO TAG-G to construct a persuasive narrative that highlights the medical, scientific and economic benefits of human genomic applications.

In addition, this communication material seeks to cultivate trust in genomics by determining its potential and outlining key considerations relevant to both individual and public health.

To build evidence of the economic value of adopting human genomic applications into healthcare across regions, several investment scenarios will be developed by WHO with input from the WHO TAG-G, taking into consideration population-specific diseases and genetic diversity, as well as available financial, technological and health systems resources. Depending on context, selected cases might include targeted prenatal or newborn screening for actionable diseases such as thalassemia and other hemoglobinopathies, reproductive or preconception carrier screening for recessive and/or X-linked diseases, testing for inherited cancer risk or pharmacogenomic testing to foster best use of therapeutics and reduce adverse events.

WHO is connecting with other relevant organizations and stakeholders in the field to strengthen collaborative initiatives in human genomics. This will map international and national expertise and ongoing efforts and emphasize alignment to prevent duplication and encourage a synergistic approach. This collaborative approach could encompass joint efforts in convening professionals, developing and implementing training programs, fostering pedagogical research, and advocating for integrating genomic and genetic counselling into the training of undergraduates and graduate health professionals. Progress has been made in genomic education and training, in which WHO is forging connections with initiatives operating in the field.

Another challenge that hinders implementation in many countries is the lack of direct access to materials, services and support from major providers of genomic equipment, reagents and analytical tools, which are primarily situated in high-income regions. Genomic entities in low- and middle-income countries rely on indirect access to these products through local distributors, resulting in higher costs and suboptimal support. To tackle such practical implementation challenges, particularly in the provision of equipment, reagents and computational infrastructure, efforts will be explored to engage with the private sector and execute plans to enhance the equitable accessibility and affordability of genomic technologies and solutions globally.

The program will foster collaboration at all WHO regions and sub-regions, by organizing consultations on human genomics involving WHO member states, local experts from healthcare and research sectors, and WHO TAG-G members. Consultations will be thoughtfully tailored to regional priorities and respond to the unique needs of each region, being firmly rooted in local expertise. Beyond nurturing collaboration within regions, consultations will serve as a cornerstone for future endeavors.

Efforts are underway to develop WHO principles for human genome data access, use and sharing in consultation with the WHO TAG-G and other experts. These principles aim to establish comprehensive global guidance to promote ethical, legal and equitable use and responsible sharing of genomic data along the data life cycle and among diverse stakeholders, fostering trust, public value, and upholding integrity and stewardship. They are intended to complement local regulations and guidelines and to be relevant across different health and research sectors.

The WHO TAG-G strongly advocates for prioritizing equity and developing an equity roadmap to implement genomic applications for global health. This should include equitable partnerships in research, fostering diversity within the genomic research workforce, ensuring representation across diverse groups in genomic studies, and facilitating the accessibility and appropriateness of genomic applications to all.

Advancing the generation of human genomic knowledge among underrepresented populations and enhancing access to genomic applications for global health requires action at the international, national and local levels. These efforts must align with the unique needs and priorities of each country and region. Without the commitment of local political leaders, advocates and communities, along with financial backing and support from governments, meaningful progress is unlikely. Failure to address disparities in access to human genomic applications will exacerbate health inequalities, leaving the most underserved communities behind. Ultimately, it is through the collective effort of all stakeholders and regions worldwide that the potential of human genomics for global health can be fully harnessed.

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