Human genomics for health Enhancing the impact of effective research

Report of the first regional meeting for the Americas

Brasília, 15–16 May 2024

PAHO
Pan American Health Organization
World Health Organization
Americas Region

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Contents

Abbreviations and acronyms

DECIT	Department of Science and Technology
HIC	high-income country
LAC	Latin American and Caribbean
LMICs	low- and middle-income countries
NIH	National Institutes of Health
PAHO	Pan American Health Organization
WGS	whole-genome sequencing
WHO	World Health Organization

Introduction

Scientists, clinicians, and public health experts across the genomics and precision health ecosystem from 19 countries in the Region of the Americas converged in Brasília, Brazil, for the first regional meeting on human genomics, held on 15–16 May 2024. The meeting was organized by the Pan American Health Organization (PAHO) with the support of the World Health Organization (WHO) and the Department of Science and Technology (DECIT) of the Ministry of Health of Brazil. This meeting provided an opportunity to reach a mutual understanding and to set the agenda for enhancing the impact of effective research for genomics applications in the Americas. To enhance communication and encourage the exchange of experiences and ideas among participants, the meeting was held in Spanish, English, and Portuguese, with simultaneous interpretation.

The objectives of the meeting were to:

1. Raise awareness among Member States and regional stakeholders about the WHO Science Council report *Accelerating access to genomics for global health: promotion, implementation, collaboration, and ethical, legal, and social issues (1)*;
2. Share experiences and best practices in the implementation of genomic evidence to improve the precision of the practice of medicine at an individual level and to inform public health strategies at a population level;
3. Identify challenges and feasible strategies for the implementation of genomics in practice and for conducting genomic research in Member States;
4. Create opportunities for linkages and collaborations.

Participants represented a broad spectrum of disciplines and contributed their expertise across relevant fields of research, including science policy, bioethics, biobanking, genetics, national genomic and pharmacogenomic initiatives, bioinformatics, cancer, rare diseases and congenital disorders, health technology assessment, and health economics. During the two-day meeting, a highly diverse group of experts shared experiences and best practices in the application of genomics in clinical care and public health, and in conducting genomic research (see details in the Appendix).

This publication provides an overview of the meeting's proceedings and summarizes the discussions of the participants.

CHAPTER 1

Background

Genomics is the study of the complete genetic material (the genome) of organisms and of the way genes and other genetic elements work and interact with each other and with the environment. It incorporates elements of genetics but, unlike the latter, genomics is concerned with the characterization of all genes of an organism, rather than individual genes. Genomics is a rapidly evolving, multidisciplinary field of science and is progressively offering insights in various areas, including clinical care, ancestry and human genetic variation, food safety, and infection control, and is a vital component of the One Health approach.

> The advances in human genetics that have occurred during the past twenty years have revolutionized our knowledge of the role of inheritance in health and disease. The genome not only determines the cause of catastrophic single-gene disorders that affect millions of persons worldwide, but also – depending on environment – puts individuals at increased risk of cardiovascular diseases, cancer, and other common diseases *(2)*.

An individual's genetic endowment has long been recognized in medicine as a key determinant of their health. It influences all aspects of health throughout a person's lifetime, including their longevity, resistance to environmental exposures, and ability to respond to therapy. There is a recognized genetic burden of disease in all human populations. In high-income countries (HICs), genetic and congenital disorders are the second most common cause of infant and childhood death, occurring with a prevalence of 52–62 per 1000 live births in Latin America *(3)*. Most prevalence estimates tend to be disease specific, precluding an examination of genetic conditions in the pediatric population overall. An estimated 3.5–5.9% of individuals worldwide have one of approximately 7000 rare or genetic conditions *(4)*. Therefore, although the prevalence of any single genetic disorder may be low, the burden of these conditions can be significant on families, public health systems, and healthcare systems *(5)*.

Genetic assessment and testing are used in all medical disciplines to meet standards of practice for the prognosis, diagnosis, treatment, and ongoing management of disease in both children and adults. Advances in human genomics are only increasing the utility of an individual's genetics in their health care.

The first human genome sequencing project culminated in 2003 and revolutionized the landscape of genomics, pushing forward the field of precision medicine *(6)*. Multiple population genomic programs have been initiated worldwide in collaborative efforts to increase the diversity in human genetic databases, which has resulted in promising outputs of the whole-genome sequencing (WGS) and microarray-based genotyping of millions of human genomes, offering breakthroughs in precision health care. The potential benefits of genomic medicine and research have become apparent for multiple countries worldwide. Such benefits include gaining a better understanding of

disease etiology, early diagnosis, drug design, pharmacogenomics, epidemiological studies, and preventive and precision medicine. Population-specific national genome programs are attracting more attention worldwide, with the aim of improving genetic diagnostics and paving the road for implementing large-scale screening and precision medicine in healthcare systems for national populations.

CHAPTER 2

Regional context

The populations of the countries of the Americas vary regionally with respect to the degree of admixture resulting from the migration of Asian, Pacific, Middle Eastern, European, and African populations to the Western Hemisphere. In addition, the indigenous populations of the Americas are genetically and historically distinct. The history and geography of the Americas does affect the genetics of its populations, and this in turn has an impact on population and individual health.

There are numerous documented instances of environmental adaptations and founder effects having an impact on the health of some populations in the Americas. However, a more comprehensive understanding of the extent of genomic variation in these populations is required to accurately identify and interpret the genomic variants associated with disease, their effects on the progression of illness, and their influence on responses to therapies.

Population stratification even within a single Member State, as has been documented, can affect the incidence of disease-related genomic variation and thus potentially disease risk. This can further have an impact on the implementation of clinical and diagnostic services and therapies. The degree of population stratification across the Americas is currently not known, even in HICs. Indigenous groups carry unique genomic information, missing from other human populations, and, currently, this can be hard to interpret in the clinical setting with respect to its impact on disease.

Knowledge sharing among all Member States and a shared understanding of the genomic architecture of the populations will be important for enhancing progress and strategizing on the best means of delivering clinical and diagnostic genomic services.

The extent of clinical and diagnostic genomic services differs between Member States. Access to clinical and diagnostic genomic services, even in HICs, is incomplete. Both public and private funding models exist, and for some Member States access to genomic diagnostic services relies on external providers. There is a shortage of trained, credentialed clinical and diagnostic personnel in all regions. The requirements for diagnostic laboratories that perform genomic testing to be accredited and to work to a recognized standard such as ISO 15189 are not consistent across Member States in the Americas.

While the value of genomic testing is recognized, it may be that the ability to efficiently implement it in clinical practice is hampered by the size of the population or the size of the region. As with other diagnostic testing, for genomic testing to be cost-effective and technically robust, a certain volume of tests must be undertaken annually. Genomic testing also requires infrastructure in terms of personnel, equipment, and information technology.

CHAPTER 3

WHO Science Council/genomics program

The WHO Science Council, in its inaugural report titled *Accelerating access to genomics for global health: promotion, implementation, collaboration, and ethical, legal, and social issues (1)*, recognized the immense potential of genomics in addressing global health challenges. Published in 2022, this report emphasizes the urgent need to ensure equitable access to genomic technologies worldwide.

The report outlines 15 strategic actions for accelerating the establishment and sustainable use of genomic technologies globally. These actions focus on four key themes: promotion, implementation, collaboration, and ethical, legal, and social issues.

Since the publication of this report, WHO has launched a strategic program of work around genomics to promote collaboration, overcome implementation challenges, and address inequitable access to genomic services. The WHO Technical Advisory Group on Genomics (TAG-G), composed of experts from diverse disciplines and regions, guides these efforts. Its goal is to advance equitable access to human genomic knowledge and technologies in clinical practice and research, benefiting individual and population health.

CHAPTER 4

Implementation of genomics in clinical medicine and public health

Findings from genomics research have applications in clinical care, namely in oncology, neurology, and rare diseases. The presenters on day 1 of the regional meeting focused on approaches for advancing genomics applications and exploring methods for effective implementation, diffusion, and sustainability in diverse clinical care settings. These included health technology assessments, economic evaluations, and regulatory pathways for the use of genomics technologies in countries of the Americas. The group discussion identified financial constraints, the scarcity of professionals trained in genomics, a shortage of coordinated infrastructure, and the lack of political will as the most prevalent challenges across countries. Below is a summary of best practices and recommendations that will contribute to increasing the access to and implementation of genomics in patient care and population settings (please see Appendix).

In most Latin American and Caribbean (LAC) countries, public health systems suffer from chronic underfunding, and, with a few exceptions, care for genetic disorders is seldomly incorporated into health systems. This is reflected in the lack of economic resources for the implementation of precision medicine. These financial constraints are intertwined with a lack of political will. Most national health authorities consider genomics applications expensive or there to be more urgent healthcare priorities to address. There are gaps in health policy, in terms of both accessibility to genomic tests and the application of the results from a clinical point of view. In some countries, there is also a lack of regulation and clinical guidelines for orienting and standardizing treatment once a diagnosis has been made (clinical pathways). This gap is more severe regarding genome-editing therapies.

Health economics assessments of genomics in medicine are needed in LAC countries. These studies may show health authorities that not all genomic interventions are necessarily out of reach of low- and middle-income countries (LMICs). The translation of knowledge in genomics research should incorporate health technology evaluation to facilitate the visibility of returns on investment and social benefits and of the importance of genomics research for both decision-makers (ministers, legislators, presidents) and the population. This will bring genomics closer to the front lines of health care. Mechanisms that facilitate communication and give value to genomic research for decision-making and support the generation of healthcare policies (e.g., availability of business case templates) should be established.

There is a need for guidance on training, credentialing, and accreditation for personnel, clinics, and diagnostic laboratories, as well as guidance on the implementation of genomics in research and clinical practice. As in research, it is also important to harmonize data collection, including genomic metadata in clinical implementation. This is important for enabling useful country-level impact comparisons to be made, validating implementation, and enabling the use of data to enact public health actions. Curricula for the training of specialists in genomics and

related sciences that have international validation should be harmonized and accredited. This requires cooperation from regional health authorities in identifying regional health priorities and funding programs. Collaboration between researchers and those in clinical/diagnostic practice is needed to increase the successful adoption of translational research.

Genomics education programs aimed at medical and research personnel need to be integrated, to increase the critical mass required to advance the field. Telematics education in genomics and bioinformatics, through international platforms such as the PAHO Virtual Campus and by taking advantage of human genetics associations, could be used to train personnel. Diverse professionals such as bioinformaticians, molecular pathologists/geneticists trained in genomic interpretation analysis, and genetic counselors have not traditionally been incorporated into the public health system in most LAC countries. Incorporating biomedical engineers, information technologists, and other relevant professionals, and utilizing hybrid (blended) models of education will strengthen interdisciplinary capacities. Member States with training programs could participate in mentorship and share training objectives, competencies, and curricula, to decrease the time needed to develop programs in other countries. In addition, Member States could "purchase" places in existing training programs as an interim measure, although it is recognized that this may be complicated by the need for return agreements with trainees.

Sequencing equipment and reagents, as well as sequencers, are still lacking in many LAC countries. It may be necessary/useful to create a regional mechanism coordinated by regional health authorities for the strategic purchasing of equipment/technology to give continuity to genomics programs (e.g., PAHO could potentially support access to a mechanism like the one used for the negotiated purchase of vaccines) and to ensure the sustainability of sequencing and genomics laboratories. It will be essential for strategic planning to favor using and sharing sequencing infrastructure: information technology hardware and software, national and regional biorepositories, regional reagent purchase agreements, and equipment service and maintenance training and agreements.

Health authorities should establish regional information forums on genomics that consider the needs of their communities and individuals, particularly those affected by rare diseases. Government and health authorities that enter public–private partnerships to provide the necessary resources for genomic medicine should guarantee the equity of the parties involved and be based on social responsibility, especially with respect to vulnerable groups in society.

CHAPTER 5

Conducting genomic research

Day 2 of the meeting consisted of presentations on large-scale genomic projects (e.g., All of Us, JAGUAR, LatinCells, Genomas Brasil) with a focus on genomics research, including data-sharing. In addition, there was a strong emphasis on the potential for collaborative efforts and the enhancement of partnerships within the Region. The discussions underscored the importance of building robust networks to support ongoing research, facilitate the sharing of resources and expertise, and develop guidance on data-sharing (please see Appendix).

Across the board, LAC countries lack research agendas that incorporate human genomics. The scientific community together with healthcare workers should identify pertinent research projects that will resonate with policymakers and the public. Instead of looking only at what seems interesting, a broader lens should be used to identify needs on the regional and global scales.

Discussing the feasibility of genomics applications and realities in healthcare settings will increase the likelihood of implementation and continuity. Therefore, early engagement with healthcare workers, genetic counselors, and data scientists should be prioritized when planning translational genomics research. Strategies to translate and disseminate the knowledge generated to reach the public domain, the integration of academia, and workflows should be discussed and developed at the conceptual stage.

Communication between politicians, decision-makers, and researchers may contribute to enhancing the much-needed political commitment. Engaging politicians and presenting information packages in line with the healthcare priorities of governments may provide common ground for discussion. The creation of a regional genomics collaboration hub will benefit genomic research by harmonizing metadata and associated clinical data for all patients with the same pathology, and through the allocation of budgets within the public health system. Public funding is required to ensure that the same individuals who have contributed to the development of genomics research/databases are able to access the genomics services that are developed. Concrete examples of the value of genomics in medicine should be presented to health authorities (e.g., decreasing the rate of neonatal malformations is important for decreasing child mortality rates), thus promoting national health policies that incorporate the need for a genomics research program in LAC countries.

Development requires innovations that countries are not ready to deal with from an ethical standpoint. The creation of legal and ethical frameworks for the management of biorepositories and data governance needs to be prioritized, and consideration needs to be given to groups such as indigenous populations and those with rare diseases. Cultural practices/beliefs may hinder use in some populations, and past practices in research and health care may also contribute to hesitancy and a lack of trust. Therefore, specific measures may be required to reach out to some populations (e.g., the consideration of existing research programs that have successfully reached indigenous groups).

Many LAC countries identified the need to build local research capacity, in terms of not only infrastructure for sequencing equipment and information technology, but also human resources. A range of strategies globally have proved successful and have been adopted to retain graduates and postgraduates in their home country.

LAC countries are in dire need of guidance and the establishment of pathways for genomic data-sharing. This includes the secure sharing of information on genomic variation discovered and the associated health-related information, and the generation of guidelines and regulatory documents for the development of genomic research that safeguards and guarantees the protection of the genomic data of the populations being researched, particularly data in repositories. This will require governments to assist with expectations and infrastructure for secure data-sharing among Member States by learning from international consortiums (e.g., Global Alliance for Genomics and Health). Guidance should be expanded to include a duty to return the genomic data and samples as a condition of participation in research or external clinical testing services to overcome the loss of genomic information and samples to external private stakeholders. The consequence of not having this policy has meant that information is not accessible for ongoing local research/clinical service analysis/use and is unavailable for patient care. There is the need for the creation of legal and ethical frameworks for the management and use of genomic data for all individuals' ethnic groups, and special attention should be given to vulnerable populations (e.g., indigenous populations, children, pregnant women, and older persons).

CHAPTER 6

Key priorities

The meeting uncovered the broad and varied expertise in human genomics across the Region, reflecting the interests and priorities of Member States in this field. The findings underscore the necessity for coordinated action at national, regional, and global levels to equitably harness the transformative potential of human genomics. Member States will focus on strengthening human resources, infrastructure and equipment, national policies and guidelines, and funding mechanisms regionally, together with PAHO, for the creation of a collaborative hub.

6.1. Strengthening human resources

Most countries applying genomics in patient care do not have the required expertise. Training programs are required for research, clinical, and laboratory diagnostic personnel at all levels; this includes creating new, innovative programs and enhancing existing programs. A credentialing program is needed for all clinical and laboratory diagnostic personnel to ensure that standards of care can be met and can generate high-quality test results. Those using genomics in the clinical/diagnostic setting must demonstrate that they have the knowledge and skills to keep patients safe and preserve their data privacy. The expectations of all clinical personnel (physicians, nurse practitioners, etc.) is that they are familiar with the features of an individual's clinical and family history that are associated with an increased genetic risk. To meet the growing demand for genetic care, clinical personnel (e.g., nurse and biomedical practitioners) must be trained to ascertain who is genetically at risk, to avoid entirely relying on physicians trained in clinical genetics.

A diagnostic laboratory accreditation program is needed in all jurisdictions offering testing for use in patient care; this is a patient safety issue and it is not currently being adequately addressed. Examples of existing programs and guideline sources are given below:

- International Organization for Standardization ISO 15189:2022 Medical laboratories – Requirements for quality and competence;
- The Institute for Quality Management in Healthcare (Canada) and the College of American Pathologists Laboratory Accreditation Program (United States of America) are based on ISO 15189:2022 standards;
- The Clinical Laboratory Accreditation Program of the Brazilian Society of Clinical Pathology;
- Guidelines for the validation and delivery of genomic services available from the American College of Medical Genetics, the Canadian College of Medical Geneticists, the European Society of Human Genetics, the National Society of Genetic Counselors, and other organizations.

As part of laboratory accreditation, laboratories are required to participate in proficiency testing programs, in particular external proficiency testing programs such as that of the College of American Pathologists or that of the European Molecular Genetics Quality Network.

There are international programs that can be adopted by Member States, and genomics experts have a role in developing and participating in those training programs. Curricula for the training of specialists in genomics and related sciences that have international validation should be harmonized and accredited. Contracts that provide guarantees of employment, salary, and a return to work for all trainees are one example of a means of encouraging trainees to return to their country. Appropriate salary compensation for healthcare professionals with diplomas or graduate-level training in genomic medicine will also encourage those in direct contact with patients to undertake continuous specialist training.

Alternative forms of education delivery for human talent should be considered according to the reality of each country and region. Member States may opt for locally provided courses and programs with in-person attendance and telematics education in genomics, bioinformatics, and ethics for training, utilizing international platforms (e.g. PAHO Virtual Campus) or hybrid (blended) models of education.

6.2. Localization (infrastructure, equipment, and funding for sustainability)

Sustainable, long-term funding is required for research, clinical and laboratory diagnostic programs, and training programs. Limited financial resources in LMICs may be further exacerbated by the higher cost of purchasing reagents and equipment than in HICs. Member States may require consortium purchasing within and between each other, including the right to repair equipment agreements with vendors. International organizations that have similar agreements/mechanisms for the purchasing of medical countermeasures and health technologies may support Member States.

Political cycles may result in inconsistent funding for research or clinical services, which may have an impact on knowledge generation, patient care, and the retention of qualified personnel. Member State population sizes (e.g., island populations) may not be large enough to sustain their own genetic/genomic services and may require interstate collaboration and data-sharing. Large geographical areas, remote populations, and differing economic means may also hinder adoption, and may require telehealth or specialist outreach services.

Funding applications for translational research can include government or regional health authority individuals who will later oversee implementation and funding in the healthcare system. As for any other clinical service and clinical diagnostic laboratory service, funding for genome-based testing should be sustainable and not subject to the influence of political cycles. Sustainable funding is needed to retain trained personnel, ensure the continuity and safety of patient care, and enable personnel to participate in regional training programs.

Strategic planning at national and local levels favors infrastructure, biorepositories, warehouses for inputs/reagents, and systems for replacing inputs/reagents due to expiration. Translational research should be directly relevant to regional and national health priorities. The relevant ministries should be included in preparing grant applications, and translational researchers should collaborate with their diagnostic and clinical colleagues to ensure that outcomes can be implemented in patient care. It is recognized that Member States may need to pursue different strategies, such as collaborating with other regions for testing or sample sharing, to realize the cost efficiencies needed, and that these may not be permanent arrangements but rather necessary steps in the process of implementing genomic services.

Technical support from companies for equipment should include a right to repair to keep equipment functional and maintain continuity. Shared endeavors among Member States may be needed as an intermediate step in implementation. With respect to local capacity-building, there should be a strategy for repatriating any testing done

abroad to Member States. This would ensure that Member States have data ownership and can realize the benefits of that. Member States should consider a standard that includes the right to return data and samples from the external laboratories, either research or clinical, that are performing testing.

Those doing translational research should engage at an early stage with the clinical and laboratory diagnostic groups that will ultimately have to implement the findings of that translational research in clinical care; this will ensure that the results of translational research are relevant and will maximize the potential for adoption.

The construction of public–private partnerships that guarantee the equity of the parties involved, based on social responsibility, especially that of vulnerable groups in society (distributive justice), is important. Consortium approaches to engaging suppliers of reagents and equipment to ensure a consistent supply and the best pricing should also be considered.

6.3. Policies and knowledge dissemination

Countries need support from health authorities to break down barriers to knowledge sharing across districts and between research and clinical/diagnostic groups. This will assist with ensuring sustainable data-sharing, implementation, and ongoing improvement in the use of genomic technology in health care.

The strategy for communication with health authorities needs to include education on the economic benefits and the medium-to-long-term value of incorporating genomics into healthcare systems While health authorities may not understand the technology, they should appreciate the impact that genomics could have on issues that are of common interest, such as child health and cancer, or issues of primary concern within the Region of the Americas. Health systems need to recognize that the technology is transferable to other more common disorders and other disciplines, including pathogen-related fields, agriculture, and environmental and ecological monitoring.

Another obligation is ensuring that minority and vulnerable groups have access to developments in genetic testing and associated care. Publicly funded strategies may be needed to ensure that those most impacted have equitable access. National health policies that incorporate the need for a genomic research program in LAC countries need to be promoted.

The generation of mechanisms for the translation of knowledge in genomic research and its importance for both decision-makers (ministers, legislators, presidents) and the population is necessary. This will bring genomics closer to primary care, alongside sufficient support and advocacy from nongovernmental organizations and the community. There is also a need to translate and disseminate knowledge in genomics that incorporates the evaluation of health technologies that facilitate the visibility of returns on investment and social benefits. Another requirement is the establishment of regional information forums on genomics that consider decision-makers, academics, healthcare officials, and communities of patients, and that prioritize those with rare diseases.

It is important to increase awareness of the benefits of human genomics and its value as a field that has multiple uses beyond understanding heritable human conditions, including among genomic researchers/clinical groups so that they can communicate with decision-makers and engage in areas of common interest with their governments.

6.4. Regional hub on genomics

Depending on the size of the country and state of development in this area, collaborating on the development of genomic testing capacity could be considered, because this would allow such development to take place in smaller steps, providing an intermediate solution. Common tools/equipment/testing/bioinformatics could be considered,

to facilitate fair and transparent collaboration among different countries. PAHO may play a role in involving other stakeholders and assisting with building consortiums and collaborations.

Taking advantage of the varied countries and expertise present at the meeting, PAHO recommends creating a regional technical group that will continue the work and coordinate four working groups with different expertise: one that focuses on a regulatory framework of research (which could include bioethics, data-sharing standards, and regulation in general); one that focuses on educational and training guidance (which could define standards for harmonizing genomics and related sciences curricula); one that focuses on genomic research (which could be divided into many subdivisions but in general would focus on research that is disease or population based); and one that focuses on clinical and diagnostic practice or research. Representatives of these four groups can further develop the terms of reference for each group as relevant and feasible. Deliverables would include the development of documents, or a white paper or a pitch package to be presented to government or an agency locally, which could then collectively be used to secure funding for a much more ambitious program in the Region of the Americas. This regional technical group will be able to bridge the two main limitations identified: lack of financial resources and lack of partnerships.

CHAPTER 7

Summary of recommendations

As was evident throughout the meeting, the rapidly evolving landscape of genomics has paved the way for exciting and innovative approaches in the fields of precision health and public health. Below are recommendations to be addressed collectively in Latin America and the Caribbean.

1. Mapping of existing resources in the Americas: create an inventory of existing initiatives, genomics applications, policies, and guidelines to support countries and research programs to find overlaps and gaps in local capacity, leverage resources, and further strengthen regional collaborations.
2. Regional advisory committee: establish a technical working group at a regional level to further support the development of solutions for each challenge identified. This small group of experts could in turn convene a larger network of regional experts to make recommendations for regions, including a list of disease priorities, in language that will be easily understood by the relevant ministries, to support workflow coordination.
3. Regional collaboration hub: establish a hub coordinated by PAHO to promote interaction among Member States, institutions, and academics from the Americas, with the aim of sharing knowledge and best practices, developing technical documents among other products, and promoting access to better conditions for acquiring and using the necessary equipment and reagents through collective bargaining and purchasing.
4. Communication material and its dissemination: develop technical documents and present them to the ministries of health of various countries seeking support for genomic research and implementation. Such documents should be as concise as possible, for instance, in the form of a pitch package, and briefly convey the necessity and value of decisively supporting genomic research across Latin America. Develop and adapt communication resources to explain to the public the value of genomics and genetics, which could be illustrated using specific examples from the region.
5. Economics of genomics: adapt or develop a costing tool that will support the sustainability of medium- to long-term genomics research programs. Develop and share strategies to secure sustained, long-term financial support for the implementation of clinical and diagnostic genomic services that is independent of any political cycles within public health systems (e.g., to create and retain capacity). The development of business case templates for scenarios that include the transition from research to clinical testing and for expansion of current diagnostic testing would ensure that all research generates a return on investment in economic and social terms. The integration of ethics analysis should be supported as part of decision-making processes aimed at determining which genomics technologies ought to be offered to the population.
6. Ethical sharing of data and samples for research: strengthen capacities for the ethical collection, sharing, using, and reusing of genetic/genomic data and samples for research. Support Member States to modify their normative frameworks, to allow ethical data-sharing for research as needed.
7. Capacity-building: develop training modules that can be adapted at country level for a variety of stakeholders (e.g., genetic counselors, bioinformaticians, healthcare workers). Utilize online training and artificial intelligence tools when needed to bridge gaps in capacity and for the creation of technical documents.

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Appendix

Agenda

The links for the recorded event stream can be found at https://www.paho.org/en/events/paho-hold-regional-meeting-human-genomics-health.

Day 1, Wednesday 15 May		Speakers/moderators
8:30	Registration and check-in for interpretation services	
Opening Moderator: PAHO Brazil		
9:00–9:30	Opening session and remarks	**Lely Guzmán on behalf of** Socorro Gross, Pan American Health Organization/World Health Organization Representative, Brazil Anna Laura Ross, World Health Organization (WHO), Headquarters Mônica Felts, DECIT, Ministry of Health, Brazil
Session 1: Introduction Moderator: Ludovic Reveiz (PAHO)		
9:30–10:15	• Presentation of the WHO Science Council report on genomics (10 min) • Introduction from WHO Headquarters about the genomics program of work and the WHO Science Council Technical Advisory Group on Genomics goals (10 min) • National Program of Genomics and Precision Health – Genomas Brasil (10 min) Group photo	Anna Laura Ross, WHO Elena Ambrosino, WHO Evandro de Oliveira Lupatini, Brazil
Coffee and tea		
Session 2A: Implementing genomics in clinical medicine and public health Moderators: Iscia Lopes-Cendes (TAG) and Vania Canuto Santos (PAHO)		
10:30–11:45	• Biomarkers and therapeutic interventions in lung cancer: personalized medicine in oncology (10 min) • Rare diseases: linking clinical, molecular, bioinformatics, and social science (10 min) • Health technology assessment experience in Brazil with new genomic technologies: from horizon scanning to access (10 min) • Managed entry agreements in Argentina and subsequent reevaluation to foster access to genomic technologies (10 min) • Building an investment case/economic evaluation for the application of genomic technologies in Colombia (10 min) Q&A	Alejandro Ruíz-Patiño, Colombia Gabriela Repetto, Chile Luciene Bonan, Brazil Manuel Donato, Argentina Adriana Robayo, Colombia

Session 2B: Implementing genomics in clinical medicine and public health – Challenges		
11:45–13:00	Interactive: **What are the specific challenges for implementing human genomics in the Region?** *The goal of this session would be to collectively develop a table that lists specific challenges (for some/all types of applications, as appropriate).*	
Lunch		
Session 2C: Implementing genomics in clinical medicine and public health – Strategies		
14:00–15:30	Breakout groups: **What are the strategies for the identified challenges?** *The groups will discuss viable strategies and experiences, addressing the specific challenges (or cluster of challenges) identified in session 2B.* Present "strategies and experiences" to their respective groups	Group A (English) Group B (Spanish) Group C (Spanish, English, and Portuguese) (S+E+P)
Coffee and tea		
Session 2D: Implementing genomics in clinical medicine and public health – Recommendations Moderators – Manuel Saborío (Costa Rica) and Simon Anderson (Barbados)		
16:00–16:45	Panel: Experiences and strategies in overcoming challenges for implementing genomics in practice	Panelists (Session 2C group moderators)
Session 3: Future of human genomics		
16:45–17:00	• The future of gene therapy: from science fiction to reality for LMICs • Conclusion • Closing session for day 1	John Tisdale, United States Secretary Carlos Gadelha, Secretary of Science, Technology, Innovation and Health Complex (Brazil)
End of day 1		

DECIT, Department of Science and Technology; LMIC, low- and middle-income country; PAHO, Pan American Health Organization; Q&A, question and answer; TAG, Technical Advisory Group; WHO, World Health Organization.

Day 2, Thursday 16 May		Speakers/moderators
8:00	Registration and check-in for interpretation services	
Session 4: Conducting genomic research Moderators – Sherry Taylor (TAG) and Ricardo Verdugo (Chile)		
8:30–9:45	• All of Us Research Program (10 min) • Human Cell Map of Latin American Diversity (10 min) • Considerations for developing a national genomic data base and sharing of data in Brazil (10 min) • Pharmacogenomics in oncology (10 min) • The Clinical Genome (ClinGen) resource (10 min) Q&A	Sheri Schully, United States Andrés Moreno-Estrada, Mexico Pablo Ivan Pereira Ramos, Brazil Luis Quiñones, Chile Erin Ramos, United States

Ethics of genomics data-sharing		
Moderator – Carla Saenz (PAHO)		
9:45–10:15	• WHO: Principles for human genome data access, use, and sharing (10 min) • Ethical sharing of genomic data (10 min) Q&A	Deborah Mascalzoni, WHO Elena Ghanaim, United States
Coffee and tea		
Session 5: Collaboration for research		
Moderators – Gabriela Repetto (TAG) and Xochtil Sandoval (El Salvador)		
10:45–12:00	• Genomic Surveillance Regional Networks for Epidemic and Pandemic Preparedness and Response (10 min) • Latin American Cancer Research Network (LACRN) (10 min) • Latin American Network on Congenital Anomalies (10 min) • Global Alliance for Genomics and Health (10 min) Q&A	Lionel Gresh, PAHO Andrea Llera, Argentina Mariela Larrandaburu, Uruguay Peter Goodhand, Canada
Lunch		
Session 6A: Conducting genomic research – Challenges and opportunities		
13:00–15:00	**Breakout groups: Challenges and opportunities** *The goal of this session would be to collectively develop a table that lists specific challenges (that may apply to some or all types of research), and to discuss viable strategies for the specific challenges.*	Group A (English) Group B (Spanish) Group C (S+E+P)
Coffee and tea		
Session 6B: Conducting genomic research – Recommendations		
Moderators: Sasha Peiris (PAHO) and Jennifer Knight-Madden (Jamaica)		
15:30–16:15	Panel: Key challenges and solutions for conducting genomic research	Panelists (Session 6A group moderators)
Conclusion		
16:15–17:00	Foresights on human genomics in 2030 Key actions for enhancing the impact of effective genomic research • The future of genomics in Brazil • The way forward for the Americas	Benilton S. Carvalho, Brazil Technical Advisory Group on Genomics Members Evandro de Oliveira Lupatini, Brazil PAHO
End of day 2		

PAHO, Pan American Health Organization; Q&A, question and answer; TAG, Technical Advisory Group; WHO, World Health Organization.

Summary of the group discussions on Day 1

Challenge	Details of the challenge	Opportunities/solutions/strategies
Lack of capacity	Enabling the logistics of sending/tracking biological samples to be processed in the sequencing technology hubs/parks/centers How to guarantee the appropriate monitoring of the services (e.g., minimum sequencing standards) and the products generated by genomics services	Look for ways to operationalize/use research technology hubs/parks in clinical practice (e.g., universities' sequencing capacities) Create regional infrastructure hubs to continue the growth of healthcare infrastructure, since political conflicts often hinder the expansion of infrastructure Encourage the involvement of all parties (including society) so that there is a greater incentive for financial investment in infrastructure
Difficulties in bioinformatics processing	Lack of professionals and lack of infrastructure for analyzing genomic data Difficulty in retaining professionals trained in bioinformatics due to the amount of grants offered in contrast to those offered by private companies Limitations in terms of the technological park, in which genetics and molecular biology are still seen as exclusively research areas	Train new professionals to work in this field Invest in artificial intelligence to be used for the benefit of genomics
Regulatory obstacles and ethical challenges	Lack of specific legislation guaranteeing the privacy of patient data Lack of specific legislation to deal with the protection of communities. Institutional review board fears of genetic reductionism and/or misuse of genetic information for discrimination	Stimulate dialogue between society, health professionals, and the scientific community to think jointly about issues involving access to genomics in public health Form groups of experts to carry out studies that indicate the costs of implementing genomics in the short, medium, and long term Train Compliance and Ethics Professionals (CEP) to work in the field of genomics Create ethical criteria for allocating resources to benefit the greatest number of people, but also consider the needs of small groups (e.g., those with rare diseases)
Lack of infrastructure and equipment	Limited regional infrastructure, especially for data storage and sharing, requiring a national public repository/population genomic database	Make the most of installed national capacity, strengthen existing regional capacities, and invest in infrastructure in regions that are in deficit It was commented that an infrastructure for storing genomic data is important, but not essential for the implementation of genomics. Among the advantages of having an infrastructure for genomic data are (1) optimization of public resources since genomic information can be relevant to health throughout a patient's life and (2) compliance with legal regulations (e.g., LGPD or GDPR)
Financial constraints	Funding is scarce. Costs are high. A maintenance fee from the hegemonic sequencer provider costs USD 10 000 or more	Find ways of communicating and sensitizing politicians to direct resources toward incorporating genomics into the health systems, showing that the gains go beyond the financial

Challenge	Details of the challenge	Opportunities/solutions/strategies
Difficulties establishing effective public–private partnerships or networks		Carry out economic/tax incentives Improve the regulatory barriers that slow down processes
Lack of workforce capacity	Lack of training and qualification of specialized professionals. Not just medical geneticists, but multiple professionals involved in the patient's journey of care and genetic counseling Absence of integration between health professionals, ministries of health, and researchers Lack of definition of the teaching competencies/ areas of work needed by professionals who will work in genomics Difficulty in linking the training of professionals with retaining them in their country of origin	Conduct training for health professionals, taking advantage of online training capacity, but also adopting teaching and service models Promote the training of new professionals to generate a specific workforce focused on the field of genomics, involving its different fields of activity Encourage integrative and collaborative work between research professionals, health professionals, and ministries of health Encourage the training of professionals from the undergraduate level by including genomics more actively in the curricula Professional master's degrees as a suggestion to overcome the immediate bottlenecks of the lack of genetic counselors Map professions and then create a matrix of courses, both virtual and face to face, focused on practical training Define basic curricular competencies and stratify them in the short, medium, and long term Rethink the training model to adapt specializations as a solution to cover the gaps in the number of professionals found, not just focusing on one single profession Reflect on hybrid training models: theoretical training through distance learning combined with application in clinical practice (professional performance of internships in service) PAHO to facilitate those already working in the field of genomics and those with an interest in this area, facilitating the sharing of experiences or giving economic incentives Focus on training new professionals and not just on training the professionals who are already in care Invest in multiprofessional residency programs Use technology (online platforms) for training, with a view to targeting countries where access to practical training is lagging behind (but always try to make practical training possible in services when possible)

Challenge	Details of the challenge	Opportunities/solutions/strategies
Society's reluctance to adopt genomic interventions	Society's lack of knowledge about the benefits of genomics Reaching culturally, regionally, or ethnically isolated populations	Develop communication mechanisms that can teach genetics to the population Empower society to understand the impact of genomics on their health Hold public hearings to involve society in the dialogue surrounding genomics and public health issues
Lack of evidence of genomic value and returns on investment		Continued need for robust clinical studies demonstrating the efficacy of genomic testing in clinical practice (financial incentive is needed to conduct these studies) Demonstrate that genomic testing leads to better health outcomes, informing society and the scientific community.
Other, please specify	Lack of political will on the part of public managers Lack of literacy among public managers in relation to genomics Lack of recognition of the values of genomics	Adopt mechanisms that demonstrate the value of genomics for the country's health system (including monetary gains) so that public resources can be allocated to this field Conduct cost-effectiveness studies involving the field of genomics Present numerical information that demonstrates the benefits and savings in resources that the implementation of genomics can generate, for example, in the reduction of infant mortality due to congenital malformations Make available evidence products (health technology) that demonstrate that genomics can reduce the impact of inequities in public health, facilitating the incorporation of genomic technologies in health Have intergovernmental institutions such as WHO and PAHO as key players in raising awareness among politicians of the importance/prioritization of genomics on national agendas Use basic indicators to demonstrate the impact of genomics on the population to raise awareness among politicians Create expert groups for Latin American regions to help formulate strategies to raise awareness among governments Aim to create subworking groups according to the area of activity to engage society and politicians

GDPR, General Data Protection Law; LGPD, General Personal Data Protection Law; PAHO, Pan American Health Organization; WHO, World Health Organization.
Source: DECIT, Ministry of Health, Brazil.

Summary of the group discussions on Day 2

Challenge	Details of the challenge	Opportunities/solutions/strategies
Lack of or limited access to research participants or their samples of data	Inclusion of vulnerable/ underrepresented populations The need for practical feedback for research participants Underrepresentation of indigenous and Afro-descendant populations in genomic studies	Decentralize research from hospitals and work on the ground in communities Offer a report followed by genetic counseling for the patients in sequencing projects; for the control group, think of a dossier with the publications made or provide information on ancestry or participation in a discussion cycle on genomics and health
Lack of research capacity	Training and establishing human resources in the field of genomics Dialogue gap between professionals engaged in clinical research and those in the pharmaceutical industry and basic research	Professionalize scholarship holders Produce local scientists with local problems in mind Generate a system of networks of researchers
Difficulties in bioinformatics processing	This is a bottleneck in genomic research, especially in poorer regions Keeping specialized professionals because super-specialized professionals are quickly absorbed abroad. In addition, there is a financial problem, because scholarships are not adjusted in line with the market value There is a gap in human biotechnology in Brazil and other Latin American countries owing to the training model	Professionalize fellows in bioinformatics and related areas Take advantage of the network of postgraduate programs that already exists to expand the possibility of training young researchers from other countries Turn the scholarship into a job Expand employment opportunities beyond the basic production of inputs in Greentech Train professionals in bioinformatics among health professionals to broaden students' contact with strategic areas Create areas of focus at postgraduate level or even undergraduate models in bioinformatics, where the professional would have a broad training and specialize in a large area Increase the synergy between what is done in the research structure and the needs of the services. Managers need to be convinced that genetics can improve the chain of care in health policies
Lack of research agenda and policies	Issues related to genomics change rapidly and legislation does not always keep up with these changes Lack of institutional policy for the treatment of hereditary diseases in hospitals	
Ethical challenges	Lack of standardization in the analysis of how projects will be conducted	Build guidelines to evaluate genetics and genomics projects
Lack of research infrastructure and equipment	The need for quality biobanks Concentration of research and idle infrastructure in certain places	Quality management of biobanks, from sample collection to sample transfer A policy of sharing research infrastructure, all of which was installed with public funds

Challenge	Details of the challenge	Opportunities/solutions/strategies
Financial constraints on research funding	Financial restrictions on conducting research into rare diseases, including pediatric cancer	Strengthen researchers as a network and engage society in the advantages of having a strong science and technology system
	Unequal distribution of resources within the same country	Use PAHO mechanisms for funding support. PAHO has two mechanisms for supporting countries: the Revolving Fund and the Strategic Fund. In the case of the latter, equipment, supplies, and reagents are made available for countries to purchase
		Build collaborative networks, including between different countries
Lack of workforce capacity	In Latin America and around the world, there is a need to expand the training of specialists in genomics, especially bioinformatics, at master's and doctoral levels	Aggressive medium-term recovery policy to make postgraduate scholarships minimally attractive to postgraduate students
	Genetic counseling as a bottleneck for work in genomics and medical genetics	Disseminate programs and technical training in genetic counseling to increase the number of professionals who provide genetic counseling in the system without compromising the quality of the service

PAHO, Pan American Health Organization.
Source: DECIT, Ministry of Health, Brazil.

List of participants

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- Christine Carrington, Ministry of Health – Trinidad and Tobago
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- María Noel Cortinas, Ministry of Health – Uruguay
- Víctor Enrique Raggio Risso, Faculty of Medicine, University of the Republic – Uruguay
- Elena Ghanaim, National Institutes of Health (NIH) – United States
- John Tisdale, National Institutes of Health (NIH) – United States (virtual)
- Sheri Schully, National Institutes of Health (NIH) – United States (virtual)
- Erin Ramos, National Institutes of Health (NIH) – United States (virtual)

Pan American Health Organization team:

- Ludovic Reveiz, PAHO – Washington, D.C.
- Sasha Peiris, PAHO – Washington, D.C.
- Vania Canuto, PAHO – Washington, D.C.
- Carla Saenz, PAHO – Washington, D.C.
- Lionel Gresh, PAHO – Washington, D.C.
- Betânia Ferreira Leite, PAHO – Brazil
- Natália Veloso, PAHO – Brazil
- Giselle Calado, PAHO – Brazil
- Gilvânia Melo, PAHO – Brazil
- Cátia Ferreira, PAHO – Brazil
- Marina Negrisolo, PAHO – Brazil
- Maria Lucia Mesa, PAHO – Colombia

World Health Organization team:

- Anna Laura Ross, WHO – Headquarters
- Elena Ambrosino, WHO – Headquarters
- Deborah Mascalzoni, WHO – Headquarters (virtual)

Resources

- WHO Science Council workshop series "Accelerating access to genomic technologies for global health" – https://www.who.int/news-room/events/detail/2021/11/05/default-calendar/who-science-council-workshop-series-accelerating-access-to-genomic-technologies-for-global-health
- WHO Science Council report *Accelerating access to genomics for global health: promotion, implementation, collaboration, and ethical, legal, and social issues*– https://iris.who.int/handle/10665/359560
- WHO Technical Advisory Group on Genomics web page – https://www.who.int/groups/technical-advisory-group-on-genomics-(tag-g)#
- WHO genomics health topic web page – https://www.who.int/health-topics/genomics#tab=tab_1

The regional meeting Human Genomics for Health: Enhancing the Impact of Effective Research was convened in Brasília on 15–16 May 2024 by the Pan American Health Organization (PAHO) with the support of World Health Organization (WHO) headquarters, the PAHO/WHO country office in Brazil, and the Department of Science and Technology, Ministry of Health, Brazil. This consultation aimed to provide a forum for the exchange of information and ideas on human genomics for health and convened over 100 participants from a broad spectrum of disciplines who contributed their expertise across relevant fields of research, including science policy, bioethics, national genomic initiatives, cancer, rare diseases, and congenital disorders. The meeting objective was to identify, discuss, and propose concrete actions for the implementation of human genomics in practice and for conducting genomic research in Member States.

During the two-day meeting, this highly diverse group of experts shared experiences and best practices in applying genomics in clinical care and public health and conducting genomic research in the Americas, highlighting both successes and obstacles encountered in their work. Detailed discussions were held on the various challenges faced in the field, on the countries' priorities, and on the opportunities for advancement of genomics. There was a strong emphasis on the potential for collaborative efforts and the enhancement of partnerships within the Region of the Americas.

This final report summarizes the reflection and discussion of the issues and challenges identified during the workshop and presents the key actions and recommendations to enhance equitable and effective genomics research in the Region, outlining a path forward for the Americas. The report underscores the necessity for coordinated action at national, regional, and global levels to equitably harness the transformative potential of human genomics in clinical care and public health.

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