Global approaches to genomic medicine implementation

Since the sequencing of the human genome, the potential of genomic medicine has stimulated global investment not only in research but also in initiatives to drive genomic medicine implementation in health care. In 2014, European chief medical officers discussed the policy implications of genomic research advances for health systems and called for coordinated engagement to develop and harmonize policies within the European Union (EU) (1). In the same year, leaders in genomic medicine from more than 25 countries discussed the range of approaches for genomic medicine implementation and identified areas that would benefit from multinational collaboration (2). This group became the Global Genomic Medicine Collaborative (G2MC; https://g2mc.org/) in 2016 and recommended that implementation projects around the world should be identified and an inventory created of the policy issues being considered. To this end, the G2MC policy working group has used several strategies to elucidate the approaches that different countries are taking to move genomics from research into routine clinical care.

Between 2017 and 2020, 65 initiatives have been identified, and a catalog of these initiatives is publicly available at genomicspolicy.org. Each catalog entry provides information on the aims and policy issues being considered and, where appropriate, details of cohorts, diseases, and testing methodologies. For initiatives undertaking genome sequencing, the size of the cohorts ranges from a few hundred individuals to large national and international cohorts comprising 1 million to 5 million persons. These initiatives are using many types of testing, including copy number variation, microarrays, gene panels, whole-exome sequencing, whole-genome sequencing, RNA sequencing, and mitochondrial DNA sequencing. The range of disease areas is also broad, involving sequencing of adult and pediatric populations with rare, common, and infectious diseases and pharmacogenomics studies. Analysis of the geographic locations, types of initiatives, and policy interests raises a number of observations about the implementation of genomic medicine.

A review of the policy themes raised by these initiatives shows considerable overlap, allowing the themes to be clustered. The most common theme (60 initiatives) is understanding the contribution of genetics to disease and the technical improvements in sequencing capabilities and infrastructure. The next four most frequent themes are data and information management (including infrastructure, tools, and governance) (41 initiatives); workforce skills (35 initiatives); ethical, legal, and social implications of genomics (33 initiatives); and public education and awareness (30 initiatives). The policy themes that are least often considered are governance and leadership (18 initiatives) and regulation (17 initiatives). A small number of initiatives also include themes that have been collectively categorized as “other”—such as plans to promote private investment in genomics and the role of biorepositories (13 initiatives). The relative frequency of each theme is likely to reflect the perceived importance of that issue in each local context and the available expertise and sphere of influence of those involved.

In terms of the geographic diversity of these initiatives, 37 different locations around the world are represented, including initiatives in each of the six regions defined by the World Health Organization (http://www.who.int/about/regions/en/). When resource availability is considered, 71% of the initiatives (46 of 65) are in high-income countries, as defined by the World Bank measure based on gross national income per capita (https://datahelpdesk.worldbank.org/knowledgebase/articles/906519). In contrast, 17% (11 of 65) are in upper-middle-income countries, 5% (3 of 65) are in lower-middle-income countries, and there are no initiatives in individual low-income countries. There are, however, multinational initiatives such as the Human Heredity and Health in Africa (H3Africa) consortium (https://h3afrika.org/) and the GenomeAsia 100K consortium (https://genomesasia100k.org) that comprise countries with a mix of resource levels ranging from low to upper-middle income, and this group makes up 8% (5 of 65).

Initiatives are also categorized by the types of approaches adopted. For this analysis, there is an additional entry because one of the initiatives requested to be reported twice. Fifty-one percent (34 of 66) of the initiatives are demonstration projects, 29% (19 of 66) are policy/implementation frameworks, 14% (9 of 66) are population-specific genetic variation studies, and 6% (4 of 66) are road maps for full integration of genomic medicine into health services. This breakdown highlights that many jurisdictions are still considering the policy issues and using demonstration projects to trial various approaches for genomic medicine implementation into clinical care. Complete integration of genomic medicine into health services as a routine practice is currently very limited.

The large number of initiatives in the catalog confirms the high level of interest and investment in genomic
medicine globally, with the push toward clinical implementation being led by high-income countries. The small number of initiatives in low- and middle-income countries mirrors an earlier finding regarding the small number of laboratories in those countries registered in the National Institutes of Health (NIH) Genetic Testing Registry (3). In 2015, 603 laboratories were registered in the Genetic Testing Registry, yet none were in low-income countries and only 20 were found in middle-income countries. A recent search of this registry shows that little has changed, with no registered laboratories in low-income countries, 25 (4%) in lower-middle-income countries, and 35 (6%) in upper-middle-income countries; 518 (90%) registered laboratories were in high-income countries.

Although a large proportion of current investment is by high-income countries, the catalog does reveal mechanisms by which the capabilities and benefits of genomic medicine can be diffused to other resource settings. One mechanism is the formation of multinational initiatives involving a mix of resource-level countries as exemplified by the H3Africa consortium. This investment of $170 million by the U.S. NIH and the U.K.’s Wellcome Trust has created a consortium of over 500 members that facilitates fundamental research into diseases on the African continent. It also develops infrastructure, resources, training, and ethical guidelines to support sustainable genomics research in Africa. The initiative consists of 48 African projects that include population-based genomic studies of common noncommunicable disorders such as heart and kidney disease and communicable diseases such as tuberculosis (https://h3africa.org/index.php/consortium/projects-2-2/#1553507718507-14bc80d-0b08). This type of initiative has helped to ensure that African countries develop the capacity and infrastructure to apply genomic science for the benefit of Africans and global health (4).

The existence of global alliances is another mechanism that can facilitate access to technology and resources and hence support the dissemination of technical and policy learnings among countries. The G2MC and Global Alliance for Genomics and Health (GA4GH) (https://www.ga4gh.org/) are two examples. G2MC is a community of global leaders fostering global collaborations in genomic medicine implementation, and GA4GH is a policy-framing and technical standards-setting organization. These alliances provide a forum for organizations and individuals from all resource settings to be involved in the development and application of genomic medicine policies and best practices.

Different policy considerations and practical solutions are likely to be needed to address the challenges of implementing genomic medicine within individual countries. These approaches need to take into account factors such as local health system infrastructure and readiness, local disease burden, and relevant regulatory frameworks. However, the recurrence of policy themes across initiatives in different countries suggests that making existing information, including tools, resources, and evidence, more accessible could allow countries to see what has been tried elsewhere and consider applying these approaches locally. A good example is H3Africa’s promotion of best practices in biobanking using the internationally accepted guidelines of the International Society for Biological and Environmental Repositories (https://www.isber.org/page/BPR). Adoption of these guidelines has enabled three regional H3Africa biorepositories to be established to support H3Africa projects and has enabled new collaborations such as the Bridging Biobanking and Biomedical Research group (http://www.b3africa.org/). This group supports research across Europe and Africa and has facilitated activities such as testing of laboratory information systems and bioinformatics tool kits developed for resource-poor settings (5).

To fully realize the benefits of genomic medicine to patients and populations worldwide will require the engagement of a diverse community of stakeholders, including scientists, bioinformaticians, clinicians, health care providers, policy analysts, and representatives from industry, academia, and government. Genomic medicine implementation is complex and multifaceted, and no one group of experts is capable of providing a complete set of solutions in isolation. The number of initiatives under way to create national implementation strategies for genomic medicine is increasing, but many are being carried out in the absence of external collaboration and understanding of other nations’ successes and failures, risking duplication of effort and slowing the pace of discovery and translation. Sharing of best practices and policy agendas that foster implementation and accelerate translation remains a real opportunity for the global genomic medicine community. The G2MC catalog of genomic medicine initiatives discussed here demonstrates the ongoing need for global collaboration and communication as genomic technologies become a growing component of health care worldwide.

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REFERENCES AND NOTES


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