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Genomics, medicine and population health to 2050

Introduction

Twenty years ago, a group of international experts met in Bellagio, Italy, to discuss the emerging area of public health genomics: to promote its goals, share knowledge and resources, and to ensure equitable access to genome-based knowledge. In the intervening years genome-based technologies have developed to the extent that genomics has entered mainstream clinical care in many countries, improving the care of patients with cancer and rare diseases, and the management of infectious diseases.

Much has changed recently in terms of the wider context beyond healthcare, including geopolitical instability, climate change, demographic and socio-political changes, and the impact of disruptive technologies such as artificial intelligence (AI). Against this background, a multidisciplinary meeting was convened in September 2025 in Ayer's Cliff (Canada), to review developments during the last two decades, reflect on where we are now, and establish some of the key action points for genomics, medicine and population health through to 2050. (Meeting participants are listed in the Appendix.)

The vision underpinning the meeting was of improved quality and equity in health across populations through the use of biomarkers and 'genome-based' technologies, in combination with other factors, (1) to generate and refine practical knowledge and (2) to guide public health interventions. These interventions can support clinical management of conditions caused by a range of genetic and/or environmental factors. There was also a recognition that while much has been achieved, some previously identified challenges remain unaddressed and some ambitions unfulfilled, while new challenges and opportunities have arisen.

Key developments in knowledge

Since 2005, key developments in genome-based knowledge and the broader knowledge landscape have included:

- Evolution of genome-based technologies, in terms of speed, accuracy, scale, scope and reduced cost. The cost of generating a raw whole genome sequence has decreased by orders of magnitude since the completion of the Human Genome Project and, depending on the sequencing depth required, is now less than USD 1,000 per genome. Studies show that the majority of sequencing costs are for technical staff and genome data analysis and data management. Sequencing technologies operate at increasing speeds and throughputs, capable of generating more than 100 whole genome sequences in less than 48 hours.
- Different sequencing technologies have become established; for example long-read sequencing has enabled analysis of previously hard-to-sequence sections of the genome, providing further valuable insights into genetic contributions to disease risk. Portable long read technologies have revolutionised the accessibility of sequencing in more challenging environments and situations, for example real time sequencing to support genetic epidemiology during disease outbreaks.
- A shift from data scarcity to data abundance has followed from the increased speed, depth and breadth of sequencing technology. This has not only increased the volume and complexity of genome data analysis, but also requirements for computational biology expertise, data storage, infrastructure and management.
- Novel molecular technologies, in particular CRISPR-Cas9 genome editing, have accelerated the speed at which some research questions can be addressed. The development of gene-edited therapies arising from this research, and the premature use of germline editing, has stress-tested the regulatory and ethical frameworks that underpin use of these technologies, with the speed of scientific development outpacing regulation.
- Rapid developments in artificial intelligence (AI) and advanced computation have generated significant promise for new insights on the causes of disease at population and individual levels, responses to therapy and population health decision making. No less important is the potential automation of significant aspects of human decision making and potential efficiency gains for health systems. However, this coincides with significant hype around potential benefits and the challenge for adopters of unpicking exaggerated claims from genuine potential.
- Technological advances have also generated new and increasingly complex considerations around utility and trustworthiness. The scrutability of AI and other sophisticated computational approaches can be challenging both in terms of understanding their scope and potential health utility, and in providing clear foundations for trust and confidence on the part of publics and professionals. The challenge of securing trust and confidence is compounded by the important role played by the private sector in these ecosystems and mixed public perceptions of their involvement.

The context

Political

The global political landscape

There are profound geopolitical shifts occurring globally with an increase in populism and transactional politics, and a trend towards nationalism, isolationism and de-globalisation. Many of these shifts are arising due to concerns around sovereignty and national security, which are having an impact on the global health landscape. These impacts include reduction or withdrawal of funding and closure of international health programmes, and the deprioritisation of health compared to other issues such as security. There seems little short term prospect of reversing the decline from the post-1945 international rules-based order, suggesting a need to develop solutions that acknowledge and engage with the new international order.

These changes are having profound impacts on the access and sharing of data. Some national or regional legal regimes are increasingly seen as restricting or discouraging data sharing and present challenges in linking datasets on a global scale.

Political responses to the impacts of climate change are also indirectly impacting health, particularly in terms of migration patterns and population demographics, which are changing globally.

Role of the state vs. non-state actors

In many countries there are changes underway in terms of the role and contributions of state actors in the health space. As a result, non-state actors such as private companies are filling the gap, generating and mining large quantities of health-related data. In many cases it is unclear what the accountability of non-state actors is in terms of the information they are generating and how it is used. The decreased involvement of the state also raises questions around ongoing investment for research and translation: where it is going to come from, and the ongoing impacts for health ecosystems.

Funding vs. time

The short-term nature of funding programmes linked to political cycles is an inherent issue in terms of establishing long-term, stable clinical research programmes and infrastructure. Combined with a failure to invest in translation as well as research, the long-term sustainability of initiatives is a challenge and there is a risk of missing the benefits that can be delivered by having access to sustainable, well curated and supported data and other health infrastructure.

Regulation and governance

Regulation and governance have a crucial role to play in this context to secure public trust and confidence while supporting the implementation of desirable measures for which a sound evidence base exists. There are multiple – and overlapping – areas of relevance, including data protection, privacy, emerging AI-regulation, biosecurity, public health law, medical device regulation, research governance and professional oversight. However, challenges arise across this landscape, particularly in relation to data access and sharing.

Although the present era is one of data abundance, a lack of standardisation and harmonisation of policy and governance surrounding data is hindering progress. A universal challenge is safeguarding an appropriate level of privacy while facilitating data access and use for health or research purposes. This is compounded by the often risk-averse tendency of data custodians and lawyers advising on compliance with local regulations. Fragmentation of the regulatory and governance framework is also a difficulty. The governance of component features (data, devices, professionals etc.) contributing to technologically supported population health approaches is not well coordinated.

There are some clear good examples of effective governance in this space. These include the efforts of the American College of Medical Genetics & Genomics (ACMG) to develop authoritative recommendations, which stimulate activity in key areas, for example in relation to secondary findings, and of the Centres for Disease Control and Prevention (CDC) Office of Public Health Genomics (OPHG) in evaluating the evidence for genomic applications to identify those (Tier 1, Table 1) with significant potential for positive impact on public health. However, there has also been a hollowing out of some of the institutions of governance in many settings (including the OPHG).

Table 1. CDC evidence-based classifications of genomic applications.

Tier 1	Supported by a base of synthesised evidence for implementation in practice	e.g., newborn screening, hereditary breast and ovarian cancer syndrome, familial hypocholesterolemia, Lynch Sydrome
Tier 2	Synthesised evidence is insufficient to support routine implementation in practice; may provide information for informed decision making	e.g., many pharmacogenomic tests
Tier 3	Evidence-based recommendations against use, or no relevant synthesised evidence identified; not ready for routine implementation in practice	e.g., direct-to-consumer personal genomic tests

*Dotson WD et al, Clin Pharmacol Ther. 2014 Apr; 95(4): 394–402.

Where regulation and governance work well they tend to be characterised by a combination of harder and softer measures, with more nimble, adaptable and interdisciplinary approaches. For example, the UK's Human Fertilisation & Embryology Authority (HFEA) has developed a successful model, working with the commercial sector as well as engaging widely with other stakeholders, adapting to scientific and societal developments and using a number of different instruments (e.g. licensing regime and code of practice) to achieve its aims. However, this model arose at a particular time in the social and political context of the United Kingdom, and may not be readily translatable to other settings.

Social

Population demographics

The global population is changing. In most of the world, societies are aging, which changes the social and cultural context of innovation, and also healthcare needs and demands. Areas of the world with younger populations at present, particularly in sub-Saharan Africa, are following this aging trajectory. Alongside this there is an increasing focus on mental health among younger generations. The impact of climate change on populations can be profound: not only can it directly impact people's health and fertility, but the wider impacts of climate change affect populations through migration.

Impact of the COVID-19 pandemic

The COVID-19 pandemic had a profound impact on societies globally, influencing peoples' relationships with each other and with the state. For some, the pandemic lockdowns were associated with a restriction in civil liberties and weakened their trust in the state. These effects, and other challenges such as misinformation and political responses to the pandemic have significantly strained aspects of social solidarity, which is needed to support population level health measures. The continually emerging scientific evidence meant that the public became much more aware of the extent of uncertainty in science in that case. This has complicated trust between citizens, scientists and politicians, and challenges the maintenance of political support for interventions, despite uncertainty being a fundamental feature of developing scientific knowledge.

There were positive impacts as a result of the pandemic. Overall, populations were exposed to knowledge about genomics and its inherent value in supporting vaccine development and managing the pandemic. There were also some positive changes in how research could be carried out to meet the needs of the global health emergency. Lessons might be drawn from this in terms of streamlining research and removing barriers to data sharing that could be modified and applied to non-pandemic research and clinical studies, though some were made possible only owing to the exceptional conditions created by the emergency.

Individual and population values

In some societies (particularly those in the West) an ongoing shift away from solidarity and towards individualism has challenged assumptions about shared values that underpin existing frameworks and created uncertainty about what societal values will hold sway in the future.

There is a need to establish values that support public health and embed these values into tools as they are built, so that they can serve the populations for whom they have been developed. Establishing an anchor in societal values will also support governments' efforts to tackle wicked problems – complex and often intractable issues – which often cannot be solved solely on the basis of evidence.

Technological

Hype around novel technologies

The hype around novel technologies can be a double-edged sword. For example, on the one hand a technology such as CRISPR-Cas9 can quickly create fundamental changes in research and medicine; on the other hand, premature and ethically unsound translation of technologies without adequate demonstration of validity can significantly impact trustworthiness.

It is likely that by 2050 there will be at least three more technologies that are as transformative as CRISPR-Cas9; the challenge lies in anticipating what these might be and how people and systems will respond to them.

While developments in the field of AI are having a significant impact in some areas, it is not clear whether it will prove to be as transformative as initially assumed in all areas. There can also be a gap in terms of interventions: while novel technologies can provide different information, their value will depend on the ability of the health system to respond and use that information.

Artificial intelligence

Artificial intelligence is a broad term used to describe a range of technologies and interventions, and it is therefore important to define clearly what is meant by AI in any particular context. These definitions matter since they have an impact on the regulatory landscape governing use of AI.

Significant uses of AI in healthcare include: data modelling and management in public health genomics; delivery of the healthcare pathway for example supporting decision making, taking notes and managing patient records and data simulations.

There are still many questions around the evidence requirements for AI and how to determine what is 'good enough', and whether the tools support the health care system in ways that are necessary or desirable. Given its rapidly evolving nature, AI development challenges the requirements for evidence and evaluation demanded by conventional regulatory approaches, and demands more adaptive governance mechanisms.

How the context is impacting genomic medicine

Social: Information engagement and understanding

There are ongoing efforts to increase public and patient engagement in healthcare decision making and research more broadly. This engagement, however, is not always straightforward and can be difficult to do well. Challenges include varying levels of wider genomic literacy, misunderstandings around genomics and disease risk, and misplaced expectations about the implications of genomics for healthcare.

In healthcare settings, there is still limited understanding of behavioural science and the potential of behavioural interventions, in terms of understanding how people behave when given information about their health, and how health systems can or should support behaviour change.

There is also a challenge around managing perceptions of genomic data. Genomic exceptionalism can have an impact by placing more demanding restrictions on access to genomic data than other kinds of health data. Public mistrust of institutions, public services and commercial actors contribute to a difficult landscape for health systems and researchers to navigate in terms of engagement.

Social: Why genomic medicine - evidence and obligations

Screening is one area of genomic medicine where there is much ongoing discussion about potential utility and the offer to citizens. A key question is clarity about the purpose of the screening and to whom it should be offered. Due to lack of data there is currently uncertainty regarding the wider impact of many proposed genomic screening programmes, and there is still much debate about how to measure the success of screening, testing or other interventions. There are also differences in how states may deliver their screening programmes, for example whether directly offered to individuals or relying on individuals to engage with the state or the health system. Differences in choice architecture create different ethical considerations around the screening programme. A challenge underlying these considerations is a perceived lack of resolution and capacity on the part of public authorities to deliver targeted disease prevention as a priority.

Another challenge is the difference between studying interventions, such as a new screening programme, in the research setting and evaluating novel services. Research allows a more detailed examination of the evidence, and more time per case, while the translation to clinical practice requires a streamlining of the process, designing it in such a way that it does not perpetuate health inequalities.

The introduction of any new genetic intervention, be it screening or more targeted testing, requires a balance to be struck between the drive to detect and treat disease early and thereby to reduce later diagnostic odysseys, and the risk of current false positives. In addressing this, there are conceptual challenges around how to value objective possibility versus the ex ante and ex post knowability of harms that may arise following testing.

In the light of the global public health success of newborn screening programmes, the use of whole genome sequencing in healthy newborns as a screening tool is currently being explored in a number of projects globally. A key question is whether the ambition to offer whole genome sequencing for all babies at birth is desirable for this, or other, purposes. Expansion of newborn sequencing also requires exploration of questions about claims and obligations relating to screening in the light of evolving technologies – of the state to children and parents, and of the 'right of the affected child to be found'. There are also questions of accountability for using and making available data that participants have consented to be shared.

Social: Engagement and equity

There are well-known issues to do with the lack of diversity in genomic datasets, which have a Euro-centric bias that is unrepresentative of the global population. While many efforts are underway to improve the diversity of datasets, this will take time. It is vital that these efforts are accelerated, since relevance of the dataset to the population has a big impact on the trustworthiness and the clinical utility of the data. Co-creation of datasets offers an approach that can not only support citizen engagement but also improve trust in the process of data collection. Engagement efforts around genomics can also benefit from making use of indigenous knowledge, perspectives and ways of thinking about data, recognising that members of many indigenous communities may want to be included in wider national resources.

Novel technologies, including AI, raise equity challenges in terms of accessibility and engagement. While there is a general public enthusiasm around new tools more generally, and some citizens will be eager to use them, others will not. Therefore the potential of technology will not be realised by every user. The question arises of how to harness existing enthusiasm but also support and engage with those who cannot or do not want to access new technology. Many novel technologies such as AI are developing rapidly, and individuals and society might not be comfortable with the pace of change, or able to adapt.

Technological: Which technology?

Which technology is most appropriate to use in genomic medicine is – or, at least, should be – driven by public value considerations such as evidence of effectiveness, cost, how invasive it is for the patient and the potential balance of harms and benefits. Potentially serviceable technologies include karyotyping, arrays, single- or multi-gene tests, exome or genome sequencing. Other factors influencing choice can also be driven by which individuals or populations would benefit most from testing. For example, offering genetic testing at a population level in a screening context may require use of cheaper and quicker technologies (e.g. single or small panel gene tests) in the interests of time and cost, while patients with rare disease on a diagnostic odyssey will benefit from thorough and in-depth whole genome sequencing.

A further influence on technology choice is around what data are required (and technology choice may then lock in the nature of further data requirements). While technology can be chosen based on the information it gives relevant to the current clinical situation, much of the drive to use whole genome sequencing has been due to demand for data in areas beyond the original clinical purpose, for example research.

Technological: How we collect, use and manage data

The data landscape is complex. It is widely acknowledged that we now exist in a time of data abundance. Despite this, there are gaps in available genomic data, for example genomic databases contain information mostly from individuals with European ancestry. Population and sub-population understanding will improve via better data collection and measurement; however, gathering these data requires careful planning and engagement with the relevant populations. Varied data sources complement the use of genomics and provide useful context to support clinical management; in many cases genomic sequencing plays the role of a supportive technology. Health-relevant data can also be collected via a range of means, such as wearables, with multimodal datasets increasingly becoming the norm.

When genetic tests are carried out, and particularly in the case of whole genome sequencing, there are questions around how these data are stored and used in the future beyond the original purpose for collection, and who can access the data both for the current purpose and in the future. A question that many are grappling with is which data to discard and which to retain, providing some future proofing while not storing unnecessary data.

Changing geopolitical contexts are having a significant impact on the data landscape. Vested interests can lead to poor utilisation of data and stifle innovation, for example if data are linked to commercial interests, or there are other political or cultural reasons to restrict sharing. There is also a need to consider how to defend public health from consumerism, where individuals are directed towards private companies who have their own agendas for wanting to collect data.

The availability of data and ease of generation is not consistent across the globe, and increasing nationalism, and concerns about privacy and security are limiting practical collaboration and data sharing. In different jurisdictions restrictions, laws and policies are contributing to a decrease in active sharing and data movement. Differences in interpretation of legislation, for example the EU's GDPR, can stymie sharing.

While secure data environments are seen as a solution, for example meeting cybersecurity and national security concerns, there are still barriers to be overcome in terms of granting access to domestic and international users. These barriers can be technical, regulatory or related to access cost. In turn, data linkage between different datasets becomes more challenging. This will have an impact on data standardisation and raises questions around how to ensure that data meet the findable, accessible, interoperable and reusable (FAIR) principles; for complex datasets, metrics will need to be agreed.

A final data consideration is around sustainability, not only the environmental impact of data storage (water and power availability, physical infrastructure), but also in terms of maintenance and curation of the datasets, who is responsible for this, and how this is funded.

Technological: The nature of evidence

The evidence on the links between genetics and disease risk is changing and diversifying. The low hanging fruit in terms of understanding genetics and risk has been collected, in the form of monogenic impacts on disease risk.

Knowledge of polygenic impacts on disease risk will alter our understanding but is unlikely to change the landscape profoundly. Significant changes could occur around the science of risk intervention, since it was acknowledged that in many cases the failure to demonstrate clinical utility of some genetic tests, such as polygenic scores, can be attributed to a lack of specific interventions. On the other hand, studies have found that most individuals have actionable pharmacogenomic results, albeit that these differ between populations.

There is also a need to question what type of evidence is needed to demonstrate clinical utility of genomics and how that evidence is collected. While pilot projects are useful, they do not always scale successfully for widespread use. The need for high quality evidence needs to be balanced against more efficient ways to gather that evidence. There is also the challenge of using genomics in isolation (whether, in an otherwise healthy patient, a genetic finding is sufficient on its own to provide a diagnosis), against considering the broader picture of phenotypic findings and other data sources.

Technological: Professional skills

The speed of development of genome-based technologies and their implementation into clinical systems has left many of these systems and healthcare professionals with a skills gap in terms of genetic knowledge and competency. This raises a number of questions around how to boost capacity within certain professional groups, such as genetic counsellors, and who might play a role in transmitting genetic information. In many countries, and particularly in LMICs, there are few genetic counsellors, so upskilling a range of clinical practitioners is necessary to support engagement with patients around genomic information. Other skills deficits are around bioinformatics, data stewardship, and the technical and security skills to develop data infrastructure environments.

Where do we go from here?

The value of preventing disease and preserving health rather than treating sickness is increasingly well recognised. The use of scientific knowledge and biomedical technologies to support this in a targeted way has great potential, yet its value remains to be proven. The discussion identified approaches that could facilitate prevention, for example:

- Behavioural science, particularly behavioural psychology
- Pharmacological interventions, informed by pharmacogenomics to prevent harm caused by drugs
- Environmental measures, including conventional public health interventions

However, how these could be made more effective by the use of genomic testing was heavily dependent on a range of contingent factors that may be specific to the technical characteristics of the test or to the context, including the social and political context, in which testing is implemented. To meet the challenges outlined in this report we can consider the following responses.

Deglobalisation and isolationism

Despite an increasing trend towards deglobalisation, multilateralism is still relevant and declarations and accords can still have impact. An example is the pandemic agreement adopted by the World Health Assembly.

International partnerships are still beneficial, particularly when focussed on areas with less visibility. Where reluctant states are unwilling to participate under international legal frameworks, ad hoc agreements and bespoke instruments can be easier to facilitate. Any agreements put in place may be strengthened by reporting obligations, with a legal basis, to hold states to account.

This kind of international cooperation can promote multidisciplinary working and also support an international coordination of standards, governance and institutions, to guard against divergence caused by competition between national and regional interests.

Solving wicked problems will involve coordination and cooperation to reconcile evidence and values-based approaches that work on a global scale. The question of which organisations can and should provide leadership in terms of responding to big global challenges remains unresolved.

Lack of knowledge around genomics

Engagement with and knowledge of genomics, genomic medicine and population health needs to take place among citizens, policy/decision makers and healthcare providers. Engagement of citizens and publics, building on the many global programmes already underway, will support efforts to improve understanding of health technologies among the general population.

Decision makers also need to be supported by those with expertise, who can provide the information they need for properly informed decision making. Developing educational and informational practices, tools and resources should ensure that knowledge, expertise and capacity are available at the right place and right time in the health system, and strengthen its ability to adapt to external events.

Access to genomic medicine

Every citizen has a right to benefit from research and from the application of this research to medicine. There is a need to recognise, however, current inequities relating to population health and to promote equitable access to the benefits and applications of scientific progress. Advocacy is required for those groups who may be left behind or whose needs are not being met by social and technological change.

When states are considering implementing population level programmes, such as genomic-based screening, they need to consider the balance of harms and benefits of implementation. The extent to which others should be required to carry costs in order to identify a person who can benefit from an intervention requires acceptable forms of collective decision making. More broadly, there are decisions to be made about how far the duties of the state, and the rights of individuals, extend when it comes to implementation of healthcare interventions.

Finally, states need to consider the consequences of using genomic medicine, and explore the potential of behavioural sciences and the use of digital technologies to support targeted behaviour change.

Uneven global accessibility to genomic technologies

To improve accessibility to genomic technologies in LMICs, support for technology transfer and lowering of barriers to access such as costs of equipment and consumables is needed. Technology choice is an important consideration given that what works in one setting will not necessarily work in another.

A clearer focus on what is needed and can be delivered, rather than enthusiasm for the most advanced technology, will lead to better solutions for LMICs. In particular, presumptions in favour of a ‘WGS first’ approach need to be challenged.

Balance is required in terms of the clinical need against the desire to generate a data resource for research and development. In some cases, focussing on the clinical need at the expense of research needs will be more beneficial.

International collaboration to set a coordinated research agenda that benefits the next generation of researchers and professionals in all countries will contribute to lowering barriers for accessing genomic technology.

Future proofing and building sustainable systems

Climate change is having a growing impact on healthcare, not only on the health of populations but also in terms of sustainability considerations for healthcare technologies, for example resource use, and water/electricity use for data centres.

Sustainability must be central to future planning of healthcare research and innovation. It can also be considered through a different lens in terms of future proofing and planning, allocating resources and funding where most long-term support is needed, for example data infrastructures.

There can also be benefits to sharing and recycling knowledge, policy and infrastructure. Understanding what worked well in the COVID-19 pandemic, without normalising exceptional measures, will help to streamline some processes and minimise duplicated effort.

On a wider scale, interdisciplinary collaboration will be essential to help solve wicked problems where issues impacting population health intersect with other policy priorities, for example housing, the food system, environment, and transport. There will be a need for visionary thinking to draw these areas together and develop coherent and innovative approaches to tackling these grand challenges that impact the health of populations.

Complex data and technology landscape

Along with continued support for current and future initiatives to improve data diversity – not just in terms of diverse populations, but also different modalities of data relevant to health – developing processes and a culture of innovation around health data collection can promote further advances through learning health systems.

Investment in organisations that work on data standards will promote the development of these standards and standardised platforms for the handling of data, which in turn fosters collaboration and sharing of data. Consideration of the data landscape needs to address data harmonisation and interoperable technology, with compatible policy supporting its use. This includes developing clarity on what the FAIR principles mean in relation to different types of health data.

More nimble approaches are needed to keep pace with the rapid evolution of AI. Mapping the steps needed to produce an AI product – design, development and field testing – against the steps needed to use it wold be an invaluable help: audit, quality control, and improvement of the product. At each stage questions around data quality and policy governing that step should be considered.

Fragmented regulation and governance

There is a need for anticipatory and adaptive governance throughout the lifecycle of technologies, which incorporates ongoing learning and foresight to prepare for developing challenges and opportunities. This includes guarding against bias, mis/disinformation, biosecurity risk (including dual use) and ensuring sustainability. Regulation should be as technology agnostic as possible in order to accommodate new developments but good governance requires proportionality to purpose and risk.

For example, consent approaches should provide a proportionate response to the risks involved and likewise privacy and data protection laws should be developed and interpreted in a risk-proportionate way. Governance should be used to facilitate overcoming the 'second gap in translation' (the delay or failure to adopt evidence-based guidelines and technologies in real-world health care) by acting as a normative instrument to produce alignment between different interests.

A multi- or trans-disciplinary approach is required to ensure regulation and governance are developed and applied with a strong grounding in the different technical, ethical, societal and professional considerations at stake.

Participatory and distributive models of governance are also likely to be required in an increasingly decentralised context, spreading responsibility among public and private actors and at different levels of activity. Empowering citizens to contribute to these processes may help address some of the issues around loss of trust and confidence in both public health policy making and regulation that have become visible following the COVID-19 pandemic.

Conclusions

The meeting in Ayer's Cliff in 2025 marks not so much the completion of an arc began twenty years ago, but a new departure for the field of population health genomics into a more complex and less certain world. At the same time, the field is being boosted in new ways by the exponent of information technologies and artificial intelligence.

While the overall vision of improving health quality and equity through genome-based knowledge remains valid and tangible, it is a more nuanced and sober vision, one to be achieved less by overarching ambition and more through finding connections, building collaboration and achieving consistency among the various contexts and uses of genome-based technologies.

The next generation of leaders must articulate a plausible and desirable vision for the future of population health that responds to these challenges, rather than allowing technology alone to drive the agenda.

Health in the new global context

Genomics cannot be treated in isolation either from the abundance of other forms of biological knowledge or from the broader sociopolitical conditions, including the global context. Genomic medicine can have a significant impact on population health but it also intersects with – and may even contribute to – 'wicked problems' such as climate change, migration, and economic instability. With the foundering of the post-1945 liberal, rules-based order in the present decade, which participants agreed was now ineluctable, there is a real danger of widening health inequalities among nations as well as within some societies.

> Health is a global public good and a critical component of global stability, security and prosperity. Withholding access to essential knowledge and available technologies should never be used as a means to achieve economic or geopolitical advantage. Meeting global health challenges therefore requires continued cooperation among international networks and the preservation of appropriate incentives for research, innovation and sharing of knowledge and health technologies (including protections for intellectual property).

Information engineering as the golden thread

The golden thread that must run through advances in population genomics is data or, rather, information: the translation of data into contexts in which it acquires meaning and delivers practical benefit. Among the most significant shifts of the last quarter century has been the transition from an era of data scarcity to one of data abundance.

The challenge for 2050 is no longer merely that of generating data but unlocking, standardising, harmonising and utilising those data effectively. If this can be achieved ethically and securely it can power dynamic and 'learning' systems continually to generate new knowledge and insight at the same time as delivering care to populations.

These systems will be increasingly underpinned by emerging forms of computational intelligence that have the power progressively to dissolve boundaries between domains of practice, in particular between research and health care, and between the historical preoccupations of public health and clinical medicine, and abridge timelines for innovation.

Such developments challenge the adaptability of frameworks of governance and raise questions of privacy and the control of data, ranging from issues of personal autonomy and the privacy of individuals, families and groups to the protection of commercial and national strategic interests.

> Anticipatory reflection and governance will be critical to managing rapid developments, especially in AI. A continual reflection on the objectives and potential consequences of these developments, including through participatory measures that empower citizens, will be essential to avoid corrosive effects of public mistrust of companies, institutions and public authorities. This must feed into adaptive governance that goes beyond technical regulation and is required to preserve the governing role of human values in an increasingly transactional world.

Ethics and values

Everyone has the right to benefit from scientific advances. Developments in knowledge and technology enlarge our capabilities to meet the challenges of disease and to improve the health of populations. Changes in the scope of what can be achieved as a result of scientific and technological progress can also affect the relations among people and alter the limits and content of moral claims and obligations. For example, the availability of effective and affordable screening may create expectations that public authorities will implement programmes to identify cases where action can be taken to prevent or manage the onset of disease.

Such innovations involve complex relations and affect the interests of multiple actors. Defining a policy or judging the appropriate action in any particular case will depend on many factors, both inherent and circumstantial, ranging from the preferences of the individuals involved to the affordability of interventions and the availability of alternatives.

Some of these factors, such as test performance, are largely technical and can be informed by evidence, but many other factors are evaluative, and depend on judgements, for example about the level of evidence deemed sufficient to support a particular course of action in the circumstances, what courses of action are acceptable to a given population, or what justification can be given for the distribution of scarce resources in favour of a new approach.

> What is required of health systems and public authorities is to match scientific advances with effective social and political technologies, including adaptive governance and participatory citizen engagement, that are adequate to the pace and complexity of innovation.

Just as the expanding horizon of technology enlarges the scope of our moral obligations to members of our communities, it also extends the scope of our moral obligations to other populations, and other nations. Since the circumstances of peoples and their nationally determined priorities may vary, appropriate technology choice should be supported and respected. This may mean that the pace of technology implementation and the nature of technologies implemented will vary according to the prevailing interests and capacities in different settings.

> What is required at an international level is to maintain engagement and the potential for exchange of scientific knowledge, technological capability and ethical governance; the facilitation of connections among international groups, of which the group convened at Ayer's Cliff is an example, can create a renewed overture in this respect, on which future leaders may wish to build.

Implementation

While the Ayer's Cliff meeting has focussed on looking forward to the next 25 years and the discoveries and inventions that may lie ahead, the inherent forward thrust of research should not detract from the implementation of the many existing applications from which benefit may be derived and for which there may already be emerging evidence of utility. Finding cases of monogenic susceptibility to common complex diseases in populations at risk ('Tier 1' applications), genetic newborn screening and pharmacogenomic tests are examples.

There may be many contingent reasons why effective and useful innovations are not adopted and widely diffused, ranging from poor definition of the target population and lack of evaluation to unresponsive or inappropriate governance. On the other hand there is a great deal of knowledge generation that lacks any realistic expectation of producing health benefits.

> There should be a focus on achieving implementation and evaluation of approaches that have demonstrated the potential for population health benefit, which should be implemented with regard to their effect on environmental and economic sustainability, and on the development of useful knowledge.

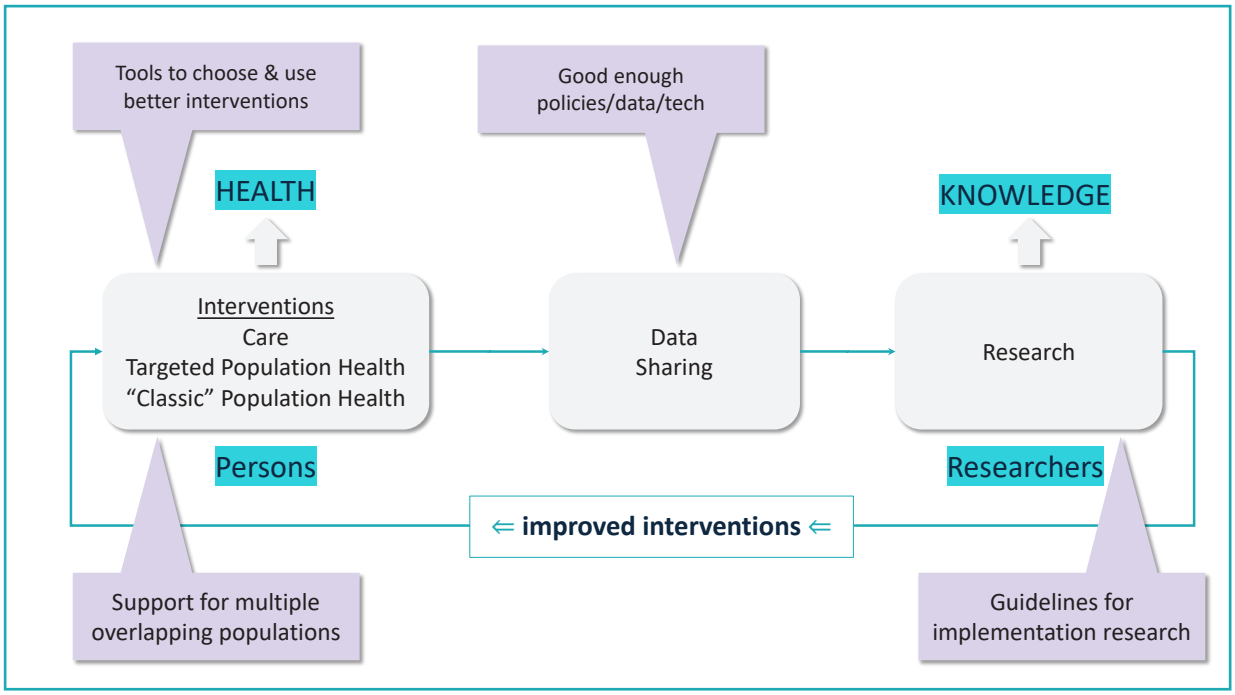
From knowledge integration to ecosystem governance

When the field of population health genomics was in its infancy, the Bellagio meeting focussed on integrating the disciplinary perspectives and contributions that were necessary for the success of the enterprise it defined as 'the responsible and effective translation of genome-based knowledge and technologies for the benefit of population health'. It acknowledged the importance of integrating both the science and the art of preventing disease, prolonging life, and promoting health, bringing technical advances into the domain of human relations.

After two decades, this public health perspective remains important but, with substantial advances in research and technological development in the intervening period, and with the establishment of new institutions and professions, there now needs to be a much greater focus on how health and research systems function as part of a concrete ecosystem to integrate knowledge into health care practice, in the context of society. This may be described as a 'learning population health service' that continually uses real world data to refine and guide the deployment of a range of available interventions. A version of this model that was discussed at the meeting is reproduced below.

> The focus for the future of genomics and population health, should be on creating dynamic, learning systems, integrating the continual cycle of generation and concretisation of knowledge in technologies, professions and institutions, and on how this is facilitated and governed by standards, protocols and codes of conduct, and anticipated and adapted through reflective practices involving ethics, regulatory science, law and democratic processes.

Figure 1. Learning population health system



Appendix – meeting participants

- Habiba Al Safar, Professor and Dean of College of Medicine and Health Sciences Medicine, Khalifa University (United Arab Emirates)
- Jeffrey Barrett, Chief Scientific Officer, Nightingale Health (Finland)
- Laura Blackburn, Head of Science, PHG Foundation (United Kingdom)
- Brian Chung, Chief Medical and Scientific Officer, Hong Kong Genome Institute (Hong Kong)
- Martina Cornel, Professor of Community Genetics & Public Health Genomics, Amsterdam University Medical Center (The Netherlands)
- Edward Dove, Professor of Law, Maynooth University (Ireland)
- David Glazer, Chief Technology Officer, Verily Life Sciences (United States of America)
- Calvin Ho, Associate Professor of Law, Monash University (Australia)
- Yann Joly, Director, Centre of Genomics and Policy, McGill University (Canada)
- Muin Khoury,* Former Director, CDC Office of Public Health Genomics (United States of America)
- Bartha Knoppers, Professor Emerita, Law and Medicine, McGill University (Canada)
- Anna Lewis, Research Scientist and Bioethicist, Harvard Medical School (United States of America)
- Katherine Littler, Co-Unit Head, Health Ethics & Governance Unit, World Health Organization (Switzerland)
- Anneke Lucassen, Director of the Centre for Personalised Medicine, University of Oxford (United Kingdom)
- Eric Meslin, Adjunct Professor, Dalla Lana School of Public Health, University of Toronto and Visiting Scholar, CGP, McGill University (Canada)
- Pete Mills, Director, PHG Foundation (United Kingdom)
- Colin Mitchell, Head of Humanities, PHG Foundation (United Kingdom)
- Nicola Mulder, Professor and Head of Computational Biology, University of Cape Town (South Africa)
- Ainsley Newson, Professor of Bioethics, University of Sydney (Australia)
- Anthony Ng, Trustee, WYNG Foundation (Hong Kong)
- Paul Pharoah, Professor of Cancer Epidemiology, Cedars-Sinai Medical Center (United States of America)

- Vasiliki Rahimzadeh, Assistant Professor, Center for Medical Ethics and Health Policy, Baylor College of Medicine (United States of America)
- Megan Roberts, Assistant Professor, Pharmaceutical Outcomes & Policy, UNC Chapel Hill (United States of America)
- Saskia Sanderson, Data Science Lead, NIHR MH-TRC Mental Health Mission (United Kingdom)
- Jeff Skopek, Professor of Law, University of Cambridge (United Kingdom)
- Ma'n Zawati, Associate Professor & Research Director, Centre of Genomics and Policy, McGill University (Canada)
- Ron Zimmern, Chair of Trustees, PHG Foundation (United Kingdom)

* Dr Khoury joined part of the meeting by videolink.

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