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# Adopting Public Health Genomics when the House Is on Fire: How Will We Navigate to 2030?

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## Introduction

At its heart, public health genomics considers how to exploit new knowledge being harvested by genomic discoveries for the benefit of the population's health [1]. Its goal is to improve health outcomes by integrating genome-based knowledge and technologies into public health practice – as well as informing, and being informed by, public health research [2]. It is useful to assess briefly the current state-of-play before embarking on the current contribution we intend to make (Box 1, sources: Khoury et al. [3]; Allen et al. [4]; Pitini et al. [5]; Stark et al. [6]; Roberts et al. [7]).

Against the backdrop of the current state-of-play in Box 1, implementation science [8, 9] can play a decisive role in advancing the public's health translating genomic information and research findings into practical, actionable public health interventions [10]. This Journal's call for *active implementation in translational genomics* takes us to a challenging space: attempting to accelerate genomic advancements for populations against the headwinds of current problems experienced by on-the-ground clinical practitioners, health policymakers, and entire health systems. Our contribution explores these key issues from a specific standpoint: how do we get to 2030, providing a better health system and more equitable, genomics-available, population-based care, given all the challenges we face today? These challenges include insufficient resources, shortages of suitably trained staff, poor digital health systems, impediments in the availability and functionality of medical products and technologies, deficits in governance structures and leadership, burnout, concerns about quality and safety, widening rather than narrowing inequities, a mismatch between system demands and service supply, poor political decision-making, ethical issues, and structural and cultural barriers to good care [8, 11–13], to name only some of the most prominent.

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**Box 1.** Brief Outline of the Current Landscape of Public Health Genomics

Public health genomics is supporting the transformation of healthcare across various fields and life stages. It is already delivering tangible public health benefits in areas such as newborn screening, hereditary cancers, rare diseases, pharmacogenetics, and infectious diseases. Specifically, there are:

- Significant advancements in bioinformatics tools for genomics data storage and analysis;
- Standards for genomics data sharing and integration;
- Decreases in sequencing costs; and
- Marked progress in data science present many further opportunities to implement public health genomics.

Challenges to progress include:

- An insufficient genomics workforce;
- Significant disparities in the supply of genomics services;
- Lack of public and healthcare providers' awareness; and
- Ethical implications of genomic medicine, including privacy, informed consent, genetic discrimination, data sharing, and inequity in access.

**Box 2.** Selected Examples of Improved Outcomes from Public Health Genomics

**Infection Control**

Since microorganisms have a genome, variants, and strains can be usefully identified from genomic testing of pathogens. This has allowed the tailoring of treatments in resistant strains of tuberculosis and accurate contact tracing in disease outbreaks including COVID-19 [14].

**Rare Disease Diagnosis**

Of the roughly 7,000 identified rare diseases listed on *Orphanet*, around 72% are genetic in origin [15]. Many more have an unknown genetic aetiology. Genomics is proving a powerful tool for identifying these rare diseases and providing more targeted management for many (e.g., in drug-resistant epilepsy) [16].

**Reproductive Genetic Carrier Screening**

Carriers of recessive conditions are often unaware of the fact. Screening identifies the chance of a couple having a child affected by one of these potentially debilitating, but hidden, conditions. In Australia, screening of 12,000 couples for cystic fibrosis, spinal muscular dystrophy and fragile X identified 610 carriers, 88% of whom had no previous family history [17–19].

**Cancer Treatments and Surveillance**

The Zero Childhood Cancer program is based on genomic testing of aggressive, atypical cancers in children to find tailored treatments. An estimated 71% of children enrolled have received a personalised treatment plan. Implications for childhood cancer are being disseminated globally [20].

## Background to Public Health Genomics

While we have opened with the manifold and daunting problems that health systems currently face, we want to frame our quest for a better health system, and the role of public health genomics in supporting it, in a positive way. We argue that harnessing genomic data and associated technologies to inform public health policies and practices, with the ultimate aim of preventing disease and improving health outcomes, is an obviously desirable pursuit and has made many gains including improved health outcomes (for four selected examples, see Box 2).

To give further encouragement, the world over the last 10 years has witnessed the rapid evolution of genomic technologies, including high-throughput DNA sequencing, which has expanded understanding of the genetic bases of diseases. To apply such knowledge to entire populations, public health genomics clearly requires a multifaceted approach. Indeed, public health genomics is centrally concerned with merging insights from many endeavours such as those discussed in Box 2 – especially genetics, but also epidemiology, bioinformatics, clinical practice, and health policy – to improve the health status of populations.

To get to 2030 with genomics playing a key role in securing better population health, however, we need to have a game plan for success, not just a hope that we will do well. Implementation science is the construct we rely on in our own work, but we often integrate it with a complexity science perspective [8, 9, 21, 22]. Our glossary of terms is in Table 1.

Many experts agree that implementation science methods can promote the systematic uptake of research findings and evidence-based practices into routine care in order to improve the quality and effectiveness of health services [9]. Implementation science attempts to address this "know-do" gap; that is, the gap between what is known from research and what is provided in practice. This field utilises various theoretical models and tools, such as the Consolidated Framework for Implementation Research (CFIR) [23], the Theoretical Domains Framework (TDF) [24], and the Reach, Effectiveness, Adoption, Implementation, and Maintenance (RE-AIM) framework [25] to understand and guide implementation activities. Figure 1 shows some of the fundamental implementation science elements; embedded in these constructs are approaches and mechanisms for taking an evidence-informed approach [26].

The Rapport et al. [26] model (shown in Fig. 1), along with Reach, Sustainability and Equity, encapsulates many of the core elements constituting implementation science.

**Table 1.** Glossary of complexity and implementation science terms

| Glossary of terms | |
|---|---|
| Complex adaptive system | A dynamic, self-similar collectivity of interacting, adaptive agents and their artefacts |
| Complexity | The behaviour embedded in highly composite systems or models of systems with large numbers of interacting components (e.g., agents, artefacts and groups); their ongoing, repeated interactions create local rules and rich, collective behaviours |
| Consolidated Framework for Implementation Research (CFIR) | A framework of five constructs that aims to describe the barriers and facilitators to an effective implementation strategy |
| Implementation science | The processes of translating research into practice, understanding what influences translational outcomes, and evaluating the adoption of interventions |
| Public Health Genomics | Incorporating genomic-based knowledge and technologies into public health for greater understanding and identification of novel and emerging threats |
| Reach, Effectiveness, Adoption, Implementation, and Maintenance (RE-AIM) framework | Five dimensions; reach, efficacy, adoption, implementation and maintenance are used to assess public health interventions at various levels; used to evaluate the impact of public health programs or policies |
| System dynamics | An analytical modelling methodology used for problem solving, which combines qualitative and quantitative data and identifies the fundamental elements of a system, and how they influence one another over time |
| Theoretical Domains Framework (TDF) | Identifies and utilises theories from implementation science to group constructs into clusters; used to stimulate theoretical advancements in implementation science |

Sources: Baker et al. [1]; Braithwaite et al. [8]; Damschroder et al. [23]; Atkins et al. [24]; Glasglow et al. [25].

Yet even with good genomics data alongside effective application of implementation science methods over an extended timescale, we know we will struggle to adopt genomics uniformly across populations in developed economies, let alone in low- and middle-income settings. Core to public health genomics is equity: the equitable access to genomics tests and data, and hence targeted treatment and precision medicine, is a constant concern. Cost is a formidable barrier, even for those with means. Poor genomic health literacy amongst policymakers, patients, and clinicians is another.

We argue that adopting genomics universally when there are so many problems and headwinds in contemporary health systems (our metaphor is the healthcare house is on fire) exacerbates this challenge into a wicked problem. To unravel some of the complexity of adopting genomics widely, we turn to a more detailed consideration of how progress might be made. In the succeeding sections we reflect on implementation, implementation science, and active implementation in genomics, and present case examples factoring in the complexity of the pursuit to illustrate our argument.

## Leveraging Implementation Science

Like public health genomics, implementation science in healthcare is a multifaceted field. As we have alluded to in our introductory remarks, it seeks to theorise, model, and pragmatically improve the uptake of evidence-based practices in healthcare settings [8]. A fundamental aim was to engage with the complex and dynamic nature of healthcare systems, acknowledging the multiple contextual factors that can influence the acceptance of innovation and change [27].

Researchers working in implementation science attempt to understand the enablers and barriers to the introduction, integration, and sustainability of local and system-based interventions and policies. They identify and account for the perspectives and insights of various

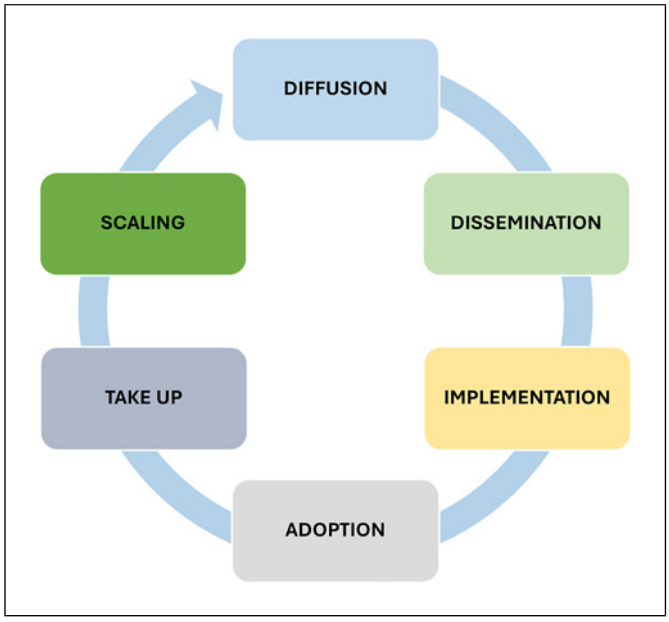


**Fig. 1.** Core elements in Implementation Science. Source: from Rapport et al. [26].

stakeholders, including healthcare providers, administrators, and patients. They design iterative strategies for the implementation of new practices, allowing for ongoing refinement and adaptation based on feedback. In this way, implementation science emphasises the importance of collaboration between researchers and practitioners, fostering a bidirectional flow of information and expertise in real-world settings [26].

As we have seen, multiple theoretical frameworks can inform the work of implementation scientists in healthcare. Many in the field factor in complexity theory which acknowledges that real-world systems are intricate, variable, and characterised by non-linear interactions and constantly emerging elements. Viewed through this lens, healthcare contexts can be understood as complex adaptive systems that can facilitate or resist new practices and policies [28–30]. Factors such as the maturity of a health context, resistance to change, cultural characteristics, and fuzzy boundaries that deter dissemination and scaling of what works across settings are central foci of implementation research. This is especially so when implementation science moves from theorising and developing frameworks to take a real-world perspective, seeking to get evidence into practice among deeply entangled systems complexity.

Implementation scientists also hold great store in being theoretically well grounded. Theories of diffusion [31], for example, which explain the dynamics of innovation-uptake over time, often guide implementation scientists in modelling the optimal spread of information, ideas, and technology through the interconnected formal and informal channels, highways, and byways of healthcare systems. The implementation science frameworks we presented above, alongside others, can provide a robust foundation for navigating the wicked problem of implementing genomics and genetics into already deeply challenged healthcare systems, at least in principle. But even with effective implementation science expertise and knowledge, implementation will always be a formidable task, in large part because of the multifaceted nature of health systems. In essence, then, implementation scientists have a core aim: to learn through research about what works, for whom, in what contexts, why and at what cost, in order to inform replication, sustainment, and scaling of evidence-based practice, with the ultimate goal of improving population health.

## The Need for Active Research in Translational Genomics

In our opening to this paper, we touched on the genomics revolution. The last 10 years has seen an explosion of applied genomic research. While this research commitment has continued in vivo and in vitro, its application in real-world settings, testing its effects on human health outcomes, has also been the focus of attention by governments, health services, and consumer advocacy agencies [32]. Research fields in clinical genomics have included its use in pharmacogenomics, somatic and germline testing of tumours in cancer, diagnosis of rare disease, and reproductive carrier screening. Research can establish the pathogenicity of a variant, then assess the clinical utility or cost effectiveness of identifying that variant through a genomic test.

Research has in addition been applied to a raft of key questions about how best to configure services to allow effective and efficient genetic testing to occur, and for the data to be used wisely. Questions arise at every stage of the testing pathway. How can clinicians be assisted to identify patients for whom a test is relevant? How much information do patients need to understand the ramifications of a test and manage expectations? How can the gigabytes of data produced by tests be curated in a timely and efficient manner? How can potentially life-changing results be appropriately shared with patients in a thoughtful way that is mindful of the familial implications for many genetic conditions? Further to these research questions are ethical, legal, insurance, and social issues, as well as topics such as workforce availability and training, data

security, service infrastructure, ongoing quality and safety, and how to sustainably and equitably fund genetic testing and advice.

The workforce training implications have a long way to go, and there are calls for hands-on learning about practical genomics application rather than formal classroom style education [33]. This necessitates a systems-level approach with new policies, curricula, and a reconfiguration of some services. As the evidence-base for what works is assembled, there is a growing need for active implementation studies to more fully realise the gains to human health that genomics technology offers.

Funding bodies can provide dedicated resources, personnel with quarantined time for research work and governance and protocols to scaffold both research endeavours and the practical application of genomics information. However, the initial burst of block research funding is coming to an end on many national genomics projects across the world, and teams must turn to more specific purpose research or ad hoc funding measures to overcome the issues created by a lack of staff, infrastructure, or funding [34].

## Strategies for Active Implementation in Genomics

We have seen that research has made substantial contributions to the advancement of genomics, but it has fallen short in addressing the issue of its widespread application. Pivoting to a robust implementation science focus, what is needed to disseminate genomics to entire populations and translate findings across populations? Key strategies in the adoption of genomic findings for the benefit of the public are being realised in several priority areas, notwithstanding that there is considerable progress yet to be made. At the patient level, initiatives include engaging community members to participate in clinical genomic projects, involving patients in implementation activities, and empowering patients to be active participants in their genomic-informed care [35, 36]. At the clinician level, initiatives include improving clinician knowledge, skills, and beliefs about clinical genomics, increasing the availability of a specialist genetics workforce, and providing the evidence base for advanced technologies, interdisciplinary, and collaborative care designs [37, 38]. At the organisational and system-level, initiatives include incentives for healthcare practitioners to screen for genetic diseases, ensuring equity in accessing clinical genomics (and especially for those living in rural and remote areas across culturally and linguistically diverse groups, and those in vulnerable populations), prioritising the integration of genomics information into electronic health records, developing effective, tailored policies, using data warehousing techniques to extract and integrate data from clinical records across organisations into a central repository, and investment in whole-of-system strategies such as Learning Healthcare Systems approaches to facilitate wide-scale education and knowledge translation at a local level [37–39].

The goal of widespread use and integration of genomic information into practice across public health settings is being advanced through organisations such as the Global Genomic Medicine Consortium. A greater focus is needed, however, on knowledge exchange and collaboration – on optimal ways to share strategies, data, and standards to foster collaborative identification of best practices using many experiences across diverse settings [36]. National genomic initiatives such as Australian Genomics [40] and Genomics England [41] are playing a vital role in presenting a unified voice to inform future policy making and service planning, yet there is more to be done [32]. There is also a need, only partly met to date, for international collaboration and communication to leverage the lessons learned on effective and sustainable implementation of genomic medicine, especially for some universal issues in genomics, such as the availability and use of comprehensive and relevant genomic reference databases [37]. Additionally, implementing genetic interventions requires the reshaping of traditional practices and organisational cultures, much like the shift in medicine from a biomedical "doctor-in-charge" model to a biopsychosocial model where shared decision-making between patients and clinicians is now best practice.

### *Studying General Practice and Consumer Involvement in Public Health Genomics*

Several of the authorship team contributed to Mackenzie's Mission [18, 42], developing a reproductive carrier screen for cystic fibrosis, fragile X, and spinal muscular atrophy. These were added to the tests that are Medicare funded (the Australian reimbursement mechanism for Government-approved services at no or low cost) in late 2023, and are available for General Practitioners (GPs) to order. Yet it remains the case that many GPs in Australia and elsewhere are inexperienced in genomic screening. At least three separate pinch-points in addition to price barriers are threatening to derail the consistent and appropriate uptake of public health genomics. Firstly, there is pressure from consumers as they become more genetically aware and request tests that may or may not be appropriate for them. Secondly, there is a bewildering array of tests now available as more and more pathology

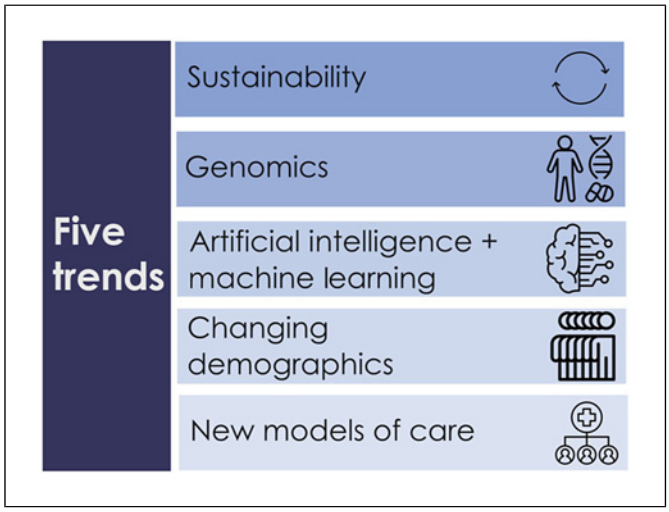


**Fig. 2.** Five trends in health systems to 2030.

service providers become accredited to provide them. Thirdly, ethical issues arise in genomic screening and testing that GPs can find distressing. All these pressures affect the ability of GPs to use genomics and integrate genomics information into their workflows and patient consultations.

A recently initiated Australian project is working to bring consistency, feasibility, and appropriateness to this area: Embedding genomics in primary care: using implementation science to design a robust national approach (MRFF2025125). Its aims are: (1) to provide high quality resources to the GPs offering tests including education modules, guidance for managing ethical issues, links to a test directory, and a real time advice service from genetic counsellors; (2) resources for consumers considering the tests, including a decision support questionnaire around whether to undergo reproductive carrier screening; (3) a health economic assessment of the value of these tests in the primary health space. All these issues need to be addressed to continue to make progress in the mainstreaming of public health genomics in primary care.

*The Narrowcast Solution: Overcoming Localised Barriers to Implementation*

Notwithstanding these exemplary gains, sweeping healthcare innovation across entire systems or populations is challenging to achieve. Effective implementation is often driven by narrowcast solutions that home in on specific contextual barriers to change [43, 44]. For example, lack of expertise and skills in genomics consistently emerges from the literature as a barrier to mainstream genomic integration [45], particularly for clinicians whose roles are not specific to the field. Multimodal education and training programs and real-time information delivery at point of patient care have been shown to enhance clinician confidence and competence and facilitate higher referral rates to genetic services [46]. Tailoring these solutions to clinicians who do not specialise in genomics may require the provision of less complex, user-friendly information about genomics, and specific guidance on the relevance, value, costs and ethical and legal implications of genetic testing for particular patient populations. Educating people requires time, however, and investments.

Predictably, competency in "doing" genomics [47] is confounded by the variability and limitations of digital resources and information technology (IT) infrastructures across different healthcare contexts. Challenges relate to the compatibility of genomic and non-genomic patient data in electronic health records, the structure, and reliability of genomic information in clinical decision-making support tools, and issues with integrating genetic test ordering into IT systems given differing laboratory formats. These barriers can be overcome through collaborations between genetic and non-genetic healthcare providers, the employment of informatics specialists to optimise workflows and the development of standardised protocols and IT policies. That, too, takes time, and adequate funding allocations.

Leadership resourcefulness is a determinant of successful diffusion, dissemination, and implementation of innovation [9, 21, 26]. Effective context-framed strategies for genomics integration should leverage the expertise of clinical leaders with IT project management experience who are aptly positioned to facilitate technological solutions [48]. This approach would complement high-level institutional endorsement and advocacy from implementation champions and early adopters working within and across formal and informal networks and multidisciplinary channels.

We can learn more about what works for whom, in what respects, to what extent, in what contexts, and how [49, 50] through long-term partnerships between implementation scientists and genomics stakeholders. Through these, tailored implementation efforts can then be further evaluated for their sustainability and their ability to be implemented at scale. Outside of targeted implementation and evaluation, when effective narrowcast solutions – specific initiatives for a designated cohort or setting – are persuasive and well-adopted in local contexts, their success can create a ripple effect across health systems. Here, positive, case-specific processes and outcomes gradually proliferate through the broader genomics ecosystem, eventually indirectly creating large-scale change [51].

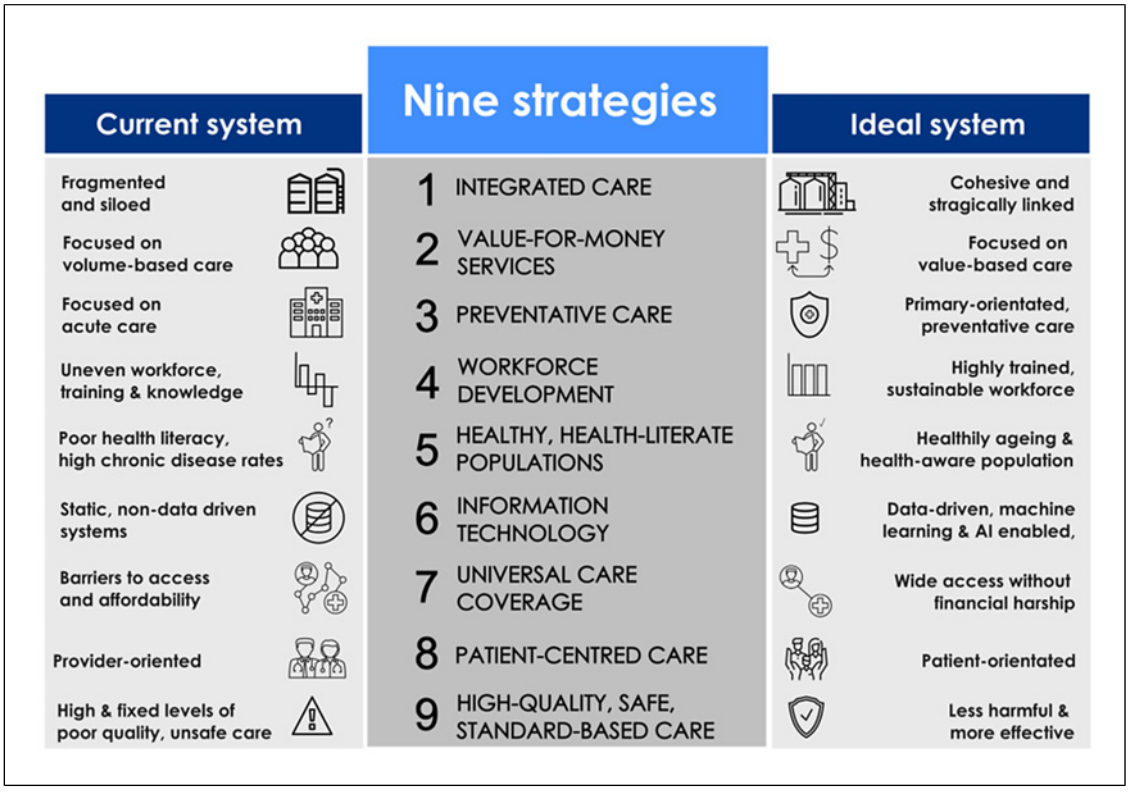


**Fig. 3.** Nine key strategies for creating better care by 2030.

*The Bigger, Transformational Solution: The Future of Healthcare to 2030*

Small-scale, bottom-up, acorns-to-oak trees change [52] such as our narrowcast examples represents one approach. But top-down strategies orchestrated, funded, and authorised from the Health Minister or government agency down into the system are also needed for transformation at scale [50, 53]. Large-scale burdens of chronic disease, fiscal constraints, disruptive new technologies, and threats of future pandemics and climate change are global drivers for change. While healthcare at the systems level learns to adapt to disruptions and key change initiatives – the rapid move to telehealth during COVID-19 is a good recent example – the solutions are often reactive and hampered by existing policies, budgets, and infrastructure which often act to constrain rather than enable change. We need an evidence-based roadmap for whole-of-system responses to future challenges to go hand-in-hand with narrowcast, localised solutions that are being implemented.

In 2018, 148 global policymakers, scholars, and experts from 152 countries came together to create the compendium *Healthcare Systems: Future Predictions for Global Care* [13]. When the fifty-seven chapters were analysed, five key, research-based trends that are shaping healthcare in the current decade emerged [54] (shown in Fig. 2).

The collective contributions of the book's editors and authors, brought together in Figure 2, make the sustained case that healthcare systems are increasingly prioritising sustainability through smarter system design, technological integration, and cross-disciplinary workforce models [13]. Genomic technologies are being taken up into routine care, albeit sporadically, with a workforce that is increasingly prepared to manage genomics risks, ethical challenges, and privacy issues [13]. Changing population demographics and disease profiles require health systems to cope with an increase in both extant chronic and emerging infectious diseases [13]. Advances in artificial intelligence and machine learning are being assessed, implemented, exploited, and belatedly governed. Central to all of these are new models of care; these are being made more adaptable to current and future pressures on health systems, as the COVID-19 pandemic showed – with adoption of telehealth and telemedicine, for example [13, 55].

The wisdom across the 536 pages of *Healthcare Systems: Future Predictions for Global Care* was analysed further. Nine fundamental strategies that are enabling healthcare systems to proactively meet the demands of these five key trends were able to be synthesised (the 5 + 9 [trends+strategies] model) [13, 56] (Fig. 3).

The book mounts the argument (Fig. 3) that health systems across the world are endeavouring to integrate services and sectors (strategy 1), fund care quality over care quantity (2), improve population health literacy (3), and prioritise the principles of patient-centred care (4). In addition, many health systems are seeking to expand preventative care (5) and develop the workforce to provide it (9). Health systems are increasingly data-driven (6). Many are striving to deliver care sustainably, and, in line with the World Health Organization's (WHO's) recommendations for universal coverage (7), to leave no-one behind who needs care (8). Finally, to be effective, health systems will need best-practice policies and standards that underpin all these strategies (9). This, of course, is an idealised picture, but one grounded in case studies across, or relevant to, the health systems of 152 countries. Each health system is, to a greater or lesser extent, working on a range of these strategies to create a better system for their citizenry. How well they do, what progress they make by 2030, and how they equitably incorporate public health genomics into the improvement equation represented by the 5 + 9 [trends + strategies] model is an open question.

We suggest, however, that planning and evaluating progress against this blueprint will help strengthen health system performance and patient outcomes, and help promote a more proactive approach, rather than being reactive to the demands every health system is facing over the rest of the decade. This 5 + 9 [trends + strategies] model is now widely available and increasingly used [54, 57–60]. And there is also a point to be made here around better alignment between health systems and universities to facilitate access to the data, the workforce, and the patients that are needed to keep pace with the science that needs to be provided to underpin the model.

## Case Studies

All that being said, we can point to progress. There are lessons to be found for public health genomics in the acute care space, e.g., from the successful implementation of ultra-rapid genomic sequencing for acutely unwell babies and children (Case 1). Genomic testing may make all the difference for these young patients who are critically unwell with a suspected genetic condition, potentially providing a diagnosis and supporting clinical management. In such instances, though, testing needs to be rapid to be useful, and available across-the-board. We can also draw lessons from a national genomic carrier screening program (Case 2).

### Case 1: Rapid Testing and Scaling to the Population

Ultra-rapid exome testing (median 3.3 days from receiving sample) was achieved by the Australian Genomics Health Alliance Acute Care Flagship [61], with 51% of patients, recruited from across Australia, receiving a molecular diagnosis, and many results influencing clinical management. Scaling this sort of intervention across a national health system has required robust clinical and laboratory pathways; considerable funding; and a participatory culture of collective learning and collaboration alongside a model of distributed leadership. The Flagship also engaged the expertise of implementation scientists [62] who supported the work, for example, by mapping barriers, enablers, and implementation strategies to the Consolidated Framework for Implementation Research [23].

### Case 2: The Australian Reproductive Genetic Carrier Screening Project (Mackenzie's Mission)

Another example is the successful implementation of the Australian Reproductive Genetic Carrier Screening (RGCS) Project, Mackenzie's Mission[1] [19]. This project provided prospective parents with information about their chance of having children with severe autosomal recessive and X-linked genetic conditions. Recruitment of couples into the program was via primary care physicians, many of whom had little experience of genetic screening. Research involved a staged approach to the implementation of Mackenzie's Mission, including pre-implementation (study design and development), followed by piloting and early take-up, before moving to nationwide implementation, assessing effectiveness and outcomes [18]. Barriers and enablers to implementation were identified early in the pilot and implementation phases of the study, with interventions being developed to overcome identified barriers [63]. Implementation science and behaviour change theories, including the theories embedded in the Consolidated Framework for Implementation Research [42] and the Theoretical Domains Framework [64], were used to study the implementation of the RGCS project. A total of 10,038

(Continued on following page)

[1] A large program of research demonstrating that reproductive carrier screening mediated by primary care physicians was feasible, acceptable, cost effective, and clinically useful at a population level.

(continued)

reproductive couples enrolled in the nationwide study; 90.7% completing screening; 175 couples (1.9%) were identified through the study with an increased chance of having children with a screened condition. The study demonstrated that the RGCS program was widely acceptable to healthcare professionals offering the screening. The approach was also feasible to deliver to Australia's geographically dispersed and diverse population and provides a model for population-based reproductive genetic carrier screening [18, 19, 65].

## Future Directions

If we take the 5 + 9 [trends + strategies] model for 2030 and map it to the problems facing extant health systems, we can begin to discern a pathway forward, and we can mobilise strategies and tactics in support of a better health system. If the current healthcare house – including public health – is indeed burning because of resource pressures, staffing shortages, clinician burnout, and issues such as low morale, quality and safety problems and poor genomics health literacy, at least we now have a target at which to aim. Table 2 summarises some of what we have suggested as a way forward.

What remains, of course, is the manoeuvring needed to make progress across rest of the current decade, and in the context of this article, to settle on understanding where genomics fits as a solution amongst the other 5 + 9 [trends + strategies]. For example, although genomics is one of the five trends running through modern health systems, it is not clear where the money will come from to pay for its availability across whole populations. The usual range of arguments to bolster public health funding include that it is an investment

**Table 2.** Six proposed solutions to challenges in adopting public health genomics

| Challenge | Solution |
|---|---|
| Lack of clinicians' expertise and skills in genomics | • Multimodal education and training programs<br>• Real-time information delivery at point of patient care<br>• Provision of less complex, user-friendly information about genomics<br>• Specific guidance on the relevance, value, costs and ethical and legal implications of genetic testing for particular patient populations |
| The variability and limitations of digital resources and information technology infrastructures across different healthcare contexts | • Collaborations between genetic and non-genetic healthcare providers<br>• The employment of informatics specialists to optimise workflows<br>• Development of standardised protocols and information technology policies |
| Need for leadership resourcefulness | • Leverage the expertise of clinical leaders with information technology project management experience who are positioned to facilitate technological solutions<br>• Providing high-level institutional endorsement<br>• Advocacy from implementation champions and early adopters working within and across formal and informal networks and multidisciplinary channels |
| Sustainability and scalability of genomics implementation | • Long-term partnerships between implementation scientists and genomics stakeholders |
| Need for large scale transformation and changes of healthcare | • Top-down strategies orchestrated, funded, and authorised from the Health Minister or government agency down into the system |
| Challenges made by existing policies, budgets, and infrastructure | • Evidence-based roadmap for whole-of-system progress alongside narrowcast, localised solutions |

Sources: Rapport et al. [9]; Best et al. [21]; Rapport et al. [26]; Schiabley et al. [46]; Ingebrigtsen et al. [48]; Pawson, [49]; Francis-Auton et al. [50]; Braithwaite et al. [53].

in the population's health rather than a cost, and therefore a desirable public good. Proponents of genomics can advance what we think are persuasive arguments about where the money can come from: for example, through increased taxes, greater contributions to health insurance premiums, co-pays and individuals with means paying more, taxing harmful products and behaviours such as sugar, smoking and vaping, alcohol consumption and fossil fuels, optimising high value rather than low value care, creating systems-level efficiencies, and, more broadly, reallocating funding from other government programs or reorienting societal priorities.

Of course, in a disputatious world, each of these funding measures will be heartily supported by some sections of society and vigorously opposed by others. Yet we cannot, even if we achieve additional funding for public health genomics at suitable levels, escape the wicked problem that does not seem to go away: expensive initiatives such as genomics are more likely to be available to those with capacity to pay. This will exacerbate existing inequities. Precision medicine and the gains we documented in our Box 2, and the case 1 and 2 examples will remain a benefit accessible only by the few without remedial action by governments and their healthcare policy and governance bodies responsible for system performance and population wellbeing, and finding ways to pay for public health genomics. While each health system has a different configuration of problems it faces, the 5 + 9 [trends + strategies] model in Figures 2 and 3 at least clarifies where attention should be, and in many settings is being, placed for a better healthcare system to 2030.

The case will need to be made for the population benefits of precision medicine by advocates for the equitable access to genomics tests and genomics-based care. Implementation science, supporting the translation of genomics data in actionable interventions and outcomes for the public, will need to be harnessed in the ways we have suggested. Table 2 summarises some of the core points we have made about what should be done and the place of the implementation science models presented earlier. And, as we have argued here, both top-down, orchestrated change, and bottom-up, narrowcast strategies will need to work in concert to achieve desired and desirable gains [66, 67].

Notwithstanding what we have put forward, there are other challenges we have touched on but others in this Special Issue will deal with in depth. Everyone who supports public health genomics knows there are ethical issues around genomics including consent, privacy and confidentiality, sharing of information, the options and decisions for policymakers, providers and patients that flow from genomics data, challenges for insurers and the insured, and risks of discrimination. These need considerable attention going forward. Patient and provider literacy about genomics and its application, too, as we have seen, is in its infancy. And the risk that the technical-scientific progress we are making in genomics continues to outstrip the capacity of the system to absorb it is omnipresent.

## Conclusion

However, we believe that if we can make the journey to 2030, accelerate efforts along the lines of the five trends and optimise progress on the nine strategies, and spread the benefits of genomics widely, applying these to whole populations, society has much to gain. On this glimpse into the future, the healthcare landscape can thrive, akin to the Australian bush's famous, almost miraculous capacity for rejuvenation in the days and weeks following a bushfire.

A gleaming multi-roomed mansion beckons, built to accommodate flourishing new services, strategies, and genomics communities, in place of the burning house metaphor with which we opened this chapter. Can we achieve this? That is one of the key questions of the genomics era. We are watchful, and, as researchers, striving to play our part.

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## Conflict of Interest Statement

The authors have no conflicts of interest to declare in relation to the work reflected in this manuscript.

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## Author Contributions

J.B. conceived the project and led the work. J.B. drafted the manuscript with input from all authors. S.S., K.L., M.V., S.S., G.F., L.P., L.A.E., K.C., and J.C.L. drafted sections of the manuscript, and J.B., R.P., N.T., S.B., and J.C.L. conducted desktop research and critically reviewed and edited the final manuscript. All authors take responsibility for the accuracy and presentation of the manuscript. The corresponding author attests that all listed authors meet authorship criteria and that no others meeting the criteria have been omitted.

## References


1 Baker KS, Jauneikaite E, Hopkins KL, Lo SW, Sánchez-Busó L, Getino M, et al. Genomics for public health and international surveillance of antimicrobial resistance. Lancet Microbe. 2023;4(12):e1047–55. https://doi.org/10.1016/S2666-5247(23)00283-5

2 Lane CR, Sherry NL, Porter AF, Duchene S, Horan K, Andersson P, et al. Genomics-informed responses in the elimination of COVID-19 in Victoria, Australia: an observational, genomic epidemiological study. Lancet Public Health. 2021;6(8):e547–56. https://doi.org/10.1016/S2468-2667(21)00133-X

3 Khoury MJ, Bowen MS, Clyne M, Dotson WD, Gwinn ML, Green RF, et al. From public health genomics to precision public health: a 20-year journey. Genet Med. 2018;20(6):574–82. https://doi.org/10.1038/gim.2017.211

4 Allen CG, Green RF, Bowen S, Dotson WD, Yu W, Khoury MJ. Challenges and opportunities for communication about the role of genomics in public health. Public Health Genomics. 2021;24(1–2):67–74. https://doi.org/10.1159/000512485

5 Pitini E, Baccolini V, Isonne C, Maran P, Marzuillo C, Villari P, et al. Public health genomics research in Italy: an overview of ongoing projects. Front Public Health. 2024;12:1343509. https://doi.org/10.3389/fpubh.2024.1343509

6 Stark Z, Boughtwood T, Haas M, Braithwaite J, Gaff CL, Goranitis I, et al. Australian genomics: outcomes of a 5-year national program to accelerate the integration of genomics in healthcare. Am J Hum Genet. 2023;110(3):419–26. https://doi.org/10.1016/j.ajhg.2023.01.018

7 Roberts JS, Dolinoy D, Tarini B. Emerging issues in public health genomics. Annu Rev Genomics Hum Genet. 2014;15:461–80. https://doi.org/10.1146/annurev-genom-090413-025514

8 Braithwaite J, Churruca K, Long JC, Ellis LA, Herkes J. When complexity science meets implementation science: a theoretical and empirical analysis of systems change. BMC Med. 2018;16(1):63. https://doi.org/10.1186/s12916-018-1057-z

9 Rapport F, Clay-Williams R, Braithwaite J. Implementation science: the key concepts. London: Routledge; 2022.

10 Taylor N, McKay S, Long JC, Gaff C, North K, Braithwaite J, et al. Aligning intuition and theory: a novel approach to identifying the determinants of behaviours necessary to support implementation of evidence into practice. Implement Sci. 2023;18(1):29. https://doi.org/10.1186/s13012-023-01284-1

11 Braithwaite J, Mannion R, Matsuyama Y, Shekelle P, Whittaker S, Al-Adawi S. Health systems improvement across the globe: success stories from 60 countries. Boca Raton, Florida: CRC Press; 2017.

12 Braithwaite J, Matsuyama Y, Mannion R, Johnson J. Healthcare reform quality and safety: perspectives, participants, partnerships and prospects in 30 countries. Farnham, Surrey: Ashgate Publishing Ltd; 2017.

13 Braithwaite J, Mannion R, Matsuyama Y, Shekelle PG, Whittaker S, Al-Adwai S. Healthcare systems: future predictions for global care. Boca Raton, Florida: CRC Press, Taylor & Francis Group; 2018.

14 Morales-Arce AY, Sabin SJ, Stone AC, Jensen JD. The population genomics of within-host Mycobacterium tuberculosis. Heredity. 2021;126(1):1–9. https://doi.org/10.1038/s41437-020-00377-7

15 Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2020;28(2):165–73. https://doi.org/10.1038/s41431-019-0508-0

16 Perucca P, Scheffer IE, Harvey AS, James PA, Lunke S, Thorne N, et al. Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Res. 2017;131:1–8. https://doi.org/10.1016/j.eplepsyres.2017.02.001

17 Archibald AD, Smith MJ, Burgess T, Scarff KL, Elliott J, Hunt CE, et al. Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. Genet Med. 2018;20(5):513–23. https://doi.org/10.1038/gim.2017.134

18 Archibald AD, McClaren BJ, Caruana J, Tutty E, King EA, Halliday JL, et al. The Australian reproductive genetic carrier screening project (Mackenzie's Mission): design and implementation. J Pers Med. 2022;12(11):1781. https://doi.org/10.3390/jpm12111781

19 Mackenzie's Mission. Providing couples with information and choices for their family planning. [Internet] [accesed 2024 14 March]. Available from: https://www.mackenziesmission.org.au/

20 Wong M, Mayoh C, Lau LMS, Khuong-Quang D-A, Pinese M, Kumar A, et al. Whole genome, transcriptome and methylome profiling enhances actionable target discovery in high-risk pediatric cancer. Nat Med. 2020;26(11):1742–53. https://doi.org/10.1038/s41591-020-1072-4

21 Best S, Stark Z, Brown H, Long JC, Hewage K, Gaff C, et al. The leadership behaviors needed to implement clinical genomics at scale: a qualitative study. Genet Med. 2020;22(8):1384–90. https://doi.org/10.1038/s41436-020-0818-1

22 Best S, Long JC, Gaff C, Braithwaite J, Taylor N. Organizational perspectives on implementing complex health interventions: clinical genomics in Australia. J Health Organ Manag. 2021;35(7):825–45. https://doi.org/10.1108/JHOM-12-2020-0495

23 Damschroder LJ, Reardon CM, Widerquist MAO, Lowery J. The updated Consolidated Framework for Implementation Research based on user feedback. Implement Sci. 2022;17(1):75. https://doi.org/10.1186/s13012-022-01245-0

24 Atkins L, Francis J, Islam R, O'Connor D, Patey A, Ivers N, et al. A guide to using the Theoretical Domains Framework of behaviour change to investigate implementation problems. Implement Sci. 2017;12(1):77. https://doi.org/10.1186/s13012-017-0605-9

25 Glasgow RE, Vogt TM, Boles SM. Evaluating the public health impact of health promotion interventions: the RE-AIM framework. Am J Public Health. 1999;89(9):1322–7. https://doi.org/10.2105/ajph.89.9.1322

26 Rapport F, Clay-Williams R, Churruca K, Shih P, Hogden A, Braithwaite J. The struggle of translating science into action: foundational concepts of implementation science. J Eval Clin Pract. 2018;24(1):117–26. https://doi.org/10.1111/jep.12741

27 May CR, Johnson M, Finch T. Implementation, context and complexity. Implement Sci. 2016;11(1):141. https://doi.org/10.1186/s13012-016-0506-3
28 Braithwaite J, Clay-Williams R, Nugus P, Plumb J. Health care as a complex adaptive system. In: Hollnagel E, Braithwaite J, Wears RL, editors. Resilient health care. Farnham, Surrey, England: Ashgate Publishing Ltd; 2013. Vol. 1. p. 57–73.
29 Braithwaite J, Churruca K, Ellis LA, Long J, Clay-Williams R, Damen N, et al. Complexity science in healthcare – aspirations, approaches, applications and accomplishments: a white paper. Sydney, Australia: Macquarie University; 2017; p. 118.
30 Taylor N, Best S, Martyn M, Long JC, North KN, Braithwaite J, et al. A transformative translational change programme to introduce genomics into healthcare: a complexity and implementation science study protocol. BMJ Open. 2019;9(3):e024681. https://doi.org/10.1136/bmjopen-2018-024681
31 Rogers EM. Diffusion of innovations. New York: Free Press; 1995.
32 Stark Z, Dolman L, Manolio TA, Ozenberger B, Hill SL, Caulfied MJ, et al. Integrating genomics into healthcare: a global responsibility. Am J Hum Genet. 2019;104(1):13–20. https://doi.org/10.1016/j.ajhg.2018.11.014
33 Long JC, Pomare C, Best S, Boughtwood T, North K, Ellis LA, et al. Building a learning community of Australian clinical genomics: a social network study of the Australian Genomic Health Alliance. BMC Med. 2019;17(1):44. https://doi.org/10.1186/s12916-019-1274-0
34 Long JC, Gaff C, Clay C. Transforming the genomics workforce to sustain high value care. Research: Deeble Institute for Health Policy; 2022.
35 Pritchard DE, Moeckel F, Villa MS, Housman LT, McCarty CA, McLeod HL. Strategies for integrating personalized medicine into healthcare practice. Per Med. 2017;14(2):141–52. https://doi.org/10.2217/pme-2016-0064
36 Sperber NR, Carpenter JS, Cavallari LH, J Damschroder L, Cooper-DeHoff RM, Denny JC, et al. Challenges and strategies for implementing genomic services in diverse settings: experiences from the Implementing GeNomics In pracTicE (IGNITE) network. BMC Med Genomics. 2017;10(1):35. https://doi.org/10.1186/s12920-017-0273-2
37 Burns BL, Bilkey GA, Coles EP, Bowman FL, Beilby JP, Pachter NS, et al. Healthcare system priorities for successful integration of genomics: an australian focus. Front Public Health. 2019;7:41. https://doi.org/10.3389/fpubh.2019.00041
38 O'Shea R, Ma AS, Jamieson RV, Rankin NM. Precision medicine in Australia: now is the time to get it right. Med J Aust. 2022;217(11):559–63. https://doi.org/10.5694/mja2.51777
39 Jones LK, Romagnoli KM, Schubert TJ, Clegg K, Kirchner HL, Hu Y, et al. Using implementation science to develop a familial hypercholesterolemia screening program in primary care: The CARE-FH study. J Clin Lipidol. 2024;18(2):e176–88. https://doi.org/10.1016/j.jacl.2024.01.001
40 Australian Genomics. Australian Genomics is a national collaboration supporting the translation of genomic research into clinical practice [Internet]. [accesed 2024 14 March]. Available from: https://www.australiangenomics.org.au/
41 Genomics England. Genomics England - Powering genomic medicine, together. [Internet] [accesed 2024 14 March]. Available from: https://www.genomicsengland.co.uk/
42 Fehlberg Z, Best S, Long JC, Theodorou T, Pope C, Hibbert P, et al. Scaling-up and future sustainability of a national reproductive genetic carrier screening program. NPJ Genom Med. 2023;8(1):18. https://doi.org/10.1038/s41525-023-00357-w
43 Best S, Long JC, Braithwaite J, Taylor N. Standardizing variation: scaling up clinical genomics in Australia. Genet Med. 2023;25(2):100109. https://doi.org/10.1016/j.gim.2022.01.004
44 Damschroder LJ, Aron DC, Keith RE, Kirsh SR, Alexander JA, Lowery JC. Fostering implementation of health services research findings into practice: a consolidated framework for advancing implementation science. Implement Sci. 2009;4:50. https://doi.org/10.1186/1748-5908-4-50
45 White S, Jacobs C, Phillips J. Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care. Genet Med. 2020;22(7):1149–55. https://doi.org/10.1038/s41436-020-0785-6
46 Schaibley VM, Ramos IN, Woosley RL, Curry S, Hays S, Ramos KS. Limited genomics training among physicians remains a barrier to genomics-based implementation of precision medicine. Front Med. 2022;9:757212. https://doi.org/10.3389/fmed.2022.757212
47 Best S, Long JC, Gaff C, Braithwaite J, Taylor N. Investigating the adoption of clinical genomics in Australia. An implementation science case study. Genes. 2021;12(2):317. https://doi.org/10.3390/genes12020317
48 Ingebrigtsen T, Georgiou A, Clay-Williams R, Magrabi F, Hordern A, Prgomet M, et al. The impact of clinical leadership on health information technology adoption: Systematic review. Int J Med Inform. 2014;83(6):393–405. https://doi.org/10.1016/j.ijmedinf.2014.02.005
49 Pawson RT, Nicholas. Realistic evaluation. London; Thousand Oaks, Calif: SAGE Publications Ltd; 1997.
50 Francis-Auton E, Long JC, Sarkies M, Roberts N, Westbrook J, Levesque J-F, et al. Four system enablers of large-system transformation in health care: a mixed methods realist evaluation. Milbank Q. 2024;102(1):183–211. https://doi.org/10.1111/1468-0009.12684
51 Pullmann MD, Dorsey S, Duong MT, Lyon AR, Muse I, Corbin CM, et al. Expect the unexpected: a qualitative study of the ripple effects of children's mental health services implementation efforts. Implement Res Pract. 2022;3:26334895221120797. https://doi.org/10.1177/26334895221120797
52 Braithwaite J, Mannion R, Matsuyama Y, Shekelle P, Whittaker S, Al-Adawi S, et al. Accomplishing reform: successful case studies drawn from the health systems of 60 countries. Int J Qual Health Care. 2017;29(6):880–6. https://doi.org/10.1093/intqhc/mzx122
53 Braithwaite J, Ludlow K, Churruca K, James W, Herkes J, McPherson E, et al. Systems transformation: learning from change in 60 countries. J Health Organ Manag. 2019;34(3):237–53. https://doi.org/10.1108/JHOM-01-2019-0018
54 Braithwaite J, Mannion R, Matsuyama Y, Shekelle PG, Whittaker S, Al-Adawi S, et al. The future of health systems to 2030: a roadmap for global progress and sustainability. Int J Qual Health Care. 2018;30(10):823–31. https://doi.org/10.1093/intqhc/mzy242
55 Braithwaite J, Tran Y, Ellis LA, Westbrook J. The 40 health systems, COVID-19 (40HS, C-19) study. Int J Qual Health Care. 2021;33(1):mzaa113. https://doi.org/10.1093/intqhc/mzaa113
56 Braithwaite J, Fisher G. Beyond the aspirational: creating the future of health care in Australia. Intern Med J. 2024;54(2):342–7. https://doi.org/10.1111/imj.16286
57 Marschall J, Snyders RE, Sax H, Newland JG, Guimarães T, Kwon JH. Perspectives on research needs in healthcare epidemiology, infection prevention, and antimicrobial stewardship: what's on the horizon-Part II. Antimicrob Steward Healthc Epidemiol. 2023;3(1):e212. https://doi.org/10.1017/ash.2023.474
58 Masoud A, Dehnavieh R, Yazdi-Feyzabadi V, Poursheikhali A, Noori Hekmat S, kazemi M, et al. What is the mind map of the hospital's future changes in a developing country like Iran? A qualitative study. BMC Health Serv Res. 2023;23(1):732. https://doi.org/10.1186/s12913-023-09507-3
59 Michgelsen J, Zonneveld N, Tesfay E, Minkman M. Measuring the impact of integrated care: from principles to real-world impact. Int J Integr Care. 2023;23(4):12. https://doi.org/10.5334/ijic.7783
60 Puddester R, Limoges J, Dewell S, Maddigan J, Carlsson L, Pike A. The Canadian landscape of genetics and genomics in nursing: a policy document analysis. Can J Nurs Res. 2023;55(4):494–509. https://doi.org/10.1177/08445621231159164

61 Australian Genomics Health Alliance Acute Care Flagship; Lunke S, Eggers S, Wilson M, Patel C, Barnett CP, et al. Feasibility of ultra-rapid exome sequencing in critically ill infants and children with suspected monogenic conditions in the Australian public health care system. JAMA. 2020;323(24):2503–11. https://doi.org/10.1001/jama.2020.7671
62 Best S, Brown H, Lunke S, Patel C, Pinner J, Barnett CP, et al. Learning from scaling up ultra-rapid genomic testing for critically ill children to a national level. NPJ Genom Med. 2021;6(1):5. https://doi.org/10.1038/s41525-020-00168-3
63 Best S, Long JC, Theodorou T, Hatem S, Lake R, Archibald A, et al. Health practitioners' perceptions of the barriers and enablers to the implementation of reproductive genetic carrier screening: A systematic review. Prenat Diagn. 2021;41(6):708–19. https://doi.org/10.1002/pd.5914
64 Best S, Long JC, Fehlberg Z, Taylor N, Ellis LA, Boggs K, et al. Using a theory informed approach to design, execute, and evaluate implementation strategies to support offering reproductive genetic carrier screening in Australia. BMC Health Serv Res. 2023;23(1):1276. https://doi.org/10.1186/s12913-023-10053-1
65 Kirk EP, Delatycki MB, Archibald AD, Tutty E, Caruana J, Halliday JL, et al. Nationwide, couple-based genetic carrier screening. N Engl J Med. 2024;391(20):1877–89. https://doi.org/10.1056/NEJMoa2314768
66 Pineault R, Borgès Da Silva R, Prud'homme A, Fournier M, Couture A, Provost S, et al. Impact of Québec's healthcare reforms on the organization of Primary Healthcare (PHC): a 2003-2010 follow-up. BMC Health Serv Res. 2014;14(1):229. https://doi.org/10.1186/1472-6963-14-229
67 Belrhiti Z, Bigdeli M, Lakhal A, Kaoutar D, Zbiri S, Belabbes S. Unravelling collaborative governance dynamics within healthcare networks: a scoping review. Health Policy Plan. 2024;39(4):412–28. https://doi.org/10.1093/heapol/czae005