POLICY BRIEF

Genomics in Healthcare

Key issues for implementation

From Genomics England to the NHS – The UK Country Exchange Visit (CEV) addressed the topics of infrastructure, including funding, sustainability, and governance; tools and training; patient awareness and engagement; health economics of genomic medicine and Implementation into healthcare systems.This visit took place virtually March 23-24 2021.

Genomics Towards Prevention The CEV to Estonia addressed the topics of personalised prevention of common and complex diseases; pharmacogenomics; population biobanks; citizen awareness. This visit took place virtually May 19-20, 2021.

Regulating the Unknown – the CEV to Finland focused on topics health sector growth strategy for research and innovation; creation of a regulatory framework; technical infrastructure; industry collaboration. This visit took place virtually June 16-17, 2021.

Genomics Medicine programs across Europe - during the three CEVs, 13 countries (Belgium, Bulgaria, Denmark, Germany, Hungary, Italy, Latvia, Lithuania, Luxembourg, Norway, Portugal, Spain, Sweden) presented ongoing genomic medicine initiatives in their respective nations.

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Implementing genomic medicine in healthcare settings can bring us one step closer to making personalised medicine a reality, bringing with it notable socio-economic benefits. Accurate, timely diagnostics, personalised treatment protocols and preventative approaches improve efficiency in health systems and patients' quality of life. Healthcare can widely benefit from genomic data analysis for diagnosis and treatment, for instance with earlier diagnosis that allow a more effective intervention, or pharmacogenomic-based treatment. Increasingly, accurate profiling of individual patients is promoting a shift of medical practice towards disease prevention.

To achieve the goal of implementing genomics in healthcare systems, countries need to establish national genomic medicine strategies. However, it is clear that European countries are currently at varying stages of maturity for using genomics in healthcare. Promoting the dialogue and cooperation among countries, for capacity building and sharing of best practices, is therefore extremely beneficial for advancing genomic medicine at the national and European levels. Some key issues to build efficient and sustainable genomic medicine strategies are:

1. Patient and citizens trust and engagement
2. Infrastructure for implementation of genomics in healthcare practice
3. Ethical and legal frameworks
4. Synergies among healthcare, research and industry
5. Training of healthcare professionals

Seeking to provide cross-border access to at least one million genomes and related clinical data, enabling secure data sharing in a trusted environment, the 1+Million Genomes Initiative (1+MG) has taken a massive step towards delivering personalised medicine to all citizens in Europe. So far, 24 European countries have committed to the initiative by signing the 1+MG Declaration of Cooperation.

In 2021, the Beyond 1 Million Genomes (B1MG) project, supporting the 1+MG initiative, organized three Country Exchange Visits (CEVs) to discuss the critical points for implementing sustainable national genomic medicine strategies. Three European countries with well-advanced genomic strategies in healthcare were invited as hosts: the United Kingdom, Estonia and Finland. Representatives of 1+MG signatory countries participated in these events, showing their commitment.

This policy brief addresses the key issues discussed at the CEVs, offering real-life examples and proposing a series of policy recommendations to successfully implement genomics in European Healthcare systems.

Patient and citizen engagement is at the heart of personalised medicine

Patient and citizen engagement

Patient and citizens' understanding and trust are critical for patient engagement in genomic medicine. Consent for data use and sharing, and support for genomic medicine policies is only possible with patient engagement and public acceptance.

Building citizen and patient trust for genomics medicine initiatives requires time and constant investment in bidirectional communication, rigorous information and public campaigns to raise awareness.

Estonia demonstrated the benefits of successful public engagement in genomics medicine. Having achieved 75% of public buy-in for personalised medicine, most patients expect their genomic data to be used for their benefit.

Estonia has only solidified their strategy after decades of communication campaigns, involving public figures, advertising, and referencing health genomics in soap operas and movies. The program also offered free genetic test results to volunteers willing to provide DNA samples to the national biobank.

It is clear that, from the very beginning, citizens and patients need to be at the center of a genomic initiative to create engagement and trust; the system must be built on a transparent basis. Patients must be represented in the established governance framework with a voice in every decision and involved in discussing the legal framework.

In the UK, patients have been involved from the start in relevant decision boards for Genomics England and the National Health Service (NHS). In Genomics England this was achieved by creating a Participant Panel, who have had significant input into the formal governance framework and implementation structure. Panel members are compensated for the time they have dedicated to representing the wider participant voice. The NHS has patient and participant involvement in all of their governance groups and a NHS People and Communities Forum for the NHS Genomic Medicine Service.

Citizens and patients also need to be offered the opportunity to consent to the use of their data for research purposes. This will increase their interest and engagement because they can contribute to the greater good.

In Finland, the Genome Act will be the legal basis for establishing a national genome centre and a centralised genomic database. This initiative has involved patients and citizens from the outset. The same procedure was taken for the Act on Secondary Use of Health and Social Data.

POLICY RECOMMENDATIONS

Establish information programs and wide-reaching communication campaigns to engage and inform citizens.

Monitor patient trust and willingness to give samples and consent for the use of genomic-based diagnosis and treatments.

Involve patient groups in all decisions and steps of implementing the genomic strategy since the outset of the programme, including discussion of the legal framework.

Ensure representation of patients in the established governance framework.

Incorporate patient representatives compensation into budgets.

Implementation in healthcare: infrastructure and regulation

High-quality data is essential for genomics in healthcare, and its collection and use for clinical and research purposes requires a safe and trusted environment. For this reason, the UK, Estonia and Finland have been taking steps towards building a secure infrastructure for managing information, with centralised governance and robust data management plans.

In the UK, Genomics England and the NHS clearly defined data access and sharing policies. The Genomics England Data Centre is a secure facility located within the NHS firewall, which stores genomic and other patient-related data.

In Finland, the National Genome Centre will be the central public authority responsible for managing the Finnish population's genome database and promoting equity and the responsible use of genomic data.

The generalised digitalisation of health and genomic data will significantly accelerate the incorporation of genomic data into healthcare. A solid investment in secure digital technologies and services, for instance in Electronic Health Record (EHR) systems, enables the combination of genomic data with clinical information from multiple sources and in variable formats, facilitating the efficient use of genomic information in clinical practice.

Collection and use of genomic and health data requires a safe and trusted environment

In Estonia, all medical records are electronic, and there is a central technical infrastructure to link information systems and ensure secure access to medical and other data. A health portal also links different databases, including clinical diagnosis, prescription, billing and disease trajectories.

Access to health data and human biological samples must be strictly regulated, with a solid ethical and legal framework that ensures the secure and transparent collection, analysis and use of data.

Estonia has prioritised the legal framework. The Human Genes Research Act in Estonia dates back to 2000, when its national Biobank was established. The national biobank has over 20% of citizens registered.

Finland has also prioritised the legal framework to ensure a trustworthy environment and establish the legal basis for implementing genomics in healthcare. Finndata, the data permit authority, implements the Act on Secondary Use of Health and Social Data. At the same time, the National Genome Centre awaits for the Genome Act to be approved.

Equally crucial is the standardisation of procedures that guarantees quality control, interoperability between services and cross-border data sharing, for clinical and research purposes, at regional, national and international levels.

The UK's genomic infrastructure associated with the NHS operates according to common national standards by an "end-to-end" ISO accredited pipeline.

Steady political commitment and sustainable funding, involving inter-ministry cooperation, are essential to support the implementation of genomics

For instance, the Finnish Health Sector Growth Strategy involves the Ministry of Education and Culture, the Ministry of Economy and Employment, the Ministry of Social Affairs and Health, Academia Finland and Business Finland, and the Innovation Funding Agency of Finland

Further development of economic models for genomic medicine is needed, including patient and family benefits. Evidence for economic value can already be found in several indicators. These can highlight the importance of genomic sequencing before even acknowledging patient and family benefits.

The NHS and Genomics England are developing solid evidence for the cost-effectiveness analysis of genomics, which can be used by policy makers to implement genomic tests in healthcare and promote wide and equitable access to citizens.

POLICY RECOMMENDATIONS

Create an infrastructure with centralised governance and a robust ethical and legal framework for secure and transparent collection, analysis and use of data.

Ensure solid investment in secure digital technologies and services. For instance, implementing electronic health record systems that combine clinical information with automated decision support action to use genomic information in clinical practice.

Seek inter-ministerial collaboration and inclusion of all stakeholders for implementing the strategy, gaining investment in public-private partnerships, and earning solid and steady political support.

Implement a standard genomic and health data management plan to facilitate sharing information for clinical and research at regional, national and international levels.

Training and capacity building

Implementing Genomic Medicine into the healthcare system requires a specialised, collaborative and well trained workforce. For this, healthcare professionals should be able to improve and broaden their competencies at any time, and for every step, process and procedure in genomic medicine. This specialist workforce must be built upon the existing workforce, and healthcare professionals must be offered the possibility to upskill their competencies through lifelong training.

The UK offers an excellent example as it has created the Genomics Education Program for healthcare professionals and has developed a competency framework – a best practice development guide to support individuals and organisations seeking to obtain more competencies in genomic medicine.

However, it is equally important to recognise the need for emerging new professions that can address the fast innovation in genomic medicine.

The UK has recognised new professions, for instance clinical scientists and medical informaticians

Communication and genetic counselling in the healthcare system is crucial to make genetic diagnosis, genetic risk scores or pharmacogenetic recommendations comprehensible and accessible to patients. Healthcare professionals are the first line of communication with patients. Some countries have already recognised the importance of communicating with patients and understanding how they deal with diagnosis and health risk information.

Finland has established several initiatives to improve health professionals competencies in communicating with patients.

A well informed and engaged workforce is vital for genomic medicine

POLICY RECOMMENDATIONS

Tailor genomic education programmes for capacity building and training the existing healthcare workforce, including:

- Online courses for professionals with limited or no genomics knowledge, with a final test and certificate
- A competency framework with different levels for individuals and organisations to evaluate the need for increased knowledge/skills

Invest in developing professionals, namely clinical geneticists and genetic counsellors, and new professions, such as medical informaticians

Define the roles of these and other professionals, such as general practitioners that specialise in clinical genetics and provide counsel related to genomic information.

Building a sustainable ecosystem for genomics in healthcare

A successful collaboration between healthcare systems, research and industry ultimately benefits patients, health services, health economy and society at large

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1+MG
1+ Million Genomes initiative

Close cooperation between research, industry and the healthcare system benefits society, health services and the health economy. Therefore, all stakeholders should be involved from the start in the design of the genomic medicine strategy.

The partnership between research and the clinics is fundamental to create a learning health ecosystem and provide sustainability.

Genomics England and the NHS developed the "infinity loop" concept to continually promote discoveries and healthcare uptake of genomic testing for diagnostics, treatment and prevention. This "infinity loop" enables close collaboration between researchers and health systems: the NHS supplies patient data to research projects and labs, which, in turn, provide the NHS with knowledge and innovative solutions for clinical care.

Genomic medicine requires constant updates of knowledge and technology, making the private sector an essential stakeholder that contributes to generating novel solutions faster. By enabling industry involvement in a transparent and regulated manner, through effective stakeholder engagement, it is possible to harness the sheer potential and value of the healthcare industry.

In Finland, the industry actively collaborates with the healthcare system. For instance, FinnGen, a public-private project, has many diverse partners from both the public and private sectors, including the biobank cooperative FINBB and 12 pharma companies.

In Wales, the Genomics Partnership Wales is based in the Cardiff centre, a hub for research and innovation, joining academia, industry and the national healthcare service.

Despite its relevance, the private sector is still not fully perceived by the public as trustworthy. Developing a strong and legal framework is thus critical. Regulation needs to allow for innovation in healthcare systems, and embrace industry stakeholders, including large pharmaceutical companies but also the small and medium technological enterprises, as key partners in the innovation process. Industry, and research should not be distinguished regarding access and usage of data, as long as they are both abiding by the same rules.

In Estonia, the newly launched National Digital Agenda 2030 will discuss how to involve the private sector and what is the right legal, technical, governance, financial, data sharing and infrastructure frameworks to do it in a way that maintains public trust and improves personalised medicine services and care.

POLICY RECOMMENDATIONS

Involve all stakeholders in discussing the legal framework to allow for innovation while building trust and avoiding data misuse.

Create an umbrella structure that allows research, clinical and industry partners to share knowledge, support each other and ensure the coordination of research and clinical outcomes.

Include industry stakeholders input in health economic evaluation.

Governments need to embrace the role of enablers of genomic medicine by:

- Encouraging the integration and management of innovation
- Promoting dialogue among all stakeholders through partnerships
- Funding initiatives that will support sustained growth and attract further funding
- Creating the conditions for research results, digitalisation and knowledge to be used for the benefit of citizens first, but also considering other stakeholders' needs.