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Deliverable description

This policy white paper provides an in-depth analysis of the integration of Next-Generation Sequencing (NGS) and Public Health Genomics (PHG) across European healthcare systems. It identifies key policy gaps, disparities in adoption, and best practices to enhance genomic technologies in healthcare. The document serves as a strategic and technical resource for policymakers, healthcare providers, researchers, and public health organizations, offering actionable recommendations to drive policy reforms, improve workforce capabilities, and promote equitable access to genomic medicine across Europe. The deliverable is derived from a study published in Critical Reviews in Oncology/Hematology, which engaged experts in NGS implementation and public health genomics: Horgan D et al. From theory to practice: Implementing next-generation sequencing and public health genomics in healthcare systems. Crit Rev Oncol Hematol. 2024 Sep;201:104433. <https://pubmed.ncbi.nlm.nih.gov/38955310/>.

Introduction: Background and Objectives

The rapid evolution of genomic technologies presents a transformative opportunity to enhance healthcare across Europe. Next-Generation Sequencing (NGS) and Public Health Genomics (PHG) hold immense potential to revolutionize diagnostics, treatment pathways, and preventive strategies, particularly in oncology and rare diseases. However, despite these advantages, their integration into national healthcare systems remains uneven.

This policy white paper addresses the urgent need for harmonized policies and frameworks to facilitate the widespread adoption of genomic medicine. It builds upon a study published in Critical Reviews in Oncology/Hematology, which engaged experts in NGS implementation and public health genomics. The findings serve as a foundation for the recommendations outlined in this paper.

Objectives of this policy paper

1. Summarize the current landscape of NGS and PHG adoption across Europe, highlighting disparities and challenges.
2. Identify key barriers, including regulatory misalignment, infrastructure limitations, workforce shortages, and data governance issues.
3. Provide actionable policy recommendations for EU and national policymakers, healthcare professionals, researchers, and industry leaders.
4. Support decision-makers in shaping policies that promote equitable and sustainable genomic integration in healthcare.

Key stakeholders

- EU and National Policymakers: Responsible for legislation, funding mechanisms, and genomic strategy development.
- Healthcare Providers (HCPs): Key actors in the implementation of NGS-based diagnostics and treatment.
- Researchers & Academia: Contribute to the evidence base and technological advancements in genomics.
- Patient Advocacy Groups (PAGs): Ensure patient-centric approaches and accessibility.
- Industry & Private Sector: Provide technological infrastructure, innovation, and investment.

By addressing these objectives, this paper serves as a roadmap for policymakers and stakeholders to accelerate the adoption of NGS and PHG in European healthcare systems.

Executive Summary

Current situation

The adoption of NGS and PHG varies significantly across European countries. While some nations (e.g., Germany, France, and Nordic countries) have successfully integrated genomic technologies into their healthcare systems, others face barriers such as:

- Regulatory misalignment
- Lack of standardized reimbursement models
- Insufficient infrastructure and trained personnel
- Ethical and legal concerns regarding genomic data use

Without targeted action, these disparities will persist, preventing patients from benefiting equally from genomic-driven healthcare.

Opportunities and barriers

Opportunities:

- Advancing precision medicine: Genomic integration can significantly improve diagnostics and treatment for oncology and rare diseases.
- Boosting research and innovation: Secure genomic data-sharing frameworks can drive medical research and innovation.
- Improving healthcare efficiency: Standardized NGS practices can reduce healthcare costs and optimize patient outcomes.

Barriers:

- Funding and reimbursement challenges: The absence of harmonized funding mechanisms restricts access to NGS testing.
- Data-sharing limitations: Legal and ethical barriers hinder cross-border genomic data exchange.
- Workforce capacity gaps: The demand for trained professionals in genomics exceeds the current supply.

Recommendations moving forward

To ensure equitable access to genomic medicine, this white paper proposes the following policy actions:

1. Harmonized EU-wide regulatory frameworks: Establish clear guidelines for NGS implementation.
2. Sustainable funding and reimbursement policies: Develop national genomic testing reimbursement schemes.
3. Investment in healthcare workforce training: Increase genomics education programs for healthcare professionals.
4. Standardized data governance: Implement robust policies for secure and ethical genomic data-sharing.
5. Public awareness and patient engagement initiatives: Strengthen efforts to educate patients on genomic medicine benefits.

By implementing these recommendations, EU and national policymakers can ensure that genomic medicine becomes a cornerstone of European healthcare.

From Theory to Practice: Implementing Next-Generation Sequencing and Public Health Genomics in Healthcare Systems

Highlights

Current state of genomic integration

- The adoption of Next-Generation Sequencing (NGS) and Public Health Genomics (PHG) varies significantly across Europe.
- Countries like Germany, France, and the Nordic nations have advanced genomic adoption, while others face barriers such as regulatory misalignment, infrastructure gaps, workforce shortages, and financial constraints.

Opportunities and barriers

- Opportunities: Genomic integration can enhance precision medicine, improve diagnostics, and optimize healthcare delivery, particularly in oncology and rare diseases.
- Barriers: Persistent challenges include fragmented policies, insufficient reimbursement frameworks, data-sharing constraints, and workforce capacity limitations.

Key recommendations for policy action

- Develop harmonized policies and funding mechanisms to facilitate genomic integration across Europe.
- Strengthen data governance to enable secure and ethical cross-border genomic data sharing.
- Invest in education and workforce training to build genomic expertise among healthcare professionals.
- Enhance patient engagement and public awareness to promote trust and acceptance of genomic technologies.

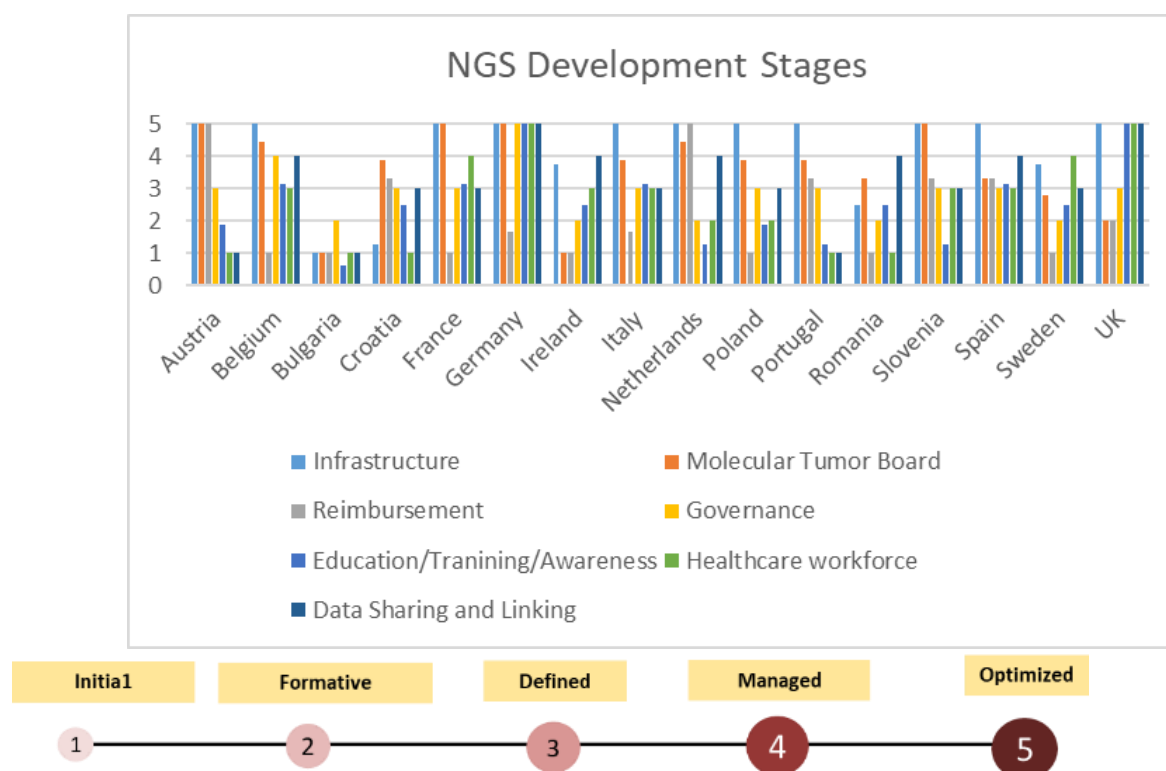


Fig 1: Development stages in NGS

An opportunity neglected?

A successful response in Europe to the growing challenges of disease depends in large measure on how far health systems adopt the technology and techniques that can enhance prevention and expedite early-stage diagnosis. At present the effective integration and exploitation of these opportunities from country to country remains hesitant and suboptimal, and health services - and health - underperform accordingly, with patients suffering as a result of continuing variations in diagnosis and in access to innovation. Conscious policy choices, accompanied by adequate resource allocation, are required to secure the potential benefits.

The degree of take-up, and crucially the state of readiness for take-up, has been tracked across European countries in a study of the influential factors and predisposing conditions, with a focus on Next Generation Screening (NGS) technologies and the development stage of Public Health Genomics (PHG). The resulting analysis of the key considerations in seamless integration offers a blueprint of how to turn theory into practice. It highlights national systems' achievements in integration, and pinpoints technical voids that inhibit adoption.

The promise of genomic data is already clear in theory, and is increasingly demonstrated in reality. NGS and PHG generate data that can bring great benefits to health. And the greater the volume of data, and the more diverse and varied the sources, better is the understanding that can flow from its analysis.

The potential gains

With the right approach, integration creates positive feedback loops. NGS supports health services, producing valuable datasets of selected populations, offering support to research, which flows back to improvements in technology in new tests and techniques through links between academia and industry and in health services. The revelations provided contribute significantly to the ongoing progress in comprehending and treating various conditions such as cancer, cardiovascular disease, rare genomic disorders, and prenatal care for newborns. PHG provides unparalleled insights into population health and trends, facilitating enhancements not only in overall citizen well-being through improved resource allocation and planning but also introducing newfound efficiencies and potential cost savings to healthcare systems. Promoting patient and public awareness opens up new avenues by building receptiveness to prevention, diagnosis and therapy, creating a basis for greater trust and confidence among health services and public authorities. Integrating these components of better health provision at national level brings benefits to national populations and helps to overcome discrepancies in health services within each country. The impact at the European level is particularly significant, presenting a pathway towards cumulative enhancements where the collective outcome exceeds the individual contributions.

Taking account of genomics in the drug development phase increases the chances of success in clinical trials, and the use of pharmacogenetics brings advantages in research and treatment, using genetic information to estimate disease risk or the efficacy of treatment. The power of genomic data is also amplified by the addition of information on the patient from medical records or the growing abundance of data from administrative sources, and on his or her context – in terms of the local and general environment. The link between data from genome sequencing and this phenotypic data can be decisive in **allowing the personalised care that is now capable of transforming treatments**, and is doing so already for many patients. By extension, this facility can also provide valuable inputs to thinking about population health. It permits **tailoring effective healthcare strategies** that can take advantage of novel but neglected possibilities, ultimately leading to **improved health outcomes** - and particularly. in the case of cancer, **a reduction in the overall societal as well as personal burden. The need now is for these practices to be more widely recognised as valuable, and for them to be implemented and to be scaled up.**

The conditions

Of course there are conditions. NGS depends not only on infrastructure, equipment, and funding, but also on its regular utilization, and the overall preparedness for its integration. For PHG, critical prerequisites include investment, adherence to data management standards, and workforce proficiency – but also on a suitable legislative framework, established governance structures, clinical organization, and public awareness. All this necessitates deliberate policy decisions. Bridging these gaps requires significant efforts in knowledge dissemination across healthcare innovation and research, enabling the application of advanced insights in both clinical practice and healthcare strategizing.

The study identified **key pillars for NGS functionality and usage** - notably the provisions for infrastructure, molecular tumor boards, reimbursement, governance, education/training/awareness, healthcare workforce, and data sharing and linking. **Readiness among healthcare systems for PHG integration** took account of governance and strategy, investment and

economic model, legislation and policy, public awareness and acceptance, workforce skills and organization, clinical organisation/infrastructure/tools, clinical genomics guidelines and infrastructure, and data management standards/infrastructure.

But also tracked was **the degree of engagement of patients**, and **the level of information** among the general public on cancer prevention and disease mechanisms – on the basis that the effective deployment of better diagnosis and therapy is not a one-way but a two-way street, including patient perspectives and the importance of patient engagement in shaping healthcare practices.

Among the factors that will influence the outcome are the success – or failure – of some changes in the environment for advanced medical care. A new system of European clinical assessment is scheduled to come into operation as from the start of 2025; final implementation is due – although seriously delayed – of legislation on diagnostics; the EU is in the process of finalising rules on secondary use of health data; and European guidelines on screening have been issued and it is now up to individual countries to apply them. More broadly, and crucially, much will depend on the attention afforded by decision makers at all levels to innovation, particularly in terms of research promotion and of take-up of promising new methods and methodologies.

Over and above ensuring the optimal implementation of new regulations, what also need to be addressed are the current disparities in diagnosis and treatment options, the wide variations in availability of quality metrics so essential for sequencing, the uneven adherence to documented best practices in sharing and linking phenotypic and genetic data, and the creation of functioning roadmaps for secure cross-border data exchange. Diverse levels of sophistication are evident in economic modeling for assessing new medications, as well as in conducting gap analyses, perpetuating a disregard for potential areas of enhancement. Taking advantage of the opportunities depends on overcoming the associated barriers. These are often largely technical – such as diverse data sets with distinct approaches, definitions and scope. In the absence of a sufficient workforce – such as in pathology, no degree of policymaking or infrastructure investment can yield optimal effectiveness. Similarly, lack of alignment among data repositories makes access to their content impractical.

Varied landscapes revealed

The picture that emerges is of **a Europe divided**. One of the most **striking divergences is in the stage of NGS integration** (Fig. 1). Austria, France, Germany, Italy, Netherlands, Slovenia, Spain, and the UK show significant progress, with established and optimized infrastructure, sufficient equipment and resources and routine utilization. They also boast functional molecular tumor boards that conduct regular consultations and discussions, indicating a high level of effectiveness and integration of genomics in cancer care, and have well-implemented educational programs and governance structures ensuring the availability of a trained workforce and of wide understanding regarding NGS testing and application. In contrast, Bulgaria, Ireland, Poland, Portugal, and Romania are at the early stages of development in most pillars, with outstanding challenges in infrastructure, reimbursement plans, and insufficient trained personnel and awareness, and only at the very beginning of developing their healthcare systems to fully integrate NGS and genomics into routine practice.

A **similar divergence is apparent in integration of PHG**, where Denmark, Sweden, Norway, and Finland constitute a vanguard, with institutionalized governance and strategy, significant investment in economic models supporting genomics, legislation and policies in place, and developed workforce skills and organizational structures. They also exhibit well-developed clinical organization, infrastructure, and tools, along with robust data management standards and infrastructure. Additionally, they have established clinical genomics guidelines and infrastructure, and public awareness and acceptance of genomics are also high.

By contrast, Latvia, Portugal, and Estonia are at varying stages of development, and while they show some progress in governance, economic models, legislation, workforce skills, and clinical organization, they lag in data management standards, genomic standards, and public awareness and acceptance.

Measures of patients' awareness levels on cancer prevention and early diagnosis reveal significant variations across European countries. While some nations exhibit high levels of understanding regarding the role of genetics, prevention strategies, and early detection technologies, others show gaps in public knowledge that may hinder the adoption of genomic medicine. The differences are particularly notable in awareness of cancer heredity, early disease mechanisms, and the use of blood tests for early screening and personalized prevention. These disparities underline the need for targeted public education campaigns and harmonized communication strategies to bridge the awareness gap across Europe.

Significant disparities were clear regarding tangible resources like adequate data storage and the availability of specialized staff such as radiologists or clinical geneticists and genetic counselors, as well as for less tangible factors such as the presence and adherence to guidelines, patient engagement, tailored consent procedures, and the intricate organization of multidisciplinary teams for comprehensive clinical follow-up. Notably, the nations with the highest scores also benefit from the crucial asset of political leadership driving practice changes and the necessary investments and commitments. Looking across the entire suite of studies, what emerges prominently is a close correlation between policy choices and the capacity and willingness to embrace innovation.

Clearly economic prosperity is a factor, since the sort of provision under discussion can have high costs. But it is equally clearly not the only influential variable. On NGS integration, Ireland too trails most of the member states. In PHG maturity level, the Nordic countries score highly, with the exception of Finland, and Spain also has a low score. But on patient awareness, Romania scores higher than Luxembourg, which has a Gross domestic product (GDP) roughly ten times higher.

The need is evident for harmonized standards and improved infrastructure to support precision oncology. The variability in biomarker testing quality and access is a particular problem that demonstrates the differences in resource availability and the need for targeted investments and policy support to bridge gaps.

Best practices already exist on sharing and linking data in both the healthcare sector and in the research setting – but they are not at present routinely followed. The blueprint approach, delineating developmental stages, makes it possible not only to identify pre-conditions, but also to monitor

advancements and potentialities from the initial introduction of innovation to the complete optimization of NGS, and to outline the pathway toward the effective implementation of PHG.

The right moment

The study takes account of the context of a rapidly changing European policy environment and the linked prompts to reconsidering how and where new tools can be integrated into healthcare systems and routine practices.

The moment is right for such a review of prospects, prerequisites and performance. There are at present **real opportunities for reconsidering health strategy** as the European Union overhauls its outdated regulatory frameworks and renews its membership of the European Parliament and the leadership of the European Commission. The possibilities are opening up of delivering the science-based medicine capable of guaranteeing a genuinely 21st century level of service. With wise choices, the outcome could prove critical in remedying the current wide inequalities in access to effective diagnosis and treatment, particularly of cancer.

The European policy landscape is evolving rapidly, creating a timely opportunity to integrate Next-Generation Sequencing (NGS) and Public Health Genomics (PHG) into healthcare systems. With the European Union revising regulatory frameworks and ushering in a new cycle of decision-making, there is momentum to address disparities in genomic medicine and enhance access to precision healthcare.

The renewal of the European Parliament and the European Commission presents a chance to prioritize policy changes that facilitate equitable adoption of genomics. Upcoming regulations, such as the European Health Data Space, revised clinical assessment systems, and finalized diagnostics legislation, further signal a shift toward fostering innovation in medical technology. Additionally, increasing awareness of advanced health technologies as crucial economic assets strengthens the case for genomic integration.

However, realizing these opportunities requires deliberate policy action and investment. Without coordinated efforts, existing gaps in workforce training, infrastructure, and reimbursement mechanisms will continue to hinder progress.

But many discrepancies have deeper roots, in national or regional differences of approach and attitude. Even in neighbouring countries, national laws differ, particularly on issues as sensitive as handling of personal data. Health service organisation and culture remain often distinct, too, and achieving new forms of cooperation can spring only from deliberate and conscious efforts. It requires pooling of information and exchanges of experiences, joint reflections on efficient data access and exchange, and on the related legal and ethical issues, on training, on building mutual confidence and trust. Real progress, with genuine Europe-wide collaboration, requires a superior level of coordination. It needs infrastructure that is not only secure at national level but can function seamlessly for cross-border data. It needs agreement on guidelines, standards and best practices for sharing genomic and phenotypic data.

The importance of trust in relation to data is starkly demonstrated by the current controversies over Europe's plans for sharing health data – the European Health Data Space. From the outset this proposal has faced strong concerns from patient and consumer organisations, who have lobbied hard for some forms of opt-out to allow for privacy over access to personal data, unconvinced by arguments from health industries and prominent elements of the research sector that access is essential for innovation.

This delicate process can succeed only if there is **engagement with policymakers, with public and patients and stakeholders in research and technology development communities, and with sources of funding**. High-level policy endorsement will come only if the benefits are seen to outweigh the costs, and further work will have to be done on demonstrating the economic merits of NGS and PHG, in a harsher public spending climate where models for HTA analysis become ever more demanded and demanding. And without closer collaboration between the public and private sectors, particularly in high technology, the chances of successful progress are dim indeed.

The European policy context is dynamic, with an amalgamation of external forces compelling European leaders to reassess longstanding strategic presumptions. Amidst this recalibration, healthcare, traditionally relegated to the sidelines among European policies, has garnered renewed importance. This shift is attributed partly to the impact of Covid19 pandemic, partly to the escalating morbidity rates within an aging demographic, and partly to the growing acknowledgment of advanced health technologies as pivotal components within Europe's economic and trade portfolio. The last five years has seen attention grow within the Council of the EU and European Parliament to the need for robust health services and systems, culminating in 2024 in formal recognition of "a changing world with new geopolitical realities and disruptive technologies, and of the necessity to further prioritise research efforts around the EU's long-term needs". The potential is obvious of a new cycle of EU decision-making that is commencing, with planning beginning for the future direction of the EU.

There is another precondition to success. The record to date of the EU in decision-making relating to health has been patchy, with legislation on issues such as clinical trials, diagnostics and data protection requiring subsequent measures to better address the ever evolving reality of healthcare and health technology. The role of stakeholders in informing decision makers in the design of policy has consequently acquired new significance, and central to that is **the supply of accurate information to guide policy formation**.

Conclusion

Policy recommendations and next steps

The integration of Next-Generation Sequencing (NGS) and Public Health Genomics (PHG) into European healthcare systems requires targeted efforts and investments to overcome existing barriers and unlock the full potential of precision medicine. Policymakers at both the EU and national levels, along with healthcare professionals (HCPs), researchers, industry leaders, and patient advocacy groups (PAGs), must collaborate to ensure sustainable and equitable genomic adoption.

Key recommendations for policy action:

To drive the successful implementation of NGS and PHG, targeted efforts should focus on the following priority areas:

1. Policy and governance:

- EU and National Governments should establish harmonized policies and regulatory frameworks to integrate genomic medicine into healthcare systems.
- Governments and regulatory agencies must streamline reimbursement mechanisms for genomic testing, ensuring sustainable financing models that encourage investment.
- National and EU policymakers should define standardized ethical and legal guidelines for genomic data sharing and secondary use, enabling responsible cross-border collaboration.
- Create national and EU-level Public Health Genomics strategies that align with cancer and rare disease plans, ensuring genomic integration into routine medical care.

2. Infrastructure and investment:

- Healthcare institutions and funding bodies should prioritize investment in sequencing infrastructure, including high-throughput NGS platforms, secure data storage, and bioinformatics capabilities.
- EU funding programs (such as Horizon Europe, EU4Health, and national research grants) should allocate dedicated funds for upgrading genomic laboratories and training centers.
- Governments should support national genomic initiatives to ensure widespread testing availability and accessibility, particularly in underserved regions.
- Public-private partnerships should be strengthened to drive investment in genomic research and its clinical applications.

3. Healthcare workforce development:

- Medical schools, universities, and training institutions should expand genomic education programs for healthcare professionals (HCPs), including oncologists, pathologists, genetic counselors, and general practitioners.
- Governments and healthcare authorities must implement continuous professional training in genomics to keep the workforce up-to-date on emerging technologies.
- Develop multidisciplinary genomic teams, integrating molecular tumor boards (MTBs) into hospital systems to improve personalized treatment decisions.

4. Data governance and interoperability:

- National governments and EU institutions should establish interoperable genomic databases that allow secure, anonymized data sharing across borders, aligned with the European Health Data Space (EHDS).
- Health ministries and IT departments must standardize electronic health records (EHRs) to include genomic data, ensuring seamless integration into clinical workflows.
- Governments should fund national genomic data repositories that support large-scale studies and allow AI-driven insights to optimize healthcare delivery.

5. Public engagement and patient advocacy:

- Patient Advocacy Groups (PAGs), healthcare authorities, and EU institutions should launch awareness campaigns to educate the public about the benefits and ethical considerations of genomic medicine.
- Ensure patient consent frameworks are clear, transparent, and inclusive, fostering public trust in genomic research.
- Increase funding for citizen-science initiatives and patient engagement programs, ensuring that patient perspectives shape genomic policies.

The urgency for action

If Europe fails to act now, the opportunity to revolutionize healthcare through NGS and PHG may be lost, leaving gaps in diagnosis, treatment, and research competitiveness. The adoption of genomics is not just a technical challenge—it is a policy imperative that requires immediate attention, coordinated investment, and long-term strategic planning.

By implementing these targeted actions, EU and national policymakers, healthcare providers, researchers, and patient advocacy groups can collectively ensure that genomic medicine delivers its full potential—improving patient outcomes, reducing health disparities, and driving forward the next era of precision healthcare across Europe.



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