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Parliamentary Inquiry Response

Innovation in the NHS - Personalised Medicine and AI

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Submission to the House of Lords Science and Technology Committee
Inquiry: Innovation in the NHS – Personalised Medicine and AI

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Executive Summary

1. This submission presents findings from qualitative empirical research examining the ethical, practical, and system-level challenges of implementing genomic medicine within the NHS. The study draws on 24 in-depth interviews with UK experts across clinical genetics/genomics, genomic science, policy and bioethics.

2. While the UK has established global leadership in genomic medicine through initiatives such as Genome UK, Genomics England and the NHS Genomic Medicine Service, the findings indicate that the primary barriers to implementation are not technological but related to system design, governance, workforce capacity, and public trust.

3. Key findings include:

- Current health service data infrastructure is not optimised for clinical interpretation and use.
- Trust, governance, and public engagement are central to successful implementation.
- There is a risk of widening health inequalities, if careful consideration is not given to managing this issue during the implementation of genomic technologies.
- Deployment challenges are not technological but primarily organisational and structural factors within the healthcare system.
- There is a risk that policy ambition to introduce genomic medicine at pace may outstrip both systems and clinical readiness.
- There are tensions between centralised genomic systems and local healthcare delivery.
- The use of different genomic data in medicine and its contextual interpretation is complicated; there is a risk that oversimplification in policy and implementation will result in unintended consequences and ethical blind spots.

4. The evidence demonstrates that successful implementation will require a coordinated, system-wide approach that integrates governance, infrastructure, workforce development, and public engagement, led by experts from different disciplines with recognition that a one-size approach will not accommodate the different uses of and contexts for genomic data within medicine.

Author Expertise and Research Background

5. This submission draws on detailed qualitative empirical research examining the ethical, practical and system-level challenges of implementing genomic medicine within the NHS. The findings are based on 24 in-depth interviews with leading UK experts, including clinical geneticists, national genomic medicine policymakers, senior academic leaders, and senior officials involved in national genomics programmes and services. This research was funded by the Wellcome Trust.

6. This independent academic research has been conducted by investigators with expertise in genomic medicine, public health, and bioethics. The views expressed are those of the authors, based upon this research, and do not represent the views of any organisation that funded or supported this research.

7. This submission is made by Dr Ingrid Slade (Principal Investigator) and Dr Glenn Simpson, who are based in the Centre for Population Health Sciences at the University of Southampton. The research was led Dr Slade, who is a medically trained public health specialist with a PhD in genetics and extensive experience in genomic medicine, public health and ethics, including leadership roles within national genomics initiatives and health and social care systems. Dr Simpson is a highly experienced qualitative methods researcher with expertise in health services research, including multimorbidity, social care and public health.

Introduction

8. Advances in genomic technologies have transformed biomedical research and hold the promise of a shift toward increasingly data-driven, personalised approaches to medicine. The United Kingdom has sought to position itself as a global leader in the integration of genomics into healthcare, supported by national strategies and infrastructure developments designed to embed genomic medicine within routine NHS care.

9. Policy frameworks, such as *Genome UK*, set out a long-term vision to create "the most advanced genomic healthcare system in the world," integrating genomics across research and clinical care to improve diagnosis, prevention, and treatment. Alongside this, the *NHS Genomic Medicine Service* has established a nationally coordinated infrastructure to deliver genomic testing and interpretation at scale, with the aim of making whole genome sequencing part of routine care and improving population health outcomes.

10. Policy developments, including the *Accelerating genomic medicine in the NHS*, further emphasise the integration of genomics across all levels of care, from population health programmes and primary care to specialist services, while prioritising equitable access, improved clinical outcomes, and the expansion of precision medicine. This is reinforced within wider NHS reform agendas, including the 10-year health plan, which highlights genomics as an enabler for more predictive, preventative, and personalised healthcare models.

11. Since the completion of the Human Genome Project, sequencing technologies have substantially reduced in costs and increased in scalability, providing the economic conditions for the integration of genomic data into routine healthcare systems (1-3). National initiatives such as the 100,000 Genomes Project and the NHS Genomic Medicine Service are born from this transition, embedding genomics within clinical care and research infrastructures at a population scale (3,4). These developments reflect the broader policy ambition to transition the NHS toward more predictive, preventative, and personalised models of care (1,5), while raising important questions regarding implementation, system readiness, and ethical oversight.

12. The move from specialist clinical genetics/genomics services to genomic data integrated through healthcare services and population-level programmes, introduces a range of complex ethical, social, and policy challenges. These include issues of data governance, consent and privacy, equity of access, and the interpretation of uncertain or probabilistic genomic risk (6-8). In addition, recent research and policy analysis highlight challenges related to public trust, data sharing, and governance of large-scale genomic datasets, particularly as genomics becomes increasingly integrated with digital health systems and artificial intelligence (9-11).

13. This submission focuses on the following questions identified by the Inquiry, although some of our responses overlap across these themes:

- Question 3: Health data research infrastructure;

- Question 5: Deployment in practice;
- Question 8: Government strategy and system design.

The submission does not directly address questions relating to the technical capabilities of artificial intelligence (AI), rather our comments are primarily focused on issues relating to the translation of genomics into the health and care system.

14. It is important to note that this research is in progress; the full data analysis will be completed by the end of May. However, we have sufficient understanding of the key themes in the data to inform this submission, and we would be happy to share additional insights if further evidence is requested by the Inquiry

Health Data Research Infrastructure (Question 3)

15. Current NHS data systems are often built around storage and data collection and not necessarily set up for clinical interpretation, data sharing and optimal use within everyday practice.

16. Trust in the use of genomic data is an enduring concern, especially around consent, privacy, data security and future data use. Given that data sharing and population-level data aggregation are key to maximising the use of genomic data within healthcare, the issues of governance, transparency and accountability and how these can be implemented with an impact on public trust, need to be carefully considered.

17. Wider public support for the collection, storage and use of genomic data within healthcare cannot be extrapolated from the rare disease context, where experts report there is a greater acceptance. Instead, it must be supported through public dialogue, clear governance frameworks, effective communication, and transparent, ethically sound decision-making processes.

18. Genomic risk prediction in common complex diseases has a far higher degree of accuracy in European populations versus other ethnicities, therefore their clinical use today would differentially benefit some populations over others and may widen existing disparities in disease prediction, diagnosis and treatment outcomes.

19. The evidence indicates that the development of genomic data infrastructure should be understood from both technical and social/ethical perspectives. Issues around data governance and management, as well as public engagement and acceptability, should also be balanced against technocratic considerations.

20. Our findings suggest that the application of genomic medicine within broader health services, such as secondary care, primary care and public health systems, requires specific translation/implementation research within those settings, which is currently lacking. As a result, there is a lack of specific implementation research to support the integration of genomics into different care pathways, potentially limiting its effective adoption beyond specialist settings.

Deployment into Practice (Q5)

Up to now, the main use of constitutional genomic testing has been in rare disease and inherited cancer predisposition diagnosis. Looking ahead, genomic data is predicted to also inform risks of both rare *and* common diseases (such as cardiovascular disease and common cancers). The transition from specialist to mainstream care and population health represents a fundamental shift in the role of genomics within the NHS. There are significant challenges to address, which are discussed below.

21. The requirement of specificity when referring to genomic medicine/data, defining the use and clinical context across different healthcare settings, was consistently emphasised in expert

responses. Participants highlighted that applications within specialist genomic services, secondary care pathways (e.g., cardiology), primary care settings (e.g., pharmacogenomics in GP consultations), and population-level programmes (e.g., screening or risk stratification), require different leadership, expertise and evidence. The blurring of these contexts, risks oversimplification, misaligned research design, ethical blind spots, unintended consequences and limited practical application of findings across the NHS.

22. Some experts expressed concern that implementation is being driven by technological capability and policy ambition, rather than patient/population need or clinical service requirement. There is a risk that the technology could be deployed too quickly and pushed too far. Currently, genomics has limited predictive accuracy in many clinical contexts, particularly outside the rare disease or inherited cancer predisposition contexts.

23. Many experts revealed that the barriers to the deployment of genomic medicine are not necessarily technological, but more often organisational, structural, and professional in nature.

24. The capacity to interpret and integrate genomic information into clinical practice remains constrained. The predictive value of many genomic variants remains uncertain; the same genetic change might mean one thing in an individual with a personal or family history and quite another in an individual from the general population. Genomic data and risk are complicated, and many clinicians outside of some specialist disciplines lack the training, confidence, or support to interpret genomic results.

25. Variation in access to genomic services was also identified as a key priority that needs to be addressed. Differences in regional and local capacity, workforce, and infrastructure development may result in patients experiencing unequal access to genomic testing and interpretation.

26. In relation to equity of access, genomic medicine has the potential to improve outcomes, but also to exacerbate inequalities if access to services is uneven or if certain populations are excluded or face challenges using services.

27. There is a need for substantive community engagement, utilising a key public health method, to ensure inclusion and reduce inequalities, so that no one is left behind.

28. Experts emphasised that genomic medicine is not a single entity, diagnostic test or intervention but encompasses a diverse range of applications across different clinical and service contexts. Greater specificity in how genomic medicine is defined and discussed is important to ensure coherent decision-making, effective evaluation and appropriate policy development.

29. Experts stressed that risk-benefit considerations at both the individual and population levels, need to be clearly thought through and assessed across all defined use-cases of genomic medicine. While genomic medicine may offer potential benefits in terms of early diagnosis, risk prediction, and targeted treatment, these benefits are often uncertain, speculative and lack evidential underpinning. On the other hand, potential harms and risks such as overdiagnosis, misinterpretation of results, and unintended consequences of a diagnosis for patients and their families, must be weighed against the potential wider population and healthcare system benefits of genomic medicine. Therefore, effective frameworks are needed to assess how risks and benefits are balanced across different contexts and populations, enabling explicit reasoning and transparency.

30. As additional uses of genomic data outside of specialist genomic medicine services, particularly within primary care and population health programmes, will become integrated into NHS commissioning, they must compete against other diagnostics and interventions within existing resource allocation decision-making priorities. Experts noted that this may lead to genomic services

being deprioritised, particularly where evidence of clinical utility and cost-effectiveness is limited, when compared with established interventions and technologies.

31. Genomics is often framed as a solution to complex health challenges and rising service demand. However, experts emphasised that complex diseases and health service demand pressures arise from a wide range of often interlinked determinants, encompassing individual biological factors, social issues such as housing, education and employment, along with broader environmental factors such as access to transport, community cohesion and the condition of local spaces (e.g., access to green spaces). Focusing solely on genomics without consideration of wider determinants of health, risks creating a disconnect between health services and the daily challenges people experience, as well as limiting complex disease impacts and population health outcomes.

32. In the context of common diseases, giving patients genomic risk information does not ensure health behaviour change. Achieving personalised prevention and a reduction in demand on health services requires genomic risk to be acted upon by individuals. Experts discussed that genomics is an additional piece of risk information and that further research is needed to understand how we can effectively support long-term sustainable health behaviour change in response to genomic information.

The Government's Strategic Approach to Innovation in the NHS (Q8)

33. Genomic medicine relies on a degree of centralisation. Large-scale data aggregation and standardisation are necessary for both interpretation and efficiency. However, healthcare is often delivered at local and regional scales, and the experts recognised a potential mismatch in scales of service delivery. Public support and patient trust in healthcare systems are often grounded in local relationships, particularly between patients and clinicians. The findings suggest that an effective strategy must balance these competing demands between centralisation and local delivery.

34. Experts in clinical genetics are, in the main, leading and governing this area of policy and implementation. However, many experts stressed the need for genomic medicine policy and implementation to be democratised, to include broad representation of clinical, public health and patient/public expertise.

35. Our work highlighted the need to manage and explicitly balance 'trade-offs' inherent in the implementation of genomic medicine. Experts emphasised that decisions often involve balancing competing priorities, such as clinical benefit, cost-effectiveness, and equity considerations. Therefore, these trade-offs need to be considered through reasoned decision-making based on agreed principles.

Conclusion

36. Implementation, not technology, is the critical challenge. The primary barriers to realising the benefits of genomic medicine within the NHS are organisational, structural and workforce-related rather than technological, requiring sustained attention to system design, governance and service readiness.

37. Public trust and governance remain foundational to success. Effective, transparent governance frameworks, alongside meaningful public and community engagement, are essential to maintaining trust and legitimacy, particularly as genomic data use expands beyond specialist settings into mainstream care and population health.

38. There is a real risk of exacerbating health inequalities. Without deliberate action, genomic medicine may disproportionately benefit some populations while excluding others, especially given

current limitations in data representativeness, variability in service capacity and uneven access across localities.

39. Policy ambition must be matched with clinical evidence and system capacity. Accelerating the deployment of genomic medicine ahead of evidential maturity, workforce capability, and infrastructure readiness risks unintended harms, ethical blind spots and inefficient use of NHS resources.

40. A differentiated, system-wide approach is required. Genomic medicine encompasses diverse applications across specialist care, primary care and population health, each requiring context-specific evidence, leadership and evaluation. A single uniform model of implementation is neither realistic nor appropriate.

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