

Public and Healthcare Professional Perspectives on Population-Level Genomic Testing

Summary Report

BY FAVA HEALTH
FOR GENOMICS ENGLAND

OCTOBER 2025



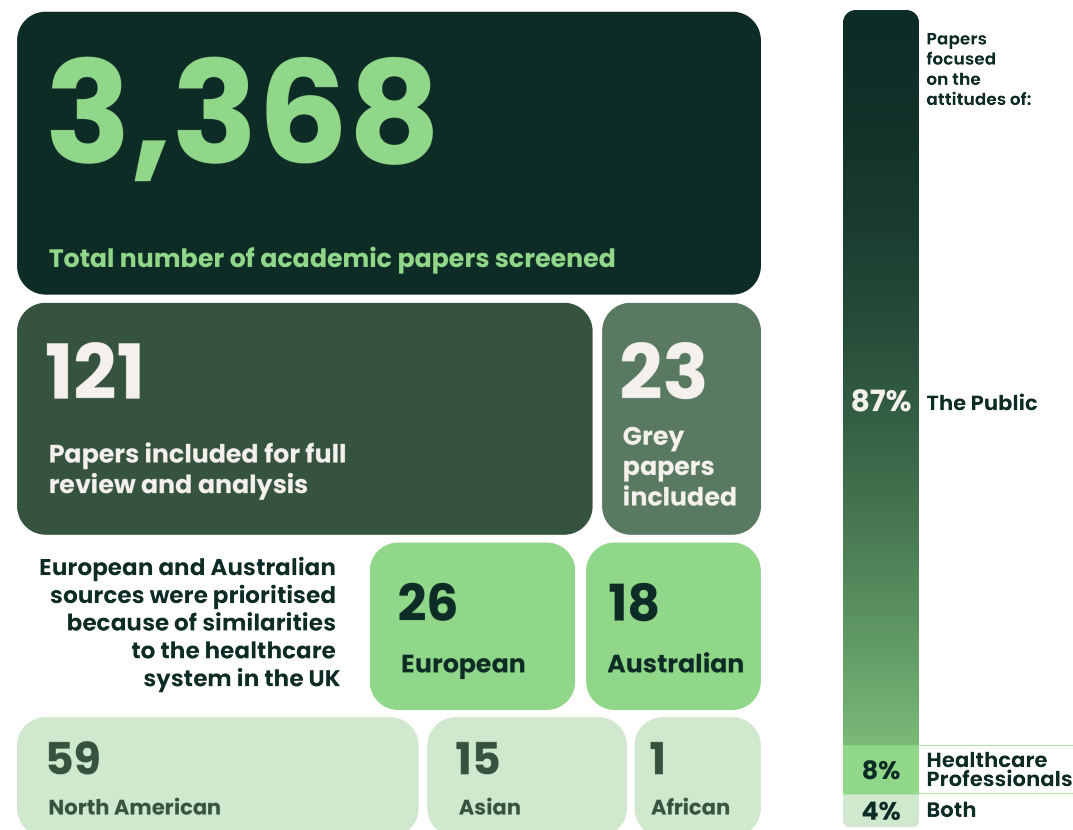
I. Introduction

Genomics England is embarking on a transformative initiative focused on integrating genomics into routine preventative healthcare for adults. The success of such an ambitious “adult population genomics” programme (hereafter referred to as the “Programme”) hinges not only on technical and clinical feasibility but, crucially, on building strong public trust, ensuring clear governance, and aligning with the expectations of both the public and healthcare professionals (“HCPs”). Understanding these perspectives from the outset is paramount to designing an ethical, inclusive, and impactful programme.

This summary report, based on the comprehensive Literature Review on Public and Healthcare Professionals Perspectives on Population-Level Genomic Testing* (hereafter referred to as “The Literature Review”), aims to distil the vast body of existing evidence into actionable insights and guide next steps. It outlines what we currently know about public and HCP attitudes, concerns, and motivations regarding population-level genomic testing. More importantly, it clearly identifies critical evidence gaps, framing them as challenge statements aimed to help design targeted follow-on work. Finally, it provides strategic recommendations to inform Genomics England’s immediate engagement plans and future programme design.

* Delivered by Fava Health in October 2025.

A Summary of the Literature Reviewed



II. What we already know



The Literature Review synthesised current evidence on public and healthcare professional perspectives on population genomic testing in adults. Consistent themes emerged across Mendelian risk (rare variants with significant impact), polygenic risk (the combined effect of many common variants), and pharmacogenomic risk (genetic factors impacting medicine response), and were structured using the Health Belief Model as an analytical framework.

The literature demonstrates that there is clear enthusiasm for the clinical and personal opportunity of population-level genomics, but this is balanced by consistent barriers that must be addressed to ensure successful adoption. Public and professional engagement will depend on building genomic literacy, providing clear and accessible communication, and embedding robust clinical support. Trust in healthcare professionals as credible messengers, equitable service design that meets the needs of diverse users, and practical integration into everyday workflows are all critical for success. Addressing these factors will be central to creating a genomic programme that is effective, inclusive, and sustainable. The following section explores each of these themes in more detail.

WHAT WE ALREADY KNOW

- 1. Clinical and personal benefits are strong motivators**
- 2. The critical role of trusted messengers and support pathways**
- 3. There are proven barriers to widespread adoption**
- 4. The importance of user-centred service design**
- 5. Addressing genomic literacy and knowledge gaps**

1. Clinical and personal benefits are strong motivators

-   Both the public and HCPs see significant value in population-level genomic testing for prevention, early intervention, and personalised health management, with healthier lifestyles, better monitoring, and more targeted care seen as key drivers of engagement.
-  Interest in testing grows when results lead to clear and actionable steps, such as prevention or treatment, with many perceiving genomics as empowering by reducing uncertainty and offering greater control over health decisions.
-  Personal and familial relevance strongly shapes motivation, with individuals more likely to engage when there is a direct connection to disease. Similarly, past negative experiences with medication emerged as key driver of interest in pharmacogenomic testing.

2. The critical role of trusted messengers and support pathways

-  HCPs are consistently identified as highly trusted sources of advice, and their endorsement strongly shapes public decisions to take part in genomic testing. Their role as credible messengers is significant to build trust and buy-in.
-  The public widely acknowledges that they would need professional guidance — particularly from their GP or another trusted clinician — to interpret complex genomic results and make informed choices about their care.
-  Testing programmes are expected to provide robust clinical support and follow-up, both to address patient anxiety and to help manage uncertainty around ambiguous or low-penetrance results, which many HCPs see as a key challenge.

KEY

Public



Healthcare Professionals (HCPs)

3. There are proven barriers to widespread adoption

-  Practical barriers within healthcare systems, such as limited consultation time, insufficient training, and uncertainty about how to integrate genomics into workflows, reduce HCP willingness to engage. Many also worry about the psychosocial impact on patients, such as increased anxiety or confusion.
-  Both the public and HCPs face significant knowledge gaps and low genomic literacy, particularly around complex areas such as polygenic and pharmacogenomic data. Many HCPs feel unprepared and often view genomic communication as a specialist responsibility rather than part of routine care.
-  Concerns about privacy, discrimination, and data misuse remain strong, with fears of insurance or employment consequences undermining trust. A lack of transparency and limited public awareness further reinforce these anxieties.
-  For the public, perceived cost is a major deterrent, with participation more likely when testing is free. Access disparities also persist, particularly among people with lower educational attainment or non-English-speaking groups.
-  Both HCPs and the public struggle to recognise the broader societal benefits of genomics in prevention and public health. Because these benefits are collective, delayed, and incremental, they often feel too distant to motivate participation.
-  Ethical considerations, data governance, and equitable access are seen as paramount, with both the public and HCPs voicing concerns about privacy, data misuse, and genetic discrimination. These worries are particularly acute around commercial involvement and represent a major barrier to trust and engagement.
-  Public health professionals and policy makers highlight concerns about resource allocation and the risk of genomics worsening health inequity, with unequal access and underrepresentation of key communities seen as both a social justice issue and a scientific risk.

4. The importance of user-centred service design

- ✚ HCPs emphasise the importance of workflow compatibility, favouring efficient turnaround times, clear reporting, and seamless integration into Electronic Health Records. When services feel cumbersome or poorly aligned with existing workflows, uptake and effective use are significantly reduced.
- 👤 Members of the public show strong preferences for non-invasive sampling methods such as cheek swabs over blood tests, citing both convenience and phlebotomy access issues. They also value the ability to access results digitally, provided this is paired with professional guidance and support.
- 👤 The public demonstrates a strong preference for genetic testing to be integrated within a clinician-prescribed model, showing caution towards direct-to-consumer genetic testing.

5. Addressing genomic literacy and knowledge gaps

- ✚ 👤 Low levels of genomic literacy remain a major barrier for both the public and HCPs. Members of the public frequently report limited understanding of genetic testing, genomics, or the role of DNA in health and drug response, and lack confidence in interpreting results without professional support. These challenges are particularly acute for complex areas such as polygenic and pharmacogenomic data.
- ✚ 👤 Clear and accessible communication is consistently highlighted as critical, with simplified, jargon-free explanations and resources seen as essential for building public confidence and understanding. For HCPs, many outside clinical genetics feel unprepared and often regard genomic communication as a specialist responsibility. Practical education and support are therefore key, with training, hands-on experience, and decision-support tools recommended to build knowledge, confidence, and self-efficacy in routine care.
- ✚ 👤 The concept of “additional findings” is also not well defined in the context of population genomics, and there is little evidence on how people understand or value such results. Traditionally, additional findings refer to results unrelated to the original reason for testing, but in a healthy population almost any result could fall into this category. What may seem irrelevant at the time of testing could become actionable in the future. This highlights the importance of co-designing pre-test information with participants and clarifying — or even re-defining — what counts as additional or actionable within a population programme.

III. What we don't know

EVIDENCE GAPS AND DESIGN CHALLENGES

While the current literature provides a good foundation and our review has identified valuable key insights, several critical gaps remain in our understanding, particularly within a UK context. These represent strategic opportunities for Genomics England to generate the evidence needed to engage the right communities, in the right way, to design an effective and equitable programme. These opportunities were identified through a Theory of Change Framework, which is described in detail in The Literature Review and framed as challenge statements with potential approaches to addressing them.

What we mean when we say...

Challenge statement:

A critical gap or unresolved question in the evidence base that needs to be addressed to ensure the Programme is effective, equitable, and trusted.

Potential approaches:

Illustrative ways such a challenge might be explored or addressed, offering starting points for engagement, research, or design activities.

WHAT WE DON'T KNOW

1. Understanding non-genetic healthcare professional views
2. Engaging underserved and marginalised communities
3. Understanding the longitudinal value of genomic testing
4. Evaluating effective engagement strategies in practice
5. Designing for user experience and interaction with a population genomics service

1. Understanding non-genetic healthcare professional views

Challenge statement:

The current literature significantly under-represents the perspectives of all HCPs and especially 'non-genetic' healthcare professionals (e.g., GPs, nurses, pharmacists, public health practitioners). How can we effectively engage these professional groups to understand their specific attitudes, concerns, existing workflows, and educational needs regarding population-level genomic testing? This knowledge is critical for designing a Programme that integrates into and adds value for routine preventative care, rather than being seen as a burdensome, 'add-on' specialist service.

Potential approaches:

Engagement and participatory co-design with primary care, nursing, and pharmacy communities — for example through professional networks, ethnographic observations, field interviews, or co-design workshops — could help uncover barriers, desired support mechanisms, and preferred models for integrating genomics into routine care.

2. Engaging underserved and marginalised communities

Challenge statement:

The existing literature, particularly UK-focused studies, often suffers from selection bias, predominantly capturing views from healthy, white, and university-educated populations. This means those who could most benefit from the Programme, especially underserved or marginalised communities, are not well represented. How can we actively and meaningfully engage these communities to understand their specific needs, attitudes, cultural considerations, and barriers to participation, ensuring the Programme is truly equitable and inclusive by design?

Potential approaches:

Targeted, community-based engagement in partnership with trusted organisations and existing programmes — for example, focus groups, town halls, or in-community workshops — could help surface culturally and linguistically relevant insights from populations historically underrepresented in genomic research and healthcare.

3. Understanding the longitudinal value of genomic testing

Challenge statement:

Most studies present genomics as a single-point intervention, focusing on initial attitudes towards a specific test, rather than as a 'lifetime genome' — a continuous relationship with evolving potential value over many years. How do public and HCP preferences, understanding, perceived impact, and value of genomic information change over time, especially as new evidence emerges or health needs change? What engagement models are required to support individuals and professionals through this longitudinal journey?

Potential approaches:

Longitudinal engagement methods — such as diary studies, serial focus groups, or cohort follow-up — could provide insights into how attitudes, preferences, and perceived value shift over time, helping to continue to evolve the design of a Programme that supports people and professionals throughout their life.

4. Evaluating effective engagement strategies in practice

Challenge statement:

While the literature reviewed highlights what people think, it rarely evaluates what effective engagement actually looks like in the context of population genomics. We lack real-world evidence on the impact of different engagement strategies. How can Genomics England assess the effectiveness of various health engagement approaches, including the role of branding, health promotion campaigns, and diverse communication channels, to guide the delivery of impactful engagement activities for the Programme?

Potential approaches:

Pilot projects and 'minimally viable' evaluations of diverse engagement strategies — for example, comparing communication channels, branding approaches, or campaign styles — could generate early practical evidence on what works best to build awareness, trust, and informed participation prior to at scale investment.

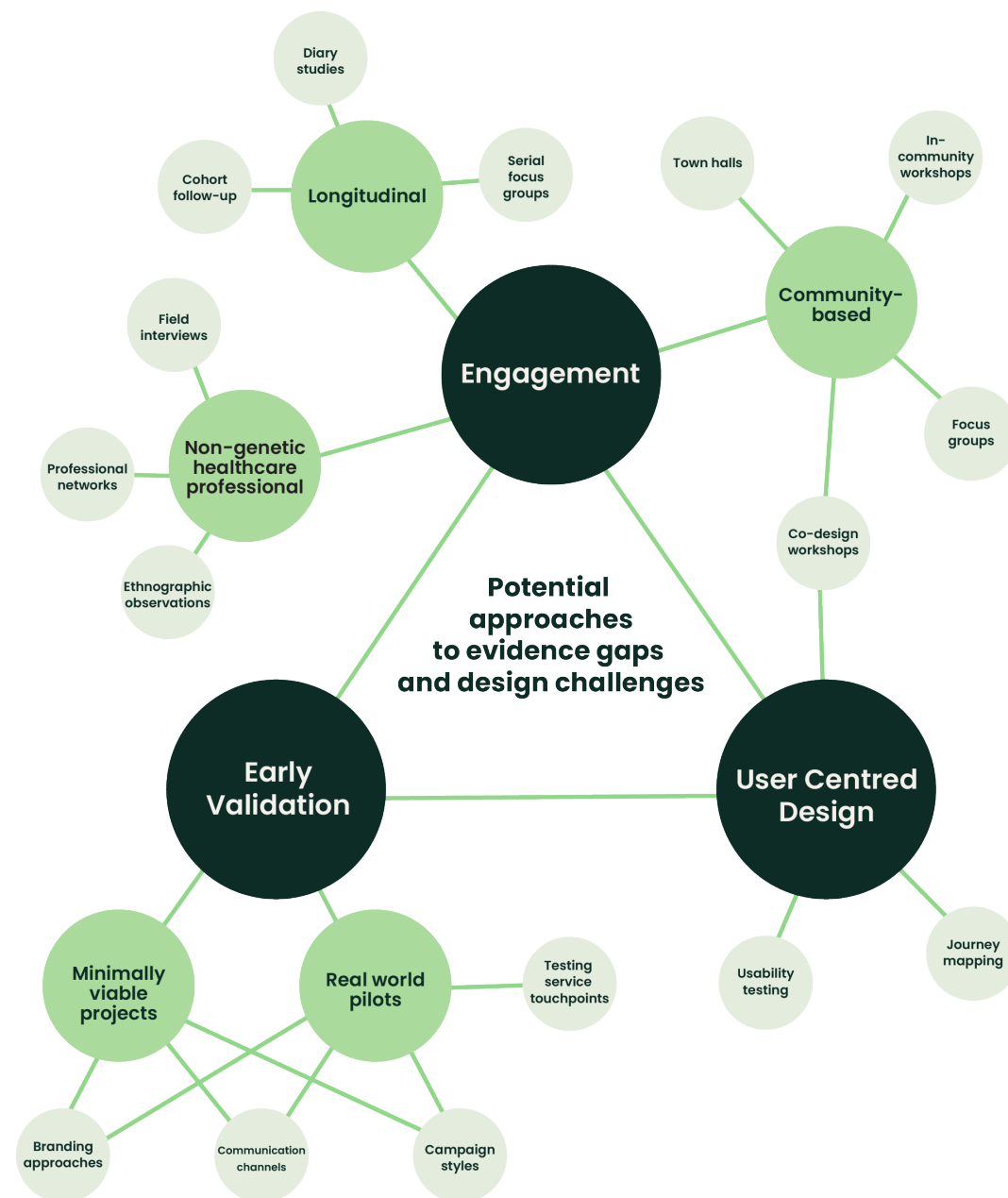
5. Designing for user experience and interaction with a population genomics service

Challenge statement:

Much of the existing literature focuses on attitudes towards health outcomes or risk, rather than directly addressing how users wish to interact with the service design and delivery of population genomic programmes themselves. How can we formalise user-centred service design methods to understand users' desired interaction points, preferred service pathways, and practical requirements for the Programme journey, ensuring it is built around their needs and expectations?

Potential approaches:

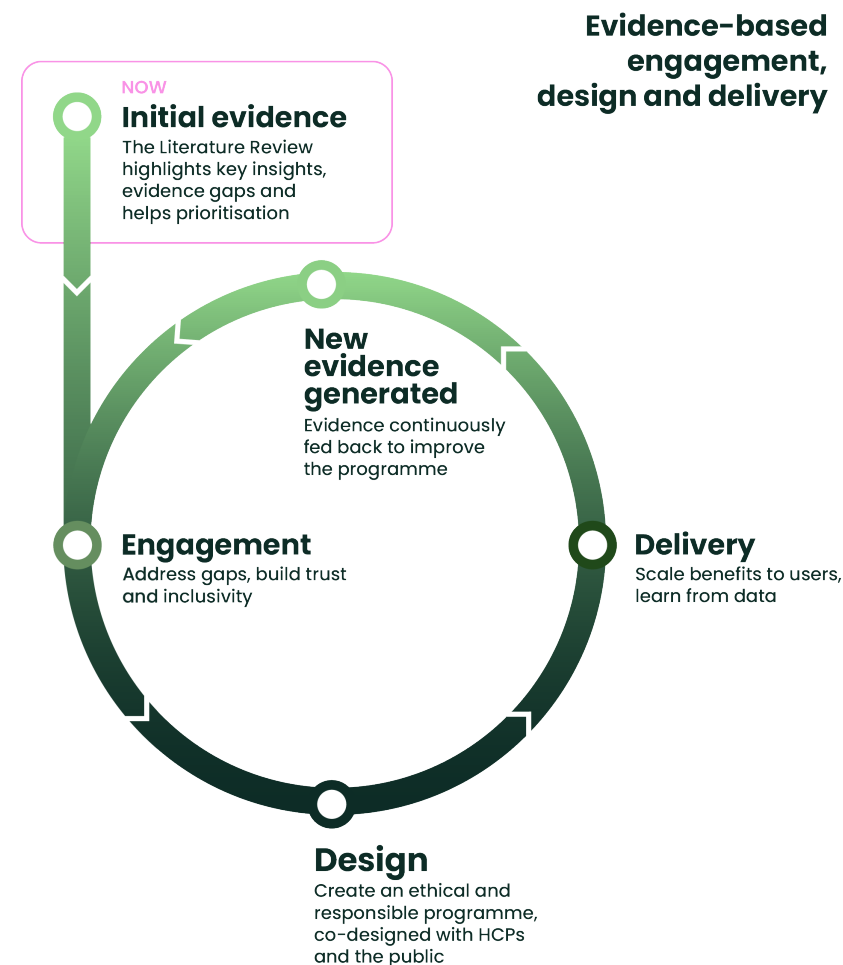
User-centred design activities — such as journey mapping, co-design workshops, or usability testing with diverse groups — could help uncover preferred pathways for recruitment, consent, sample collection, results delivery, and follow-up, ensuring the programme is intuitive, accessible, and responsive to user needs.



IV. Looking ahead

The evidence gathered in this review makes clear that the success of an “adult population genomics” programme hinges on building trust, inclusivity, and meaningful engagement. Public and professional perspectives provide both encouragement and challenge: there is strong enthusiasm for the promise of genomics to shape healthier futures, alongside clear reminders of the ethical, practical, and social responsibilities that must guide programme design.

This strategic briefing has highlighted the most immediate insights for action, while the full Literature Review provides further detail and nuance to support Genomics England as it moves forward. Together, these resources offer a strong foundation for creating a programme that is not only clinically effective but also socially responsive and globally leading. Looking ahead, the opportunity is to transform these insights into a national initiative that inspires confidence, empowers individuals, and ensures the potential benefits of genomics are realised at scale.





Thank you.

© FAVA HEALTH LTD.

info@favahealth.com
www.favahealth.com