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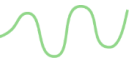
Public and healthcare professional perspectives on population-level genomic testing *A Literature Review*

A REPORT PRODUCED BY FAVA HEALTH
ON BEHALF OF GENOMICS ENGLAND

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01 Executive summary



This report presents the findings of a literature review on public and healthcare professional (HCP) perspectives on population-level genomics, highlighting insights for effective engagement strategies for Genomics England's Programme to explore the use of genomics for preventative care in adults.

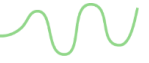
We found that both the public and HCPs see significant benefits from genomic testing, largely driven by the opportunity for prevention, early intervention, and personalised health management. The public's interest increases with perceived personal or familial disease links and the availability of clear, actionable results. Similarly, HCPs recognise the value in identifying high-risk individuals and informing preventive care.

However, considerable barriers exist. For the public, these include cost, complexity of information, privacy concerns, and fears of discrimination or anxiety. Many also lack confidence in understanding results without professional support. HCPs face major barriers due to insufficient genomic knowledge, lack of training, limited time, and concerns about psychosocial impacts or integrating testing into clinical workflows. They often view detailed genomic communication as a specialist role.

Key insights

- Communication around genomics is often complex, and both the public and HCPs report challenges in understanding polygenic and pharmacogenomic data without professional support.
- Many healthcare professionals, particularly GPs, highlight the need for targeted, practical training and digital tools to build their confidence in using genomics in practice.
- Wrap-around clinical support and follow-up are seen as essential to address anxiety and ensure participants can make informed decisions.
- Service design strongly influences uptake, with HCPs preferring efficient turnaround times and EHR integration, and the public showing clear preferences for non-invasive sample collection.
- Concerns about privacy, discrimination, and data misuse remain significant barriers, with transparency about data use and protection critical to building trust.
- Clinicians are trusted messengers whose endorsement strongly shapes uptake, and their role could be reinforced by equipping them with the right communication resources.

02 Introduction



2.1 Background and context

Genomics England is embarking on an ambitious population-level genomics programme aimed at building the evidence and digital infrastructure for the next big shift in genomic healthcare – its routine use in preventative care in adults. Year one of such an ‘adult population genomics’ programme (hereafter referred to as ‘Programme’) is focused on public engagement, design, and proof-of-concept, with a view to consciously create a programme built on trust, transparency, and collaboration with patients, the public, and healthcare professionals.

To support this ambition, Genomics England commissioned Fava Health to deliver a structured literature review exploring public and healthcare professional perspectives on population-level genomic testing in adults. The aim of this work is to understand the existing evidence base and generate insights to help shape Genomics England’s engagement strategy for the programme. In particular, it identifies what is already known about attitudes, concerns, expectations, and motivations around population genomic testing, and highlights where gaps in understanding remain.

Embedding genomics into routine healthcare requires technical and clinical feasibility, but equally, strong public trust, clear governance, and alignment with public and healthcare professional expectations. The successful integration of population-level genomic testing for preventative care depends on anticipating and addressing the concerns and needs of all stakeholders.

This report provides a comprehensive synthesis of peer-reviewed and grey literature, structured into key insights, evidence gaps, and recommendations. The findings are intended to inform Genomics England’s public and clinical engagement strategy for 2025/26 and to support wider strategic decision-making for the Programme.

2.2 Project aim

This project aims to conduct a structured literature review of public and healthcare professional perspectives on population-level genomic testing in adults. The objective is to synthesise current knowledge about attitudes, concerns, expectations, and motivations—including around the return of results—and identify evidence gaps. The review will generate actionable insights and recommendations to inform Genomics England's engagement strategy for this programme, ensuring it is evidence-based, ethical and inclusive.

2.3 Scope and definitions

To ensure clarity and consistency in our analysis, we have defined key concepts that frame this literature review. These definitions guided our search strategy, inclusion criteria, and thematic analysis..

Table 1: key concepts and their applied definitions

Concept	Applied definition
Population level	Initiatives designed to improve health outcomes across entire populations or specific subgroups, rather than focusing on individuals.
Return of results	The process by which genomic test results are communicated to individual participants or to the healthcare system or clinicians involved in their care. This includes personal health-related findings, secondary or incidental findings, and considerations such as content, timing, method of delivery, and clinical implications of disclosure.
Attitudes	The beliefs, feelings, and predispositions that individuals or groups hold toward aspects of a programme—such as its goals, methods, or outcomes. This includes cognitive, affective, and behavioural components.
Adults	Individuals 18 years of age or over.
Public	The broad population or community that the programme is designed to serve. This may include patients (i.e., those with a disease or experiencing healthcare) but not exclusively.

Healthcare Professional	Individuals who are trained to provide care and who are being asked to consider their involvement or interaction with the genomic testing programme.
Healthcare Stakeholders (Non-Clinical)	Individuals involved in the governance, management, or policy aspects of healthcare, including those who influence or oversee the implementation of genomic testing but are not directly involved in clinical care.
Genomic Testing	Any methodology which results in the generation of genetic or epigenetic data, undertaken within a healthcare system or accessed directly by consumers.
Trust	The confidence that individuals or groups have in the integrity, competence, and intentions of those delivering or governing genomic testing programmes.
Genomic Literacy	The degree to which individuals have the knowledge, skills, and confidence to understand, interpret, and use information related to genomic testing. This includes understanding of genetic risk, inheritance patterns and limitations.
Data Literacy	Understanding of how genomic and healthcare data is collected, stored, shared, and governed, including awareness of privacy protections, data rights, and regulatory frameworks.
Engagement	The ways in which individuals or groups are involved in, consulted on, or contribute to the design, delivery, or oversight of genomic programmes. This may include passive information provision, active participation, or co-design activities.
Equity	The fairness and inclusivity of programmes in addressing the needs of different population groups, particularly historically underrepresented or disadvantaged in healthcare or research.
Informed Consent	The process by which individuals are provided with comprehensive information about genomic testing, including its purpose, procedures, risks, benefits, and alternatives, enabling them to make voluntary and informed decisions about participation.
Preventative Care	Healthcare services and interventions aimed at preventing disease, detecting conditions early, or reducing risk factors before symptoms appear or conditions worsen.

03 Methodology



3.1 Overview

A structured literature review was undertaken to summarise the existing evidence related to public and healthcare professional perspectives on population-level genomic testing. A tiered multi-modal search strategy was used to systematically identify publications of interest, including Boolean search, bi-directional citation search, and structured site search of grey literature sources. This was followed by evidence synthesis via thematic analysis using the Health Belief Model (HBM) as the theoretical framework, and quality appraisal using the Critical Appraisal Skills Programme (CASP) Qualitative Studies Checklist.

3.2 Inclusion and exclusion criteria

Studies were included if they presented original research published in peer-reviewed journals or findings published in grey literature that described public or healthcare professional attitudes or perspectives towards population-level genomic testing. The broader definitions of "population level", "genomic testing", and "attitudes" used for this analysis are defined in Table 1. No restrictions were placed on study design or geographical location to ensure comprehensive coverage of available evidence.

Systematic reviews, meta-analyses, and opinion pieces were excluded from the final analysis. However, original studies referenced in identified systematic reviews were captured through hand-searching of reference lists and assessed for inclusion. Conference abstracts and dissertations were excluded due to limited methodological detail and lack of peer review, while non-English language studies were excluded due to limited accessibility.

3.3 Literature search strategy

3.3.1 Boolean search

The literature pool was identified via a pragmatic Boolean search performed on 1 July 2025 via Web of Science, MEDLINE (via OVID), Embase (via OVID), and PubMed Central. The search function was defined using predefined concepts and definitions with overlapping

terms (Table 1). A date limit of 1 January 2010 was chosen to ensure comprehensive coverage of literature which remains relevant to current practice, considering that this date approximately coincides with the emergence of the availability of next generation sequencing technologies in clinical practice.

All identified citations were entered into Covidence, a collaborative systematic review tool. In Covidence, for each article, preliminary relevance was determined by review of the title and abstract. A full manuscript review was then undertaken for all of these preliminary manuscripts to determine the final literature pool. Both the preliminary and full text reviews were undertaken independently by two authors. Where there was disagreement, a third author would act as adjudicator. Inter-rater reliability was assessed via Cohen's Kappa (κ) statistic.

3.3.2 Bidirectional citation search

A secondary bidirectional citation search strategy was then used to ensure the evidence pool was fully saturated. This literature searching strategy provided a systematic approach to maximise the identification of relevant articles in interdisciplinary topics, such as population-level genomics, using a starting pool of five "initial pearls" identified from the Boolean search strategy. Articles were chosen as initial pearls based on their breadth and quality, determined in part by pre-existing awareness of the literature and ensuring balance between articles describing research in primary and secondary care settings.

This approach involved both backward and forward citation tracking. Backward citation searching involved reviewing the reference lists of key articles to identify earlier studies relevant to public and healthcare professional perspectives on adult population-level genomic testing. Forward citation searching involved identifying more recent publications that had cited these key articles, using Scopus. This approach enabled the identification of both foundational and emerging literature, ensuring a more comprehensive and contextual understanding of the evidence base, particularly in areas where terminology may vary or relevant studies may be dispersed across disciplines

3.3.3 Grey literature search

To develop the grey literature search strategy, we undertook an iterative process of refining key concepts and search terms relevant to public and healthcare professional perspectives on population-level genomic testing. Initial broad terms such as "genomics" and "genetics" were combined with synonyms capturing stakeholder views, including "attitudes," "views," and "perspectives." A list of grey literature sources was developed in

collaboration with the Genomics England team and included Think Tanks, Research Institutes, and NGOs as well as public bodies and government organisations. A full list of sources can be found in the supplementary materials.

Given the dispersed nature of relevant documents across multiple official organisational websites, we employed Google's site-specific search functionality to systematically identify pertinent grey literature. We deliberately kept the search broad to account for the likelihood that relevant information might not always be explicitly labelled or easily discoverable; a tighter search risked overlooking valuable content embedded within a variety of documents and formats. This approach enhanced the precision and relevance of results while maximizing capture of diverse reports, policy documents, and engagement materials.

The final search string for Google site search was: site:genomicsengland.co.uk ("genomics" OR "genetics" OR "genomic testing" OR "genetic screening") AND (attitudes OR views OR perspectives OR perceptions OR opinions OR beliefs OR acceptance OR trust).

3.4 Data extraction and management

Data extraction was conducted using a predefined, structured template designed to capture key information from each included study. This template was aligned with the study objectives and included fields for study characteristics (such as geography, publication year, and methodological design), population details (including sample size, demographics, and healthcare context), and relevant findings related to genomic testing, public and healthcare professional attitudes, and engagement strategies.

We used a semi-automated, prompt-based approach aligned with the study protocol, key concept definitions and thematic framework. Full-text PDFs of all eligible articles were downloaded and renamed using the first author's surname and year of publication. We then developed three structured prompts using Google AI Studio, reflecting the study aims, inclusion criteria, and HBM model constructs. These prompts were designed to: (1) confirm article eligibility, (2) extract relevant data, and (3) capture CASP checklist items. Each article was uploaded individually to Google's NotebookLM tool, where the prompts were applied. Extracted content was then reviewed for accuracy and copied into a structured Google Sheet for further synthesis and analysis. Extracted data from each study was independently reviewed, with ongoing checks to ensure consistency and accuracy across included studies. This approach enabled an efficient extraction process while maintaining transparency and fidelity to the predefined protocol.

3.5 Thematic analysis

For thematic analysis, we drew from the Health Belief Model (HBM) of behaviour change. The HBM is a conceptual model and psychological theory developed to explain and predict health behaviours by focusing on individuals' attitudes and beliefs. It is considered highly applicable for studying responses of individuals to new health interventions, such as population-level genomic testing.

Themes were developed from the constructs within the HBM, adapted depending on whether the study investigated public or healthcare professionals' attitudes. Several questions were developed for each, which were used to extract data (Table 2).

Table 2: HBM themes and key constructs

HBM Themes	Public Constructs	HCP Constructs
Perceived Susceptibility	- Do they believe genomics contributes to health risk? - Do they see genomics as relevant to their own health? - Do they perceive genetic risk in the same way as other health risks?	- Do they believe genetic variation can inform the risk of disease and/or be used to guide treatment? - Do they view genomic testing as broadly applicable or only for high-risk groups?
Perceived Severity	- How serious do they think the role of genomics is in their health risk? - Do they fear the implications of health risks identified through genetic testing? - Do they believe early detection of risk matters?	- How do they perceive the clinical impact of identifying risk? - Do they believe genetic risk and/or diagnoses affect outcomes or management plans?
Perceived Benefits	- Do they believe testing helps prevent or manage disease? - Does it help them feel more informed or empowered?	Do they believe testing improves care quality or outcomes?

Perceived Barriers	- Concerns about data privacy, discrimination, or stigma? - Do they feel confident understanding results? - Do they trust the system offering the test?	- Concerns about time, resources, or workload? - Do they feel confident interpreting and explaining results? - Are there systemic or ethical concerns?
Cues to Action	- Have they received prompts (e.g., media, GP, family history)? - Was uptake reactive or proactive? - Difference of uptake across different groups	- What prompts them to offer testing (e.g., guidelines, alerts)? - Would testing be driven by patient demand? - Does genomics reduce further health needs?
Self-Efficacy	- Can they understand results and make informed decisions? - Are they able to navigate follow-up or seek help?	- Do they feel sufficiently trained and competent? - Are they confident in counselling and follow-up processes?

3.6 Focus of analysis

Based on priorities discussed with the Genomics England Engagement team, this report focuses on areas of the literature that provide the highest value insights to inform engagement strategies and decision-making. In line with this, we prioritised UK, European, and Australian studies and excluded literature on direct-to-consumer genetic testing. Studies were further categorised by the type of genetic risk they explored: 1) Mendelian (rare variant), 2) Polygenic (common variant) and 3) Pharmacogenomic (Table 3).

Table 3: risk types and applied definitions used to report preliminary findings.

Risk type	Applied definition
Mendelian (rare variant)	Genetic risk caused by a single rare variant that can significantly increase the chance of developing a condition (e.g. BRCA1/2 and breast/ovarian cancer)
Polygenic (common variant)	Genetic risk based on the combined effect of many common variants, each with small effect, used to estimate a person's overall risk for common diseases (e.g. heart disease, diabetes)
Pharmacogenomic	Genetic risk that affects how the body responds to certain medicines, used to guide safer and more effective prescribing (e.g. avoiding side effects or treatment failure)

04 Results

4.1 The literature pool

The boolean search retrieved 1,049 unique records, all of which underwent screening via abstract and title review. 136 records were screened via a full text review, leading to 90 records being added to the final literature pool from the boolean search (Figure 1). The inter-rater reliability showed strong agreement with a Cohen's kappa statistic of 0.84.

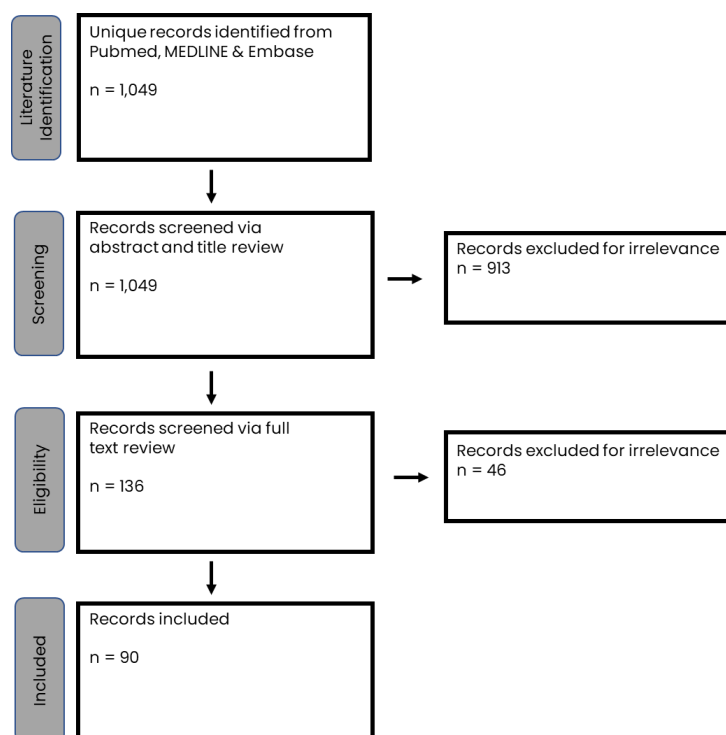


Figure 1. Boolean search strategy

Five of the articles identified in the boolean search were chosen as initial pearls for the bidirectional citation search (BDCS). Five screening rounds were required to complete the bidirectional citation search which involved the review of 2,319 abstracts. A further 31 relevant publications were identified for inclusion in the final literature pool (Figure 2). Using both search approaches, the final literature pool for analysis was 121 manuscripts.

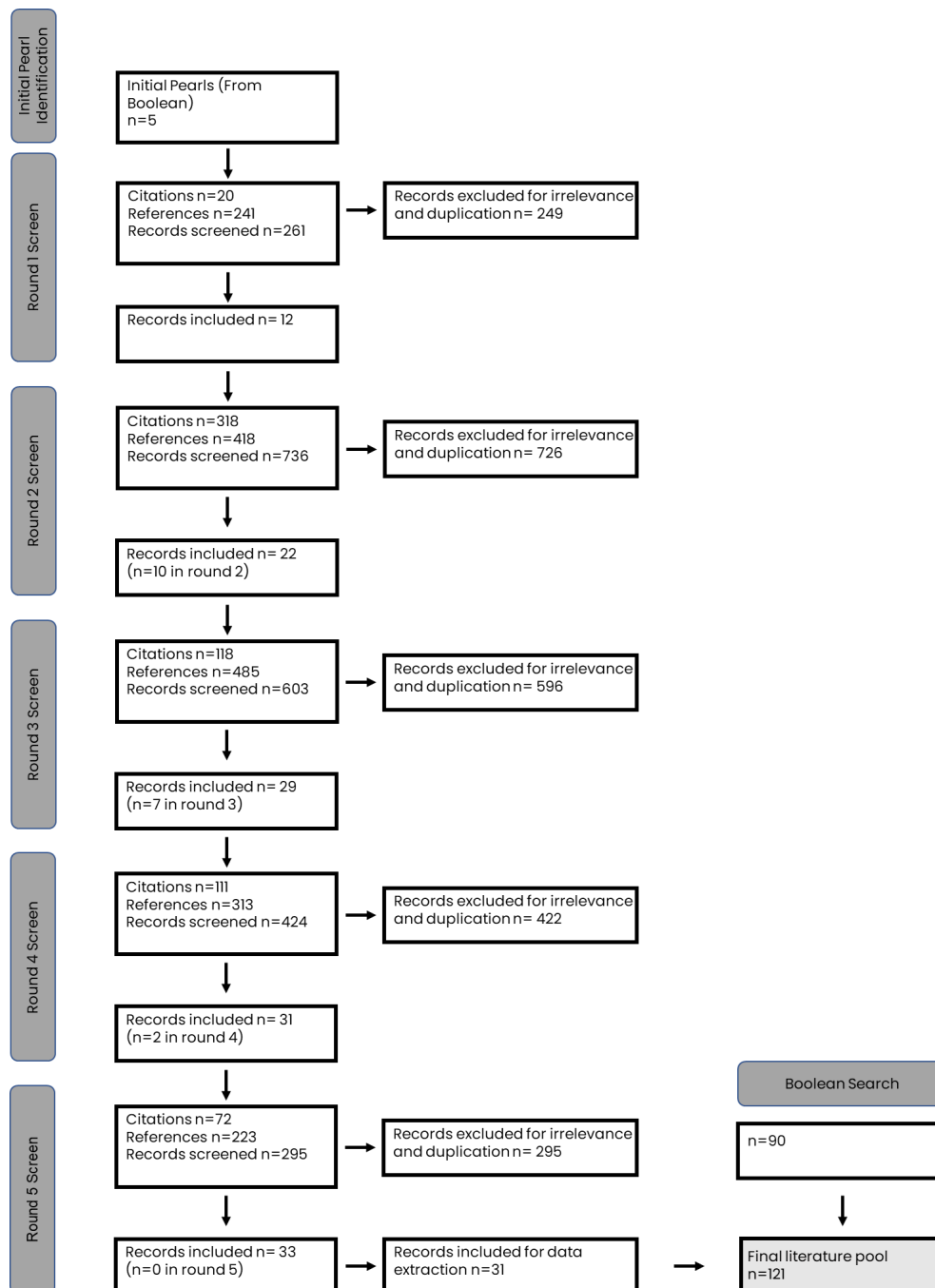


Figure 2. Bidirectional citation search and final literature pool.

4.2 Characteristics of the literature

The maximum number of publications in a single year was 11 (2017), the minimum was 3 (2022). Despite an increasing interest in population genomics from global health systems, there was no observable increase in the annual number of publications which met the inclusion criteria in this literature search (Figure 3).

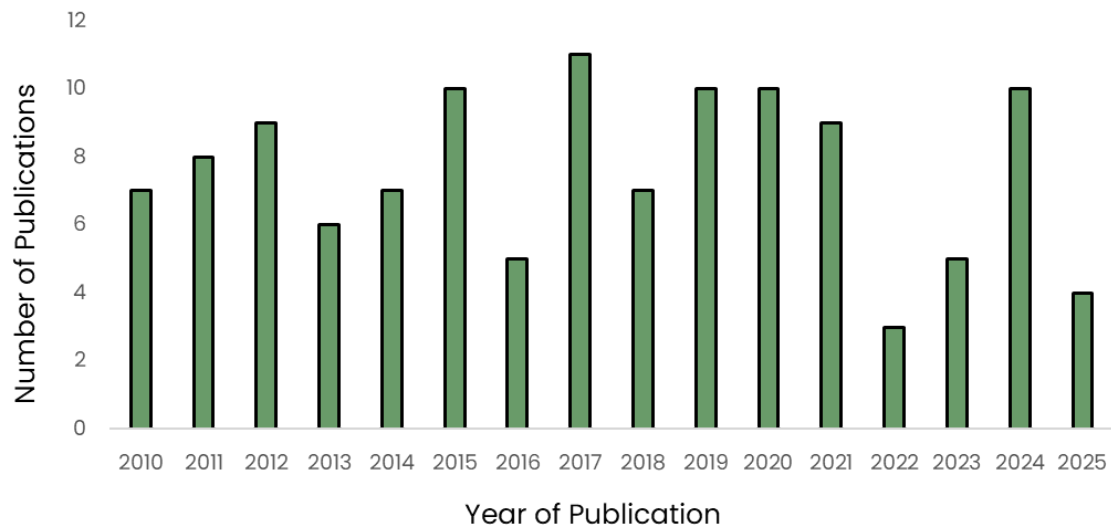


Figure 3. Publications per year

The majority (87%) of publications were focussed on the attitudes or views of public stakeholders, with only 8% specifically addressing the needs or requirements of healthcare professionals (Figure 4A). A small number (4%) of manuscripts considered the views of both healthcare professionals and the public in the same article, but not necessarily using identical methodologies.

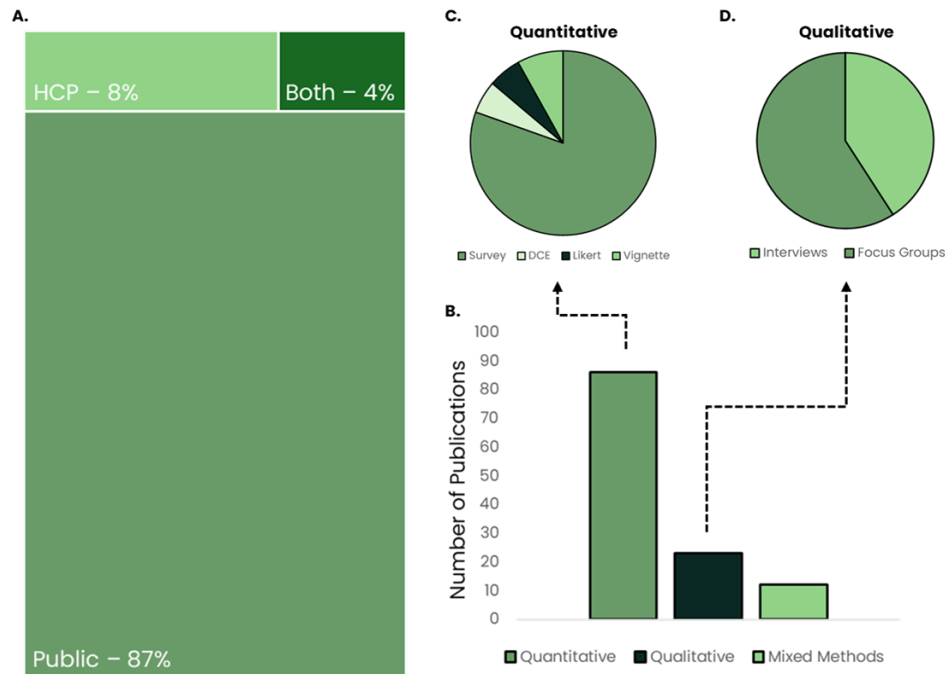


Figure 4. A) The proportion of studies which addressed healthcare and professional stakeholders views. B) The types of studies used, with a breakdown of detailed methodological approach for C) quantitative and D) qualitative studies.

Quantitative approaches were common, with over 80% of manuscripts making use of these approaches (Figure 4B). Surveys were the most frequently used quantitative tool, but a number of other sophisticated methodological designs were identified in the literature including discrete choice experiments and vignette studies (Figure 4C). In the 20% of manuscripts where qualitative methods were deployed, both interview and focus groups were described (Figure 4D).

Literature was identified from 19 countries in total, with a large proportion (48%) emerging from North America, and the US (41%) specifically (Figure 5). The global south was underrepresented, with only one publication from Africa and none identified from any South American nations. Thirteen publications were identified as having been published from the UK.

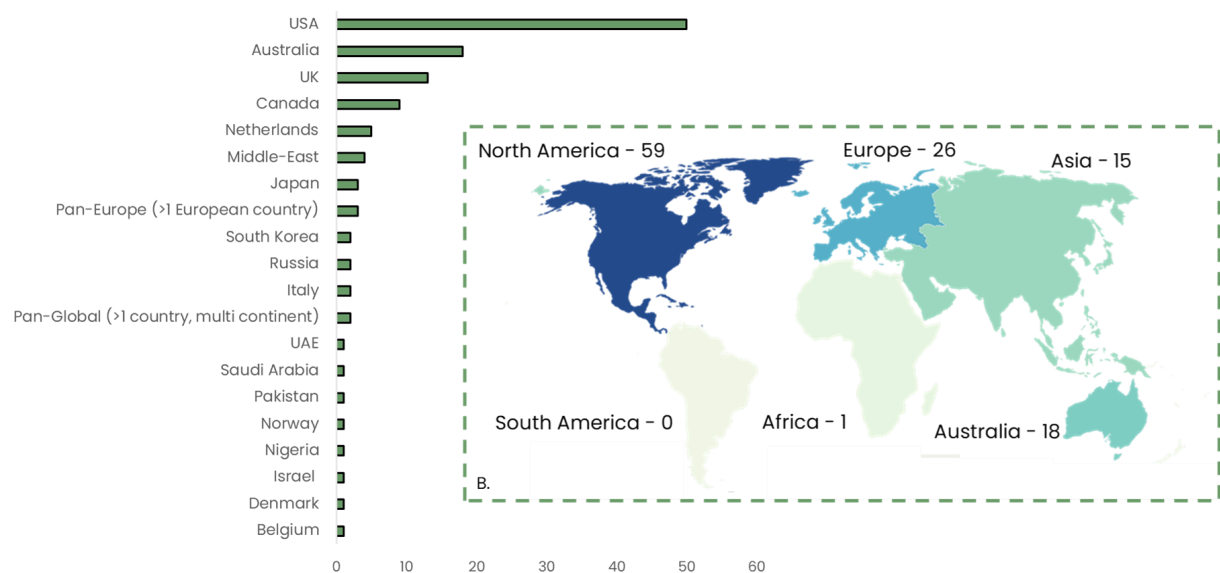


Figure 5. A) Publications by country and B) by continent.

Identified manuscripts were sub-divided by the type of risk they explored with their participants; mendelian, polygenic, or pharmacogenomic. Just under a third (32.3%) of all studies focused specifically on mendelian risk, 21.5% explored attitudes towards polygenic risk, whilst 14% focussed on pharmacogenomic risk. 20.7% of manuscripts explored different types of risk within the same analysis.

Fourteen manuscripts did not explicitly specify the type of genetic risk they were studying in their manuscript nor was it possible to define this from the article itself. In some cases, this was a conscious choice and part of the methodology with several vignette or survey based studies subtly altering aspects of the genomic intervention to assess how this altered preference or uptake. In these cases, the lack of definition was advantageous and provided meaningful insights for the development of broad and multifaceted

interventions such as those being considered by the Programme. However, in some cases the lack of clarity was not purposeful, and “genomics” was treated as a homogenous concept with the public asking for their attitudes towards this relatively intangible and nebulous intervention.

4.3 Grey literature results

We searched a total of 35 organisational websites using the Google Site Search Tool, screening 4,286 webpage titles and descriptions. From this process, 28 documents were identified for full-text review. After removing duplicates and assessing eligibility, 23 grey literature sources were included in the final analysis. The most common sources were from Genomics England, the Public Health Genomics Foundation, and the Global Alliance for Genomics and Health. The included grey literature was sub-divided based on type of publication, including (1) public engagement reports, (2) policy briefs, (3) research and literature reviews and (4) theses and dissertation. The included documents were subsequently thematically analysed to provide complimentary insights to findings from the academic literature.

05 Analysis



5.1 Key emerging themes

Key emerging themes are presented according to the three risk types—Mendelian, polygenic, and pharmacogenomic—for both public and healthcare professional perspectives, structured using the domains of the Health Belief Model (HBM). This is followed by an overview of key evidence gaps and strategic recommendations.

5.1.1 Mendelian (rare variant) risk

Definition: Genetic risk caused by rare variants in a single gene that can significantly increase the chance of developing a condition (highly penetrant) e.g. BRCA1/2 and breast/ovarian cancer.

Table 4. Summary of mendelian risk findings

Public views	Healthcare professionals' views
Interest in testing increases when people perceive a personal or familial link to disease, especially when it relates to conditions they already know about – <i>highlighting relatable scenarios or family health contexts may help make testing feel more relevant.</i> Concerns about data misuse, discrimination, and lack of transparency reduce trust in testing programmes – <i>providing clear, simple and honest explanations about how data is used and protected can help build public confidence.</i> People are more motivated to engage with testing when they believe results will lead to clear actions or benefits, such as prevention, treatment, or self-control – <i>framing genomics around its actionable value can improve engagement.</i>	Professionals recognise genetic testing can identify high-risk individuals for serious diseases (like hereditary cancers), but many lack detailed knowledge about which conditions are evidence-supported – <i>providing targeted condition-specific education can improve confidence in recommending testing.</i> HCPs worry about negative consequences of genetic testing, such as anxiety, uncertainty, or unclear clinical action – <i>engagement strategies should acknowledge these concerns and clarify how to manage uncertain or low-penetrance results.</i> Perceived benefits include empowering patients and improving decision-making, especially when linked to actionable outcomes – <i>communications should emphasise the real-world impact of testing</i>

Many individuals lack confidence in understanding and acting on genetic test results, especially without clinical support — *ensuring access to professional guidance and using plain, accessible language can boost confidence and reduce anxiety.*

Healthcare professionals are highly trusted sources of advice, and their endorsement significantly influences decisions to test — *supporting clinicians with tools and training can strengthen their role as effective engagement messengers.*

on prevention and control.

Lack of time, genomic literacy, and doubts about clinical utility are major barriers, especially among GPs — *capacity-building through continuing professional development and decision-support tools could reduce resistance.*

Prior training and hands-on involvement with genomics strongly predict positive attitudes and confidence — *offering practical learning opportunities and undergraduate exposure can foster long-term engagement and adoption.*

Public (22 papers)

Perceived susceptibility

Public perception of their susceptibility to health risks, particularly those linked to rare genomic variants, is significantly influenced by a blend of existing health concerns, family history, and the anticipation of actionable results. A widespread belief exists that genetic factors play a crucial role in health [Taylor 2011], leading to a strong interest in understanding personal disease risk [Calabrò 2024, Henneman 2013, Manchanda 2019, Wilde 2011 (2)]. This interest is often driven by a perceived personal relevance, whether it is to adopt a healthier lifestyle or to gain more self-knowledge [Cherkas 2010]. Notably, family history emerges as a powerful predictor of perceived susceptibility and a key driver ([for interest in genetic testing [Cousens 2017, Fehlberg 2025, Manchanda 2019, Wilde 2010, Wilde 2011 (2)]. Individuals are significantly more interested in testing if a condition "runs in the family" [Wilde 2010] and, conversely, may not feel a sense of urgency for screening if there is no family history of a particular cancer, such as breast cancer [Fehlberg 2025]. Younger adults also show a particular interest in testing when family history is present [Cousens 2017].

However, baseline perceived susceptibility for specific conditions might be low, as many have not considered their personal risk for conditions like ovarian cancer [Hann 2018, Meisel 2013, Meisel 2016]. Despite this, an acknowledged role of genes in disease development means that when specific risk information is presented, or a higher risk result is received, it can act as a "wake-up call" for preventive action [Meisel 2013, Taylor 2011]. A genetic predisposition to familial hypercholesterolemia directly leads to a higher

perceived comparative risk for cardiovascular disease [Claassen 2012]. Furthermore, individuals who self-estimate their risk for conditions like major depressive disorder as higher than average are significantly more willing to engage in testing and subsequent interventions [Wilde 2011, Wilde 2011 (2)]. When presented with hypothetical scenarios, the desire to understand risk and undergo genetic testing is elevated when the probability of illness increases [Tiller 2023, Wolff 2011]. While the public generally expects to be informed about incidental findings [Ballard 2019] and understands the importance of genetic information for themselves [Savard 2014], there can be misunderstanding regarding the familial relevance of genomic results, with some participants viewing additional findings as personal rather than also being relevant to relatives [Ballard 2020].

Ultimately, the perceived susceptibility to genetic health risks is a strong motivator for engaging with genomic testing, particularly when results are perceived as actionable [Manchanda 2019]. The desire to know disease risk is often directly linked to the availability of effective prevention [Calabrò 2024] or the motivation to adopt healthier lifestyles [Cherkas 2010].

Perceived severity

The public generally acknowledges the serious nature of conditions that mendelian genomic testing aims to address, viewing information about such risks as critically important for health outcomes. The perceived seriousness of conditions like ovarian cancer, including its symptoms and impact on life expectancy, significantly shapes individuals' reactions to receiving genetic test results [Meisel 2013]. There is an implicit understanding that early detection of conditions, particularly those that can be prevented or treated, can be "life-saving" [Tiller 2023], influencing a willingness to participate in screening [Meisel 2016]. Similarly, the willingness to undergo testing even if it might lead to risk-reducing surgery suggests a recognition of the severe consequences of the diseases in question [Meisel 2016].

However, the perceived severity also encompasses significant psychological and social dimensions. Participants express concerns about the potential for anxiety upon receiving test results [Fehlberg 2025, Gerdes 2021, Hann 2018, Wilde 2011 (2)], and the prolonged worry that can occur between taking a test and receiving results [Hann 2018]. For some, knowing a genetic risk can evoke fears of fatalistic thinking or an inability to cope with the information [Wilde 2010]. Beyond the individual, there are social implications, with concerns that high-risk results or the need for surgery could jeopardise marriage prospects, particularly for younger women in certain cultural contexts [Cousens 2017, Hann 2018, Manchanda 2019]. The term "worrying" is a chosen word by a notable percentage of the public when considering genomic sequencing [Ballard 2019, Ballard 2020].

Despite these concerns, the potential to mitigate or prepare for severe health outcomes remains a strong underlying motivator. The desire for early intervention and prevention to avoid the full impact of serious illness reinforces the perception of severity as a driving factor for engagement with genomic testing [Meisel 2013, Wilde 2011]. This nuanced understanding of severity, encompassing both health consequences and personal/social impacts, is crucial for effective communication strategies.

Perceived benefits

The public perceives numerous and significant benefits from mendelian genomic testing, largely centred on empowerment, proactive health management, and a deeper understanding of self and family. A primary benefit is the opportunity for prevention and early intervention [Calabrò 2024, Cherkas 2010, Meisel 2013]. Testing is seen as a means to "adopt a healthier lifestyle" if found to be at high genetic risk [Cherkas 2010]; to gain "awareness" and "opportunities for early intervention" [Meisel 2013]; and to "know how to prevent" a condition [Hann 2018]. The belief that early detection can be "life-saving" [Tiller 2023] further reinforces this. It allows for "better and more targeted care" [Meisel 2013] and the implementation of preventative techniques [Wilde 2010].

Beyond direct health benefits, participants value the reassurance and reduction of uncertainty that testing can provide [Manchanda 2019, Meisel 2013, Wilde 2011 (2)]. Many express a desire for "peace of mind" [Wilde 2011 (2)] and relief from not being a carrier [Cousens 2017]. The information gained is also highly valued for personal knowledge and family legacy [Ballard 2019, Cherkas 2010, Savard 2014]. Genomic sequencing is viewed as "informative" and a "family legacy" [Ballard 2019], allowing individuals to "learn more about myself" [Cherkas 2010]. The ability to convey genetic risk information to children [Cherkas 2010, Manchanda 2019] is also a significant motivator, underscoring the perceived intergenerational benefit.

Furthermore, the public believes genetic testing can enhance medical management and a sense of personal control. It enables doctors to "monitor my health more closely" [Cherkas 2010] and is seen as empowering individuals to feel "in control" over cancer development [Meisel 2013]. Even for complex traits like depression, one study which investigated attitudes towards 5-HTTLPR genotyping found that early psychological support informed by this testing would be viewed as a potential benefit [Wilde 2011 (2)]. These perceived benefits contribute to overall positive attitudes towards genomic testing and strong anticipated uptake [Meisel 2016, Taylor 2011].

Perceived barriers

Despite the perceived benefits, several significant barriers hinder public engagement with Mendelian genomic testing, ranging from psychological and practical concerns to issues of privacy and understanding. A major psychological barrier is anxiety related to receiving results or living with high-risk information [Fehlberg 2025, Gerdes 2021, Hann 2018, Wilde 2011 (2)]. Participants express "worrying" as a negative association with genomic sequencing [Ballard 2019, Ballard 2020] and fear fatalistic thinking or an inability to cope with genetic risk information [Wilde 2010].

Practical and informational barriers include the cost of testing, which is a significant deterrent, with interest rising dramatically if tests are free [Cherkas 2010]. There is low public awareness of personal genome testing [Cherkas 2010] and widespread acknowledgement of insufficient knowledge about genetic testing and omics sciences [Calabrò 2024]. This lack of understanding can lead to misconceptions about results, such as misunderstanding complex genetic concepts [Gerdes 2021] or viewing additional findings as purely personal rather than familial [Ballard 2020]. Many participants do not recall or fully understand consent details, including familial implications [Ballard 2020]. Less educated and ethnic minority groups may also face barriers related to information needs and attitudes towards potential interventions like surgery [Meisel 2016].

Furthermore, ethical and social concerns represent considerable barriers. Privacy concerns about data security are prevalent [Calabrò 2024, Fehlberg 2025, Gerdes 2021, Tiller 2023], as is the fear of genetic discrimination by insurers or employers [Cousens 2017, Manchanda 2019, Taylor 2011, Tiller 2023, Wilde 2010, Wilde 2011 (2)]. Social and cultural factors also play a role, with some younger women expressing concerns about test results jeopardising marriage prospects [Cousens 2017, Hann 2018, Manchanda 2019]. There is also opposition to Direct-to-Consumer (DTC) testing due to credibility and security concerns, and the absence of face-to-face counselling [Wilde 2010]. These multifaceted barriers underscore the need for comprehensive, culturally sensitive communication and robust policy frameworks to support equitable uptake.

Cues to action

Various cues act as significant motivators, prompting the public to consider or pursue Mendelian genomic testing, often stemming from personal relevance, trusted advice, or information exposure. A primary cue is a recommendation or suggestion from a healthcare professional [Metcalfe 2019, Wilde 2010, Wolff 2011]. Physician suggestion significantly increases willingness to undergo testing [Wolff 2011], and the desire for GP interpretation of results is high [Cherkas 2010]. Pre-test counselling is also a critical cue, facilitating informed decision-making [Manchanda 2019].

Personal and familial health circumstances serve as powerful internal cues. The presence of a family history of a condition is a major driver for interest in testing [Cousens 2017, Wilde 2010]. Additionally, receiving a higher-than-average self-estimated risk for a condition, such as major depressive disorder, acts as a cue for greater willingness to engage in testing and interventions [Wilde 2011, Wilde 2011 (2)]. The hypothetical availability of a genetic test itself can serve as a cue, prompting consideration [Wilde 2011 (2)]. For individuals already seeking answers for chronic health issues, the search itself can be a cue leading to genomic testing [Metcalfe 2019].

External factors also play a role as cues. Public awareness campaigns and media discourse about genomics can increase public expectations and knowledge [Ballard 2020, Fehlberg 2025]. For example, the Angelina Jolie effect may have influenced awareness and attitudes towards BRCA testing [Meisel 2016]. Community-based group sessions and local media are suggested avenues for awareness campaigns to act as cues [Hann 2018]. Finally, the perceived actionability of results, such as the availability of effective prevention tools, serves as a strong cue, directly linking the desire to know risk to the possibility of tangible health improvement [Calabrò 2024].

Self-efficacy

Public self-efficacy regarding Mendelian genomic testing is multifaceted, encompassing confidence in making lifestyle changes, understanding complex information, and navigating the healthcare system. A strong sense of self-efficacy in adopting healthier lifestyles is evident: 93% of individuals state they would adopt healthier habits if found to be at high genetic risk [Cherkas 2010]. Participants express confidence in their ability to engage in preventive action and feel "in control" over cancer development, especially when provided with lifestyle advice [Meisel 2013]. Willingness to engage in health behaviours is also strongly linked to perceiving risk as "modifiable" and understanding gene-environment interactions [Wilde 2011].

However, self-efficacy regarding understanding and interpreting genetic information appears to be lower. Many individuals feel they would need the help of their GP to interpret test results [Cherkas 2010], and some report problems understanding reported medical implications, such as the difference between monogenic variants and SNPs [Gerdes 2021]. There is a widespread acknowledgement that citizens are not adequately informed about omics sciences and direct-to-consumer genetic testing [Calabrò 2024], suggesting low perceived knowledge and a need for greater self-efficacy in comprehension.

Despite these gaps, factors that enhance self-efficacy in engaging with genomic services include receiving counselling from healthcare professionals [Fehlberg 2025], which can instill confidence. Trust in healthcare providers and large projects also contributes to a

sense of security and willingness to participate [Ballard 2020]. Furthermore, higher BRCA-related knowledge is positively associated with increased interest, intention-to-test, and actual uptake [Manchanda 2019], indicating that greater understanding boosts an individual's confidence in their ability to process and act on the information. The public's overall willingness to engage in interventions also implies a degree of confidence in their ability to take action [Wilde 2011].

Healthcare professionals (2 papers)

Perceived susceptibility

Healthcare professionals (HCPs) largely acknowledge the potential for genetic testing to identify individuals at high risk for certain Mendelian conditions, such as ovarian cancer [Hann 2017]. A significant majority of HCPs agreed that genetic testing would assist in pinpointing those with an elevated risk of ovarian cancer [Hann 2017]. Similarly, Public Health (PH) professionals showed high agreement that they should consider genetic susceptibility when implementing evidence-based genomic applications [Rosso 2020]. This indicates a general understanding among professionals that some individuals are indeed susceptible to rare genetic conditions.

However, the precision of this perceived susceptibility varies. Only a minority of European PH professionals (28.9%) could accurately identify clinical conditions like hereditary ovarian cancer or Lynch syndrome for which there is strong evidence supporting genetic testing for risk prediction [Rosso 2020]. This suggests that while there's a broad acceptance of genetic susceptibility, the detailed knowledge of which conditions are amenable to current genetic risk prediction, and thus who is susceptible, may be limited within certain groups of professionals.

Perceived severity

HCPs perceive potential negative impacts associated with genetic testing, reflecting a recognition of the severity of outcomes beyond the direct health risk itself. A majority of HCPs believed that genetic testing "could have a negative impact on some of my patients" [Hann 2017]. This concern is further elaborated by genetics clinicians, whose hesitation to broadly offer testing stems from an awareness of variants of uncertain significance (VUS), lower penetrance of some genetic variants, and potential psychosocial impacts on patients [Hann 2017]. These concerns underscore the perceived severity of the consequences of genetic information on individuals' well-being and mental health.

Beyond individual patient concerns, PH professionals also raised significant worries about the broader systemic impact of investing in genomics. Over 60% of these professionals expressed concern that such investment might "divert economic resources from social and environmental determinants of health" [Rosso 2020]. This highlights a perceived severity regarding resource allocation, suggesting that the expansion of genomic services could negatively impact other vital public health initiatives. Conversely, the high agreement (over 70%) that genetic tests for diseases with "fatal outcome," such as BRCA testing, should be publicly funded, implicitly acknowledges the severe nature of these diseases and the importance of interventions [Rosso 2020].

Perceived benefits

Healthcare professionals generally perceive substantial benefits from genetic testing for Mendelian risk, particularly in risk identification and disease prevention. Most HCPs agreed that genetic testing would be instrumental in identifying individuals at high risk for conditions like ovarian cancer [Hann 2017]. A primary motivation for discussing interventions like surgery was the belief that it "would benefit patients by reducing risk and preventing ovarian cancer" [Hann 2017]. This directly links the testing to tangible, life-saving outcomes.

Furthermore, HCPs believe that genetic testing can empower patients, enabling them to make "good healthcare decisions" and providing them with a "sense of control" through risk stratification [Hann 2017]. This highlights the perceived non-clinical benefits of autonomy and informed choice. Public Health professionals also largely hold positive attitudes towards the implementation of genetic testing and services [Rosso 2020], agreeing that they should implement evidence-based genomic applications [Rosso 2020]. This broad support for implementation further underscores the perceived public health benefits of integrating genomics into practice, especially for conditions with severe outcomes, with many agreeing that publicly funded testing is warranted for those who could benefit [Rosso 2020].

Perceived barriers

Despite the recognised benefits, several significant barriers hinder the widespread adoption and support for Mendelian risk testing among healthcare professionals. A major barrier is the lack of high support for population-based genetic testing, with less than half of HCPs endorsing it [Hann 2017]. Clinical Geneticists, in particular, were the least willing to offer genetic testing for ovarian cancer risk to all adult female patients [Hann 2017]. This reluctance stems from practical and professional concerns, including lack of time, unproven screening effectiveness, or it not being considered part of their job [Hann 2017].

Furthermore, insufficient knowledge poses a substantial barrier, especially among European Public Health professionals, who exhibited poor genomic knowledge [Rosso 2020]. A minority (28.9%) could correctly identify conditions where evidence supports genetic testing for risk prediction [Rosso 2020]. This lack of foundational knowledge of genetic testing applications directly acts as a barrier to its effective integration [Rosso 2020]. Another important barrier is the concern among over 60% of PH professionals that investing in genomics might "divert economic resources from social and environmental determinants of health" [Rosso 2020], highlighting a resource allocation apprehension. Lastly, geneticists' hesitation also arises from the complexities of genetic information, such as the implications of variants of uncertain significance (VUS), incomplete penetrance, and potential psychosocial impacts, which they perceive as significant challenges [Hann 2017].

Cues to action

Several factors act as cues encouraging healthcare professionals to engage with and integrate Mendelian risk testing. The need for targeted training and education is a strong cue. Both studies highlight the importance of training, particularly for primary care HCPs, if population-based testing is to be widely rolled out [Hann 2017]. This suggests that the availability and provision of specific training programmes act as a prompt for professionals to increase their involvement.

Moreover, direct engagement with Public Health Genomics (PHG) activities and undergraduate training on genetic testing are significant cues. Positive attitudes towards PH professionals' roles in genomics are strongly associated with prior undergraduate training on genetics and direct involvement in PHG activities [Rosso 2020]. This indicates that formal education and practical experience serve as powerful motivators for professionals to embrace genomics. The existence of evidence-based applications for specific conditions, such as hereditary ovarian cancer and Lynch syndrome, also acts as a scientific cue for professionals to recognise the utility and necessity of genetic testing in their practice [Rosso 2020].

Self-efficacy

Healthcare professionals exhibit varied levels of self-efficacy regarding Mendelian genetic testing. Generally, HCPs reported high self-efficacy in conducting cancer risk consultations [Hann 2017]. Most were confident in explaining risk levels to patients once they had been informed of the boundaries of the risk assessment [Hann 2017]. This suggests that with adequate information and frameworks, HCPs feel capable of communicating complex genetic risk.

However, this self-efficacy is not uniformly distributed. General practitioners (GPs) showed significantly lower self-efficacy compared to other HCPs in conducting these consultations [Hann 2017]. This lower confidence among GPs is attributed to their broad practice and highlights a crucial need for specific training to enhance their self-efficacy regarding population-based testing [Hann 2017]. Similarly, Public Health professionals, despite generally positive attitudes towards genomics, acknowledged a lack of sufficient genomic knowledge, with nearly all PHG-involved professionals agreeing that specific training is needed to evaluate genetic service quality [Rosso 2020]. This consensus on the need for specialised training underscores a perceived gap in their current capacity or self-efficacy to fully engage with and implement genomic services without further education.

5.1.2 Polygenic (common variant) risk

Definition: Genetic risk based on the combined effect of many common variants, each with small effect, used to estimate a person’s overall risk for common diseases (e.g. heart disease, diabetes)

Table 5. Summary of polygenic risk findings

Public views	Healthcare professionals’ views
The public strongly believes genetics influences health and value personalised risk information, especially when they have a family history—<i>emphasise personal relevance and tailor communication to individual risk profiles to deepen engagement.</i> Conditions linked to polygenic risk (like cancer, heart disease) are seen as serious, and risk awareness often drives preventive action, even among those with fatalistic attitudes—<i>frame polygenic risk communication around empowerment and actionable steps to reduce risk for serious disease.</i> People perceive understanding their polygenic risk as empowering and linked to better decisions, early detection, and	Healthcare professionals recognise the value of polygenic risk for identifying at-risk individuals and informing preventive care, but need clearer integration into clinical workflows—<i>position polygenic risk as a routine part of personalised prevention to support everyday decision-making.</i> Concerns about psychosocial impacts, insurance implications, and family dynamics make professionals cautious about communicating risk, especially in primary care—<i>develop clear guidance and tools to support sensitive and responsible communication of genomic risk.</i> Professionals see major benefits in prevention, screening, and behaviour change, and believe genomic risk should be

reassurance, though there remains uncertainty whether people will act on this information – *highlighting the practical health and lifestyle benefits in public messaging.*

Barriers include cost, complexity of information, health system constraints, and fear of discrimination, with low genomic literacy particularly affecting equity—*simplify risk communication, and ensure materials are culturally and linguistically inclusive.*

Personalised risk feedback, family history, and endorsement by trusted healthcare professionals are powerful cues to action, but people still want guidance to interpret results—*train GPs to act as credible intermediaries and provide clear, supportive pathways to counselling or follow-up.*

part of public health strategies—*frame genomic engagement around its alignment with preventive health goals and population health management.*

A major barrier is professionals' lack of knowledge and confidence, due to inadequate training and complex risk information, which limits their willingness to engage—*prioritise targeted genomics education and practical training for both public health and primary care professionals.*

Cues to engage include postgraduate education, CME, patient questions, and media attention, with a growing sense that genomics will become part of future responsibilities – *leverage existing training systems and professional expectations to normalise genomics as core clinical knowledge.*

Public (17 papers)

Perceived susceptibility

The public perception of susceptibility to diseases influenced by polygenic risk is varied, yet a general belief in genetic influence on health is evident [Taylor 2011]. A significant majority (77%) of the public have reported to believe that genetic factors play a substantial role in their health [Taylor 2011]. This perceived likelihood drives a desire for personalised risk information, as it makes the risk feel tangible and more relevant than generic health advice [Smit 2020]. Individuals are keenly interested in understanding genetic heritability and their own specific risk, particularly when there is a family history of a disease [Saya 2012, Smit 2020(2)].

While there's a strong interest in personalised genetic risk, public understanding of its precision can be variable. Many participants in one study initially overestimated their colorectal cancer risk, highlighting that while there's a general recognition of risk, a precise understanding may be lacking [Saya 2020]. However, after receiving personalised information, average-risk individuals improved the accuracy of their risk perception [Saya 2020]. The public generally desires to know their disease risk to enable prevention and

gain more control over their lives [Bíró 2020], and there has been an increasing curiosity in genetics over time [Henneman 2013].

Perceived severity

The public generally perceives the conditions for which polygenic risk information is offered, particularly various cancers, as serious. The paramount importance placed on sensitivity in cancer screening programmes, equated with detecting cancers and saving lives, underscores this perception [Dennison 2023]. Even when individuals hold fatalistic views about past exposures, such as sun exposure and melanoma, the seriousness of the disease still motivates them towards behavioural changes [Fenton 2019]. Adjusting an individual's prior risk of a disease through genomic testing could therefore impact behaviour.

Knowing one's susceptibility to a serious condition like breast cancer can induce anxiety and worry, with some women expressing that such knowledge might make them "feel ill" [Henneman 2011]. Furthermore, even a low-risk result for breast cancer is considered "still a risk," which can create sustained anxiety and drive a desire for further monitoring, such as mammography [Henneman 2011]. Despite these potential anxieties, studies show that the psychosocial impact of receiving polygenic risk information for conditions like colorectal cancer tends to be low, suggesting that while the conditions are perceived as serious, the distress from receiving this type of risk information can be manageable when communicated effectively [Saya 2020]. However, most of these studies look only at a snapshot in time.

Perceived benefits

The public identifies numerous benefits associated with receiving polygenic risk information, primarily centring on empowerment, informed decision-making, and proactive health management. Key perceived benefits include the ability to adopt a healthier lifestyle and to support screening if required [Cherkas 2010]. Knowing one's risk provides "new knowledge" to help manage health, leading to feelings of happiness, reassurance, and being better equipped to address health concerns [Fenton 2019].

Moreover, receiving this information is perceived to empower individuals to make decisions to minimise potential risks, fostering a proactive approach to health management, encapsulated by phrases like "forewarned is forearmed" and "information is power" [Smit 2021, Smit 2015]. For average-risk individuals, personalised risk information can also reduce cancer-specific anxiety and improve the accuracy of their risk perception, leading to more positive attitudes towards screening behaviours [Saya 2020].

Perceived barriers

Despite the potential benefits, several significant barriers may impede the uptake or experience of polygenic genomic testing. Low public awareness and a perceived need for specialist interpretation are consistently reported issues [Cherkas 2010]. Public understanding of complex genetic information can be challenging, with a limited understanding of the difference between monogenic variants and SNPs [Gerdes 2021].

Concerns about adverse psychological impacts are prevalent, including anxiety, worry, disappointment, or hyper-vigilance from high-risk results, which could "freak you out" [Henneman 2011, Smit 2015, Smit 2021]. Conversely, low-risk results could lead to false reassurance or apathy, with individuals incorrectly believing they will "be fine" or that their current behaviours are sufficient, thus limiting further positive change [Smit 2015, Smit 2021]. Broader societal concerns include fears of discrimination by employers or insurance companies, as well as worries about the implications for life insurance eligibility or premiums [Henneman 2011, Smit 2015, Saya 2012, Taylor 2011]. There are also concerns about the capacity and cost implications for the health system [Smit 2015, Smit 2021]. Additionally, issues related to GP knowledge and training in genetics, limited time in consultations, and access to genetic counsellors are perceived as barriers [Smit 2020]. Disparities for lower-educated or non-English-speaking individuals represent another barrier to equitable access [Saya 2020, Saya 2012].

Cues to action

Various factors act as cues that prompt individuals to consider or engage with polygenic testing. Personal motivations and experiences are especially important cues. A desire for cancer prevention, familiarity with a disease (e.g., breast cancer), or a personal search for causes of chronic health issues can prompt interest in testing [Henneman 2011, Metcalfe 2019]. A strong family history of disease is a key motivator for individuals to seek testing, as they are interested in the genetic component and their own inherited risk [Saya 2012, Smit 2020, Stewart 2018]. The intervention of trusted healthcare professionals, such as GPs, who can endorse and facilitate appropriate screening, acts as another important cue, leveraging existing trust in the healthcare system [Saya 2020, Metcalfe 2019, Venning 2022].

Self-efficacy

Reported self-efficacy regarding polygenic risk information is mixed, generally reflecting high confidence in their perceived ability to enact behavioural changes but lower confidence in interpreting complex genetic data independently. In Australia, individuals who received personalised melanoma risk information felt "armed with that knowledge"

and confident in making changes like increasing their use of sunscreen and seeking shade [Fenton 2019]. There is a general belief that if information helps prevent cancer, "there's always something you can do" [Smit 2020]. Whether these beliefs translate into long-term behaviour change remains unproven.

There is a perceived need for healthcare professional assistance in interpreting test results [Cherkas 2010, Gerdes 2021]. While some women felt that a high genetic risk of breast cancer could motivate them to engage with more regular check-ups or healthy lifestyles, others were unsure how results would impact their lives or what actions to take [Henneman 2011]. Studies suggest that this counselling may only need to be brief, with a short discussion around colorectal cancer risk tests leading to most participants demonstrating a good understanding of the test and felt able to make informed choices [Saya 2020, Saya 2012].

Healthcare professionals (2 papers)

Perceived susceptibility

Healthcare professionals (HCPs) generally acknowledge the utility of predictive genetic testing in identifying individuals at higher risk of developing disease, particularly chronic conditions [Marzuillo 2014]. Almost all public health professionals surveyed felt that such tests could help identify individuals with increased susceptibility [Marzuillo 2014]. General Practitioners (GPs) also view polygenic risk information as a valuable tool for overall risk assessment for preventable or detectable diseases [Smit 2019]. Specifically, they believe it is useful for preventive care and informing screening strategies [Smit 2019].

Perceived severity

From a public health perspective, although there is limited detail provided on HCPs perception of individual disease impact in relation to polygenic risk, it implicitly links the "clinical utility" of genetic tests to guide the management of chronic diseases and their "power to improve a subject's health status" [Marzuillo 2014]. This suggests that the lack of health improvement, or the presence of a chronic disease state, is viewed as severe enough to warrant interventions, which genomic testing could facilitate [Marzuillo 2014].

Perceived benefits

Healthcare professionals perceive substantial benefits from utilising polygenic risk information, primarily in enhancing prevention and motivating behavioural change. Public health professionals largely hold positive attitudes towards predictive genetic testing for chronic diseases [Marzuillo 2014]. A significant majority (76.5%) agreed that these tests

increase prevention opportunities, and an even higher proportion (90.5%) believed they should be integrated into wider prevention strategies [Marzuillo 2014]. This indicates a strong belief in the public health utility of polygenic risk information for proactive health management.

GPs also identified several key benefits. They believe genomic risk information can be useful for preventive care and for informing screening frequencies, particularly when effective prevention strategies are available [Smit 2019]. Furthermore, they see potential in motivating health behaviour changes in patients [Smit 2019]. This suggests that professionals view polygenic risk data not just as diagnostic, but as an actionable tool to empower patients and guide clinical interventions for better health outcomes.

Perceived barriers

Despite the recognised benefits, several significant barriers impede the effective integration and communication of polygenic risk information. A primary concern is the inadequate knowledge among healthcare professionals. In Italy, approximately 80% of public health professionals considered their knowledge of predictive genetic testing for chronic diseases to be insufficient [Marzuillo 2014]. Australian GPs also reported feeling unprepared and had limited confidence in discussing genomics with patients [Smit 2019]. They face difficulty explaining complex risk information, and most view genomic risk communication as a specialist responsibility [Smit 2019]. This perceived complexity and the position that the communication of detailed genomic information falls outside their current scope of practice, both act as significant barriers.

Cues to action

For public health professionals, exposure to postgraduate training and time spent on Continuing Medical Education (CME) were all predictors of increased knowledge, which in turn strongly predicted positive attitudes towards genomics [Marzuillo 2014]. The widespread belief that their knowledge should be improved, with 86% advocating for specific post-training courses, highlights that the availability and emphasis on such training serve as strong cues for action [Marzuillo 2014].

In primary care, patient-initiated inquiries about genetic risk (including cancer) are a key cue for GPs to discuss genomics [Smit 2019]. Additionally, increasing public awareness and media attention around genomics prompt GPs to engage in these discussions [Smit 2019]. The belief among GPs that communicating genomic risk "may become a responsibility within primary care" also acts as a forward-looking cue, suggesting an anticipation of future professional obligations that will necessitate increased engagement with this area [Smit 2019].

Self-efficacy

Healthcare professionals exhibit varying levels of self-efficacy when it comes to managing polygenic risk information. The aforementioned strong self-perception of insufficient knowledge directly impacts HCP self-efficacy in effectively utilising and integrating genomic information for public health initiatives [Marzuillo 2014]. While they possess the "necessary attitudinal background," there is a clear perceived need for "more methodological training" to enhance their capabilities and allow them to integrate this into routine practice [Marzuillo 2014].

While HCPs may feel confident with basic explanations, many HCPs view discussing genomic interventions as something which should be undertaken by "other professionals" [Smit 2019]. This highlights a need for engagement with these groups, the development of appropriately designed training programmes and, critically, tools to ensure these interventions are made to feel like part of their existing practice – not simply an extra task to perform.

5.1.3 Pharmacogenomic risk

Definition: Genetic risk that affects how the body responds to certain medicines, used to guide safer and more effective prescribing (e.g. avoiding side effects or treatment failure)

Table 6. Summary of pharmacogenomic (PGx) risk findings

Public views	Healthcare professionals' views
Past negative experiences with medication drive public interest in pharmacogenomic testing, as people feel personally at risk of side effects or ineffectiveness—engagement strategies should emphasise personal relevance and real-life examples of how genomics can improve medication safety. The public sees pharmacogenomics as a tool for gaining control and improving treatment outcomes, valuing access to personalised information and safer prescribing—highlighting how PGx testing can empower individuals and support shared decision-making.	HCPs view pharmacogenomic testing as highly relevant for identifying patient risk and guiding safe, effective prescribing – emphasis on clinical relevance and practical prescribing benefits in engagement materials to reinforce perceived value. Service design is a critical factor for uptake, especially turnaround time, result format, and EHR integration – co-design PGx services with HCPs to ensure compatibility with workflows and existing digital systems. There is strong support for PGx testing, but concerns remain about misinterpretation and data overload – PGx results should be

Low awareness of DNA's role in medication response and a lack of accessible patient education are major barriers, despite overall interest—important to prioritise clear, non-technical communication materials and work with trusted professionals to explain PGx simply and effectively.

Lived experience with adverse drug reactions and preferences for non-invasive, digital tools strongly shape testing intentions—design services around ease of use (e.g. cheek swabs, online portals) and frame them as solutions to everyday medication challenges.

People want to understand their PGx results but know they need help doing so, signalling both a desire and a need for structured guidance—build engagement mechanisms that combine digital tools with healthcare professional support to build confidence and understanding.

structured and clear with decision support tools to increase confidence and minimise risk of misinterpretation.

Limited training and uncertainty in interpreting complex genomic data undermine confidence in using PGx – invest in targeted training and practical tools (e.g. embedded guidelines in EHRs) to build self-efficacy among professionals.

Demand from patients and public interest in personalised care could be a motivating force for HCP engagement – equip professionals with communication tools and educational resources to respond confidently to patient interest in PGx.

Public (11 papers)

Perceived susceptibility

The public demonstrates a significant awareness of their susceptibility to varied responses to medication, with 89% of respondents understanding that individuals can respond differently to the same medication [Magavern 2025]. This understanding is profoundly influenced by personal experience, as 59% of individuals reported having experienced either no benefit or a side effect from prescription medication [Magavern 2025]. Such lived experience is a powerful motivator, nearly doubling the likelihood of an individual wanting a pharmacogenomic test (Odds Ratio 1.94) [Magavern 2025].

This keen interest in pharmacogenomic testing aligns with a broader curiosity about genetic predispositions to disease [Chokoshvili 2017, Henneman 2013, Ilyas 2021, Gerdes 2021]. Individuals are driven by a desire to obtain actionable results that could inform prevention and enhance preparedness for future health challenges [Tommel 2023]. In pharmacogenomics, this translates into a perceived susceptibility to medication

inefficacy or adverse drug reactions, motivating individuals to seek genetic insights for proactive health management and to make informed healthcare and lifestyle decisions [Charbonneau 2020, Metcalfe 2019]. There is limited exploration in the literature regarding how individuals consider their susceptibility might change over time.

Perceived severity

Public concerns regarding potential misuse of pharmacogenomic data exist, but they are not perceived as significantly higher than those for other routine medical data [Magavern 2025]. Specific worries related to pharmacogenomic data, such as concerns about police access, ethnicity identification, or family relatedness, were marginally elevated (1–3% more worried) compared to general healthcare data [Magavern 2025]. The public clearly recognises the potential for serious consequences related to medication toxicity, valuing pharmacogenomic information for its ability to inform treatment and avoid such harm [McDermott 2024].

More broadly, perceptions of severity extend to potential social and psychological consequences. These include fears of losing control over information, privacy breaches, confidentiality issues, and risk of discrimination [Tommel 2023]. The potential for non-actionable results or incidental findings, which could cause unnecessary worry and distress, also contributes to perceived severity [Tommel 2023, Gerdes 2021]. A substantial minority are worried that genetic testing could stigmatise people with disabilities or create an inferior image of affected individuals [Chokoshvili 2017].

Perceived benefits

The public holds a strong positive view of pharmacogenomic testing, primarily perceiving it as a valuable tool for improving health outcomes and optimising medication use [Magavern 2025]. A majority (72%) of respondents in one study were motivated by the potential for enhanced drug effectiveness, while 63% saw the benefit in reducing adverse drug reaction (ADR) risks [Magavern 2025]. Furthermore, 67% believed testing would increase medication adherence [Magavern 2025]. This positive outlook reflects a general belief that genetic factors influence health and that genetic testing is a positive advancement [Taylor 2011].

Beyond clinical benefits, the public believes pharmacogenomic testing empowers patients through better access to their own health data [McDermott 2024, Tommel 2023]. There is strong expectation that testing should be standard within the healthcare system, with 85% believing the NHS should offer it to those regularly taking many medicines, and 58% advocating for it to be available to everyone [Magavern 2025]. This reflects a desire

for tailored treatment and continuity of care [Tommel 2023], viewing pharmacogenomics as contributing to a more efficient and comprehensive system [Tommel 2023].

Perceived barriers

Despite high public support, several barriers impede adoption. A crucial barrier is low awareness and understanding of DNA's role in medication response, with only 52% aware of its role in drug benefit and 48% in ADR risk [Magavern 2025]. Critically, 93% of participants had not encountered patient materials explaining pharmacogenomics, highlighting a major gap in public education [Magavern 2025]. Limited public knowledge also extends to availability and legality of genetic tests [Savard 2014].

Practical barriers include reliance on blood testing for genetic samples, which presents access issues, with strong public preference for non-invasive methods like cheek swabs [McDermott 2024]. Privacy concerns are paramount, encompassing fears of data misuse, leaks, and a lack of trust, particularly regarding commercial entities [Tommel 2023, McDermott 2024]. Past failures in national health data programmes in the UK have also eroded trust [McDermott 2024]. There is a strong preference for hospital-based, doctor-initiated testing, with overwhelming opposition to internet sales of tests in one study (95.5% against), reflecting a desire for professional oversight [Chokoshvili 2017]. Cost is also a substantial deterrent, with interest notably higher if tests are offered for free [Cherkas 2010].

Cues to action

Several factors strongly prompt consideration of pharmacogenomic testing. The most prominent cue is personal experience with medication issues, such as no benefit or side effects from prescribed medication, which significantly increases the likelihood of wanting a test [Magavern 2025]. This direct experience highlights personal relevance as a driver.

Broader cues include curiosity about genetic make-up [Henneman 2013, Gerdes 2021, Cherkas 2010]. The prospect of a free test boosts interest [Cherkas 2010]. Actionability of results is a crucial driver, as interest is considerably higher when conditions are treatable or preventable, or when high-risk results are communicated [Chokoshvili 2017, Charbonneau 2020, Cherkas 2010, Tommel 2023]. The desire for reciprocal data sharing, where individuals feel informed and partnered in the process, also acts as a cue [Tommel 2023]. External cues such as GP recommendations or advertising can also encourage uptake [Metcalfe 2019].

Self-efficacy

While the public expresses confidence in adopting healthier behaviours if informed of high genetic risk (93% would adopt healthier habits) [Cherkas 2010], self-efficacy in interpreting complex genetic data, including pharmacogenomic results, is low [Cherkas 2010, Charbonneau 2020, Gerdes 2021, Tommel 2023]. There is a strong desire for assistance from GPs or other expert healthcare professionals for interpretation and explanation [Cherkas 2010, Ilyas 2021, Mcdermott 2024, Tommel 2023, Metcalfe 2019]. Many individuals acknowledge that their personal capacity may limit their ability to make fully autonomous decisions regarding genomic data [Tommel 2023].

The lack of readily available patient materials explaining pharmacogenomics [Magavern 2025] further indicates low self-efficacy in building understanding independently. Respondents express a preference for interactive online portals to access complex data [McDermott 2024], suggesting recognition of the need for structured, user-friendly support to enhance understanding.

Healthcare professionals (2 papers)

Perceived susceptibility

Healthcare professionals consistently recognise the value of pharmacogenomic testing in identifying patient susceptibility to specific drug responses. Both general practitioners and pharmacists show a strong preference for pharmacogenetic testing over standard care, indicating a clear perception of its relevance for informing risk and guiding treatment decisions [McDermott 2024 (2)].

Specifically, the perceived susceptibility is linked to the potential for pharmacogenomics to guide prescribing for improved safety and effectiveness of medicines [McDermott 2024]. This demonstrates an understanding that an individual's genetic makeup can make them susceptible to varying drug outcomes, highlighting the utility of identifying these predispositions to optimise patient care and prevent negative events [McDermott 2024].

Perceived severity

Healthcare professionals implicitly perceive the severity of ADRs and ineffective treatments, which drives their interest in pharmacogenomic testing. The increased uptake of pharmacogenetic tests is directly associated with their ability to improve medicines effectiveness or reduce adverse drug reactions [McDermott 2024]. This strong preference

indicates that HCPs view these negative outcomes (ADRs and ineffectiveness) as sufficiently severe to potentially warrant preventative genetic testing.

However, within the broader context of genomic medicine, HCPs did express concerns that patients might misinterpret complex genetic results, leading to potentially severe consequences such as stopping necessary medical support or experiencing unnecessary worry [Tommel 2023]. While this concern relates to genomic information generally, it could extend to pharmacogenomic results, where misinterpretation of drug-response predictions could lead to dangerous self-medication changes or anxiety about prescribed treatments [Tommel 2023]. The need for careful explanation of results and clear instructions underscores the perceived severity of misunderstood genetic information [Tommel 2023].

Perceived benefits

Healthcare professionals perceive substantial benefits from integrating pharmacogenomic information into clinical practice, primarily centered around optimising medication use and patient outcomes. There is a strong consensus among GPs and pharmacists that pharmacogenetic testing is preferable to no testing [McDermott 2024]. This highlights the perceived benefit of tailoring treatment based on an individual's genetic profile.

Moreover, the integration of pharmacogenomic data, often as part of broader personalised genomic medicine, is seen to enhance various aspects of healthcare. HCPs believe that data sharing contributes to complete records and more efficient, better healthcare by facilitating the tailoring of treatment [Tommel 2023].

Perceived barriers

Significant reported barriers impede the widespread adoption and effective implementation of pharmacogenomic testing, largely revolving around service design and clinical integration. HCPs' uptake of pharmacogenetic testing is highly sensitive to how the service is designed [McDermott 2024]. Specifically, increased turnaround time for results significantly reduces uptake for both GPs and pharmacists [McDermott 2024]. This logistical constraint points to a practical barrier in including pharmacogenomic in fast-paced clinical workflows.

Furthermore, there are clear preferences regarding the format and delivery of results that act as barriers if not met. GPs prefer specific clear actionable results to avoid information overload or irrelevant data [McDermott 2024]. GPs report disliking receiving results via post, email, or web-portals, preferring Electronic Health Record (EHR) integration, which, if

unavailable, serves as a significant barrier to efficient result utilisation [McDermott 2024]. Beyond pharmacogenomics, broader genomic data barriers, such as concerns about data misuse, privacy, and leaks, as well as issues with complex infrastructure and low patient literacy/skills, could also hinder the effective use of pharmacogenomic information [Tommel 2023].

Cues to action

Several factors act as cues prompting healthcare professionals to engage with and adopt pharmacogenomic testing. A primary cue is the optimisation of service design, as HCP preferences for specific service attributes, such as EHR integration for results, and faster turnaround times, are identified as crucial for maximising HCP uptake for population-level services [McDermott 2024]. This indicates that well-designed, user-friendly pharmacogenomics services are a strong motivator for adoption.

Additionally, the fundamental recognition that pharmacogenetic testing is preferred over no testing by all respondents signals a general positive disposition and an inherent cue to action within the professional community [McDermott 2024]. Patient demand for personalised genomic information is also identified as a driver that could push HCPs towards greater engagement with these services [Tommel 2023].

Self-efficacy

Healthcare professionals' self-efficacy regarding pharmacogenomic testing is implicitly linked to their preferences for service design and their perceived need for support in managing complex genetic information. The strong preference for results to be returned within the EHR suggests a desire for integrated, accessible information that enhances their ability to act on results efficiently [McDermott 2024]. The negative preferences for less integrated methods (post, email, web-portal) imply that these cumbersome methods reduce their perceived capacity to effectively use the information [McDermott 2024].

More broadly within genomic medicine, HCPs recognise the crucial importance of their own competencies, including knowledge and communication skills, to effectively explain and manage genetic data [Tommel 2023]. They stress the need for patient support in understanding results, genetic counseling, and follow-up, indicating that they may not feel fully equipped to handle all aspects of complex genomic interpretations, including those for pharmacogenomics, on their own [Tommel 2023].

5.2 Analysis of grey literature

Grey literature was analysed by segmenting the different sources based on the type of document. These included (1) public engagement reports, (2) policy briefs, (3) research and literature reviews and (4) theses and dissertations. The following consistent themes were identified across all of the grey literature and key insights from each type of document were analysed under the following headings:

- Trust, data governance, and ethical safeguards
- Equity, diversity, and inclusion
- Communication, education, and understanding
- Benefits, motivations, and participant agency

5.2.1 Public engagement reports

The following sources are included in this analysis:

1. Genomics England, 2018. *100,000 Genomes Project Black African and Black Caribbean Communities: A Qualitative Exploration of Views on Participation*.
2. Genomics England & Sciencewise, 2018. *A Public Dialogue on Genomic Medicine: Time for a New Social Contract?*
3. Genomics England & Ipsos MORI, 2025. *Almost 90% of People Would Agree to Genetic Testing to Tailor Medication Use, Survey Finds*.
4. Genomics England & Muslim Census, 2022. *Attitudes Towards Health and Medical Research Within the Muslim Community*.
5. Innovate UK & Sciencewise, 2014. *Developing Stratified Medicine: A Public Dialogue on the Human Issues Raised by the Development and Implementation of Stratified Medicine*.
6. Ada Lovelace Institute & Nuffield Council on Bioethics, 2024. *Predicting: The Future of Health? Assessing the Potential, Risks and Appropriate Role of AI-Powered Genomic Health Prediction in the UK Health System*.
7. Innovate UK & Sciencewise, 2014. *Stratified Medicine: A Public Dialogue – Final Report*.

Trust, data governance, and ethical safeguards

Several public engagement reports state that trust in institutions and robust data governance frameworks are foundational to public willingness to engage in genomic initiatives. From a diversity perspective, historic abuses, such as the Tuskegee Study in Alabama from the 1930s to 1970s and the use of cancer tissue taken from Henrietta Lacks

in the 1950s in the USA, continue to fuel distrust among some communities, particularly Black African, Black Caribbean, and Muslim communities (UK).

Across reports, concerns about privacy and security are consistently raised. Participants identify strong “red lines” around commercial misuse of data and express fears about access by pharmaceutical companies, insurers and employers, and even government or police. A clear desire for personal control over data is evident, increasingly through personal digital devices, alongside calls for transparency and robust safeguards.

Equity, diversity, and inclusion

Concerns about fairness and equity are reported widely. Reports warn that genomic programmes could exacerbate existing health inequalities, creating unequal access or a “postcode lottery”. Fears of genetic discrimination – especially by insurers – are repeatedly highlighted.

A recurring issue is the underrepresentation of key communities in genomic research, notably Black African, Black Caribbean, and Muslim communities (UK). This lack of inclusion is framed as both a social justice problem and a scientific risk, potentially leading to biased datasets and findings or treatments less effective for certain groups. Recommendations emphasise the need for greater visible diversity in workforce and leadership, as well as culturally competent engagement approaches.

Communication, education, and understanding

Low public awareness and understanding of genomics is a consistent theme. Communication materials are often described as overly technical, wordy, or unclear, with terminology itself creating barriers – “stratified medicine” being criticised for elitist connotations, while “personalised” or “targeted medicine” were preferred.

The public expect trusted messengers – particularly GPs, the NHS, and charities to help with education and understanding. Reports recommend communication that is clear, consistent, visually representative, and realistic about the limits of genomics. Importantly, when given time and accessible information, participants demonstrated they could grasp complex concepts and form sensible, balanced opinions.

Benefits, motivations, and participant agency

Despite their concerns, the public expresses strong enthusiasm for the personal and societal benefits of genomics. Anticipated benefits include more accurate diagnoses,

personalised treatments, fewer side effects, and improved health outcomes. Preventative healthcare is also viewed positively.

Altruism is a strong motivator, with many participants keen to contribute to research for the benefit of future generations. At the same time, people want to be active partners in their care: accessing their own genomic data (e.g., via the NHS app) and making informed choices as “partners, not passengers”. This reflects a public eager to engage — provided that trust and equity are ensured.

5.2.2 Policy briefings

The following sources are included in this analysis:

1. PHG Foundation, 2014. *Genomics and the Boundary Between Research and Clinical Care and Treatment*.
2. GA4GH, 2023. *Public Attitudes for Genomic Policy Brief: Familiarity*.
3. GA4GH, 2023. *Public Attitudes for Genomic Policy Brief: Return of Research Results*.
4. GA4GH, 2022. *Public Attitudes for Genomic Policy Brief: Trust and Trustworthiness*.
5. GA4GH/Ipsos MORI, 2023. *Public Attitudes for Genomic Policy Brief: What Does the UK Public Think About the Storage and Use of Its Genetic Data?*
6. PHG Foundation, 2021. *The Ethical and Legal Framework for a Genomics England and Sano Genetics Participant Engagement Platform*.

Trust, data governance, and ethical safeguards

Trust is repeatedly identified as a prerequisite for meaningful public engagement in genomics. Policy briefs stress that this trust depends on robust governance, clear privacy protections, and strong safeguards against misuse. Concerns about commercial exploitation and weak regulation highlight the importance of explicit frameworks to reassure participants that their data will be managed responsibly. Another recurring issue is the blurred boundary between research and clinical care, which risks creating “therapeutic misconception” among participants. Reports recommend making this distinction explicit to avoid confusion and preserve trust.

Equity, diversity, and inclusion

Policy briefs emphasise that genomic programmes must not exacerbate existing inequalities. Risks include digital exclusion for people with low digital literacy or limited economic resources, potentially creating new barriers to accessing research and clinical benefits. At the same time, genomics is recognised as a powerful tool to address current disparities in representation and outcomes — particularly for minority and

under-researched groups. By intentionally designing inclusive programmes, policy frameworks aim to advance equitable access and ensure that the benefits of genomics contribute to improved health for all.

Communication, education, and understanding

Low public familiarity with genomics is seen as a persistent barrier. Policy documents call for clear, consistent, and transparent communication to clarify the purpose, benefits, and limitations of genomic initiatives. This is especially critical for supporting informed consent, where the complexity of genomic data makes misunderstanding likely.

Returning results through digital platforms adds to this challenge, since genetic counselling has traditionally relied on in-person conversations to explain findings. Without careful design, digital engagement risks leaving participants underinformed or misinformed.

Benefits, motivations, and participant agency

Policy briefs highlight that engagement is motivated by both altruism (supporting societal good) and anticipated personal benefit. A central issue is the return of results, with participants more willing to contribute data when they expect actionable findings to be shared back. Reports recommend platforms that empower participants to shape their involvement – including setting preferences, choosing what information to receive, and having an active voice in research priorities. This emphasis on agency reflects a shift towards viewing participants as *partners rather than passengers* in genomic research and healthcare.

5.2.3 Research and literature reviews

The following sources are included in this analysis:

1. Genomics England/Ipsos MORI, 2019. *A Public Dialogue on Genomic Medicine: Time for a New Social Contract? Final Report*.
2. PHG Foundation, 2008. *Evidence and Evaluation: Building Public Trust in Genetic Tests for Common Diseases*.
3. Ethox, 2014. *Finding People Who Will Tell You Their Thoughts on Genomics – Recruitment Strategies for Social Sciences Research*.
4. PHG Foundation, 2021. *Implementing Polygenic Scores for Cardiovascular Disease into NHS Health Checks*.

Trust, data governance, and ethical safeguards

Trust emerges as a foundational condition for the integration of genomics into healthcare. This trust is closely linked to the generation of robust clinical evidence for genetic tests, with near-universal agreement on the need to improve the evidence base and crucially, make it accessible to clinicians, patients, and consumers. Concerns include potential direct harms from poorly validated tests, such as misleading results or unnecessary follow-up procedures.

Public support for data use is also conditional: participants accept data sharing when safeguards are in place, including consent processes, de-identification, and explicit protections against misuse. Persistent anxieties focus on commercial exploitation, surveillance, and predictive insurance testing, reflecting broader concerns about profiteering and inappropriate data use.

Equity, diversity, and inclusion

Reviews highlight consistently low baseline awareness of genomics among the public. While participants show the ability to understand complex concepts when they are clearly explained, much of the available information is seen as too technical or inaccessible in its current format. Knowledge gaps extend to healthcare professionals, who report difficulties in keeping pace with the growing body of evidence and in evaluating the utility of genetic tests.

Improving “genomic literacy” is identified as critical for both the public and professionals, requiring communication that is clear, realistic, and delivered by trusted messengers. For professionals specifically, significant training is needed to build confidence in applying genomic knowledge, including emerging tools such as polygenic risk scores. The “failure to educate staff” is noted as a major risk to successful implementation.

Communication, education, and understanding

Concerns about fairness and equity are prominent across the reviews. Fears include “dystopian” outcomes such as a stratified society or new forms of disenfranchisement if genomic data is misused. Even where polygenic tools perform equally across ancestry groups, they risk exacerbating health disparities because of broader structural inequities in healthcare delivery. A further limitation of the evidence base is demographic bias in research studies: participants are often disproportionately white, female, and highly educated. This highlights the need for more targeted efforts to engage non-white groups, men, and individuals with lower educational attainment or socioeconomic status to ensure diverse representation in both research and implementation.

Benefits, motivations, and participant agency

Despite these concerns, reviews highlight broad optimism about the benefits of genomics. Anticipated personal gains include more accurate diagnoses, personalised treatments, improved management of long-term conditions, and even potential cures. Altruism is again seen as also a powerful motivator, with many participants expressing a willingness to contribute to research for the sake of advancing science and benefiting future generations. New tools, such as polygenic scores, are seen as promising for providing more refined risk assessments and enabling earlier prevention. However, realising these benefits depends on responsible implementation, including clear clinical guidance, major infrastructural investment, and stronger regulatory systems to address existing weaknesses. A strong public preference for agency is also evident, with participants expecting shared decision-making, access to their results, and support for behaviour change within trusted and equitable systems.

5.2.4 Theses and dissertations

The following sources are included in this analysis:

1. Atutornu, Kafui Jerome, 2024. *Attitudes of Ghanaian People Towards Genomics and Sharing Genomic Data*.
2. Bartos, Meghan N., Mount Sinai Graduate School of Biomedical Sciences, 2021. *Attitudes on Pharmacogenetic Results as Secondary Findings Among Genetics Providers*.
3. Duggan, Estelle, University of Tasmania, 2021. *Factors Associated With Interest in Using Genetic Testing to Identify Predisposition for Psychological and Physical Health Conditions*.
4. Rao, Nandana D., 2022. *Population Genetic Screening in Adults: Implementation and Health Equity Considerations*.
5. Barnard, Pepita M., School of Computer Science, University of Nottingham (in partnership with Nottingham University Hospitals NHS Trust), 2021. *Young Adults' Considerations for Whole Genome Sequencing*.

Trust, data governance, and ethical safeguards

Trust is consistently identified as a fragile but essential condition for public engagement with genomics. Theses highlight that historical injustices, such as eugenics and unethical experimentation (e.g. Tuskegee Study), continue to shape perceptions of genomics and foster deep mistrust in institutions, particularly within communities such as the Ghanaian public. Concerns about privacy, surveillance, and misuse of data are widespread,

encompassing fears of commercial exploitation, bioweapons, and discrimination in employment or insurance. Some communities conceptualise DNA as *communal property*, creating tension with Western models of individual consent and underscoring the need for stronger protections in what is described as a “post-trust era”.

Equity, diversity, and inclusion

Equity concerns are a strong theme across the theses, with repeated warnings that genomic programmes risk reinforcing existing disparities. Underrepresentation of African populations in genomic research is viewed as a threat to social justice, with fears that this exclusion could lead to biased science and ineffective treatments. Stigmatisation and discrimination are also highlighted: sickle cell status, for example, can negatively affect marriage prospects or employment, while stigma around genetic testing for mental health conditions contributes to reluctance to participate. Evidence of lower enrolment among African Americans in population screening initiatives suggests these programmes risk replicating historical patterns of unequal access to genetic services.

Communication, education, and understanding

All five theses underline the persistence of low genomic literacy among both the public and healthcare professionals. Although public familiarity with genomics is often limited, many people draw on broader “science capital” from their everyday experiences and cultural context, enabling them to engage meaningfully in genomic discussions when the information is presented in accessible ways. Concerns remain about misinterpretation of results, information overload, and the psychological impact of genomic information. Calls are made for culturally and linguistically appropriate communication, using relevant metaphors and accessible terminology. At the same time, genetics providers themselves may feel unprepared to interpret and communicate genomic results, including pharmacogenetic findings, highlighting a professional training and support gap.

Benefits, motivations, and participant agency

Despite these challenges, the theses also reveal strong public interest in the potential benefits of genomics, driven by both personal and altruistic motivations. Anticipated benefits include more accurate diagnoses, personalised treatments, better health outcomes, and disease prevention. Altruism continues to play a central role, with individuals expressing willingness to donate DNA to advance science and address diseases prevalent in their communities. Alongside these motivations, there is a clear demand for agency and control: participants want transparency about how their data will be used, choice over what results are returned, and opportunities to shape their

involvement. For younger adults, engagement with genomics also extends to self-discovery and life planning, reflecting broader motivations beyond health alone.

5.3 Summary

Taken together, the grey literature paints a consistent picture of a public that is both hopeful about the promise of genomics and cautious about the conditions under which it is delivered. Across engagement reports, policy briefs, reviews, and theses, four key themes recur:

- trust and robust governance are indispensable but fragile, shaped by historical injustices and fears of misuse;
- low genomic literacy among both public and professionals requires clear, culturally competent communication;
- equity and inclusion must be actively safeguarded to avoid exacerbating disparities; and
- enthusiasm for the benefits of genomics is tempered by demands for agency, transparency, and meaningful involvement.

While the documents vary in scope—from direct public dialogue to policy frameworks and in-depth doctoral research—they converge in emphasising that successful population-level genomic testing depends not only on technical readiness but also on embedding fairness, trust, and shared decision-making at its core.

06 UK-specific findings



6.1 Overview

Thirteen UK-based studies, published between 2013 and 2025, were identified. Eleven explored public perspectives and two examined healthcare professional views. The studies were generally of good methodological quality, with large sample sizes across many of the quantitative analyses. However, there was a clear skew towards particular groups and methods, which limits representativeness.

- Stakeholders: public (11), healthcare professionals (2)
- Approach: quantitative (10), qualitative (2), mixed-methods (1)
- Method: surveys (5), discrete choice experiments (3), Likert-based questionnaires (2), focus groups (2), mixed (1)
- Risk types: Mendelian (7), pharmacogenomics (3), polygenic (1), mixed (1), not specified (1)
- Sample sizes: 22,012 participants across the 13 studies, ranging from small focus groups (n=62) to large online surveys (n=6,500)

6.2 Key findings

Across the UK studies, both the public and healthcare professionals reported generally positive attitudes to genomic testing, particularly when it was presented in terms of prevention or personalised care. At the same time, there were clear barriers that will need to be addressed if population genomics is to be implemented successfully.

6.2.1 Public attitudes

- Interest in testing increased when results were considered actionable or personally relevant, such as in the context of family history or previous adverse drug reactions. The literature pool explicitly excluded research focussing on populations where there was a clinical ascertainment (i.e. family history clinics, or personal history of cancer), where we would expect uptake and engagement to be higher.
- Concerns were frequently raised about privacy, data use, and potential discrimination.
- Engagement was stronger when trusted clinicians were involved in communication and interpretation of results.

6.2.2 Healthcare professional attitudes

- Healthcare professionals recognised the clinical value of genomic testing, particularly for pharmacogenomics and high-risk Mendelian conditions.
- Barriers included lack of time, limited training, and challenges around managing uncertain or ambiguous results.
- Positive attitudes were closely linked to prior training or direct hands-on experience with genomics.

6.3 Limitations of the UK evidence base

Despite relatively strong methodological quality, the UK literature is limited by its representativeness.

- Online surveys and discrete choice experiments, although often large in scale, were affected by selection bias. Participants tended to be healthier, white, and more highly educated than the general population.
- Communities with the poorest health outcomes, who may stand to benefit the most from population genomics, were not well represented.
- Better representation was achieved in curated focus groups, for example studies involving South Asian and Ashkenazi Jewish participants, but these remain limited in number.

6.4 Implications

To ensure that the evidence base more accurately reflects the diversity of the UK population, future research and engagement work should:

- Develop inclusive recruitment strategies that reach underserved groups.
- Combine large-scale quantitative approaches with targeted qualitative methods to capture depth, context and nuance.
- Provide training and practical support for healthcare professionals outside clinical genetics, particularly general practitioners and pharmacists, who will play a central role in programme delivery.

Taken together, these findings highlight both the opportunities and challenges for population-level genomics in the UK, and they provide a foundation for the strategic recommendations that follow.

07 International Literature



7.1 Overview

The main analysis in this review focussed on literature from the UK, mainland Europe, and Australia (Section 5) with UK-specific findings highlighted separately (Section 6). This decision was taken as findings from these manuscripts were judged to have greater relevance to the Programme and the immediate ambitions of the delivery team.

The broader international literature spans North America, Asia, and the Middle East, with substantial heterogeneity in study aims, sampling frames, and how “genomics” is presented to the public and healthcare professionals. Across regions, attitudes are shaped not only by perceptions of genetic risk and utility but also by the organisation and financing of healthcare, cultural norms, and national genomics initiatives.

Survey-based designs still dominate, often with large online samples; however, qualitative studies add context on trust, data governance, and familial decision-making. The framing of genomics—as a consumer service, a public health tool, or government-led infrastructure—strongly conditions expectations and concerns.

The description and design of these services, specifically pharmacogenomic programmes, have already been summarised [McDermott 2022]. We previously identified 40 international pharmacogenetic programmes across 104 publications, highlighting the diverse ways health systems have approached design, governance, and integration into electronic health records. We do not aim here to reproduce those descriptions; instead, our focus is on summarising international literature on public and professional attitudes to genomics, which offers complementary insights into likely opportunities and challenges for the Programme.

7.1.1 North America

North America’s health systems combine a largely insurance-based US model with a publicly funded Canadian system. Most North American literature arises from individual institutions within the US. This context ensures that cost, coverage and discrimination are highly relevant to public and professional attitudes, and clearly distinct from a UK model.

As in other regions, public awareness of specific genetic tests is modest, yet willingness to engage can be high when perceived benefits are clear or cost barriers removed. For

example, awareness of BRCA1/2 among Black women was low, but interest was strong when access was facilitated [Adams 2015]. In US national data, roughly a third reported awareness of direct-to-consumer tests, concentrated among more affluent, educated respondents, indicating the interaction between socio-economic status and exposure [Agurs-Collins 2015]. Perceived benefits commonly include prevention and empowerment, whereas concerns cluster around privacy and potential misuse in consumer or employer contexts [Boeldt 2015].

Cost, coverage and discrimination risks are recurrent determinants of attitudes. Willingness to undergo genetic testing is sensitive to out-of-pocket payments, and participants flag insurer or employer discrimination as key barriers [Botosaneanu 2011; Armstrong 2012]. Mistrust and perceived disadvantage are particularly important for some communities, notably Black African immigrants and refugees, where fear of exploitation or stigma dampened willingness to participate [Buseh 2014].

Healthcare professional perspectives emphasise the need for clearer clinical pathways and evidence before routine recommendation. Canadian primary care providers reported limited personalised medicine experience and a requirement for more training and implementation clarity, mirroring caution in US settings [Carroll 2016]. Acceptance is strongest where results are actionable and workflows or liability are clear; where these are uncertain, clinicians remain hesitant to adopt testing [Baker 2024].

7.1.2 Asia

Asian health systems are diverse, spanning mixed public-private financing and state-led precision medicine initiatives. The dataset includes primarily quantitative surveys and biobank-linked participant studies in Japan, with multi-group comparisons in Korea and a national survey in Pakistan.

In Korea, awareness of genetic testing was high, with most indicating they would test if costs were removed. Clinicians were comparatively more cautious and tended to raise privacy and discrimination concerns; among the public, paternity emerged as a common association with genetic testing, reflecting a different reference point for genomics in everyday life [Eum 2018]. In Pakistan, willingness to participate was strong despite only about half reporting familiarity with personal genomics, suggesting enthusiasm is outpacing literacy and underscoring the importance of education [Ilyas 2021].

Japanese studies show more reserved participation intentions in population-level initiatives, especially as burdens or risks increase. Many participants were unaware of large-scale biobank projects, and willingness to join sequencing studies decreased when additional commitments were introduced, including increased travel time, higher costs or

invasive testing [Miyamoto 2016; Okita 2018]. Nevertheless, when asked about return of results, most biobank participants indicated a preference to receive findings, particularly those of clear clinical value [Yamamoto 2017].

7.1.3 Middle East

In the Middle East, large government-led genome programmes shape expectations around stewardship, data centralisation and national benefit. The dataset includes mainly quantitative public and clinician surveys, with one qualitative study; several are explicitly linked to national programmes, including the Saudi Human Genome Program [Alrefaei 2022; Uddin 2024] and the Emirati Genome Programme [Ramadan 2025].

In Jordan, baseline awareness of genetic testing was reasonable but actual experience low. Support was strongest for initiatives already established in the region, such as premarital screening [Ahram 2015; Khdaier 2021]. In Saudi Arabia, surveys reported moderate awareness of the Saudi Human Genome Program, broad support for national initiatives, and gaps in understanding of data protection practices, highlighting the need for clearer privacy communication [Alrefaei 2022; Uddin 2024].

High willingness to participate was also observed in Qatar, where about 70% indicated readiness, particularly among those with higher education and trust in institutions [Rahim 2020]. Similarly, in the UAE, overall knowledge was described as optimal but accompanied by persistent misconceptions, suggesting targeted education should accompany rollout under the Emirati Genome Programme [Ramadan 2025].

7.2 Key differences and learning points

The international literature highlights how attitudes to genomics are shaped by local health system structures, cultural norms and national strategies. For the Programme, several learning points can be drawn:

7.2.1 Framing and funding

In the US, cost and insurance coverage dominate attitudes, whereas in the UK these concerns are less central. However, clarity around the long-term impact of these results on an individual's life insurance coverage should be clarified at the outset.

Middle Eastern programmes demonstrate how state-led, national framing can strengthen trust. The Programme can learn from this by emphasising NHS stewardship, public benefit and national equity as core values.

Asian experiences show that cost sensitivity and government endorsement strongly shape uptake. The UK should ensure messaging highlights that the initiative is endorsed by trusted institutions.

7.2.2 Consent, family and social norms

In parts of Asia and the Middle East, family obligations and stigma appear to influence decision-making. While UK frameworks tend to centre on more individualist models, the Programme can learn from these contexts by providing clear guidance on familial implications of results, including when and how to share findings with relatives.

Public associations of genomics with paternity testing in Korea highlight the importance of clarifying what population genomics is – and what it is not – in public-facing communication.

7.2.3 Data governance and trust

In the US, fears about commercial misuse of data limit engagement. The Programme should pre-empt similar concerns by foregrounding NHS governance, statutory safeguards, and strict limits on commercial access

In the Middle East, trust is often anchored in visible government leadership. The Programme can learn from this by making governance processes transparent and visibly accountable. However, attitudes towards government and deference to public figures vary considerably.

In Asia, willingness to participate can be high even where literacy is low. This underscores the need for simple, accessible explanations, backed by trusted institutions, so that engagement does not depend on specialist knowledge.

Overall, while structural differences limit direct transferability, the international evidence provides valuable lessons. The Programme should frame genomics as a national health asset, communicate clearly about scope and limits, address privacy and discrimination transparently, and ensure both individual autonomy and family implications are respected in practice.

08 Strategic recommendations



8.1 Key insights

Below are key strategic recommendations, structured around key insights, evidence gaps, and potential actions. These are presented using a theory of change logic model to link the conditions required for change, with engagement mechanisms at both local and system levels.

1. Communication around genomics and population testing is often complex, and both the public and HCPs find polygenic risk and pharmacogenomic information particularly challenging without professional support. Clear, accessible explanations are therefore essential for confidence and understanding.
2. Many healthcare professionals, especially GPs, report low confidence in genomics and highlight the need for targeted, practical training and digital tools to support their role.
3. Concerns about privacy, discrimination, and data misuse remain major barriers to engagement, with the evidence showing that transparency and clarity around data use and protection are central to building trust.
4. Service design strongly influences uptake, with HCPs preferring genomic services that integrate smoothly into existing workflows and public participants expressing clear preferences for non-invasive sampling methods and efficient turnaround times.
5. Both the public and HCPs are more motivated to engage with genomic testing when it is framed around clear, actionable health benefits such as prevention, early intervention, and more personalised care.
6. The evidence points to the importance of wrap-around clinical support and follow-up, both to reassure participants and to help manage uncertain or low-penetrance results that HCPs find difficult to interpret.
7. Clinicians are consistently viewed as trusted messengers whose endorsement influences public uptake, and their effectiveness depends on being equipped with

the right resources and communication tools. Public awareness can also be supported by other credible voices and influential figures.

8. Cost is a significant barrier to participation, with evidence showing that interest in testing increases substantially when it is available free of charge.
9. Psychosocial impacts such as anxiety, worry, and fatalistic thinking are frequently highlighted as risks of genomic testing, underscoring the importance of clear, accessible, and co-designed pre-test information.
10. Equity and inclusivity emerge as cross-cutting concerns, with evidence warning that population genomics programmes risk widening health inequalities if they do not consciously design for fairness and proactively address the needs of underrepresented groups.

8.2 Evidence gaps

Our review identified several important gaps in the current literature, particularly within a UK context. Most notably, healthcare professional (HCP) perspectives—especially those outside clinical genetics—are under-represented. To address this, we recommend targeted engagement with “non-genetic” HCPs to better understand their attitudes, concerns, and preferences. This may be facilitated by partnering with Royal Colleges, professional bodies and existing clinical networks.

There is also a lack of insight into the views of underserved or marginalised communities. To ensure inclusivity, engagement strategies should include focused community-based approaches, such as partnering with existing programmes (e.g. Genes and Health or Black Health Legacy) and hosting town halls or in-community workshops tailored to these groups.

Many studies present genomics as a single-point intervention, often tied to a specific test or moment, rather than as a longitudinal, lifetime relationship that creates potential value in the long term. Future qualitative work should explicitly encourage participants to consider the ongoing nature of population genomic programmes and how preferences, impact and value may evolve over time.

In particular, the concept of an “additional finding”, which has been extensively explored in the context of traditional rare disease or cancer genomics, is not well defined in this literature. Nor is there good quality research exploring individual’s attitudes towards these results within population level genomics programmes. An additional, or secondary,

genomic finding traditionally refers to a result which is of potential medical value yet is unrelated to the initial reason for examining the person's genome. Where the individual tested is apparently healthy, all findings could be considered as "additional" or "secondary" using its traditional definition, unless the scope of the test is clearly defined at the outset. What may be considered an irrelevant finding at the time of testing could be of significant actionability in the future. As such, it will be important to co-design pre-test information with participants and to clarify, or even re-define, the definition of both additional and actionable findings for a programme such as this.

As well as being relevant to an individual across their life, genomic results may be of value to other members of the family. This is straightforward to conceptualise in the context of results such as a *BRCA1* variant, but less straightforward for polygenic risk or pharmacogenomic variants. Depending on the final set of results returned to participants, and their care providers, engagement work should explore views around sharing information with relatives and define pathways to facilitate this in collaboration with healthcare professionals and relevant care organisations.

The evidence reviewed does not address the leadership and organisational structures required to deliver population-level genomics, which by nature is a cross-industry and cross-organisational endeavour. Ensuring public trust, equitable access, and effective user experiences will demand visibly diverse leadership that is both professionally and ethnically representative. Building such leadership capacity should be seen as integral to the success and legitimacy of the programme.

Furthermore, current literature tends to focus on theoretical perspectives, with little real-world evidence on the impact of population genomics initiatives. The Programme has the unique opportunity for continuous engagement throughout the programme to look at evolving views and generate timely evidence that can iteratively inform service delivery. We encourage a design-led mindset, with an ongoing relationship with participants that drives improvement.

We also note that while the reviewed research explores views and attitudes, it rarely evaluates what effective engagement actually looks like. Our review did not include reviewing effective engagement approaches. Thus, the insights generated should be combined with evidence-based strategies for effective engagement. There may be value in understanding the role (and importance) of branding, health promotion campaigns, and communication channels—to guide future engagement design.

Finally, much of the literature is framed around attitudes to health outcomes or risk, rather than to the design and delivery of services themselves. We recommend placing greater emphasis on understanding how different stakeholders wish to interact with population genomic programmes, such as the Programme. Focussed user research and service design methods are likely to add early value in both shaping engagement, as well as the delivery of the programme itself.

8.3 Theory of Change actions

To translate these evidence gaps into practical next steps, we have applied a Theory of Change and logic model approach. This framework helps distinguish between the conditions that need to change, and the mechanisms—whether localised or system-wide—that can achieve them. The following tables illustrate how specific barriers or opportunities can be mapped to proposed engagement mechanisms for the design of an effective and equitable Programme

8.3.1. Understanding non-genetic HCP views

Insights on perceived attitudes	Barrier to overcome	Condition for change	Engagement mechanism	Outcome
Limited representation of non-genetic HCP perspectives in current evidence	Lack of understanding of clinical realities and attitudes from front-line staff	Need to capture views from GPs, nurses, pharmacists, and public health practitioners across diverse settings	Local level: qualitative interviews, focus groups, surveys within primary/community care networks. System level: professional body consultations, integration of findings into workforce planning and speciality guideline development	Clearer understanding of HCP needs and concerns to inform design of scalable genomic programmes
Genomics perceived as a “specialist” or “add-on” service	Low integration into routine workflows and clinical identity	HCPs need to see genomics as relevant, actionable and achievable within their current role	Local level: co-design workshops linking genomic use cases to everyday care scenarios (e.g. cardiovascular prevention, prescribing). System level: inclusion of genomic content in professional development, educational curricula, and clinical decision support tools	Shift in perception of genomics from specialist to mainstream preventive care
Low genomic literacy and confidence among non-genetic HCPs	Limited training, time pressures, and competing priorities	HCPs need accessible, practical education tailored to their role	Local level: peer-led training sessions, case-based learning. System level: national e-learning modules, endorsed by professional regulators and integrated into mandatory training	Increased confidence and competence to apply genomics in everyday care
Concerns about patient impact and service burden	Anxiety about causing confusion, inequity, or adding workload	HCPs need clarity on support pathways, communication tools, and service design	Local level: pilot clinics with embedded evaluation, patient communication toolkits. System level: development of national guidance, referral pathways, and resource allocation for “wrap-around” care	Reduced anxiety among HCPs, improved willingness to offer genomic testing within routine care

8.3.2. Engaging underserved and marginalised communities

Insights on perceived attitudes	Barrier to overcome	Condition for change	Engagement mechanism	Outcome
Existing studies skew towards healthy, white, educated populations	Lack of representation and selection bias	Need to actively include underserved and marginalised groups in evidence generation	Local level: targeted focus groups, town halls, workshops co-delivered in community spaces. System level: partnerships with established programmes (e.g. Genes & Health, Black Health Legacy, etc) to scale inclusive recruitment	More representative evidence base that reflects diverse needs and values
Communities may feel excluded, mistrustful, or culturally disconnected from genomics research	Low trust and cultural relevance	Communities need to feel ownership and see genomics framed in ways that resonate with their lived experience	Local level: co-designed engagement with trusted local leaders, use of culturally and linguistically tailored materials. System level: national campaigns designed with input from community representatives	Improved trust, cultural relevance, and willingness to participate
Potential benefits of genomics not well understood or seen as relevant	Limited awareness of personal and community-level value	Communities need clear, tangible examples of how genomics can improve their health outcomes	Local level: storytelling, case studies, peer advocates. System level: integration of findings into public health messaging and service design, including representative visual materials	Increased awareness of relevance, reduced barriers to participation

8.3.3. Understanding the longitudinal value of genomic testing

Insights on perceived attitudes	Barrier to overcome	Condition for change	Engagement mechanism	Outcome
Genomics often framed as a one-off event (single test)	Limited understanding of genomics as a “lifetime resource”	Public and HCPs need to see genomic information as something that can gain value and relevance over time	Local level: longitudinal qualitative research (e.g., diary studies, repeat focus groups). System level: design of national programmes that emphasise lifetime use and evolving value of genomic data	Greater recognition of genomics as a long-term, evolving asset
Shifting attitudes and needs across life stages not captured in current evidence	Lack of longitudinal data on changing perceptions	Need for engagement approaches that follow participants through different health journeys	Local level: cohort studies with periodic check-ins. System level: integration of longitudinal engagement into programme evaluation frameworks	Deeper understanding of how genomic attitudes and values evolve over time
HCPs uncertain how to integrate genomic insights that change as new evidence emerges	Limited training and adaptive infrastructure	HCPs need ongoing support and updated guidance to incorporate new genomic evidence into care	Local level: clinician discussion groups, feedback loops in pilot sites. System level: regular updates to CPD modules, adaptive clinical decision support	Increased HCP confidence to use evolving genomic knowledge in practice

8.3.4. Evaluating effective engagement strategies in practice

Insights on perceived attitudes	Barrier to overcome	Condition for change	Engagement mechanism	Outcome
Current literature describes opinions but not what works in practice	Lack of real-world evidence on effective engagement	Need to systematically test and evaluate different approaches to engagement	Local level: pilot engagement projects using varied formats (e.g., workshops, community campaigns, digital outreach). System level: independent evaluation frameworks to compare impact across sites and populations	Evidence base for “what works” in genomic engagement, reducing reliance on assumptions
Branding, campaigns, and communication channels not systematically tested in genomics	Limited understanding of the role of messaging and media in shaping perceptions	Need to understand how branding, framing, and channels affect awareness, trust, and uptake	Local level: A/B testing of materials, community co-design of branding. System level: national pilots comparing communication channels (e.g., NHS campaigns, social media, trusted community voices)	Identification of effective comms strategies tailored to diverse audiences
Engagement often seen as a one-off consultation rather than an iterative process	Lack of ongoing monitoring and feedback	Need to embed continuous evaluation into programme delivery	Local level: post-engagement surveys, rapid feedback loops in pilots. System level: long-term tracking of awareness, trust, and participation across demographic groups (including building longitudinal digital relationships)	Ongoing improvement of engagement strategies based on real-world evidence

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

Insights on perceived attitudes	Barrier to overcome	Condition for change	Engagement mechanism	Outcome
Literature focuses on attitudes to outcomes, not on desired service journey and interactions	Limited understanding of how people want to engage with programme processes	Need to capture user preferences on consent, sampling, results, and follow-up	Local level: journey mapping, co-design workshops, usability testing with public and HCPs. System level: integration of insights into national service design and existing digital tools/infrastructure	Service pathways that reflect real user needs and expectations
Current service design often top-down, not co-created	Risk of poor uptake or dissatisfaction if services don't align with user needs	Need for participatory design methods that embed user voice throughout development	Local level: prototyping sessions, iterative feedback cycles. System level: adoption of user-centred design principles in programme governance and leadership	Greater usability, accessibility, and satisfaction with genomic services
Practical requirements (time, accessibility, digital literacy) not systematically considered	Risk of exclusion or friction in user journey	Need to design flexible, intuitive pathways that accommodate diverse needs	Local level: targeted testing with underserved groups to identify barriers. System level: embedding inclusive design standards across the Programme	Equitable, intuitive, and integrated user experience across diverse populations

09 Limitations and potential biases



This review was designed to be systematic, transparent and pragmatic, balancing rigour with the need to deliver timely insights for the Programme. The use of multiple databases, bidirectional citation searching, and grey literature sources strengthened the comprehensiveness of the evidence base. However, it is important to be aware of limitations and potential biases.

Some limitations arose from deliberate scoping choices (such as language and timeframe restrictions) made to ensure relevance and feasibility, while others reflect the methodological tools adopted (such as the use of structured templates and a single theoretical framework). These choices were appropriate for this review, ensuring that the analysis was focussed, relevant and delivered in a relevant timeframe.

9.1 Literature identification and search

This review made use of a structured, multi-modal search strategy, covering MEDLINE, Embase, Web of Science, and PubMed Central. The decision to focus on biomedical databases was pragmatic, reflecting the immediate need to capture literature most closely aligned with health service implementation. However, this choice could have led to under-representation of sociological or psychological perspectives.

We limited the review to English-language sources. This was necessary given time and resource constraints, but it introduces language bias and may have excluded relevant studies from non-English-speaking contexts. However, we would expect these sources to be of less relevance to the Programme.

The grey literature search focused on organisational websites and Google site-specific searches. While efficient, this approach risks overlooking perspectives from patient advocacy groups, charities, and other informal sources.

Finally, the decision to restrict the search to studies published from 2010 onwards was justified by the emergence of next generation sequencing in clinical use from this period. While this ensured relevance to contemporary practice, it may have excluded earlier but still influential research on public attitudes to genetic testing.

9.2 Screening and data extraction

Screening and extraction were performed using a structured template and supported by semi-automated methods, including NotebookLM prompts. This approach allowed for efficiency and consistency, which was critical given the volume of material and the timeframe for reporting. However, it is possible that this structured approach could constrain the capture of unexpected findings or nuanced insights.

9.3 Synthesis and theoretical framing

The Health Belief Model (HBM) was chosen as the primary framework for synthesis, given its established use in explaining health behaviours and its fit with programme needs. While this provides coherence and comparability across studies, it inevitably restricts interpretation within predefined constructs such as susceptibility, severity, and self-efficacy. Other theoretical lenses, such as trust frameworks or sociotechnical approaches, may have captured additional dimensions of public attitudes. Future engagement work could triangulate HBM-based synthesis with alternative models to build a richer, more holistic understanding.

The main analysis also narrowed the scope to UK, European and Australian studies and excluded direct-to-consumer genetic testing. These scoping choices were made to focus on evidence most immediately relevant to the Programme. However, this may exclude research which, regardless of its geographical setting, may nonetheless inform public perceptions of an initiative like the Programme.

9.4 Interpretation process

As this review was commissioned to inform the development of an active programme, there is an inherent risk of interpretive bias towards findings that align with immediate engagement needs. While efforts were made to report both enablers and barriers in general, some perspectives may have been underemphasised. This limitation can be addressed through ongoing engagement with diverse stakeholders to test and challenge the interpretation and gaps presented in this work.

10 Conclusion



This literature review reveals a clear enthusiasm from HCPs and members of the public for the clinical and personal opportunities which could arise from population genomics, largely driven by the potential for prevention, early intervention, and personalised health management. Both the public and HCPs perceive significant benefits, especially when testing leads to clear, actionable results or addresses personal and familial health needs. HCPs recognise the value in identifying high-risk individuals and informing preventive care, while the public views genomics as empowering, providing reassurance and control.

However, these positive perspectives are balanced by consistent and significant concerns. Both groups highlight substantial knowledge gaps and low genomic literacy, particularly concerning complex polygenic and pharmacogenomic data. The public harbours anxieties about privacy, data misuse, and potential discrimination, while HCPs grapple with insufficient training, limited time, and challenges in integrating testing into existing clinical workflows.

Critically, this report has identified key areas of future work, including the need to understand non-genetic HCP views, engage underserved communities, and design for user experience and service interaction. The ultimate delivery of success in this ambitious programme is intrinsically dependent on building strong public trust, ensuring clear governance, and fostering robust partnerships across all stakeholder groups from the outset.

Moving forward, success will require not only addressing these identified challenges but also maintaining a commitment to consistent and longitudinal engagement. This involves understanding how preferences and perceived value of genomic information evolve over an individual's lifetime and as new evidence emerges. This Programme has a unique opportunity for continuous engagement to iteratively inform service delivery and drive improvement, ensuring fairness, trust, and shared decision-making. By actively testing and evaluating different engagement strategies and adopting user-centred design principles, the programme holds significant potential to improve health and wellbeing, while positioning the UK as a global leader in large-scale genomics innovation.

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