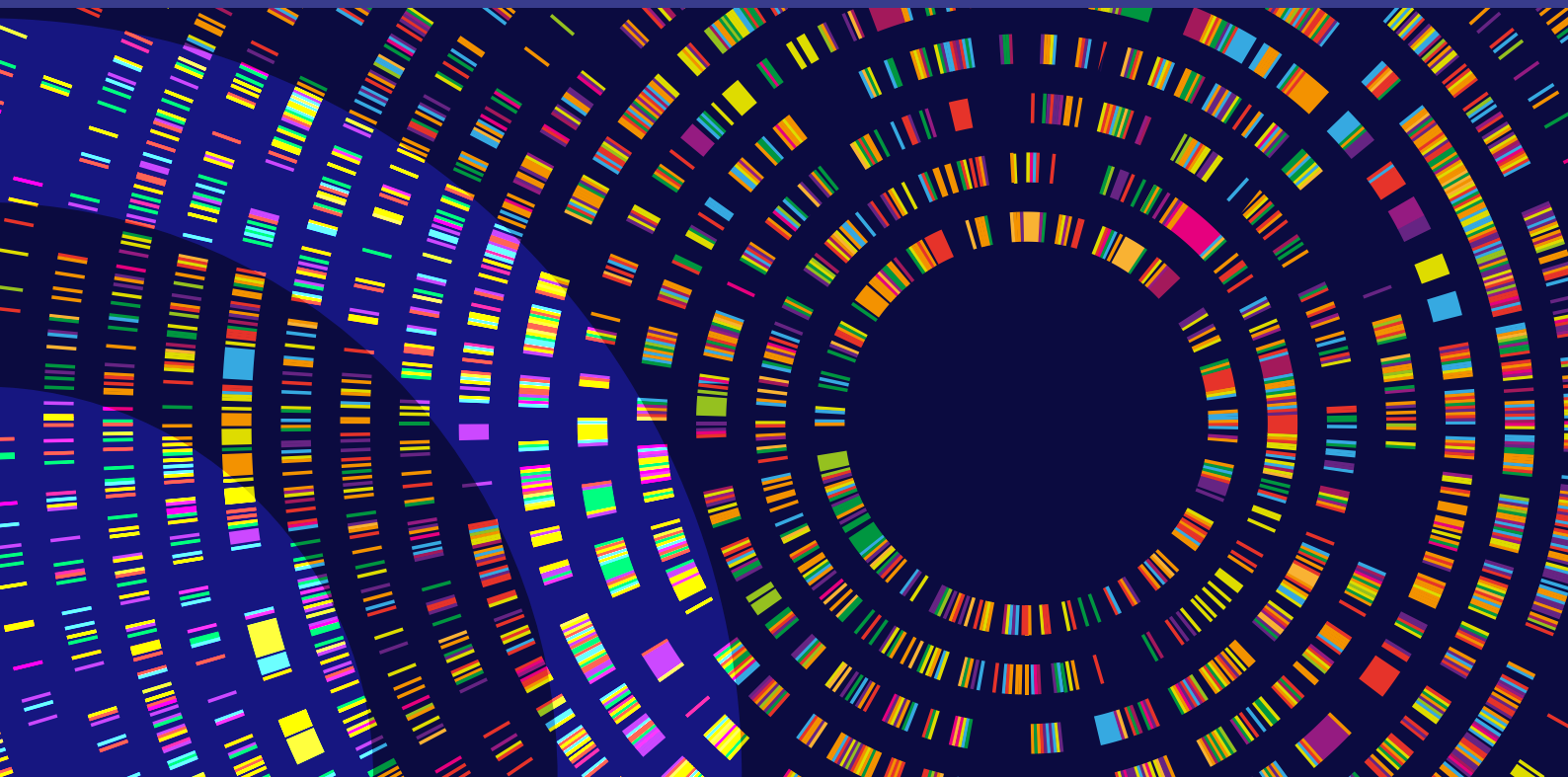


ETHNIC INEQUITIES IN GENOMICS AND PRECISION MEDICINE



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FOREWORD

Genomic medicine is still a relatively new service within the NHS; however, we have found that elements which reinforce ethnic health inequalities, including structural, institutional and interpersonal racism, are similarly entrenched in this service as they are in maternity care or in mental health services. Therefore, embedding genomics medicine within the NHS does not only signal the next big step in healthcare innovation, but also presents a real opportunity to instigate meaningful change early to help ensure no single community is left behind.

We know new frontiers in genomics and precision medicine have the ability to revolutionise the delivery of healthcare, and that accelerating genomic medicine is therefore vital for the future of healthcare in the NHS. There is a risk, however, that these advances could leave behind those communities that already experience ethnic health inequalities. A lack of diversity in datasets is a well-documented challenge not only in genomics and biomedical research, but across healthcare research in general. Historically most human genomic studies have been performed on populations of European ancestry. This underrepresentation limits the generalisability of research findings, as well as the viability of using genomics in the clinical care of persons of non-European ancestry, therefore exacerbating health inequalities.

Our goal is to work in partnership with leaders in this field to develop and implement actionable policy recommendations that will help ensure we have a truly equitable service available to all. This report provides the foundation of our determination to reduce ethnic health inequalities in genomics and precision medicine. It builds on our seminal report, *'Ethnic Inequalities in Healthcare'* and sets the scene for our upcoming projects: *'Understanding Ethnic Health Inequalities in the Genetic Testing and Diagnosis of Familial Hypercholesterolemia'* and *'Improving the utility of dihydropyrimidine dehydrogenase genetic testing in the NHS'*.

In examining ethnic inequalities in genomics and precision medicine, this report is the first of its kind investigating, exploring or addressing the NHS; however, turning the insights in the report into actions will be critical because we know that delivering equitable genomic testing for improved outcomes in cancer, in rare, inherited and common diseases, will play a pivotal role to reduce health inequalities and improve patient outcomes across all communities.

Professor Habib Naqvi, Chief Executive Officer, NHS Race & Health Observatory
Dr Veline L'Esperance, Senior Clinical Advisor, NHS Race & Health Observatory



OUR APPROACH TO LANGUAGE

Throughout this report we have adopted The NHS Race and Health Observatory approach to language when talking about race and ethnicity ².

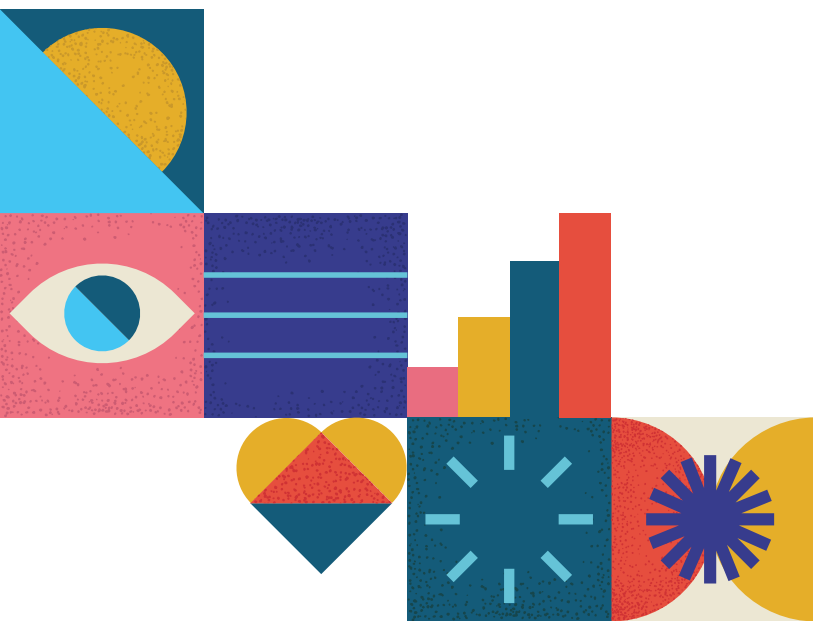
- Where possible we have been specific about the ethnic groups we are referring to.
- We have avoided the use of acronyms such as 'BME' or 'BAME'.
- We have used the terms 'ethnic minority' or 'minority ethnic' interchangeably when we are not referring to a specific ethnic group.
- We have been transparent about the language used within this report. We acknowledge that acceptable terminology will evolve over time and will be adaptable to changing terminology in our future work.

It is important to note, within this report, we present studies which involved reporting on previously published documents and research. The original terminology related to ethnicity and race used to describe included participants have been reported to ensure that the studies' results are accurately presented. In addition, direct quotations from stakeholders interviews may also use terms that may not be the preferred terminology of the authors nor the NHS Race and Health Observatory.

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 - Nottingham Muslim Women's Network
 - Moseley Community Hub
 - Loughborough Gurdwara
 - South Asian communities of Leicester, Loughborough and Nottingham



ABBREVIATIONS

UK	United Kingdom
NHS	National Health Service
GLH	Genomic Laboratory Hub
GMSA	Genomic Medicine Service Alliances
GWAS	Genome-Wide Association Studies
PRS	Polygenic Risk Scores
PHGF	Public Health Genomics Foundation
NHS RHO	National Health Service Race and Health Observatory
AI	Artificial Intelligence
NHSE	National Health Service England
NCIG	National Centre for Indigenous Genomics

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EXECUTIVE SUMMARY

BACKGROUND

Precision medicine strives to understand human genetic variation in populations and individuals, and how these patterns influence disease pathology and treatment, aspiring to develop tailored therapies that are most efficient. Precision medicine advances have resulted in personalised treatments becoming more embedded within healthcare. However, black and ethnic minority groups are hugely underrepresented in genomics and precision medicine research, with evidence further downstream of inequities in access to genomic medicine services that are developed as a result. These ethnic inequalities need to be better understood and addressed, to ensure individuals are not excluded from shaping and informing this field, and to ensure existing health disparities are not further exacerbated.

The objectives of the review were:

The headline objective of this review was to appraise and understand ethnic inequalities in precision and genomic medicine.

The specific objectives were to:

1. Understand current priorities in achieving ethnic health equity in precision and genomic medicine services, by reviewing policy and guidance documents.
2. Identify ethnic inequalities in recruitment for biomedical research and in patients' access to precision medicine, through academic evidence synthesis.
3. Identify educational and service needs for better implementation, by assessing stakeholders' knowledge and practice in promoting access to precision medicine services and assessing likely barriers and facilitators to access.
4. Investigate equity of access and uptake of genomic testing, by exploring current practices in data recording of protected characteristics (ethnicity) and evaluating key barriers and facilitators in data monitoring.

5. Identify information, education, and service needs for promoting equitable access among individuals from diverse ethnic backgrounds, by exploring the knowledge, attitudes and engagement of such individuals with precision medicine services/ research.

The research presented in this report employed a mixed methods approach underpinned by the pragmatic paradigm, choosing methods most suitable to address the above-stated objectives. The research comprised:

- A UK-based policy and guidance document review, which sought to explore the extent to which ethnic inequities are acknowledged in policy and guidance documents related to the field of precision and genomic medicine.
- An academic evidence synthesis to identify ethnic inequalities in precision medicine, focusing on recruitment biases in biomedical research and patient access to genomic medicine services.
- A programme of qualitative research, designed to gather the views of stakeholders representing clinicians, policymakers, academics, and NHS Genomic Medicine Service Alliance representatives, regarding current knowledge, understanding and practice relating to genomics/precision medicine research and genomics service provision. This exercise also explored views on promoting diverse access to genetic medicine services, and likely barriers and facilitators to access. Task group methodology was used to understand the views of individuals representing different ethnic groups on knowledge, understanding, attitudes and engagement with precision and genomic medicine research and health service provision.

Findings from each of the research workstreams were triangulated to inform recommendations for research, policy and practice, and stakeholder engagement – with the overarching goal to identify information, research, educational and service needs to promote more equitable access by these groups.

The work presented in this report was led by academics (including those with a clinical background) at The University of Nottingham and overseen by Dr Veline Lesperance, Senior Clinical Advisor, NHS Race and Health Observatory.

KEY FINDINGS

Policy and guidance document review

The sampling of documents focused on organisations and think tanks with a remit in the planning, or delivery of precision medicine and genomics-related healthcare, or for having an advocacy or priority/agenda-setting role within the field. Only documents published in the last five years (2017-2022), were included.

In total, 70 documents were identified and reviewed. These were published by one of the following: HM Government, the NHS, NHS Health Improvement, Public Health England, the Public Health Genomics Foundation (PHGF), Genomics England, Public Policy Projects, and The Kings Fund. Documents encompassed formal documents (such as official policies, guidance, and strategies) and organisational materials not published formally, such as reports, blogs, evaluations, information on webpages and white papers. Of these 70 documents, 50 contained textual information specifically related to precision medicine and/or genomics, which were extracted and analysed using inductive thematic analysis.

The results from the document review highlights that there is under-representation of ethnic groups in genomic datasets. These data limitations have far-reaching consequences for ethnic groups, regarding understanding on genetic variation and the development of optimal genomic medicine services, be it in terms of improving early detection and diagnostic tools and more timely interventions. Considering precision medicine research, a lot of the work currently being done on advanced analytic techniques - such as artificial intelligence-based risk prediction tools and the development of polygenic risk scores - is limited due to ethnic representation being poor in datasets. As such the predictive utility of such tests among diverse ethnic groups is reduced.

Efforts to involve diverse communities and individuals in public engagement work has been undertaken to inform genomic initiatives/action plans, but it is piecemeal and details on how this work is happening on the ground, is rarely acknowledged – though co-design and co-production is recommended by almost all organisations, in at least one document. A concerted effort is required to understand the needs of different ethnic minority communities to educate them on the concepts associated with genomic and precision medicine to enable informed contributions to the field. These concepts are not always well understood, and time and resources are essential to facilitate the education required.

Health service and system level improvements are needed to address current ethnic inequities in precision medicine, such as evaluation and monitoring of genomics services and initiatives. Diversity within the healthcare workforce was also advocated in documents, which is considered key to addressing historical barriers to access around mistrust and suspicion among ethnic minority communities. Training health service staff

to foster cultural awareness and understanding and embedding diversity within clinical care guidelines were also areas discussed as positive steps towards improving ethnic inequities in precision and genomic medicine. However, the nature and content of such training requires further unpicking, including more public engagement activity with different ethnic minority groups.

In conclusion, improved collaboration is warranted, among the research and academic community, and other relevant stakeholders (e.g. sectors involved in service design, delivery and recruitment, healthcare, analysts), government and public stakeholders (public, patients, community and faith groups) representing diverse ethnic groups.

Systematic review

The aim of the review was to explore ethnic inequalities in precision medicine, focusing on recruitment biases in biomedical research and genomic medicine and patient access to precision medicine. Four databases were searched with no temporal limitation up until October 2022. Out of 10,984 titles identified from the databases, 143 studies were included (137 quantitative and 6 qualitative). Most of the included studies provided basic descriptive information about ethnic minorities in the databases. As most studies used UK Biobank this information was identical in several papers. In terms of socioeconomic profile of ethnic minorities in the database, the actual genomic analysis of minority groups appears rudimentary, and findings were rarely interpreted in results and discussion sections. Although ethnic minority groups are incorporated into analysis, this is usually simply as a covariate in multivariate analysis rather than trying to identify clinically meaningful differences between ethnic minority groups and White European groups. 91.1% (n=123) of studies include ethnic minorities within the text of respective result sections. Of these, 106 (78.5%) performed statistical analysis of genomic data involving ethnic minority participants and 17 (12.6%) studies only stipulated ethnic minority study participants within demographic data without further statistical analysis. However, the lack of ethnic minority data was often recognised as a limitation.

Qualitative studies reported that ethnic minority groups had concerns about providing their samples for biobanking research or were reluctant to participate, due to negative historical experiences with researchers leading to questions around how their communities would benefit from such research. Addressing ethnic health inequalities in precision medicine is crucial for achieving more equitable healthcare outcomes. This requires efforts to increase ethnic representation in research studies, which will help to bridge existing gaps and ensure equal access to genomic services.

Stakeholder interviews

Semi-structured interviews were conducted with clinicians, researchers, community engagement representatives and policymakers. These interviews explored knowledge, attitudes, understanding and practice in promoting access to biomedical research and genomics medicine services for ethnic minority groups. Focus groups or semi-

structured interviews were also conducted with public stakeholders from different ethnic minority groups. These considered knowledge, awareness and attitudes towards genomics medicine services and precision medicine research. Informal focus groups and interviews were conducted with Genomic Medicine Service Alliance representatives to explore the monitoring of access and uptake of genomics medicine services by ethnic minority group. An inductive approach to the analysis of qualitative data was undertaken.

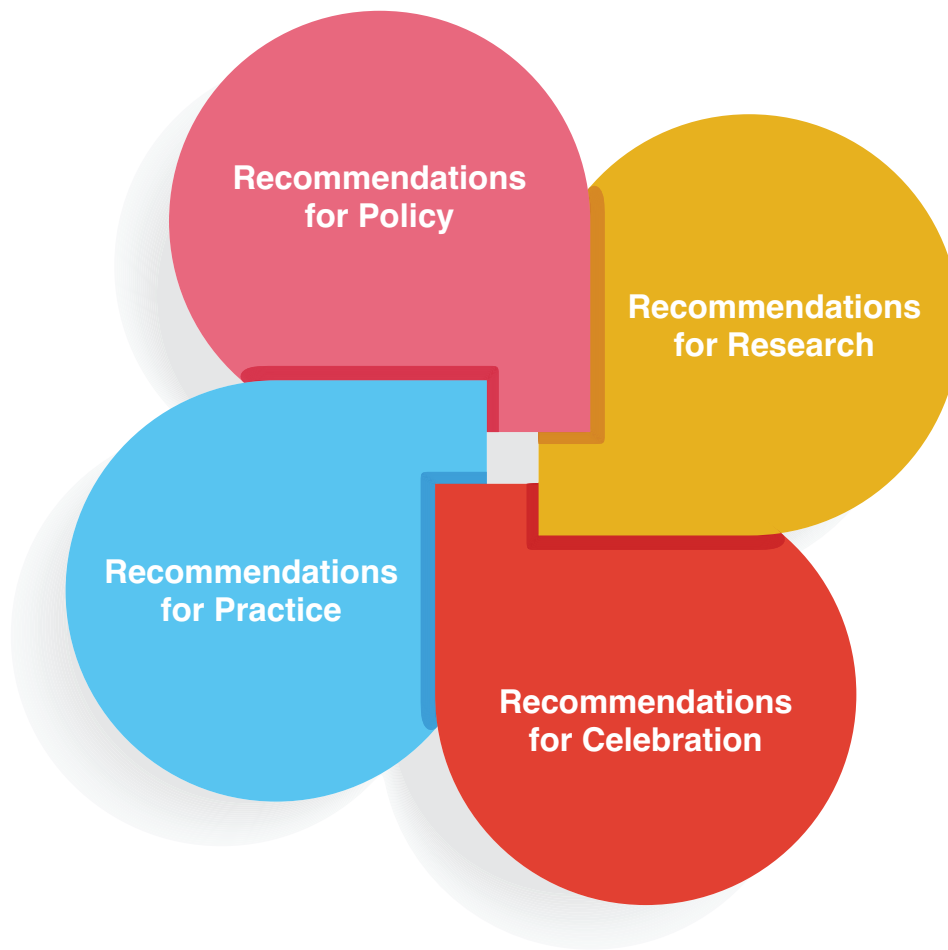
A total of 20 professional stakeholders were interviewed:- 8 were healthcare professionals, 5 were academics working in genomics and precision medicine research, 3 were policymakers, 3 were community engagement representatives and 1 participant was from industry. Representatives from 5 of the 7 Genetic Medicine Service Alliances participated in informal online focus groups or one-to-one interviews.

Ninety eight participants from ethnic minority groups participated in online or face-to-face focus groups or interviews. Participants were from Black African, Black Caribbean, Indian, Pakistani, Bangladeshi, Arab, and mixed ethnicity groups. Through the interviews and focus groups several themes pertaining to knowledge and awareness of genomics and precision medicine, key barriers, and facilitators to access and engagement and workforce training needs were identified.

Current knowledge and awareness about the links between genes and our health, terms such as genomics and precision medicine were variable across public stakeholders. Public stakeholders also queried knowledge and awareness of genomics and precision medicine research and the genomics medicine service among healthcare professionals working outside the field. Both public and professional stakeholders discussed ways to improve knowledge and awareness of genomics, precision medicine across ethnic minority groups. Healthcare professionals were seen as the first point of call for information. Additionally, faith or community leaders were seen as key points of contact to share information within ethnic minority communities. The need for tailored strategies for different communities and sustained messaging was also discussed by all stakeholders to improve knowledge, awareness, and engagement.

Barriers to accessing services and research included language, which encompasses challenges around accessing translators and accuracy of translation. Additional barriers included, mistrust, fear and suspicion of healthcare professionals, health systems and research. Acknowledging and understanding the reasons for mistrust were seen as key facilitators to improving engagement with services, testing and research.

Community engagement was raised by all stakeholders as key to improving equity of access and participation in research. A tailored and sustained approach to engagement is likely to be most effective. Workforce training needs were also identified. All stakeholders highlighted the need to improve genomics education across the healthcare workforce and other professionals involved in health decision-making. Cultural awareness and competency training for healthcare professionals and researchers not just in genomics and precision medicine but across the healthcare system is also needed.



RECOMMENDATIONS

Recommendations for meaningful community engagement and building trust

1. There is a need for meaningful, sustained, and tailored community engagement activities across the healthcare systems with NHS England to ensure all benefit from new advances and with researchers/research councils to ensure all communities can engage in genomic/precision medicine research.

Community engagement activities must:

- Focus on improving knowledge and awareness of genomics services and research through tailored engagement approaches enabling communities to make informed contributions to the dialogue (including e.g. via the NHS GMS People and Communities Forum, and NHS GMS Alliances). Examples of community engagement activities such as 'Genetics in Communities' and Genomics England's initiative to improve genomics literacy across England provide a blueprint for how to do this at a local level using co-design and co-production (see

Chapter 5).

- Include the development of an engagement space or platform that brings together communities to inform them about genomics and its potential benefits. Further work is needed to tease out the format of this platform and how this could be built and implemented.
- Inform the development and implementation of inclusive and accessible service provision for ethnic minority groups.
- Build trust and overcome barriers relating to mistrust (discrimination, fear, suspicion, past trauma, lack of understanding of genomics and precision medicine). To do this, key stakeholders including policymakers, researchers and healthcare service providers must listen to and acknowledge the challenges ethnic minority groups continue to experience collectively as a community and as individuals.
- Involve reaching out to and including underrepresented ethnic minority groups using tailored communication channels that are appropriate to different communities.
- Include clearly defined plans for implementation and must include mechanisms for monitoring and evaluation. In tandem with evaluating engagement initiatives, the sustainability and scalability of these approaches need to be assessed.
- Ensure public engagement is built into the development of future advances of health technologies within the fields of genomics and precision medicine. This will help to prevent further inequity and begin to build equity for ethnic minority groups.
- Are well supported with sustained financial, personnel and time investment.

Recommendations for policy and practice to ensure equitable access

2. All patients and healthcare professionals must have access to interpreters who are qualified and able to communicate complex medical terminology.
3. There is a need to develop a national Equality Diversity and Inclusion Framework with all relevant agencies in the NHS Genomics Medicine Services. The framework should also consider data governance and sharing policies that improve access to patient-level monitoring data for organisations key to implementing the NHS Genomics Medicine Services so that:
 - GMSAs can negotiate local data-sharing agreements with local trusts and Genomics Laboratory Hubs (GLHs) to obtain data on access and uptake of genomics services and testing for different communities.
 - GMSAs can evaluate current inequities in access by monitoring data relating to availability of tests, numbers and proportions of patients referred, ethnicity of patients accessing services and turnaround times for test results.
 - GMSAs and other relevant stakeholders can monitor care outcomes between different ethnic minority groups.
 - GMSAs can develop targeted community engagement strategies to improve knowledge, awareness and accessibility of services and genomic testing.

4. Regular monitoring, evaluation and publication of projects by NHS England Genomics Policy Unit which aim to address inequities in genetic medicine services and testing uptake must be routinely published and publicly accessible.

This should be routinely published, publicly available and shared across the NHS Genomics Medicine Services. Public authorities working in genomics, such as the NHS England Genomics Unit and NHS Genomic Medicine Service Alliances must hold key stakeholders to account through regular monitoring and evaluation of action and implementation plans.

5. NICE clinical care guidelines (e.g. implementation of pharmacogenetic testing) should acknowledge how population diversity relates to testing outcomes, health and disability. Where evidence shows ethnic differences, this should be included in NICE recommendations along with implementation tool to enable healthcare professionals to embed strategies to help facilitate equitable access into practice.
6. Better representation of ethnic minority groups within workforce across the genomics medicine services, precision medicine research and more generally across the healthcare service, including at leadership and decision-making levels. Further work is needed to explore how increasing diversity of the workforce can be achieved, perhaps through diversification of entry routes into medicine and applied healthcare training.

Recommendations for research: diversifying research participation

7. Governments, research bodies and funders should ensure research databases hold genetic information that is representative of our diverse population, with appropriate coding and recording of ethnicities. Work to increase representation of those that take part in research in genetic and precision medicine should be prioritised.
 - To improve understanding of genetic variation (according to ethnicity).
 - To improve subsequent development of genomic medicine services.
 - To ensure GWAS, PRS and other measures of risk are inclusive of different ethnic groups.
 - This should be underpinned by engaging with different ethnic minority communities (see recommendations on community engagement).
 - If ethnic bias is not addressed, ethnic inequities in genomic and precision medicine will be exacerbated. Oversampling of ethnic minority groups is recommended across genomic medicine research.
8. Ethnicity coding needs to be inclusive and consistent between different health services and electronic patient record systems:
 - Ethnicity coding should be developed in consultation with communities to ensure

- inclusivity and avoid mislabelling or arbitrary grouping.
- There needs to be joint efforts across the health service to improve data recording practices for protected characteristics such as ethnicity.
9. Lived and historical experiences of unethical research practices need to be addressed and factored in when developing genomics research with ethnic minority groups. To ensure that this is done in a sensitive and meaningful way, researchers must engage with communities and ensure that research practices are sensitive to the needs of participants from different ethnic minority groups.
 10. Research culture needs to change and develop more inclusive recruitment methods and research processes (e.g. informed consent, delivery of participant information):
 - In preparation for study recruitment, research teams need a clear plan to engage ethnic minority groups. This should include public engagement through tested communication channels and use of established community engagement models and support networks.
 11. When researchers apply to use established databases (e.g. UK Biobank, Genomics England databases) or apply to funding bodies for research grants (e.g. Medical Research Council, Wellcome Trust, National Institute for Health Research), research approval committees should appraise the study proposals on how they will incorporate information on ethnic minority groups in the overall sample.
 12. Legislation or official guidance for the UK pertaining to making research procedures and genomics research accessible for ethnic minority groups needs to be enacted. Lessons should be taken from the US where The National Institute for Health Clinical Diversity Act (2022) requires funding applications to provide clear plans for addressing accessibility and inclusion of diverse populations in clinical trials.
 13. Genetic ancestry should not be used as a surrogate measure of race and ethnicity in genomic research; however, ancestry does provide insight into genetic predisposition.

Recommendations for workforce training and education

Several training needs for the workforce have been identified that should be considered as part of the national strategy to embed genomics medicine services across the NHS. The training needs include:

14. A drive to improve the genomic education of health professionals from undergraduate to advanced postgraduate training and for healthcare professionals

currently working in the health service.

- For health professionals currently working in the health service training needs to highlight how healthcare professionals may already be interacting with genomics and precision medicine and show the relevance to their practice and their patients.
- Training for all healthcare staff must cover the implications of genetic diversity and cultural awareness. This training should address potential conscious and unconscious biases held by healthcare workers that may be affecting the quality-of-care patients receive. This will equip the workforce with an understanding of the needs of different groups, how to apply this knowledge to tailor conversations and inform interactions with patients. This will help to ensure that people from ethnic minority groups are receiving equitable care and support.

15. Providing general training around genomics services and precision medicine to non-healthcare workers involved in decision-making around healthcare such as social workers and other professionals (e.g. chaplains) involved in providing support to patients is key to ensure that minority groups can access accurate and reliable information.

16. Training around data collection for ethnicity and protected characteristics also needs to be developed and rolled out across the service. Healthcare workers across all levels who interact with patients as part of their role need to understand the importance of why this data needs to be collected and how to have a conversation with patients in a meaningful way.



1. INTRODUCTION

1.1. BACKGROUND

Precision medicine has the potential to transform health and medical care through “... *the provision of the right drug at the right dose to the right patient*”⁽³⁾. It rejects the notion that “one size fits all”, and thus strives to understand human genetic variation in populations and individuals, and how these patterns influence disease pathology and treatment - aspiring to develop tailored therapies that are most efficient. Precision medicine approaches have enabled the early detection of disease with personalised treatments becoming more embedded within healthcare^(4, 5). For instance, genotype information is used to make decisions about the best dosage of warfarin⁽⁶⁾. Precision medicine has also improved outcomes for cancers, for example, genomic profiling of breast and lung cancer tumours can help target treatment⁽⁷⁾. While these advances are promising, there are concerns that they may worsen health disparities,^(8, 9) particularly where race, ethnicity and ancestry are concerned^(10, 11). Diverse ethnic groups are hugely underrepresented in precision and genomic medicine research, with evidence also showing inequities in access to genomic medicine services that are developed as a result. Unsurprisingly, the engagement of some ethnic groups in all aspects of precision medicine research is also suboptimal. Additionally, such groups are rarely engaged, or when they are, this often seems tokenistic. Consequently, individuals representing the different ethnic groups are not part of shaping or informing the focus of this area of research. There is an urgent need to address these inequities to ensure that future advances in precision medicine research, and downstream healthcare developments are accessible to all.

1.2. THE GENOMICS LANDSCAPE IN THE UNITED KINGDOM

The United Kingdom (UK) is positioned as a global leader in precision medicine and genomics advances. In 2020, the UK government published a 10-year strategy to create the world’s most advanced genomic healthcare system to deliver better health outcomes at lower cost⁽¹²⁾. This strategy focuses on three key areas: diagnosis, predictive and preventative care, creating a seamless interface between research and healthcare delivery. Alongside these key areas are five cross-cutting themes comprising of engagement and dialogue with the public, workforce development, supporting industrial growth in the UK, maintaining trust and delivering nationally co-ordinated

approaches to data and analytics. In 2018, NHS England launched the NHS Genomic Medicine Service (NHS GMS) enabling the NHS to harness the power of genomic technology and science to improve the health of patients and the population served and to deliver on the genomics commitments in the NHS Long Term Plan (LTP) and the Accelerating Genomic Medicine in the NHS strategy, published in October 2022.

Genomic testing for the NHS in England is carried out by a national network of seven NHS Genomic Laboratory Hubs (NHS GLHs), with a shared aim to standardise testing, reduce variation, ensure equity of access, meet growing demand and provide access to the latest genomic technology.

Through the NHS GLHs, the NHS GMS delivers over 800,000 genomic tests every year for common and rare and inherited disease, pharmacogenomics, and cancer, as outlined in the National Genomic Test Directory and through a range of techniques, from WGS to the latest Next Generation Sequencing (NGS) technology.

The NHS GMS also includes seven NHS GMS Alliances, who work closely with the NHS GLHs and Clinical Genomics Services in their region, to drive the strategic systematic embedding of genomics into mainstream end-to-end clinical specialities and pathways. They also have responsibility for workforce development, delivering transformation and supporting the NHS Genomic Networks of Excellence and reviewing equity of access to genomic testing.

The patient pathway to accessing genomics services and testing involves a referral to clinical genomics services, where a clinical geneticist can request a test with their local GLH, pathologists or phlebotomists then take a sample from the patient and send it off to the lab for processing. At the stage of undergoing genomics testing, patients are also approached with regards to research involvement and inclusion of their samples in genomic databases.

1.3. RACE, ETHNICITY AND ANCESTRY

The conceptualisation and definitions relating to race, ethnicity and ancestry are ever evolving and the associated heterogeneity may lead to confusion within scientific and public realms. However, there are distinguishing factors and important implications separating the three terms within the genomic field ⁽¹³⁾. In precision and genomic policy documents and research, the terms are often used interchangeably, failing to recognise distinct differences.

Race is a largely socially constructed notion which categorises individuals and populations based on common phenotypes - namely skin colour. The term does not have a genetic basis and may vary significantly, geographically and temporally ⁽¹⁴⁾. The United Nations (UN) provide insights by broadly defining 'ethnicity' as relating to the social and cultural aspects of identity and is therefore a social construct. These aspects

are influenced by historical ties, geographic origins or shared experiences. Given the UN's broad definition, defining ethnicity can be largely affected by migration ⁽¹⁵⁾.

Ancestry refers to an individual's lineage and identifies the inherited origin of an individual. In genomics research (and over-the-counter genetic tests), there is a tendency for this to be based on the DNA analysis in the individuals. Unlike, 'race' and 'ethnicity', the term ancestry is a genetic/biological concept and hence is not affected by social or political constructs ⁽¹⁶⁾. For example, an individual could self-identify as Black race, with ethnicity as Latino of Spanish-Caribbean origin and Northern European genetic ancestry ⁽¹⁷⁾. There are concerns when genetic ancestry is considered as a proxy of ethnicity in genomic research ⁽¹⁷⁾.

1.4. SOCIAL DISADVANTAGE, INTERSECTIONALITY, HISTORICAL AND CONTEMPORARY ISSUES

Social disadvantage, intersectionality, historical and contemporary issues are often cited as factors contributing to ethnic inequity. These, therefore, offer some insights into why some of these ethnic inequities in healthcare and genomics, specifically, exist. This section considers these concepts and theories briefly, focusing on intersectionality, structural and systemic racism, literacy theory, the resistance model and competency theory (many of which have been studied or noted to play a role in relation to other ethnic inequities in other health fields).

1.4.1. Ethnicity and intersectionality

Acknowledging the role of intersectionality in relation to ethnicity must be noted too, as ethnicity does not necessarily provide the entire picture when attempting to understand inequities in precision and genomic medicine. Intersectionality is where different forms of discrimination and disadvantage (due to race, gender, class and other social identities) intersect and can lead to certain experiences of oppression or privilege. Individuals representing different ethnic groups may experience discrimination and disadvantage at the same time, causing exacerbation of disparities.

1.4.2. Structural and systemic racism

Racism operates through multiple avenues to predispose ethnic minority populations and individuals to health inequalities. An understanding of these avenues is important to address underlying nuances and prevent further exacerbation of inequalities. A proposed framework to consider the implications of racism involves the following classification: structural racism, interpersonal experiences of racism and institutional racism ⁽¹⁸⁾. Structural racism relates to the mechanisms through which societal structures, civil institutions or policies disadvantage minority ethnic groups. Systemic racism is a broader concept, as it not only engulfs structural racism but also cultural norms, societal beliefs and values ⁽¹⁹⁾.

Structural and systemic racism pose a significant barrier to ethnic minority groups accessing and being involved with genomic medicine services ⁽¹⁹⁾. This relates to a variety of often intersected dynamics. Historical unethical human experimentation on minority populations, such as the Tuskegee syphilis experiment have created significant distrust from within ethnic minority populations towards biomedical research ⁽²⁰⁾. The legacy of historical exploitation has led to underlying scepticism among minority populations fearing that genetic data and materials might be misused ⁽²¹⁾. Moreover, ethnic minority populations often face systemic inequities to accessing healthcare more broadly. Limited access to healthcare may lead to reduced awareness of genomic research and thereby limit engagement. Economic inequalities affect ethnic minority populations disproportionately and represent significant financial barriers, these may include transportation costs, time off employment and other study specific expenses ⁽²²⁾.

Implicit bias among genomic medicine practitioners and researchers potentially present a further barrier for involvement among ethnic groups in the field ⁽²³⁾. This highlights the importance of due assessment of implicit biases during study design and study implementation processes by research practitioners.

1.4.3. Literacy theory

Literacy theory highlights how differing levels of health literacy and language barriers among individuals may hinder access, comprehension and informed decision-making concerning involvement in genomic research ⁽²⁴⁾. Genomic research may include complex notions and terminologies which may lead to misunderstanding or reluctance to participate; thereby potentially preventing critical perspectives and evidence from being generated ⁽²⁵⁾. A potential strategy to address this barrier involves the translation of research materials (consent forms and participant information sheets) into languages better suited to the study participants ⁽²⁵⁻²⁷⁾.

A modern implication of the literacy theory relates to digital literacy ⁽²⁸⁾. Genomic studies may adopt online platforms to recruit participants, for data submission and communication purposes. Some individuals from ethnic minority groups may not have access to the required technology and are hence excluded; others may lack digital literacy, which thereby poses a significant barrier to access and involvement ⁽²⁹⁾. Lack of digital literacy affects elderly ethnic minority participants disproportionately ⁽²⁹⁾.

1.4.4. The resistance model

The resistance model refers to the notion that ethnic minority individuals from marginalised or disenfranchised populations may resist participation in genomic research specifically, and healthcare research altogether ^(30, 31). The model deliberates on historical and current inequities, imbalanced power dynamics, ethical concerns and cultural insensitivity in healthcare research ⁽³²⁾. Researchers ought to reflect on cultural insensitivity as a potential barrier which may alienate ethnic minority populations ⁽³²⁻³⁴⁾. Written and verbal communication between researchers and participants should be

culturally sensitive and reflect due consideration of participants' norms and beliefs, to promote engagement of ethnic minorities ⁽³⁵⁾. Promoting diversity and representation within genomic research teams help to recognise the needs of ethnic minority groups and would enable community partnerships to be built with ethnic minority communities ⁽²⁵⁾.

1.4.5. The competency theory

The competency theory relates to the cultural knowledge, attitude and practises upheld by research practitioners to address the needs of all populations in an equitable manner. This theory is underpinned by a collaborative approach throughout the decision-making processes of the research itself and the interventions/services proposed to ethnic minority populations ^(36, 37).

Ethnic minority groups participating in genomics research and services may add key knowledge, insights and perspectives which can enrich the methodology, results and recommendations of genomic studies ^(33, 36). The competency theory aims to recognise ethnic minority groups' expertise and agency to empower both researchers and participants to shape culturally sensitive and targeted research.

1.5. UNDERSTANDING ETHNIC INEQUITIES IN GENOMIC AND PRECISION MEDICINE. THE LITERATURE

The following section maps ethnic representation in genomic databases and genetic research, genetic testing and counselling, and finally, in relation to genome wide association studies, polygenic risk scores and other measures of risk.



1.5.1. Limited representation of diverse ethnic populations in genomic databases and genetic research

Genetic diversity is lacking in research and clinical endeavours^(38, 39). Research shows that certain populations are underrepresented in genomic and related large population databases which contributes to biases in precision medicine services, therefore also limiting the value of resulting treatments. This underrepresentation is not a new problem, it exists across biomedical research and is a limitation across healthcare systems generally (e.g. poor recording of ethnicity). Most genetic studies within the field of precision medicine utilise samples representing European ancestry, for example in the 500,000 UK Biobank^(10, 40, 41). Another issue is that ethnicity data are missing altogether, for a sizeable number of patients e.g., in the Prostate Cancer Biobank⁽⁴²⁾.

The varied ways in which race, ethnicity and ancestry data are collected, categorised and subsequently utilised within the field of genomic medicine leads to limitations in the use of such data within precision medicine and research. For instance, European populations have specific descriptions/categories such as “Scandinavian”, “Northern European”⁽⁴³⁾ however, categories for non-Europeans such as “Black”, “African ancestry”, lack geographical specificity^(11, 44). Moreover, where descriptors are used, they are often inconsistent. For instance in their review of GWAS studies, Mills and Rahal report 26 terms were used to describe African ancestry⁽⁴⁵⁾. As such, some researchers have called for guidelines standardising data collection and reporting within the context of health disparities⁽⁴⁶⁾.

Reasons for poorer representation of ethnic groups in genetic databases and research are well documented. Qualitative research highlights that whilst some ethnic minority individuals acknowledge the benefits of precision medicine, concerns exist, for instance, about the use of personal information and the likelihood of racial discrimination, as a result^(47, 48). Furthermore, differences between ethnic groups have also been reported. Rosa et al. report that African Americans and American Indigenous communities had more concerns about precision medicine than Latino and Asian Americans, with respect to unfavourable historical experiences of biomedical research argued to account for such differences⁽⁴⁷⁾. Further research also shows that whilst some ethnic groups do acknowledge the benefit of precision medicine, questions on whether all ethnic groups would benefit equally were conveyed⁽⁴⁸⁾. For instance, Yeh et al’s findings raise concerns, in the USA, about whether African Americans or Hispanics would benefit, due to problems with healthcare settings which were based on prior experiences of receiving health care and socioeconomic barriers^(48, 49). Other reasons for non-participation of ethnic minority groups in pharmacogenomic and other genomic studies were attributed to the amount of blood to be taken, concerns about who may access genetic information, privacy of data (e.g. use by third parties outside healthcare systems), and general mistrust of research^(49, 50). Where gender and age effects were also reported; higher percentage of men and older persons had concerns compared with women and younger people^(49, 50). Concerns around taking samples have also been reported in the English Longitudinal Study of Ageing and the Whitehall II Study of civil servants, where individuals representing ethnic minority groups were more likely to refuse providing a DNA sample compared to White participants⁽⁵¹⁾. Recommendations

are therefore proposed that collaboration with ethnic groups are necessary, to overcome barriers around mistrust, in order to encourage participation ⁽⁴⁹⁾.

1.5.2. Genetic testing and counselling

The literature is mixed regarding ethnic differences in genetic testing ⁽⁵²⁾. For instance, Yu's review found that compared with White women, those from Pakistani, Indian and Bangladeshi backgrounds are less likely to be offered prenatal diagnostic testing for thalassaemia. This is despite other studies reporting differences in attitudes towards prenatal screening among Pakistani and White women; though issues around asking the former for consent for thalassaemia testing have been reported ⁽⁵²⁾.

In contrast, there is evidence that suggests some groups feel that health professionals impose their own views, for example around prenatal screening, upon people from ethnic minority groups. This "directiveness" then steers these individuals' decisions ⁽⁵³⁾. This has been reported with South Asians in research on cancer genetics services ⁽⁵⁴⁾. Inability to speak English fluently or a poor understanding of the healthcare system has also been reported to hinder women's interactions with GPs about antenatal care ⁽⁵⁵⁾ especially with respect to decisions about genetic screening ⁽⁵³⁾.

Research on Down's syndrome antenatal testing highlights that ethnic minority groups had less capacity than White women to make an informed choice about such testing. This is due to lacking knowledge about testing, evaluating the positives and negatives and how these mapped to their own attitudes about the test ⁽⁵⁶⁾. Low awareness of cancer genetic services among different ethnic groups affects equitable access, with suggestions that White patients were also more likely to be referred to such services, which may also reflect differences in assessment/recording of family history ⁽⁵⁴⁾. However, many of these studies are either of low quality or lack details on differences according ethnic groups (such as often analysing or presenting data for all groups together).

Efforts to recruit ethnic minority groups exist, but are limited. For instance, the Genes and Health study (UK), ⁽⁵⁷⁾ has recruited large numbers of Pakistani and Bangladeshi people from community settings in East London, Bradford and Manchester, through bilingual health researchers. However, other genetic research, such as the Genetics of Mortality in Critical Care Consortium, which investigated the predictive value of genetic markers in predicting the need for critical care among COVID-19 patients, does not appear to have attempted to recruit an ethnically diverse population ⁽⁵⁸⁾.

1.5.3. Genome wide association studies, polygenic risk scores and measures of risk

GWAS have led to the identification of thousands of genomic variants associated with disease, through the vast numbers of populations that have facilitated the sequencing of the human genome⁽⁵⁹⁻⁶¹⁾. However, the representation of ethnic groups in GWAS is poor, globally. For instance, Landry et al. examined the populations included in genomic studies in the Genome-Wide Association Study Catalogue and the database of Genotypes and Phenotypes, and found significantly fewer studies of African, Latin American, and Asian populations compared to European populations⁽¹⁰⁾. In recent years though, participation of individuals from non-European ancestry has improved to approximately 20 per cent⁽⁶²⁾. Other researchers analysing GWAS in specific fields also show they are dominated by individuals of European ancestry (86% of total samples)⁽⁴⁵⁾. For example, Fitpaladi and Franks mined the NHGRI-EBI GWAS Catalog (2005–2022) for the most burdensome non-communicable causes of death worldwide, which investigated ethnic diversity (among other factors)⁽⁶³⁾. Most GWAS have been conducted in people of European ancestry, with East Asians being the second most represented ethnicity and African ancestry least represented in the field. The authors also found that GWAS research in diabetes, chronic kidney disease and digestive diseases are the most ethnically diverse.

GWAS have enabled the development of polygenic risk scores (PRS) that “*aggregate the effects of many genetic variants across the human genome into a single score*” and have been shown to improve prediction of future health outcomes e.g. cardiovascular disease^(64, 65). However, research inequities have resulted in poor utility of PRS in non-European populations, with reports that published PRS are three times more accurate for European ancestry compared to individuals of other ancestries⁽⁶⁶⁻⁶⁸⁾. Duncan et al. analysed polygenic scoring studies from 2008-2017 and found 67% of studies included exclusively European ancestry participants, 19% included only East Asian ancestry (e.g. Chinese and Japanese) participants, with only 3.8% of studies conducted among cohorts of African, Hispanic, or Indigenous peoples⁽⁶⁶⁾. Studies have shown that combining PRS from different ethnic group populations improves the predictive accuracy of models, for instance in type 2 diabetes and irritable bowel disorder; though the latter still has lower predictive utility for non-Europeans^(69, 70). Similarly, other studies for PRS in predicting age-related hearing impairment and prostate cancer were found to have less predictive accuracy among ethnic populations^(71, 72). This matter requires urgent attention to ensure that this does not result in a healthcare inequity, where at present, White individuals would benefit most in terms of identifying risk of disease and any resulting advances in precision medicine healthcare⁽⁶⁷⁾.

1.6. CLOSING REMARKS AND OVERVIEW OF WORK TO BE CONDUCTED

It is evident that there are various ethnic inequities in genomic and precision medicine which require further investigation to facilitate the development of recommendations to address the many limitations outlined in this introductory section. There is a need to understand the discourse on ethnicity in relation to genomic and precision medicine within policy and guidance documents, the academic literature and from the perspectives of various stakeholders representing individuals from Black, Asian and minority ethnic groups, policy makers, academics, clinicians and those working within the genomics medicine service.



2. OVERALL METHODOLOGY

2.1. PROJECT AIMS AND OBJECTIVES

Aim: to appraise ethnic inequalities in precision and genomic medicine.

The specific objectives were to:

1. Examine policy and guidance documents to grasp current priorities and strategies to ensure ethnic health equity in the development and implementation of precision and genomic medicine services.
2. Conduct an academic evidence synthesis review to identify ethnic inequalities in precision medicine, focusing on recruitment biases in biomedical research and genomic medicine and patient access to precision medicine.
3. Assess the views of stakeholders representing clinicians, policymakers, academics and NHS Genomic Medicine Service Alliance representatives to explore current knowledge, understanding and practice in promoting diverse access to precision and genetic medicine services. This includes deliberation of likely barriers/facilitators to access, to identify educational and service needs for better implementation practice.
4. Explore the current practices around data recording of protected characteristics including ethnicity as well as the key barriers and facilitators to monitoring of this data to assess equity of access and uptake of genomic testing.
5. Understand the views of individuals representing different ethnic groups on knowledge, understanding, attitudes and engagement with precision and genetic medicine services/research to identify information, educational and service needs to promote more equitable access by these groups.

2.2. METHODOLOGICAL APPROACH

A mixed methods approach was used comprising a review of policy and guidance documents, academic evidence synthesis, and interviews and focus groups with stakeholders. The document review was conducted first, which helped inform the search strategy for the academic evidence synthesis. The evidence synthesis, qualitative interviews and focus groups were conducted in parallel. Methods relating to each component are detailed in their respective chapters.

Outlining the philosophical paradigm underpinning this mixed methods study is also important. The pragmatic paradigm underpins this study ⁽⁷³⁾. The pragmatic worldview is not aligned with a single philosophy. Our position is that the mixed methods research conducted here is pragmatic in that the chosen approach was deemed most suitable to address the objectives ⁽⁷³⁾. The essence of the pragmatic paradigm is that it is problem-centred, where the most suitable approaches are employed to best address the research problem. Hence, it was considered the most useful and practical methodology for this piece of research ⁽⁷⁴⁾. Data from each of the studies are interrelated. Findings from all components have been integrated at the level of interpretation and reporting, where recommendations have been proposed together at the end.

2.3. STAKEHOLDER ENGAGEMENT ACTIVITY

A variety of engagement activities have taken place over the course of the project. First, the proposed study was presented to the RHO's stakeholder reference group, which provided an opportunity to discuss the study aims and methods in a broad sense, including terminology to be used (e.g. around precision medicine, genomics, underrepresented groups). Discussions also addressed practicalities to consider in conducting focus groups with different ethnic minority groups (e.g. separate gender groups, male/female facilitators, online versus in-person, different languages, incentives). This was followed by a separate meeting with members of the group that worked with specific communities (e.g., Roma, traveller and gypsy communities), and a group of ethnically diverse nurses who reviewed study recruitment approaches including consent process and topic guides for focus groups.

Following this, the research team identified gatekeepers for community groups representing different ethnic groups, who were contacted about the proposed work. These endeavours comprised either informal discussions or presentations providing an overview of the project, with a focus on the qualitative work to be conducted with individuals of different ethnic groups. This enabled the research approach to be adapted; for instance, we made changes to enable verbal consent to be obtained as this was considered important to facilitate engagement by different ethnic groups.

Following initial public stakeholder engagement, we also decided to change the approach from traditional focus groups to task group methodology, as community gatekeepers highlighted knowledge and understanding of concepts related to genomics and precision medicine were likely limited, and thus some education was required to facilitate more informed contributions by ethnic minority groups. Additionally, we offered both in-person and online sessions via Microsoft Teams and Zoom platforms to ensure that digital preference was not a limiting factor to participation. We also worked with gatekeepers to offer focus groups in different languages, where some kindly provided this service themselves (e.g. Arabic and Portuguese).

Interim findings were presented to the RHO's Academic Reference Group towards the end of the project. Preliminary findings from the qualitative work were presented back to one of the community groups who participated in the research, to sense check that preliminary themes resonated with individuals and discuss the recommendations that were being posited as a result of the project overall. This served as respondent validation (member checking) and an opportunity to provide feedback on the progress of the project to participants.

2.4. ETHICAL APPROVAL

The research conducted as part of this project was approved by the University of Nottingham's Faculty of Medicine and Health Sciences Ethics Committee (Study Reference: FMHS 32-0722).

3. UNDERSTANDING COVERAGE OF ETHNICITY IN POLICY AND GUIDANCE DOCUMENTS. A DOCUMENT ANALYSIS

3.1 INTRODUCTION

Health disparities according to race and ethnicity are well documented but the picture in relation to precision and genomic medicine requires further study. The material covered in the Introduction section of this report highlights that ethnic representation (and recording) is poor in genomic databases, in GWAS, and in informing the development of PRS and other measures of risk. Moreover, ethnic inequities have been documented in research that have explored access to genetic testing and counselling, including findings that fewer referrals to such services are made by health professionals for ethnic minority groups, and that health professionals hold stereotypical views and biases that can impact their practices.

Such disparities need to be better understood before they can be addressed both at a policy and clinical level, and there are suggestions that much of the work needed relates to limitations within existing genetic data sets ⁽⁷⁵⁾. Mistrust among ethnic minority groups, cultural and language barriers, lack of awareness and education, inequitable access to healthcare, are also reported to account for why representation of ethnic groups is poorer in genomic and precision medicine. Some of these are argued to stem from historical events, structural and systemic racism which can result in resistance among ethnic minorities to engage, and competency and literacy issues. All of these have been exacerbated by the socio-political context. Furthermore, the collection and use of personal data and the benefits of individuals sharing information (with public authorities and for research) is often poorly communicated and thus misunderstood among ethnic groups ⁽⁷⁶⁾.

The overarching aim of this study was to explore the extent to which ethnic inequities are acknowledged in policy and guidance documents related to the field of precision and genomic medicine, and the nature of this content. The specific objectives were:

1. To review policy and guidance documents to understand how ethnicity is acknowledged in relation to precision medicine and genomics.
2. To explore how ethnicity is considered in the development and delivery of precision medicine services.

3. To identify priorities, best practice guidance, barriers and facilitators related to ethnicity in precision medicine and genomics.

3.2 METHODS

3.2.1. Design

Document analysis was used to understand how ethnicity is covered and framed within the context of precision medicine and genomics, in official documents (policies or policy directives including strategy and implementation frameworks), official reports and in strategies for sectors on specific health problems, from a UK setting perspective.

3.2.2. Search strategy, materials and data collection procedure

Sampling of documents combined both purposive and snowballing approaches. Searches and identification of documents took place between September and December 2022 and were revisited again in February 2023. Initially, a list of organisations and Think Tanks were chosen as a starting point for searches to be conducted. These were chosen based on them having a remit in the planning, or delivery of precision medicine and genomics-related healthcare or having an advocacy or priority/agenda-setting role within the field. This step also involved experts in the field and NHS RHO to identify any policies or guidance they thought relevant for inclusion.

Therefore, we purposively searched for documents published by HM Government, the NHS, NHS Health Improvement, Public Health England, the Public Health Genomics Foundation (PHGF), Genomics England, Public Policy Projects and the Kings Fund.

The search focused on identifying documents that were classified as formal documents (such as official policies, guidance and strategies) and grey literature. This ensured inclusion of organisational materials not published formally, such as reports, blogs, evaluations, information on webpages and white papers. With health policy changing frequently, only documents published in the last five years (2017 until Feb 2023), were included.

Desktop searches were conducted through the worldwide web using Google Chrome and via organisational webpages, respectively. The latter included searching sections on webpages, along with conducting searches in the 'search and find' toolbar (on organisational websites), for each organisation using a set of keywords. Documents were read through in their entirety, and text related to '*ethnicity*', '*ancestry*', '*BAME/BME*', '*race*', '*diversity*', '*health inequalities*' '*health inequities*', within the context of precision medicine (including the terms 'personalised', 'stratified') and/or genomic medicine (including '*genetics*', '*genes*', '*DNA*' and so on) were extracted for analysis.

It is acknowledged that use of BAME/BME acronyms should be avoided; however, we found mention of these in some documents and thus included in searches, to ensure relevant text was not omitted. The terms used to decide on inclusion of extracts evolved iteratively, following preliminary scoping, and reviewing of documents, which ensured relevant data was not missed. Snowball sampling was incorporated to include further documents that were cited in those identified during the first steps, and these were subsequently reviewed using the same steps outlined previously.

To organise the data in a systematic and meaningful manner, information about each document was listed in Appendix 1: Policy documents summary. Information for each document was recorded, which covered: document title, document type, publication date, source, format (online or print including number of pages), general notes on the purpose of the document or broader contextual information about the document, and whether there was any relevant information on ethnicity (Yes/No). Text extracts that covered ethnicity (or associated terms as above) in relation to precision medicine and genomics were recorded in Appendix 1, for further analysis.

3.2.3. Analysis

Data extracts were coded inductively using a thematic approach. To generate themes to represent the nature of data extracts, Braun and Clarke's framework for thematic analysis was followed, ⁽⁷⁸⁾ where the stages of the process were supplemented by additional reading ⁽⁷⁹⁻⁸¹⁾. Initially, extracts were read and reread to facilitate data familiarisation, where understanding the semantic nature of extracts was realised. Further readings enabled the generation of initial codes, using an open and inductive approach. Subsequent readings facilitated analysis at a more interpretive, latent level, where initial codes were grouped together due to some shared meaning/relatedness, which led to the beginnings of theme generation, where a theme was characterised by a pattern of codes that shows something significant or of interest about the data ⁽⁸⁰⁾.

Initial themes and corresponding sub-themes were developed by MB, where the review involved critically considering that these were coherent and reflected the data, as well as being distinct from one another. To enhance the validity of the thematic framework and the trustworthiness of the findings, LJ and IB double coded a sub sample of extracts ⁽⁸⁰⁾. Themes were compared and minor refinements (e.g., organisation of sub-themes), were agreed. This process, referred to as investigator triangulation, ensured the analysis process was valid, collaborative and reflexive in nature ^(79,80). Following this, the final framework of themes and sub-themes was applied to the entire data set. This also enabled frequencies for each theme to be recorded, where some articles either covered multiple themes or the same theme more than once.

3.3. RESULTS

In total 70 documents were identified and reviewed (summarised in Appendix 1: Policy documents summary). Of these, 50 contained textual information specifically related to precision medicine and/or genomics. These extracts were included in the qualitative thematic analysis.

Qualitative themes

Four themes were generated from the data, each with corresponding sub-themes: 1. Ethnicity in relation to precision and genomic medicine, 2. Data recording on ethnicity, vision and the way forward, 3. Efforts to engage ethnic minority groups and, 4. Health service and system level recommendations. Each theme is covered in turn with supportive excerpts provided, throughout.

3.3.1 Ethnicity in relation to precision and genomic medicine

There were few examples or mentions of differences relating to ethnicity within genomics and precision medicine, in general terms. For example, documents published by the PHGF referred to ethnic minority groups being at risk of certain diseases such as cancer, breast cancer and cardiovascular disease. Ethnicity was considered in relation to risk or prevalence of certain rare diseases or conditions, particularly in documents published by PHGF:

In addition to age, smoking, obesity, and hypertension, risk of AMD (acute macular degeneration) is also associated with ethnicity. For example, research found that AMD prevalence was highest in people with European (12.3 %) and Hispanic (10.4 %) ancestry compared to Asian (7.4 %) and African (7.5 %) ancestry.

(PHGF, Age-related macular degeneration and genomics, 2021).

There was also a general acknowledgement that, *'Individuals may respond differently to a vaccine because of variations in immune response due to sex or ethnic background.'* (Vaccinomics, PHGF, 2021, webpage).

The role of ethnicity in relation to breast cancer was considered broadly in another PHGF document, and the importance of this within the context of identifying population sub-groups for directing prevention efforts, was warranted:

All individuals are at risk of developing breast cancer. However, some groups are at an increased risk due to factors such as age, biological sex, ethnicity, family history, genetics and lifestyle. Decision-making for individuals and planning at the population level is based on an understanding of risk profiles, and evidence-based consideration of preventive options together with the capacity of the sub-group with a specified level of risk to benefit. Identifying sub-groups for each preventive initiative is therefore important.

(Personalising breast cancer prevention – bridging the gap between research and policy, PHGF, 2020, p18).

Other documents explicitly noted that within genomics, inequalities relating to race and ethnicity have been deliberated upon. In a Public Policy Project report, it was concluded that, ‘... providing equitable access to genomics is a global problem that every country in the world struggles with; individual countries must endeavour to make genomics accessible to their entire population, with particular consideration shown for minority and Indigenous Communities.’ (Equity of Access and Return in Global Genomics, Public Policy Projects, 2022, p-28).

Initiatives and screening for diseases more inherent among ethnic minority groups such as sickle cell disease and thalassaemia were discussed as being important. One example, considered these inequalities in a global context too, suggesting the gravity of the matter in relation to genomics:

On a global basis, the countries that would be most able to benefit from genomics at a population level are the least equipped to do so. Further, most of the ‘genetic diseases’ such as hereditary cancers or inherited cardiac diseases have hitherto largely been studied in white Caucasian populations, meaning that variants that may be pathogenic (disease causing) in non-Caucasian populations may not ‘show up’ on the standard current comparison databases and vice versa, or at the very least there is less information available concerning the variants found in individuals from non-Caucasian populations.”

(Health technologies and social impacts, PHGF, 2019, p16)

3.3.2 Data recording on ethnicity, the vision and the way forward

Data recording on ethnicity was the most prominent theme running through many of the documents. Therefore, several sub-themes were identified. The first sub theme covers the need for more ethnically diverse data in genomic medicine. The second sub-theme focused on ethnic representation in artificial intelligence and machine learning and polygenic risk scores. Data recording, privacy and consent was also identified from sources as a matter to consider, especially as understanding about data collection and use is already remarked as being perceived as unclear among ethnic groups. When lack of diversity in data was considered in documents, research recommendations and the vision for tackling and improving such data was acknowledged, resulting in a final sub-theme that encapsulates these suggestions.

3.3.2.1 The need for more ethnically diverse data in genomic medicine

The ethnic diversity of data in relation to genomic medicine was documented, with acknowledgement that there is, '*an ethnic bias in most large genetic datasets and the bioinformatics tools used in healthcare.*' (Genome UK the Future of Healthcare, 2020, p39). Similarly, the PHGF highlights that databases are, '*...drawn predominantly from European and North American populations and may not reflect the genomics of other populations so well.*' (International lessons for personalised medicine, PHGF, 2021, Blog). Therefore, these limitations result in lack of understanding on diversity in genomic medicine, which in turn, makes it difficult to develop useful tests.

As such, various documents highlighted that datasets need to be more diverse, and that this was holding back progress within the field. Moreover, this under-representation was argued to be contributing to health inequalities, misdiagnoses, and poor healthcare, for ethnic groups:

The overwhelming majority of reference genomes globally are from Caucasians, while many ethnic groups are simply not represented⁽⁴⁶⁾. This is holding back progress in genomic medicine and compromising the quality of care for non-Caucasian patients, by increasing the chances of them receiving inconclusive results or erroneous interpretations of genomic variants⁽⁴⁷⁾.

(The Operationalisation of Precision Medicine, Public Policy Projects, 2020, p-28).

The extent of the problem was underscored consistently across different documents, and often more than once, in the same document:

The overrepresentation of populations from 'WEIRD' societies (western, educated, industrialised, rich, and democratic) in genomic databases has resulted in misdiagnoses, poor understanding of conditions and inconsistent delivery of care, as well as mistrust amongst excluded communities on the collection and use of their genetic data. As a result, genomic medicine does not always benefit all people equally.

(Diverse Data Vision, Genomics England, website)

Europeans represent 78% of people in genome-wide association studies (GWAS). Polygenic risk scores are 4.5x more accurate for people of European ancestry than of African ancestry. 7% of significant associations have been discovered in individuals of African ancestry. While only 2% of genome-wide association studies (GWAS) participants are of African descent.'

(Diverse Data Vision, Genomics England, website).

Reasons for this bias is argued to be multifaceted, where the availability of data, how data are prepared and amalgamated, how questions are formulated and pre-existing prejudices within society, are implicated. Moreover, the need to address the matter is underscored, or else existing health disparities for underserved groups would be further exacerbated. Despite widespread recognition that datasets need to be more

diverse, challenges were referred to, namely that curating, cleaning and preparing data was a time-consuming process, and thus there was not a quick fix. This was further compounded by the fact that,

'In reality, healthcare datasets are noisy, complex, heterogeneous, poorly annotated and generally unstructured'⁽³²⁾. In some cases, valuable health data may not even be captured digitally – the most fundamental prerequisite for building AI models. For these reasons, extensive effort has to be expended on collating, cleaning, standardising, and formatting datasets before they are used to develop algorithms... Genomic data has several sources of error and biases including those stemming from differences across various laboratory sequencing kits, methods and technologies, as well as technical sequencing artefacts.

(Artificial intelligence for genomic medicine, PHGF, 2020, p38).

3.3.2.2 Artificial intelligence (AI) and machine learning and PRS

Documents (note these were predominantly those published by PHGF) considered ethnicity in relation to AI and machine learning and PRS in more detail, therefore this warranted a standalone sub-theme.

Several documents stated that limitations inherent within existing datasets make machine learning algorithms less effective, as they are based on samples comprising predominantly European ancestry individuals. It was also advocated that more transparency was needed about data limitations too. As such, documents were unanimous in stating that these AI models, *'...will be less effective than those trained on a fully representative dataset, and potentially even incorrect and harmful, if used to make predictions on individuals of non-European descent.'* (Artificial intelligence for genomic medicine, PHGF, 2020, p39). With this said, commitments to addressing the issue were also reported, with reference to the UK's five million genomes programme and in the US, the National Institute of Health's 'All of Us' program which is striving to sequence a diverse sample; suggesting that this is a matter to be addressed on a global scale. Another UK example, suggests that despite such issues/challenges there was a sense of progress related to a couple of AI initiatives, which were focusing on ethnicity:

'The NHS AI Lab and the Health Foundation have awarded £1.4 million in funding to 4 projects to: understand and enable opportunities to use AI to ensure innovation happens in response to the health needs of ethnic minority groups; contribute to improving the quality, availability and appropriate use of datasets to account for ethnic diversity in the development of AI models; improve the development, testing and deployment of AI models across patient populations to reduce bias; and improve the performance and accuracy of emerging and existing tools for different subpopulations...'

And the same document goes on to state, *'(in the UK Biobank section) a University College London study into factors increasing risk of dementia. Researchers were able to study participant blood samples to identify how genetic makeup affects risk. The*

diverse participant pool has enabled the research team to explore ethnic differences in dementia risk' (Data Saves Lives: Reshaping Health and Social Care with Data, 2022, website).

PRS and ethnic representation were unpicked in more explicit detail across a variety of documents, particularly those published by PHGF. First, it was suggested that the purpose of tests needs to be clear, along with how such models have been developed, as these have implications for their relevance and utility. It was consistently mentioned that these risk scores are not based on datasets representative of ethnic minority groups, and therefore their predictive utility was scrutinised. Unsurprisingly, these limitations were attributed to the fact that datasets containing genomic information on individuals from different ethnic backgrounds are scarce. Thus, the need for increased diversity in data to generate PRS with better predictive utility, to better understand genetic variation and early detection for improved diagnostics and more timely interventions.

Most of the documents that explicitly touched on ethnicity and polygenic risk stated,

Polygenic scores have mostly been developed in exclusively or majority European populations. Some preliminary assessments have demonstrated that scores can still discriminate between high and low risk groups in other ethnicities, but that they don't perform as well...'

(Polygenic scores, risk and cardiovascular disease PHGF, 2019, p50).

A more recent document also published by PHGF, suggests that these limitations raise ethical concerns, '*...datasets in which polygenic score models are constructed and validated need to be representative of the population in which their use is intended. Differences between the two, as a result of ethnic mix or different health and age profiles can lead to poor performance or inability to generalise the model*'⁽⁶²⁾. *In some instances, models have been developed that function best in specific populations, which raises questions as to the ethics of implementing polygenic score models that are not generalisable*'⁽⁶³⁾. (Polygenic scores and clinical utility, PHGF, 2021, p34).

The areas of dementia, breast cancer and cardiovascular disease were also discussed in several documents (published by PHGF), suggesting that these models were further along in terms of considering ethnicity.

For dementia, it was acknowledged that demographic models do include ethnicity, and that prognostic models that aim to predict dementia risk have been validated in different ethnic groups. However, further testing among different ethnicities was still required (due to insufficient ethnic representation in data, across different ethnic groups and ages) (Dementia risk prediction, PHGF, 2019). The Confluence Project was mentioned briefly in one document, which is a breast cancer GWAS working to improve polygenic risk prediction across ethnic groups and different breast cancer types. Whilst this document suggests this is '*Providing new avenues to engage with specific populations...*', (Personalising prevention for breast cancer, PHGF, 2019, p50) how this will be done, particularly among ethnic groups is not explicit. When considering cardiovascular disease, ethnicity is recognised as a risk factor, which is likely to be due to, '*biological,*

environmental and cultural factors', (Polygenic scores, risk and cardiovascular disease PHGF, 2019, p31). Another document focused on how PRS for cardiovascular disease could be implemented into the NHS Health Check Programme to analyse utility. However, again, the need for more work with ethnic minority groups was advocated and recognised as a limitation. The recognition of this limitation renders these tools inefficient for implementation into clinical practice and questions why such plans are being proposed when the tools are inefficient:

This report therefore addresses how polygenic score analysis for CVD could be implemented into the NHS Health Check programme as a potential route to providing polygenic score analysis at scale within the population of adults in England and Wales. It assumes that the mechanism for delivering polygenic scores to individuals will be primarily using existing NHS Health Check pathways, infrastructures and workforce. In this report we make the assumption that the use of polygenic score analysis in this population has clinical validity and utility. Although the evidence base concerning the clinical validity and utility of polygenic score analysis for CVD is growing, more work is needed to prove demonstrable validity and utility, especially across the diverse ethnic groups within the population

(Implementing polygenic scores for cardiovascular disease into NHS Health Checks, PHGF, 2021, p50).

Examples of current initiatives to attempt to bridge the gap were commented on, but details on implementation to improve ethnic representation were lacking:

Current initiatives such as plans to sequence the genomes of up to five million individuals over the next few years from UK Biobank and NHS patients, and to develop a cohort of healthy participants as part of these sequencing efforts, are important opportunities to address this need. Going forward, it will be important to ensure these cohorts contain the appropriate population mix for validation of robust polygenic scores.

(Polygenic scores, risk and cardiovascular disease PHGF, 2019, p70)

Genomics England is undertaking a diversity in genomic data initiative, which has been created with the purpose of enriching genomic datasets by engaging with relevant communities, sequencing consented cohorts from diverse backgrounds and developing analytics to derive the most value possible from the data.

(Accelerating genomic medicine in the NHS, NHSE, 2022, webpage).

Other UK-based initiatives were provided as examples too, that were striving to recruit diverse participants (e.g., Accelerating Detection of Disease Challenge), which also builds on the UK Biobank programme. Another example of how to achieve this was via future GWAS. There were even examples of how the UK was seemingly a global leader in the field of genomics and that diversity was a key component of either existing or planned programmes, and that this vision was already being implemented:

...Greater diversity in the dataset will lead to greater scientific understanding. Examples of programmes that are seeking to rectify the diversity disparity within existing genomic datasets such as Genomics England's 'Diverse Data Initiative' and ELIXIR's 'Beyond 1 Million Genome Project', mark the recognition that more needs to be done. However, such effort require coordination and cannot be left to emerge from uncoordinated decisions by national and independent institutions. (Bringing the Benefits of Genome Sequencing to the World, Public Policy Projects, 2022, p-16).

3.3.2.3 Data recording, privacy and consent

A couple of documents such as those published by Public Policy Projects highlighted that processes were needed to reassure ethnic minority individuals about data privacy and storage, to overcome historic barriers to participation. Other documents debated the anonymity/identifiable nature of genomic data (where ethnicity was mentioned broadly). For instance, a PHGF document on Identification and genomic data states, *'Our position is that genomic data are not exceptional but sometimes do possess characteristics that challenge the law and policy relating to anonymity* ⁽³⁾*... (p12). In the same document it is suggested that certain endeavours, will require appropriate consent practices, which would also help to address historic barriers around ethnic groups not sharing information or being suspicious of research. This does also link to data from public stakeholders, that suggest clear information on data practices needs to be provided from the outset. The importance of this matter was further emphasised when considering the use of phenotype data:*

More recently, researchers have claimed to predict biometric traits using whole genome sequencing, detailed phenotyping and statistical modelling in a cohort of participants of diverse ancestry ⁽⁷⁵⁾*. Although the significance of these findings have been challenged (76,77), this work raises the possibility that associating deidentified genomic data with phenotypic measurements such as height, skin and eye colour, facial structure and voice might have implications for personal privacy and consent practices.*

(Identification and genomic data, PHGF, 2017, p32)

3.3.2.4 Research recommendations and ambitions

When considering the improvements required in terms of data diversity, research recommendations were routinely presented around the need for clinical research to be more diverse. Ensuring data diversity in relation to ethnicity was dependent on concerted efforts between industry, academia, funding bodies and the NHS. Suggestions have been made for implementation of, *'Genomics England's proposal for a mandatory equality impact assessment for clinical research, to quickly and decisively expand the diversity of the genetic database'*, (The Operationalisation of Precision Medicine, Public Policy Projects, 2020, p-5), where this same document also advocated for oversampling of ethnic minority groups. Reaching and inviting underrepresented

groups into research, and explaining the benefits and risks, have been highlighted as means of addressing biases in data; otherwise, we would be faced with 'health data poverty' (Genomics Revolution, Public Policy Projects, 2021). Communication needs to be delivered through appropriate language mediums and platforms that are amenable to diverse groups. In contrast, another document argued that addressing the data diversity issue was not enough, and that a more diverse research culture was also warranted:

Change, however, will not simply be achieved by creating a more diverse database; there is also a global need to create a more diverse research culture. This will require a different approach to the peer review process, allocation of funding grants, selection of cohorts and editorial boards as well as building capacity locally.

(Bringing the Benefits of Genome Sequencing to the World, Public Policy Projects, 2022, p-28).

Important to note is that these sorts of recommendations were not only related to genomic medicine, but that clinical research in general needs to, 'Reflect the diversity of the UK's population – system partners, including the medical research charities, will work together to proactively increase the racial, age, gender, and geographic diversity of clinical trial participants and those in real world data sets.' (Life Sciences Vision, 2021, p20).

As such, many of the documents that made such recommendations, often presented these as either 'visions' or 'ambitions' for the future. For example, The Women's Health Strategy for England (2022) added that their 10-year ambition was that '...research is representative of society, with increased participation of women and other groups who have historically been under-represented in research. Funders and researchers address barriers that may prevent under-represented women from participating in research, including women from ethnic minority groups...', (p65). There were examples of this being somewhat limited in scope, or of these plans/priority setting being in their infancy, where research was required to help understand and delineate areas of focus. Other documents went a little further to identify specific populations of interest:

...The key populations of interest vary by both condition and the research focus of the major programmes (which will be refined based on a collaborative priority-setting exercise), but broadly, our communities of interest include: Who: Multi-ethnic cohorts; Our best proxy: People with ancestry from India and Bangladesh (other communities with strong representation in the UK include Philippines, Sri Lanka, Turkey, Iran, Iraq, Malaysia, Afghanistan, Brazil, Nepal); Why: High levels of differentiation between different ethnic groups studied and different linkage disequilibrium patterns...

(Diverse Data Strategy, 2022, p11).

One aspect that was referred to more consistently, was a commitment to invest in the field of prevention and early detection, focusing on at-risk and diverse populations. For example, Genome UK highlighted that, 'We will use genomics to accurately predict

the risk of chronic diseases and our national screening programmes will use genomics to identify at-risk populations. Preventing disease before it begins is key to our future healthcare system and requires the right technology, large diverse datasets, and validated analytical tools to predict the risk of disease.' (Genome UK the Future of Healthcare, 2020, p27)

3.3.3 Efforts to engage ethnic minority groups

A few documents mentioned some public engagement activities that had either been conducted or were recommended, to inform the design of action plans, commitments (services to be developed) or actual precision medicine/genome programmes. Inclusivity was suggested as something that patient and public involvement (PPI) needed to champion, and that '*...appropriate accommodations (be) made for those with additional support needs and involving participants who reflect the diversity (in every sense of the word) of the population that the NHS serves... The types of considerations that might be needed include: How best to recruit a diverse range of participants, especially participants from marginalised groups.'* (Better, Broader, Safer: Using Health Data for Research and Analysis, 2022, p165).

Two sub-themes were derived. The first sub-theme relates to public engagement activity that has used/advocates use of co-design/production methods. The final sub-theme covers broader level public engagement in relation to informing action plans, future commitments and programmes. It should be noted that quite often, terms such as "diverse" and "marginalised" groups were referred to more broadly when these documents covered public engagement. The exact proportions involved in public engagement activity, was rarely detailed.

3.3.4 Using co-design methods to engage

There were some examples of efforts to engage with diverse communities and individuals, to inform the development of genomics programmes to ensure inclusivity (across various characteristics, rather than ethnicity specifically). A Public Policy Projects report (The Operationalisation of Precision Medicine, 2020) referred to encouraging outcomes resulting from public engagement activity undertaken by Genomics England to understand the attitudes of people from Black African and Black Caribbean communities regarding participation in the 100,000 Genomes Project. In short, this work highlighted biases in culture, systems and practice which undermined diversity in research participation, leading to the development of a mandatory equality impact assessment for clinical research.

Other examples of public engagement activity were also evident. For instance, the Newborn Genomes Programme (2021) had a section on public engagement, that involved a '*diverse range of communities and groups*' (p6), to inform the pilot, which included reference to ethnic minorities (p6). However, there was no information on the proportions of individuals that represented ethnic minority groups (N=130 for the

work). Encouragingly, this initiative went on to mention that, *'Co-designing the pilot with patients, families, and clinicians will ensure that we understand their needs and concerns, and act on experience.'* (p1).

Whilst The Diverse Data Vision (Genomics England webpage) did not explicitly refer to co-design and production, some of the vision statements proposed infer this is likely needed, to address some of the inequities evident in genomic medicine:

'Close the gaps, together - Convene and work with patient, genomic and data communities to design, develop and implement equity-enhancing strategies... Products, tools, & behaviours: Bridge the data gap - Work with clinicians, analysts, researchers, patients, and community groups to develop tools, and processes to improve research, service-delivery practices, recruitment and care...'
(webpage reference?).

The Women's Health Strategy for England (2022) document also suggests that co-production is involved in relation to addressing disparities within the maternity system including those related to ethnicity:

'To address these disparities, local maternity systems received £6.8 million of funding in 2021 to 2022 to co-produce and implement their equity and equality action plans, including their implementation of continuity of care for black, Asian and mixed ethnic groups, and those living in the most deprived areas.' (p95).

3.3.5 Effective engagement to inform action plans

A couple of documents underscored the importance or made recommendations about engaging those from ethnic minority groups, in precision medicine. Several documents, including many of those published by Public Policy Projects, stressed the need to recognise the historical trauma and colonialism that have led to mistrust and *'lack of willingness to engage with science, and more specifically medicine and genomics'* (Socialising the Genome: Communications, Public Trust and Engagement, Public Policy Projects, 2021, p-5).

There were also some suggestions that engagement activity needs to be, *'...truly investing in a shared story about what genomics can offer for all populations and specifically what it can do to reduce inequalities directly. Taking those conversations forward and building trust with communities that genomics can make a real difference in their lives is hard, but we must commit to doing it.'* (Equity of Access and return in Global Genomics, Public Policy Projects, 2022, p-22).

For example, one document stated that precision medicine should lead to individuals receiving better targeted interventions. Therefore, finding the best means of conveying concepts of genomics needs careful consideration, time and resources to ensure individuals can be active participants/contributors in the discussion. Making use of case

studies that resonate with different ethnic groups was suggested as a potential way to achieve this:

One approach could be to link the benefits to individuals and society to advances in healthcare treatments, to help patients understand that sharing their data could contribute to advances in treatments that could ultimately benefit them or their loved ones...Many of the concepts around genomics are intangible, and clear, real-life case studies demonstrating clinical utility and how patient data has contributed to advances in treatments could contribute to building support for data-sharing frameworks.

(Genomics Revolution, Public Policy Projects, 2021, p-20).

These sorts of approaches may go some way in helping to address why some patients may refuse initiatives, for instance, due to lack of understanding (My healthy future – person centred healthcare, 2019, p12). The importance of understanding the needs of groups and the role of certain factors such as ethnicity in decision making, was also implicated in other sources. This suggests that groups need to be engaged to help frame conversations to help towards achieving the most optimal outcomes:

If clinicians know that particular groups are less likely to make demands, they can adapt their conversation in order to address this. Decision-making should incorporate social, gender and other relevant factors such as ethnicity. We can do more to tackle inequalities if we understand what the barriers to particular groups are, and what interventions work. Stratification, not just by disease but by beliefs/values and social characteristics would also help to promote equity.

(My healthy future – person centred healthcare, PHGF, 2019, p13).

Public engagement was also discussed in relation to health technology advancements, suggesting that inclusivity was important to ensure historic barriers and inequities do not persist:

We need to support as wide a spectrum of people as possible to be involved in early adoption and ongoing development of health technologies, particularly those from ethnic minorities and disadvantaged socio-economic groups. Research planning should address this equity issue, and improvements to social infrastructure (such as internet access) should remove practical barriers to technology.

(Control of patient information in the COVID-19 era, PHGF, 2021).

There were several examples of how public engagement was being carried out to inform action plans, the design of future programmes and even resources and materials to be used. It was apparent that most of these documents tended to refer to engagement with 'diverse' and 'marginalised' groups more broadly, rather than stating ethnicity more explicitly. Moreover, where ethnicity was referred to, the level of detail regarding the groups involved and the proportions representing ethnic minority groups, and the precise nature of such activity varied across documents.

The UK Rare Diseases Framework (2021) and the England Rare Diseases Action Plan (2022), provide several comprehensive examples of community engagement activities, and how these interactions have informed action plans and so on. The former specifically sets out the importance of involving patients, families and their carers, and the organisations that represent them to inform the ‘commitments’ that are developed, where ethnic involvement is highlighted:

Therefore, any commitments will be developed in consultation with patient representatives, giving particular consideration to ensuring representation from those whose voices can often go unheard, including patients from Black and Minority Ethnic (BAME) or disadvantaged backgrounds.

(The UK Rare Diseases Framework, 2021, p17).

In addition, responses were received from 48 rare disease patient organisations, who represent the wider rare disease community, including organisations representing primarily BAME individuals living with a rare disease.

(The UK Rare Diseases Framework, 2021, Annex A).

While the more recent England Rare Diseases Action Plan (2022) did not explicitly refer to ethnicity, the document did outline the engagement work conducted with the support of Genetic Alliance UK, which referred to including individuals from diverse and marginalised groups affected by rare diseases as well as organisations representing the rare disease community. This engagement activity was carried out to help provide input on draft actions and an action plan. Details around the topics discussed were also given:

‘Participants shared their lived experiences on issues relating to each of the framework priorities, with discussion questions including “what are your experiences of receiving or waiting for a diagnosis?”, “how much do you feel healthcare professionals understand about you and your condition?”, “what is your experience of care coordination?” and “is access to specialist care, treatments and drugs equitable?”. Participants further discussed the larger question of how to create a fairer system which meets the needs of a diverse community.’

(England Rare Diseases Action Plan 2022, p39).

This document went on to provide the main themes to be generated from these initiatives, including how these findings would be utilised to ensure appropriate resources and promotional materials are developed, which was something other documents failed to set out:

'The Breaking Down Barriers workshop and stakeholder publications such as the "Whose Voice is it Anyway?" meeting report (summarising the findings of an NHSE/ engagement session hosted by RareQoL and Medics 4 Rare Diseases) highlighted a number of common themes, including the need for: a holistic approach to care and support, which considers the needs of the whole family, both at the point of diagnosis and over the longer term, accessible resources, taking into account people's lived experiences, and the challenges associated with communicating complex medical terms across cultural and language barriers, developing and maintaining trust in healthcare professionals. With this in mind, Health Education England is committed to ensuring that the resources and promotional materials it develops reflect the diversity of the UK population, both in terms of design and in the use of inclusive language.'

(England Rare Diseases Action Plan 2022, p48-49).

In a couple of their documents, Genome UK also appeared to show a commitment to ensuring engagement with ethnic minority groups, and went further to state that this public involvement was imperative to help address the broader issue surrounding underrepresentation of ethnic minority groups in genomic datasets:

'With this implementation plan, we set out our priority actions for the financial year 2021 to 2022 which include the following key commitments: a major drive, led by Genomics England, to improve the diversity of genomic data, addressing the historic under-representation of data from ethnic minority groups in genomic datasets, which results in health inequalities. The work will include widespread community engagement alongside sequencing and analytic tool development...'

(Genome UK: 2021 to 2022 implementation plan, 2021, webpage).

The Genome UK Future of Healthcare (2020) document also expressed commitment to developing more robust systems of outreach and communication to facilitate diversification of genomic datasets and to, *'ensure that we increase equity of access as much as possible...'* (p40). Engagement was considered key in helping to realise this, though details on how to do this were not given:

'Engagement and dialogue with the public, patients and our healthcare workforce, placing the patient and the diverse UK population at the heart of this journey.'

(Genome UK The Future of Healthcare, 2020, p7).

Finally, the Diverse Data Vision (Genome UK webpage), suggests that some form of engagement platform was required as a channel to discuss and inform underserved groups on the importance of genomic medicine.

3.3.6 Health service and system level recommendations

Across some documents, with varying degrees of detail, recommendations for service and system level improvements to address ethnic inequities were proposed. Some of these extracts were broad over-generalizations around the need to embed equality

and inclusivity within systems and services. Many of these examples related to the general health context, rather than genomic or precision medicine specifically, the latter is highlighted where applicable. Several sub-themes were identified: a call for better planning and evaluation of health systems and services, the need for a diverse workforce, supported with appropriate training, and suggestions that clinical guidelines needed to be customised based on ethnicity.

3.3.6.1 A call for better planning and evaluation of systems and services

A few documents proposed that each locality needed to produce a plan to improve the health outcomes of ethnic minority communities (along with other groups), whilst also considering broader national priorities. Other examples appeared to make a declaration that, *'The NHS will contribute to and review the evidence generated from key research initiatives to inform any future decisions regarding commissioning of services.'* (Accelerating genomic medicine in the NHS, NHSE, 2022, webpage)

However, more details on how and the conduit through which to do this, was not always apparent:

Each locality would be invited to develop a joint local plan to access the funds setting out how they would improve those with worst health and address the poor health of BAME groups and some deprived white communities, reflecting both national and local priorities.

(Levelling up Health, 2021, p14).

In addition to better planning, there was one suggestion that to improve access, experiences and thus outcomes among ethnic minority groups, evaluation and monitoring needed to be ingrained within systems.

Improve access, experiences and outcomes of NHS, local government and integrated care systems commissioned services by BAME communities including: regular equity audits; use of health impact assessments; integration of equality into quality systems...

(Beyond the Data: Understanding the impact of COVID 19 on BAME groups, 2020, p10).

Hence, monitoring equity of access was advocated within the field of genomic medicine. Appropriate measures and timelines were required to set out how this would be realised, though the challenging nature of this task was also acknowledged:

Equity of access is key to the entire NHS genomic medicine programme and is arguably the biggest challenge. Timescales need to be announced for when comprehensive monitoring will be in place. The data gathered, identifying key measures such as availability of tests, numbers and proportions of patients referred, turnaround times and quality metrics, need to be published. The Genomic Medicine Service should lay out plans for ensuring variations in access are addressed quickly and effectively with the support of industry.

(The Operationalisation of Precision Medicine, Public Policy Projects, 2020, p-19).

3.3.6.2 The need for a diverse workforce, supported by appropriate training

Several documents, including those on genomic medicine (e.g., Genomics England, PHG Foundation) underscored the importance of having a diverse workforce, with ‘... good representation of black and minority ethnic communities among staff at all levels...’ (Beyond the Data: Understanding the Impact of COVID-19 on BAME Groups, 2020, p10). A few documents argued that in doing so, historical barriers to service access and engagement by ethnic minority communities, such as mistrust and suspicion among communities could be addressed, though working with community partners and faith communities was also considered key to realising this goal.

In contrast, The NHS Long Term Plan (2019) recognises that the diverse workforce is not being utilised in the most optimal manner and that there is inequality in treatment of healthcare staff who represent diversity, which also needs to be addressed, and that this is not a quick fix:

‘The NHS draws on a remarkably rich diversity of people to provide care to our patients. But we fall short in valuing their contributions and ensuring fair treatment and respect. Through the Workforce Race Equality Standard, we are making progress in addressing these issues from the perspective of BAME staff. However, two years is not long enough to achieve the necessary change and so NHS England will invest an extra £1 million a year to extend its work to 2025. Each NHS organisation will set its own target for BAME representation across its leadership team and broader workforce by 2021/22...’

(NHS Long Term Plan, 2019, p87).

A couple of documents also highlighted the need to train the healthcare workforce to foster awareness and understanding of the differences that exist, according to characteristics such as ethnicity. For example, one document cited that ‘Health Education England’s competency frameworks include information on cultural awareness, and healthcare professionals can access a suite of online resources to help them support patients and their families through the Genomics Education Programme.’ (England Rare Diseases Action Plan 2022, p48-49). The urgency in addressing such inherent issues is emphasised by an extract from a document published by Genome UK. The said document suggests lack of understanding and bias among healthcare professionals is likely to be affecting clinical practice in relation to engagement with ethnic minority communities, for instance within genomic medicine:

In 2017, Genomics England commissioned a qualitative review of people from black African and black Caribbean backgrounds' views on participation in the 100,000 Genome Project. It identified some suspicion and distrust within many of these communities and also highlighted that some healthcare professionals may assume refusal from ethnic minorities and are less active in recruiting from these populations.

(Genome UK The Future of Healthcare, 2020, p39-40).

Following training of the healthcare workforce, there were also a few extracts that showed a desire and commitment to work with The National Institute for Health and Care Excellence in ensuring clinical care guidelines begin to incorporate and acknowledge the diversity among the population and how it relates to health conditions and disabilities; where the evidence-base is proven so that healthcare professionals are made aware:

Our 10-year ambition - curricula, further education and training, and NICE guidelines that reflect the diversity of society – for example, by reflecting sex or ethnicity-based differences in symptoms, or response to treatment for general health conditions or disabilities. Where there are clear gaps in the evidence base in these areas, we want to see work to fill these gaps and ensure the findings are effectively communicated to frontline healthcare professionals.

(Women's Health Strategy for England, 2022, p57).

Provide best practice for the inclusion of known health disparities, including those experienced by ethnic minorities, in clinical care guidelines. Work closely with the National Institute for Health and Care Excellence (NICE), and other bodies, to ensure all guidance includes information on disparities as standard.

(The Report of the Commission on Race and Ethnic Disparities, 2021, p20).

The PHGF, makes recommendations that clinical guidelines for pharmacogenetic testing should be customised for patients from different ethnic groups. For instance, in a PHGF blog on international lessons for personalised medicine it was acknowledged that internationally, *'Health professionals and policymakers are already well attuned to the variable healthcare needs of different groups within populations, especially ethnic groups, and this often informs local provision. At a national level, understanding the genetic variation present in a population is equally important for optimising relevant clinical guidance.'* (International lessons for personalised medicine, PHGF, 2021, Blog).

3.4 SUMMARY

The document review highlights that diverse datasets are needed, and that current underrepresentation of ethnic groups is contributing to health inequalities and ethnic inequities in precision and genomic medicine. Data limitations have far-reaching consequences for ethnic groups, regarding understanding on genetic variation and the development of optimal precision medicine services, be it in terms of improving early detection and diagnostic tools and more timely interventions, that are applicable to diverse ethnic groups. A lot of the work currently being done on advanced analytic techniques such as AI-based risk prediction tools and development of PRS is also limited due to ethnic representation being poor in datasets, and the predictive utility of such tests among diverse ethnic groups, is therefore limited. However, there are initiatives underway to address this matter, though the precise nature of this activity, is unclear.

The PHGF, the leading UK organisation evaluating genetic healthcare policy, and the NHSE funded research organisation, Genomic England, has recognised the importance of considering underserved ethnic minority groups in their policy reviews and research implementation.

Efforts to engage diverse communities and individuals for public engagement work has been undertaken to inform genomic initiatives/action plans. However, such efforts are piecemeal and details on how the work is happening is rarely acknowledged, though co-design and co-production are commented on, albeit briefly. There were valuable points made about how engaging with ethnic minority groups requires appropriate time and resources, and that employing examples that would resonate most with them, was needed to encourage participation. Concerted efforts are required to really listen to and understand the needs of different ethnic minority groups, to educate them on the concepts associated with genomics and precision medicine, and to enable them to make informed contributions to the field.

Health service and system level recommendations are also proposed. For instance, to improve access, experiences and thus outcomes among ethnic minority groups, evaluation and monitoring needs to be ingrained within systems. However, this brings attention to the unavailability of data due to poor recording, or underrepresentation of ethnic groups in the first place. There were general suggestions around the healthcare workforce needing to be more diverse and that they are treated more equally too. A more diverse healthcare workforce is argued to be key in addressing issues of historical barriers to access, and mistrust/suspicion among communities.

Training health service staff to foster cultural awareness and understanding, and embedding diversity within clinical care guidelines would be positive steps towards improving ethnic inequities in precision and genomic medicine. However, the nature and content of such training would require unpicking, as this was another idea proposed without any suggestions on the modality.

In conclusion, better collaboration was advocated. For instance, pooling together of different datasets and improving data diversity is key (e.g. among the research and academic community). Advancement in this area also depends on the engagement of the wider research community and relevant stakeholders (e.g. sectors involved in service design, delivery and recruitment, healthcare, analysts), government and public stakeholders (public, patients, community and faith groups) representing diverse ethnic groups.

4. HEALTH INEQUALITIES IN PRECISION MEDICINE. MIXED METHODS SYSTEMATIC REVIEW

4.1 AIM

To explore ethnic inequalities in recruitment for biomedical research and patient access to genomic medicine services.

4.2 METHODS

A systematic review was undertaken. The review protocol was published on the PROSPERO database (CRD42022371245)

4.2.1 Inclusion criteria

Condition/Phenomenon of interest: The review included studies about precision medicine (defined as genomics or genetic services which focus on the development and application of genomic advances and big data analysis in diagnosis, treatment of illness, and predictive or preventative care) that refer to at least one ethnic minority group.

Participants/population: Healthcare service users.

Exposure: The exposure of interest is at least one ethnic minority group (either presented as aggregated or disaggregated groups). Ethnicity includes reference to ethnic or race groups and can be self-reported or reported by another party, based on recognised categorisation (e.g., census, healthcare administrative system), place of birth, nationality, or migration status.

Comparator: If available, was a non-ethnic minority group.

Context: The studies were in any healthcare setting, including primary, secondary and tertiary care in the UK or other high-income countries with similar healthcare systems (e.g., Australia, Canada, the Netherlands).

Outcomes: The included outcomes were ‘representation in genetic databases’ and ‘polygenic risk scores’ (how likely you are to get a specific disease due to combination of your genes) or similar measures of risk.

Study design: The review included studies that employed quantitative, qualitative or mixed methodologies. Included studies were randomised control trials (RCTs) or observational studies (comparative or non-comparative).

4.2.2 Search strategy and study selection

A comprehensive search was undertaken for both academic and grey literature. Peer reviewed/published literature were identified by searching MEDLINE (OVID), EMBASE (OVID), PsycINFO (OVID), and CINAHL (EBSCOhost). Unpublished literature were identified from searching ProQuest Central (ProQuest), ASSIA (ProQuest) and Scopus, to ensure all existing relevant studies were captured. Studies were included from database inception to October 2022.

A combination of medical subject headings (MeSH) and key text words were used. An example search strategy is given in Appendix 2- Search Strategy- see attached document.

4.2.3 Study selection and data extraction

Two authors independently screened titles and abstracts (ZH and IB or MB or LJ), and then the full texts, of potentially eligible studies identified from the searches (ZH and MB). Disagreements in eligibility were discussed and resolved through consensus or with the help of a third author (NQ). Two authors (ZH, SH) extracted data from studies that met the inclusion criteria, using a pre-piloted data extraction form tailored for quantitative and qualitative study designs. Any disagreements were discussed within the whole research team to reach consensus. The reference lists of previous systematic reviews were used to identify further eligible studies.

The data extraction form included information on study methodology; study aim; whether a database was used and the nature of it; whether and how ethnic minorities were identified in the database/sample; how authors incorporated ethnic minorities in the analysis, and whether this information was given in the main text or supplementary section of the paper; number of total participants; number of included ethnic minorities; whether ethnic minorities were included in demographics sections; whether any data about deprivation in ethnic minorities were included; whether ethnic minorities were included and reported in data analysis; whether data on ethnic minorities were interpreted in the discussion section were interpreted in the discussion section; and whether lack of collation and/or analysis of ethnicity was discussed as a limitation.

Relevant [PROGRESS-Plus] characteristics, such as measure of deprivation, were extracted together with data related to the study characteristics, population, exposure,

outcome measures, findings and limitations of the studies ⁽⁸²⁻⁸⁴⁾. Where studies did not have ethnicity in the results, the methods and discussion sections were examined to see if they had recognised the lack of information about ethnic minorities as a limitation in their work. Characteristics such as measures of deprivation, were extracted together with data related to the study characteristics, population, exposure, outcome measures, findings and limitations of the studies ⁽⁸²⁾.

4.2.4 Data synthesis

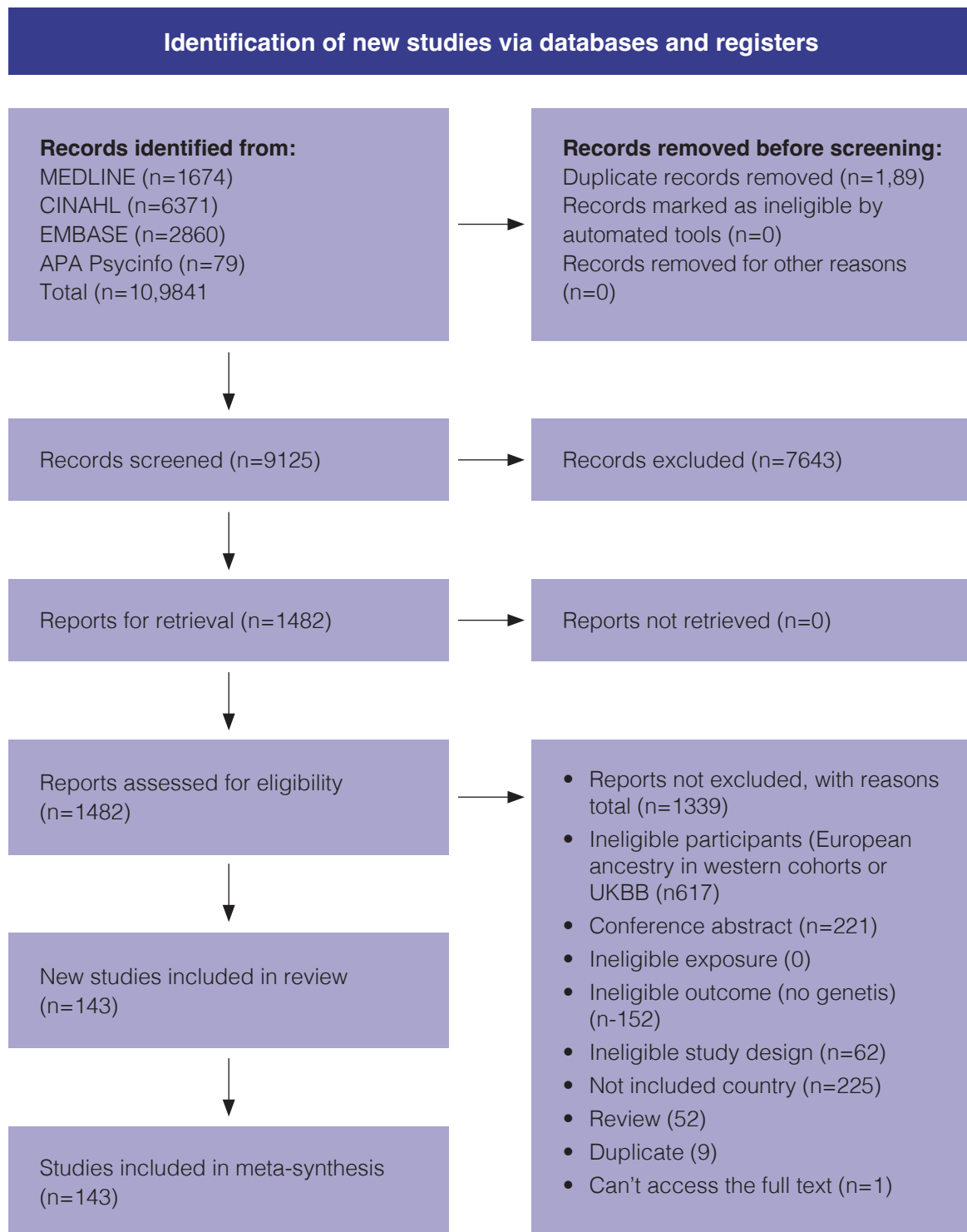
A narrative synthesis of the included quantitative and qualitative studies was performed. Due to heterogeneity of the included studies' methodologies and participants, no further sub-group analysis was possible.

4.3 RESULTS

4.3.1 Study inclusion

A total of 10,984 titles were identified from the databases, unpublished sources, and grey literature searches. After removing duplicates, 9125 titles and abstracts were screened. Of these, 1482 full-text papers were identified as potentially eligible for inclusion and assessed for eligibility. 1339 papers were excluded, due to either ineligible population (n=617), conference abstract (n=221), ineligible outcome (n=152), ineligible study design (n=62), reviews (52), duplicate (n=9) or no access to the full text (n=1). See Figure 1 for an overview of the study selection and inclusion process. 143 studies met the inclusion criteria and were included in the current review. Among those, 137 studies were quantitative, and 6 studies were qualitative.

Figure 1. PRISIMA flow chart, search results and study selection and inclusion process.



4.3.2 Narrative synthesis of quantitative studies

Characteristics of included studies

A total of 137 quantitative studies were included, which were published in 2021 or 2022. Two of the studies were interventional studies ^(85, 86), 5 were meta-analyses ⁽⁸⁷⁻⁹¹⁾, 19 were case-control studies ⁽⁹²⁻¹¹⁰⁾, 48 were cohort studies ⁽¹¹¹⁻¹⁵⁸⁾, 2 were cross-sectional studies ^(151, 159) and 61 were observational studies analysing genetic databases ^(103, 160-217).

The included studies addressed diseases and health conditions in various medical specialties, with a considerable number falling under cardiology, oncology, pulmonology (particularly COVID-19), endocrinology, neurology, psychological medicine and rheumatology.

The total number of participants from all the included studies was over 64 million, ranging from 17 to 3,902,748 participants (mean 1,461,803). Out of these 64 million participants from all included studies, about 6.1 million (<10% of total participants) belonged to ethnic minority groups, with this value ranging from 5 to 622,604 participants across the various studies (mean 48,421). However, it is worth noting that given that many participants originated from the UK Biobank, some participants would have been counted more than once. Regarding the 137 included quantitative studies, we have 18 studies comparing UK cohorts with international cohorts from countries that have similar healthcare systems such as the Netherlands, Germany, Canada and Australia; 96 studies comparing genetic profiles of ethnic minority versus white populations; 13 studies reporting genetic profile of ethnic minority populations only; 6 studies focusing on means of accounting for ethnic minorities in genetic research; and 4 studies that addressed precision medicine approaches to stratified care.

The studies sourced data from various databases. Of the 137 included studies, 99 sourced data from the UK Biobank, either alone or in combination with other databases such as 100,000 Genomes Project, Biobank Japan, Trans-Omics for Precision Medicine (TOPMed) Program, Million Veteran Program (MVP) and cohorts from other countries such as Bangladesh, India, Uganda, Korea, Finland, China, Turkey, and Iran.

Some of the studies relied upon patients' hospital records for research data and others used primary data from participants recruited specifically for the study. Some studies integrated data from UK-based cohorts ^(175, 176) such as Southall and Brent Revisited (SABRE) cohort, Born in Bradford (BiB) cohort, and the database of University College Hospital, London to explore various health aspects, including cancer risk and rare genetic variants. The means of identification of participants' ethnicity was by self-reported ancestry, either used alone or alongside principal component analysis of genetic data. Appendix 3: Quantitative studies characteristics provides the full details of the 137 quantitative studies included in this review.

Seven of the included studies did not specify the entire number of study participants nor those from ethnic minority groups, while nine studies did not specify the number

of participants belonging to ethnic minority groups though they reported figures for their total sample size. For these studies, main text and supplementary materials were searched by two independent reviewers but the details on number of participants were not found.

Descriptive overview of the methodology of the included studies

All the databases/data sources reported inclusion of ethnic minority populations, but many studies did not include them in the main analysis. One-hundred and thirty-nine studies identified ethnic minorities via self-reported ancestry ^(85-89, 91-126, 128-131, 133-138, 140-165, 167-170, 172-270), one via the discovery phase (participants from the Qingdao twins registry) ⁽¹³⁹⁾, one via electronic/hard copy patient records ⁽¹⁶⁶⁾ and three studies did not report how the authors identified ethnic minorities in the database/sample ⁽¹³⁶⁾.

Several studies incorporated ethnic minorities in the analysis. Types of analysis employed across the various studies include regression analysis, genome-wide association analysis, meta-analyses, chi-squared test for association, cross-ancestry validation, etc. (see Appendix 3 for further details) ^(85-97, 99-101, 104, 105, 107, 108, 110, 113-117, 120, 122-124, 126, 134, 135, 137-141, 143-151, 154-156, 159-162, 164, 165, 168, 169, 173, 176-178, 180, 181, 186, 188, 189, 191, 192, 194, 196, 198, 200-202, 204-207, 212, 213, 215, 217, 221, 249, 254, 264, 268, 269, 271).

4.3.3 Descriptive overview of the findings of the included quantitative studies

Sample sizes and proportion of ethnic minority participants in studies

Among the 137 included studies, 94.9% (n=123) reported total participant numbers, while 88.3% (n=121) reported specific ethnic minority sample sizes. 83.2% (n=121) of included studies reported the proportion of ethnic minorities. As most included studies derived details from the UK Biobank genetic database, similar details on proportion of ethnic minorities were reported. In contrast, 5.1% (n=7) of studies only highlighted ethnic minority involvement by presenting 'non-Caucasian' or 'non-European' participant data without stipulating specific ethnic backgrounds. 11% (n=15) of included studies reported sample sizes including only ethnic minorities. Globally, ethnic minority participants accounted for 6.56% of the total participant population across all included studies. Across all studies, the average number of all participants in individual studies was 218,083. The average number of non-ethnic minority participants was 203,785. In contrast, the average number of ethnic minority participants in individual studies was 14,298.

Demographic characteristics of ethnic minority participants

57.7% of studies (n=79) reported demographic data relating to ethnic minority participants. Common demographic parameters described by studies that reported on ethnicity included age (20.4%, n=28), sex (16.8%, n=23) and BMI (9.5%, n=13). Furthermore, 16.8% (n=23) of studies reported study specific demographic data relating to ethnic minority participants - for instance, parity, level of education, physical activity and diet. 6% (n=8) of studies reported ethnic minority participants as belonging to an 'Other' ethnicity category without further ascertainment of participants' specific ethnic background. 4.4% (n=6) of included studies reported the income status of ethnic minority participants, four of which utilised the Townsend deprivation index, and the remaining two studies adopted their own specific decile system to stratify income levels among ethnic minorities and non-ethnic minority participants.

Analysis of ethnic minority participants in included studies

96.4% (n=132) of included studies reported analysis of ethnic minority participants. 77.4% (n=106) included ethnic minority participant information in the main results section (usually in demographics table), 11.7% (n=16) stipulated analysis by ethnicity within supplementary materials and 7.3% (n=10) reported analysis using ethnicity within the main analysis and supplementary materials. 87.6% (n=120) of studies included ethnicity within the text of respective results sections. Out of these 120 studies, 100 (73.0%) performed statistical analysis of genomic data involving ethnic minority participants and 20 (14.6%) studies only stipulated ethnicity of study participants within demographic data without any further statistical analysis.

Discussion of ethnic minority participants in included studies

70.1% (n=96) of included studies considered ethnic minority participants in discussion sections, however, the nature of this varied. 43.8% (n=60) reported statements suggesting lack of ethnic minority involvement as a limitation in the field of genomics. These findings are also reflected in our review, as ethnic minority participants comprised 6.56% of the total participant population across all included studies. The importance of ethnic minority involvement in genomic studies was further emphasised in the conclusion of most studies as 31.4% (n=43).

4.3.4 Narrative synthesis of the qualitative studies

Characteristics of included studies

Out of 143 included studies in the current review, 6 were qualitative ^(227, 234, 240, 248, 272, 273), which were published between 2021 and 2022, and 4 were from the UK ^(240, 248, 272, 273), and 2 were from Australia ^(227, 234). For the Australian studies, participants were from the Australian Aboriginal ethnic minority group ^(227, 234). For the UK studies, not all the

participants were from ethnic minority groups. For instance, in the study by Dennison et al., 29 participants took part, of which 31% were from ethnic minority groups: 14% from Asian/Asian-British ancestry; 10% from Black/African/Caribbean/Black-British ancestry; and 7% were of mixed/multiple ancestries or categorised as others⁽²⁷³⁾. In Gaba et al's study, two of the nine participants were of Southeast Asian ancestry and one was of Jewish ancestry⁽²⁷²⁾. Similarly, in Kinsella et al's study, 4 of the 19 participants were from ethnic minority groups⁽²⁴⁰⁾.

In five of the six studies, ethnicity was reported based on self-reported ancestry^(227, 228, 230, 234, 240). Three studies included ethnicity information in the demographics section^(228, 230, 240). When considering methodology, two referred to using grounded theory^(234, 272), whereas design was unspecified in the remainder^(227, 240, 248, 273). The data collection methods used were primarily semi-structured or in-depth interviews and most used thematic analysis.

4.3.5 Descriptive overview of the findings of the included qualitative studies

Barriers to access and engagement

Dalach et al. examined the experiences of Aboriginal and Torres Strait Islander people who had attended mainstream clinical genetics services in the past either as patients or caregivers⁽²²⁷⁾. These participants expressed that while genetic services were cost-free, affordability remained a significant barrier because of other costs associated with attending the appointment⁽²²⁷⁾. Moreover, the study found evidence that many participants were unaware of the free transport options available to Aboriginal and Torres Strait Islander patients to enable them to attend medical appointments, such as the Patient Assistance Transport Scheme or their Aboriginal Community Controlled Health Services⁽²²⁷⁾. Patients from regional and remote areas who accessed the Patient Assistance Transport Scheme frequently relied on the referring practitioner's knowledge of the system and the ability to advocate on the patient's behalf.

There was also evidence to suggest the lack of Aboriginal support services during clinical genetics appointments, even though these services were available within the hospitals where the clinics were held. Findings from the study revealed that none of the participants were provided with support from an Aboriginal Liaison Officer during their clinical genetics session⁽²²⁷⁾. One participant stated: *"... If I had the option of having an Aboriginal liaison officer with me, I would have said yes, every time. I've got eight kids, so you can imagine how many times I'm in the hospital and having that person there that understands their culture and how some things are different in their culture to Europeans, that would just be amazing."*^(227,p.6)

Dalach et al. concluded that the lack of Aboriginal support services might mean important socio-cultural issues were not being recognised or managed⁽²²⁷⁾. For instance, gender may impact patient-provider relationships among Aboriginal and Torres Strait Islander women, depending on cultural norms, personal preference, and

lived experience. Hermes et al. conducted a study in 2021 that qualitatively explored the perspectives of Australian Aboriginal people whose tissue or those of their ancestors were stored in the biobank of the National Centre for Indigenous Genomics (NCIG) ⁽²³⁴⁾. Some participants expressed optimism that biobanking research could result in medical advancements and potential remedies for medical issues affecting their ancestry and communities. However, some Australian Aboriginal people had concerns about keeping their samples in the NCIG collection or wished to withdraw their samples with the intention of disposing them.

Some people were hesitant to accept that samples remain in the biobank, because samples had been taken without their consent and because of historical experiences of abuse and exploitation of Aboriginal and Torres Strait Islander people. Other people were reluctant to participate in the biobank because of negative past experiences with researchers and an understandable lack of trust that their community would benefit from such research ⁽²³⁴⁾. The main motivation for one community to withdraw their samples from the biobank collection was the cultural taboo around blood and concern for the spiritual afterlife, as several members of the community requested that the samples be returned to the community for burial or disposal. *“As Aboriginal person... that blood sample, sacred sample...once its brought back, then we might get rid of it. In proper way. Not just chuck it in the, anywhere in the ground.”* ^(234,p.1430)

Including ethnicity within risk prediction models

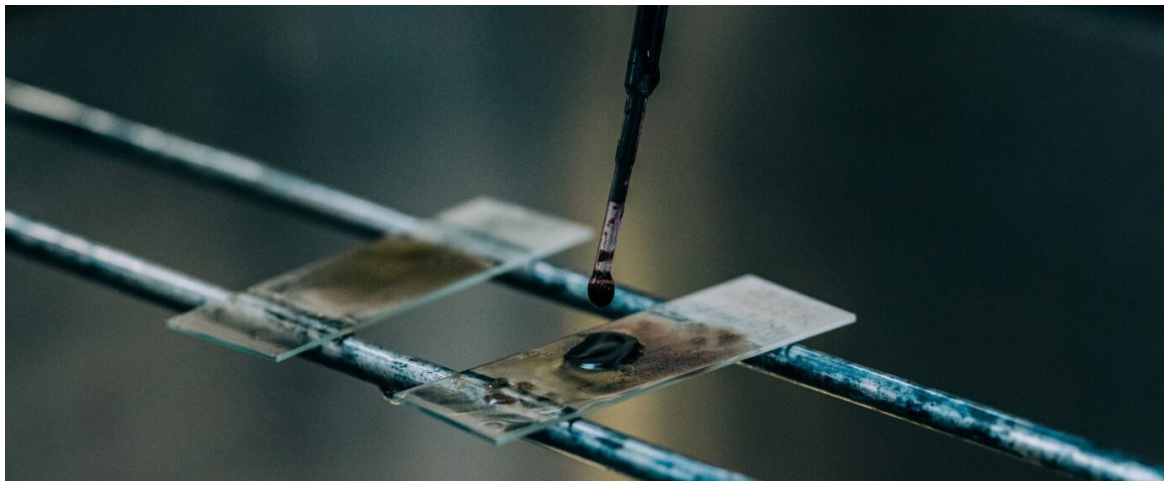
Gaba et al. explored the experiences and impact of undergoing genetic screening for personalised ovarian cancer risk on the emotional well-being of women from the general population ⁽²⁷²⁾. The main findings related to ethnicity reported that a Jewish participant was aware that being Jewish was a strong motivator for undergoing risk stratification and testing as individuals of this ethnic minority group were more likely to be carriers of certain genetic diseases ⁽²⁷²⁾. A similar study by Dennison et al. explored the acceptability of using risk stratification to determine eligibility for cancer screening, using three community online juries. Juries one and two supported including ethnicity within risk prediction models, provided it was clearly justified and communicated. These participants considered ethnicity to predict cancer risk and to be closely linked to genetics and family history ⁽²⁷³⁾.

Kinsella et al. explored the views of members of the public without direct experience of cystic fibrosis to determine if there was a preference for maximising the sensitivity or the specificity of cystic fibrosis screening ⁽²⁴⁰⁾. The authors reported that to ensure that new screening approaches will better reflect the ethnic mix in the UK population, more participants from ethnic minority groups and those of younger adults were needed. Polchar et al. report a case of a young girl of South Asian descent who presented at the hospital at 23 months with gastroenteritis, dehydration, and faltering growth due to feeding difficulties ⁽²⁴⁸⁾. At age six, after extensive testing and ruling out various differential diagnoses, the patient and her parents underwent whole-exome sequencing as part of the ‘100000 Genomes Project’, which identified homozygous variants in Glycerol-3-phosphate dehydrogenase 1 (GPD1) deficiency.

4.4 SUMMARY

Research studies often underrepresent certain racial and ethnic groups, leading to a limited understanding of how different populations respond to specific treatments or interventions. Therefore, efforts should be made to increase diversity in research studies by actively recruiting participants from underrepresented communities. This would help generate more robust data on how different population groups respond to various treatment modalities ^(200, 234). The review found some evidence of ethnic inequalities in attitudes towards accessing, and access to, genetic services, this included the direct and indirect cost of accessing genetic services. There was evidence in the review that ethnic minority groups should benefit from liaison officers when needed. Additionally, patients are unaware of the full range of the cultural, health and social support services available to them ⁽²²⁷⁾. Unawareness of these services was linked to the attitudes, knowledge and behaviours of healthcare providers who rarely made efforts to refer patients to services available for ethnic minorities, thus making it difficult for them to navigate unfamiliar services. The review suggested the central importance of making service providers responsible for proactive and immediate changes at the individual and service level to ensure that inequitably distributed benefits of genomic healthcare do not exacerbate the existing gap in health outcomes ^(227, 234, 240, 272, 273).

The review also found evidence that ethnic minority groups were generally uncomfortable allowing their samples to be stored in a Biobank, this may be borne out of mistrust, disrespect, discrimination and cultural insensitivity. It also provided insight into the lived experience of unethical research practices and of researchers' frequent failure to return results to communities, these factors appear to undermine trust and feed fear, which in turn are described as resulting in poorer access to, and engagement of these services. This suggests the importance of building a trusting relationship between researchers and ethnic minority communities and the need for researchers to respect people's culture, traditions and values ⁽²⁷⁴⁾. However, like many areas within the healthcare system, genomic medicine also faces challenges in addressing ethnic health inequalities. It is essential that the benefits of advances in technology do not widen the existing gaps among communities already experiencing ethnic health inequalities because such groups are already less likely to engage with healthcare services.



5. EXPLORING KNOWLEDGE AND AWARENESS OF GENOMICS, THE BARRIERS TO AND FACILITATORS OF ENGAGEMENT, ACCESS AND UPTAKE IN GENOMICS SERVICES AND RESEARCH.

5.1 BACKGROUND

Access to emerging genomics services (e.g. counselling and testing) is not equal for ethnic minority groups. Additionally, precision medicine services that incorporate advanced data processing methods such as machine learning to develop diagnostic tools such as PRS may have limited utility in ethnic minority groups as the datasets used may not be representative of the population. To develop effective action plans and implement solutions to address inequities in service access, uptake of testing and participation in research relating to genomics and precision medicine, it is important to understand the views of key stakeholders regarding the current challenges for communities, healthcare professionals and the health service. This chapter presents findings from interviews and focus groups with public stakeholders from ethnic minority groups and key professional stakeholders working within the fields of genomics and precision medicine.

5.1.1 Aims and objectives

Aim: This qualitative study delves into the intricate web of genomics, aiming to elucidate the multifaceted perspectives of public stakeholders from ethnic minority groups and key professional stakeholders working within the fields of genomics and precision medicine

Objectives:

1. Explore knowledge, understanding and attitudes towards genomics and precision medicine services and biomedical research among different ethnic minority groups.
2. Explore key barriers to and facilitators of access and uptake of genomics and precision medicine services, as well as participation in genomics research for ethnic minority groups.
3. Explore knowledge, understanding and current practices among stakeholders (public stakeholders from ethnic minority groups, healthcare professionals, researchers, policymakers and service providers) to promote access and equity in biomedical research, genomics and precision medicine services.

5.2 METHODS

We conducted qualitative semi-structured interviews with clinicians, individuals involved in the delivery of precision medicine research and policymakers to assess the knowledge, attitudes, understanding and practices in promoting access to biomedical research and precision medicine services, by ethnic minority groups. With these stakeholders, we also explored current practices and challenges around ethnicity data recording and analysis.

We also interviewed individuals representing different ethnic minority groups to map knowledge, understanding and attitudes regarding awareness of and engagement with biomedical research and precision medicine services. Finally, we explored barriers to and potential facilitators relating to accessibility and uptake of genomics services and involvement in precision medicine research.

5.2.1 Study recruitment and sampling

We recruited and interviewed study participants representing:

- i) Clinicians, representatives from GMSAs, policymakers and individuals (e.g., academics) overseeing current or recent genomic and machine-learning research. Recruitment was multi-faceted, clinicians, policymakers and researchers were recruited through co-investigators' networks, NHS GMSAs, Genomics England, social media advertising via Twitter and LinkedIn, and relevant professional organisation webpages or newsletters. A snowballing approach was also used, whereby we asked current participants who identified relevant individuals to share the study within their networks. Irrespective of stakeholder type, contact was initiated via e-mail or advert, where the purpose of the study was briefly outlined. A detailed information sheet and consent form was also attached for perusal. Individuals were then able to contact the research team to express interest in participating.
- ii) For individuals representing different ethnic minority groups, we identified and approached local grassroots community organisations and community leaders across the Midlands and Northwest to recruit individuals representing ethnic minority groups. We used different methods of contact including a gatekeeper who signposted individuals to the research team, individuals themselves who gave permission to be contacted by the research team directly, individuals who contacted the research team directly in response to adverts in emails, newsletters, community settings, social media (Twitter and LinkedIn) or word of mouth.

5.2.2 Sampling strategy and justification

Sampling in qualitative research is complex and challenging ⁽²⁷⁵⁾. Determining sample size *a priori* for qualitative research does not lend itself to a prescribed or formula-based approach, and only provisional numbers can be specified at the outset of a study ⁽²⁷⁵⁻²⁷⁷⁾. Therefore, for all stakeholders, our sampling approach combined both convenience (self-selecting individuals) and purposeful strategies to ensure the inclusion of a wide representation of the five groups outlined. We purposefully invited individuals representing specific criteria, to ensure we captured a range of perceptions and experiences.

For clinicians, GMSA representatives, individuals involved in research and policymakers, we sought to purposefully sample a range of individuals representing:

- Different job roles e.g., GPs, community nurses, genetic nurse specialists, genetic and other medical specialists.
- Those with involvement in biomedical and machine-learning research and/or those involved in development/running of genomic medicine services.

For individuals representing ethnic minority groups:

- Representation of different ethnic minority groups, age, gender and socioeconomic status.

5.2.3 Data collection

We designed separate semi-structured interview guides for each stakeholder group. The semi-structured approach was adopted because it helped to ensure the interviews were participant-led, consequently giving the interviewer ample opportunity to further explore the contextual meaning of participants' responses ⁽²⁷⁸⁾.

As regard to the interview settings, focus groups and one-to-one interviews were used, with the option for them to be held either physically or virtually. A one-to-one approach was deemed most appropriate for our clinician, research and policymaker stakeholder groups, in terms of practicalities e.g., getting a group of clinicians together for a focus group was likely to be a challenge. Informal focus groups were used for our GMSA representatives stakeholder group. For our ethnic minority stakeholder group, we adopted a more flexible approach and offered either one-to-one interviews or focus groups, depending on what was considered most appropriate by our target population and following advice and engagement with community organisations. The focus groups and interviews with the ethnic minority stakeholder group used a task group format whereby participants were presented with information about genomics and precision medicine. The purpose of using a task group approach was to stimulate conversation and provide participants with knowledge of the topic area.

For clinicians, researchers, policymakers and GMSA representatives, examples of key areas for discussion included: (1) knowledge and understanding regarding ethnic equity in biomedical research and precision medicine; (2) awareness, views and experiences of promoting diverse access to biomedical research and precision medicine service, including unpicking of examples e.g. perception of best practice; (3) perceived barriers, facilitators and opportunities to promote recommendations for improving ethnic equity in biomedical research and precision medicine e.g. education needs, service development and implementation and (4) current practices around ethnicity data recording and analysis and current challenges.

For individuals representing ethnic minority groups, examples of topics covered included: (1) knowledge, understanding and views on biomedical research and precision medicine; (2) perceived importance of ethnic minority representation and involvement; (3) barriers, facilitators and opportunities to improve ethnic inequalities in biomedical research and precision medicine; (4) recommendations for information/ education and service needs to better engage ethnic groups.

5.2.4 Data analysis

Audio recordings were transcribed verbatim by an external specialist transcription service, approved by the University of Nottingham. Following receipt of transcripts, data were checked for accuracy and personal identifiers removed. Data was analysed using the framework approach, which is a hierarchical, matrix-based method developed for applied research ^(279, 280). NVivo 12.0 was used to facilitate data management and analysis.

The framework approach enabled mapping of thematic differences/similarities within and between groups, such as by stakeholder type or ethnicity. Data was coded both deductively - according to a priori themes (based on aims and discussion topics) - and inductively. Initial readings of the transcripts facilitated familiarisation and led to the generation of initial codes. Further reading and immersion resulted in more substantive themes and sub-themes, resulting in the generation of an analytical framework. Data were then indexed according to the identified thematic framework. A sub-sample of data was also double coded to ensure validity of interpretations ⁽²⁸¹⁾.

We also incorporated a feedback loop and respondent validation whereby we shared initial themes and findings with a subset of public stakeholders and invited them to check the initial accuracy of transcripts, add to them after a period of reflection, and review whether mapped themes were a reasonable reflection of their data. Finally, themes were discussed and agreed between the research team, allowing clarification of the final framework that was then applied across all the transcripts. Data was then charted according to each theme to facilitate interpretation, synthesis, and reporting. Demographic data from public stakeholder participants were summarised using descriptive statistics.

5.3 RESULTS

5.3.1 Participant characteristics

Public stakeholders

In total, 98 participants from different ethnic minority groups participated in either face to face or virtual focus groups (n=10) or one-to-one interviews (n=1), between March and July 2023. On average, each session of focus group and interview lasted for 70 minutes, (ranged from 50 to 97 minutes). Ninety-three participants completed a demographics survey (Table 1: Public stakeholder demographics.). Participants ranged in age from 18 to 85 with a mean age of 47. Public stakeholder participants were from Black, Asian and other minority groups. Thirty-eight participants reported a degree level qualification or higher (Appendix 4 Public stakeholder graphs) and employment status varied across the group (Appendix 4 Public stakeholder graphs). Participants reported speaking a range of different languages including English, Arabic, Urdu, Portuguese, Yoruba and Swahili to name a few (see Figure 3).

Table 1 Public stakeholder demographics.

FG N	N participants	M	F	Ethnicity N
FG1	7	-	7	Indian = 7
FG2	12	-	12	Arab = 12
FG3	12	-	12	Arab = 12
Interview	1	-	1	Black Caribbean
FG4	4	4	-	Indian = 3; Pakistani = 1
FG5	7	7	-	Arab = 7
FG6	8	7	1	Pakistani = 2; Indian = 1; White = 1; Asian - Other = 3; Mixed - white and Asian = 1
FG7	10	-	10	Pakistani = 7; Bangladeshi = 1; Indian = 1
FG8	21			<i>Black African = 7; Black Caribbean = 2; Black or Black British - Other = 1; Mixed White and Black African = 5; White = 2; White Other = 2; Pakistani = 1; Any other group = 1</i>
FG9	8	2	6	<i>Black African = 7; Black Caribbean = 1</i>
FG10	8	1	5	<i>Black African = 6</i>

Figure 2 Public stakeholders spoken languages.



Professional stakeholders

Twenty professional stakeholders were recruited and interviewed. Interviews averaged 56 minutes (ranged from 29 to 76 minutes). Most participants were female (16 [80%]). Two participants were clinical genetics consultants, 6 were nurses working within genomics medicine services and the genomics medicine service alliances, 1 participant was from industry, 3 participants were community engagement representatives, 5 participants were academics working in genomics or precision medicine research and 3 participants were policymakers. Additionally, we also conducted 3 informal focus groups and 2 informal interviews with representatives from 5 of the 7 genomics medicine service alliances in England. Representatives included consultant geneticists, data analysts, patient and public involvement and engagement leads.

Table 2 *Thematic Framework.*

Theme	Sub theme 1	Sub theme 2
Current levels of knowledge and awareness	Genomics	
	Personalised medicine	
	Health inequalities	
Strategies for improving knowledge and awareness	Role of healthcare professionals	
	Role of faith and community leaders	
	Presence in community spaces and events	
	Role of lived experience	
	Multigenerational approach	
	Multimedia approach	
	Sustained messaging	
	Tailored approaches for communities	
Shared barriers to and facilitators to access and engagement with genomic medicine services and research	Challenges accessing healthcare services	
	Limited knowledge and awareness	
	Literacy skills	
	Socioeconomic factors	
	Language	Accessing interpreters
		Translation accuracy
		Language impacts engagement
	Mistrust	Fear
		Acknowledging the past
		Government policies and politics
		Building trust

	Representation	Diversity in materials
		Workforce diversity
		Representation of the messenger
Specific barriers and facilitators to genomics services	Limited understanding of cultural and religious differences	
	Healthcare professionals knowledge and confidence	
	Healthcare professionals' biases (e.g. stereotyping)	
	Gatekeeping by healthcare professionals	
	Negative feelings (stigma, shame, guilt, fear)	
	The implications of testing outcomes	
	Waiting lists and a busy NHS	
Specific barriers and facilitators to genomics research	Lack of diverse data	
	Current recruitment methods	
	Concerns around how data will be used and managed	
	Increase awareness of research (talk to people)	
	Close the loop	
	Inclusive research processes (go to the communities)	

	The value of being involved and the consequences of not participating	
Community engagement	Sustained engagement	
	Strategies to improve engagement	
Monitoring equity of access in genomics medicine services - the barriers and facilitators	Current data recording practices	
	Current test ordering process	
	Data sharing, access and governance	

	GMSA EDI strategy development	
	Collaboration	
	Projects and initiatives aimed at addressing inequities	
Workforce training	Genomics medicine training for healthcare and other professionals	
	Cultural and religion awareness	
	Community engagement training for researchers	
	Importance of ethnicity data recording and having the conversation with patients	

5.3.2 Thematic framework

There were 8 themes and corresponding sub themes arising from stakeholder focus groups and interviews (see table 2). The themes are presented in text below and supporting quotes have been tabulated (see Appendix 5 Stakeholder themes with supporting quotes). Participant ID codes were used to show the different stakeholder who participated in interviews and focus groups to share their views (see table 3).

Table 3 Key detailing participant ID codes for stakeholder type

Code	Stakeholder type
C#	Healthcare professional
R#	Researcher
PE#	Public engagement representative
I#	Industry representative
FG#	Public stakeholders
GMSA FG#	GMSA representatives

5.3.3 Current levels of knowledge and awareness

Genomics

Genomics literacy varied across the public stakeholders. Some were aware of the terms 'genes' and 'DNA' in reference to paternity, genealogy, and inheriting conditions such as diabetes, breast cancer and autoimmune conditions, while others had not heard the terms and did not know what they meant (See Appendix 5.1 Knowledge and awareness themes).

"For me, genes and genome are something that is passed down from your parents, and it is the genetic makeup of who you are physiologically, and it's basically your biological makeup or like the coding that you receive from your parents." FG3

"We inherit most of our genes, well what I think I know is we inherit most of our genes from our parents, so half from our mum and half from our dad. I know certain genes can affect say how likely we are to have certain diseases, or how likely you are to not have certain diseases, which can affect our health in the future. I know doctors can inform us of this so that we're aware of the possible dangers to our future health, there are some doctors that give advice based on your genetics that can help you watch out for these certain things that can damage your health." FG10

Some participants who had some knowledge of the terms genes and DNA said they knew of the terms through TV shows and movies.

"We mostly hear those terms in movies or TV shows when they want to determine the biological parent of the baby or by doing paternity tests by matching the baby's genes with the parent's genes. That is what I know about DNA." FG3

Others highlighted they wanted to know more about their genes and their risk for health conditions such as diabetes and cancer. Participants also queried whether their GP or healthcare professionals knew what it was, as the feeling amongst these groups was that healthcare staff provide very little information about anything or often do not know themselves about certain conditions and treatment options available to their patients.

When speaking with professional stakeholders, the feeling among participants was that genomics literacy was poor among healthcare professionals working in other therapeutic areas and other relevant professionals such as social workers, chaplains and counsellors. Additionally, these stakeholders also felt that genomics literacy was poor among minority groups and pointed to a bigger issue of poor health literacy within these communities.

Personalised medicine

Knowledge of personalised medicine amongst all public stakeholders was varied. Most participants reported they had never heard the term before and did not know what it was. One participant's view of personalised medicine was that of the family doctor

model they had experienced, whilst another participant saw personalised medicine as person centred care.

“Well I’m old enough and have been in this country for long enough to have experienced using a family doctor. A family doctor, as soon as he saw you, he knew everything about you. He knew your parents. He knew your brothers and sisters, what sort of ailments you had. And we haven’t got that anymore. That’s personalised medicine for me in very simple terms.” – FG6

“Personalised medicine is like saying personal centred care. Now, everybody knows that certain medications, like general for a general set of people, maybe black people can’t tolerate this particular medication, and there’s particular medication for high blood pressure that it’s just like generic, given to all black people. But there are people within this same group of people that probably would react to this medication. So you cannot give just a general medication to everybody.” – FG9

Health inequalities in precision medicine

Information around health inequalities, the differences in health outcomes for different communities and differences in service uptake of precision medicine services such as cancer screening and genetic testing was also discussed with public stakeholders. Some public stakeholders said they were not aware that differences in outcomes and service access differed between groups. Most public stakeholders were aware there were differences in health outcomes and access to service among different minority groups but queried whether this was an issue of uptake or not being offered services in the first place. These participants further asked about the measures in place to address the inequalities.

I thought the treatments spread it out equally. Well, I didn’t know about higher chances of them. I think it’s where it depends on geography, where they live or something. - FG5

Are the figures so low for the ethnic groups because they didn’t take up the services or were they never offered them? - FG1

5.3.4 Strategies for improving knowledge and awareness

Public and professional stakeholders discussed strategies for improving genomics and healthcare literacy among underserved communities. The following themes emerged from the interviews and focus groups (see Appendix 5.2 Improving knowledge and awareness themes for supporting quotes):

Role of healthcare professionals

All participants felt that healthcare professionals such as GPs, midwives and health visitors can play an important role in increasing genetic literacy among their patients who are from minority groups. Members of the public often felt they did not get much information from their health professional, and that receiving more information about genomics and personalised medicine from these individuals would be a good start. A few members of the public highlighted that midwives and health visitors could play an important role in teaching young families about genes and their family's health as well as the genomics services and testing available.

Healthcare professionals also reflected on the role that they could play in improving genomics and personalised medicine for these communities. Participants highlighted providing general information to build a base level of knowledge among communities would be helpful and make the process for individuals easier to understand when they are referred to and need to access genomics services. In professional stakeholder interviews, participants highlighted a current project called Genes and Communities, running in the North of England, which is providing genetic education for underserved communities and working to better support health professionals with engagement (see Case Study 1).

Genetics in Communities

Genetic in Communities is a community based education and awareness programme to improve genetic literacy among minority groups within the local community in the North of England. This initiative is funded by Bradford Public Health and hosted by WomenZone. WomenZone is a registered charity and community based organisation in Bradford. The organisation aims to empower, inspire and enrich the lives of women, many of whom are from underrepresented communities, taking a whole family approach to achieve this.

The Genetics Communities program was developed through collaboration between public health consultants, clinicians, researchers, teachers and community consultation. The aim of the project is to improve genetic literacy, awareness of genetic testing and services and provide underserved communities in Bradford with accurate information about how our genes influence health, the causes of genetic conditions, and the implications of consanguineous marriages. The program involves both group workshops and tailored one-to-one sessions for individuals and couples. Workshops and one-to-one sessions are interactive, include physical models and visual diagrams of the body, cells and DNA and are delivered in English and Urdu. The program commenced in March 2021 and will continue to run until March 2024. To date, the program has engaged with 300 people in the community and 200 professionals.

CASE STUDY ONE

Role of faith and community leaders

Both public and professional participants highlighted that faith and community leaders may play an important role in providing communities with health information. Some participants reported that faith leaders may be more trusted in their community to provide them with accurate information and would actively listen to the information they shared.

Professional participants also highlighted this and felt that working with community and faith leaders may be a useful way to improve genetic and health literacy within minority communities. Professionals cited community champion approaches, whereby leaders are trained by health professionals to provide health information to the communities they serve.

The community champion approach is now being adopted by Genomics England and their community ambassadors. Genomics England is in the process of setting up a training initiative for faith leaders from the Muslim community in England, where Genomics England and their community Ambassadors provide training on genomics and health to faith leaders so that they can provide members of their community with reliable information and signpost them to appropriate services for additional support (see case study 2).

Presence in community spaces and events

Public stakeholders generally reported the format of the focus group, which involved providing participants with information about genomics and health inequalities, to be beneficial and would prefer sessions of this format within their local community spaces.

Public stakeholders recommended that healthcare professionals and researchers should get involved in cultural or religious festivals that are important to the communities they want to reach. One participant provided a suggestion of participating in Diwali festivals to start the conversation and share information with lots of people at the same time. One professional stakeholder recalled attending a community Eid festival with the purpose of engaging the local community and raise awareness of genomics medicine services.

CASE STUDY TWO

Genomics England is a company set up and owned by the Department of Health and Social Care. It was first set up to deliver the 100,000 Genomes Project with the NHS. Since then Genomics England is now working with the NHS to deliver services which support the whole genome sequencing service, accelerate research and to deliver a number of proof of concept studies.

Genomics England works with ambassadors who are leaders or representatives within different communities. The ambassadors help share content and information about genomics within their communities and contribute to the national conversation. Genomics England has partnered with community ambassadors to increase genetic literacy in Muslim communities in England. Community Ambassadors hold information stalls in youth centres and mosques up and down the country.

The stalls provide communities with information about genes and their health while having a volunteer present for their community to talk to and signpost to further resources. Genomics England is also working closely with their ambassadors to develop first responder training for community and faith leaders. This approach involves providing community and faith leaders with training around genes and health and equipping them with the knowledge and confidence to have conversations around genomics with members of their community.

Role of lived experience

Public stakeholders suggested that people from their community who were willing to share their experiences of genomics services or research would be an effective way to engage minority communities. Hearing from a person's lived experience of how genomics services and research have benefitted their health, who are from the same or similar communities, may be more relevant to people from minority groups, prompting them to seek more information and access services.

Multigenerational approach

Public stakeholders highlighted the need for a multigenerational approach to improving genetic and health literacy among their communities. They highlighted not only should this information be shared with older people, but also with young people who are embarking upon starting a family. The information can be provided through family planning clinics, midwives and health visitors.

Additionally, public participants stressed the need for genomics and its impact on health to be taught in schools, whether in Personal Social and Health Education lessons or science lessons. By teaching children this information, they can take the information they learn back home to parents and grandparents, thus sharing knowledge and awareness across multiple generations.

Multimedia approach

Public and professional stakeholders recommended that information should be shared through multiple mediums. Having written information in different languages was seen as a start, but all participant groups highlighted that there are differences in literacy skills, thus written information alone, even in different languages would mean information is still inaccessible for some people. Stakeholders identified the use of social media such as community Facebook groups to share information, sending out information via WhatsApp to community groups, auditory and video methods (such as YouTube) as ways of communicating information about genes, health and related healthcare services that are available.

Sustained messaging

All public stakeholders and some professional stakeholders stressed that information needs to be shared with different groups in a sustained way over time. One-off public health campaigns to improve knowledge and awareness are likely to not be effective and quickly forgotten. Consistent messaging over time was thought to be the best approach to improving genetic and health literacy in underserved communities.

Tailored approaches for communities

Stakeholders also highlighted the need for tailored approaches for different communities. While some communities and some age groups prefer to attend in-person information sessions, others used local radio, social media and WhatsApp to receive and share information. Some professional stakeholders also provided details of current community engagement projects aimed at improving genetic literacy among ethnic minority groups (see case study 1 and case study 2).

5.3.5 Shared barriers to and facilitator to access and engagement with genomic medicine services and research

In addition to varied knowledge about health inequalities in genomics medicine, the following barriers and facilitators contributing to inequitable access and engagement common to both genomics medicine services and research were identified (see Appendix 5.3 Services and research themes for supporting quotes):

Challenges accessing healthcare services

All public stakeholders discussed and shared personal experiences with challenges accessing healthcare services. All participants reported challenges accessing GP services and felt that the quality service received was inadequate. Participants highlighted their understanding and awareness about the increased work pressure faced by GPs, nonetheless, public stakeholders reflected on personal experiences with GPs and other healthcare professionals such as nurses, and midwives, where they felt they were not treated well. Particularly, one public stakeholder highlighted that this could be due to potential biases the healthcare professional may unconsciously hold about people from specific groups.

Limited knowledge and awareness

All stakeholders highlighted the knowledge and awareness around services, genetic testing and research was limited and highly variable. A few public stakeholders who were aware of genomics services, came to this knowledge through their personal or their families' experiences with cancer and genetics services. Public stakeholders also mentioned that they did not know much about research and many participants said they had not been asked to participate in research studies before.

Literacy skills

All stakeholders reported that literacy skills are likely to vary within underserved communities, which can be a considerable barrier to accessing healthcare services. Public stakeholders highlighted that information about appointments, tests and other healthcare-related activities are usually sent to people via letter, in English. For those with low literacy skills, regardless of their spoken language, receiving communications in this way only will affect how well people are able to engage with service and their healthcare professionals. All stakeholders also highlighted that research participation often involves reading lengthy documents such as participant information sheets, consent forms, and surveys, which may exclude those who may not be confident with their English or literacy skills.

Socioeconomic factors

Professional stakeholders discussed the impact of socioeconomic status on engagement with services and research. People from lower socio-economic areas may struggle to afford time off from work attend appointments or travel to clinics for appointments and additional research-related activities. Providing remuneration for expenses relating to time lost from work and support with transportation should be implemented to overcome these barriers. One healthcare professional mentioned that as part of the 100,000 genomes project, reimbursement for travel expenses were provided for participants who required it.

Another healthcare professional discussed how patients often had to travel quite far to undergo medical tests which took significant time commitment and being able to afford transport to travel into clinic. This was a barrier for those from lower socioeconomic groups. To overcome this barrier, they adopted a hub and spoke model so that patients could attend clinics for testing closer to home.

Language

All participants discussed language barriers at length throughout the focus groups and interviews. Language barriers were seen to widely impact access and engagement with healthcare services and research including the fields of genomics. The following sub-themes pertaining to language barriers were identified (see Appendix 5.4 Language themes for supporting quotes):

Accessing interpreters

All participants discussed the challenges around accessing interpreters and felt this impacted the quality of the interactions between healthcare professionals and patients. Healthcare professionals discussed how lack of translators, and reliance on family or friends to translate made it difficult for them to support and communicate complex information regarding genomics to their patients.

Translation accuracy

Professional stakeholders in clinical genetic services expressed concerns about the accuracy of information relayed when communicating with patients through a translator, be it a professional translator, family member, or friend. They questioned if patients truly understood their intended message. Public stakeholders also voiced concerns, emphasising that there were instances where they felt translators were unable to accurately convey their messages to healthcare professionals or accurately relay the advice given by healthcare professionals back to them.

Language impacts engagement

Professional stakeholders felt that terminology relating to genomics can be difficult to communicate and complex to understand which can impact engagement in services and research. A few public stakeholders felt that terms relating to genomics and the way in which professionals talk about genomics was more medical language and people find it difficult to understand, so it needs to be translated to plain English. Additionally, with different languages it is possible that the terminology related to genomics does not exist, making it difficult to communicate which may also hinder engagement.

Mistrust

All participants highlighted the role, mistrust of healthcare professionals, systems and research within ethnic minority communities plays in access and uptake. Participants reflected on where this mistrust comes from and ways in which healthcare professionals, researchers and leaders can work with ethnic minority groups to build trust. The

following subthemes relating to mistrust emerged from discussions with stakeholders (see Appendix 5.5 Mistrust themes for supporting quotes):

Fear

Public stakeholders from Arab, African and Caribbean communities discussed that there was fear among their communities, which may explain why the uptake of services and involvement of their communities in research was limited. One participant from the Arab community said that there may be fear within their community about how samples or data they provide for research will be misused and therefore people may be reluctant to come forward and get involved. Participants from African communities reflected on how people were suspicious about what their samples or their data would be used for. Professional stakeholders also discussed mistrust, fear and suspicion of healthcare system and healthcare professionals and talked about the need to be aware of historic trauma people have faced, explore and understand why people are experiencing mistrust.

One researcher suggested that better communication around the purpose of collecting information and how it will be used would be helpful in addressing people's fears, especially for people from minority groups who may have been treated differently due to their background. Another clinician felt fear and mistrust combined with socio-economic challenges may lead to decline in individuals from minority groups in participating in research, as they perceive it as irrelevant to their needs. Hence, having a conversation about the purpose of services and research and the impact on their health has been effective for people from minority communities to appreciate that it is relevant for them.



Acknowledging the past

To build trust, public participants highlighted that the past needs to be understood and acknowledged and the lessons learnt discussed. Public stakeholders from African and Caribbean communities reflected on the community history and real-life examples such as the Syphilis studies and other unethical practices conducted on Black people. Public stakeholders also reflected on personal past experiences that many people from minority communities may have had with healthcare professionals and services which had led to mistrust. Thus, the public stakeholders highlighted the need to acknowledge these stories and learn from them moving forward.

Government policies and politics

Politics and government policies around immigration, for example the Windrush scandal, have contributed to mistrust of the government and related institutions such as the health service among ethnic minority communities. Additionally, the rhetoric and stances taken by Governments on key issues relating to foreign policy also contributed

to mistrust within diaspora communities living in the UK. One participant highlighted how Government stances on foreign issues in countries where people were of a similar race to them impacted their trust in government-related institutions. A healthcare professional reflected on how stances taken on specific cultural practices such as consanguineous marriage has also contributed to mistrust and suspicion among those communities. One public stakeholder also highlighted how more recently, the discussion around COVID-19 and the disproportionate impact on Black and ethnic minority groups contributed to mistrust among these communities as they felt they were being blamed.

Building trust

To build trust, public stakeholders discussed how this needs time and how sharing information is a key first step.

Trust is earned over time. For them not to want to trust you, they know what they have seen. They know what they have heard. Let's make a reference to COVID. So, people felt like they were going to be killed by the vaccine and stuff like that. So, I don't know how to end that, but I just know that you don't force people to trust you. You earn it over time. Information is one of the first ways to – and trust. People need to know; people need to be carried along. People need to understand why certain things are being done. Not everyone is out to hurt you. But, like I said earlier, you earn trust. You don't force it. - FG9

Representation

All stakeholders discussed the role of representation in increasing engagement within both services and research as well as the ways representation can be achieved.

The following sub themes relating to representation were noted (see Appendix 5.6

Representation themes for supporting quotes):

Diversity in materials

All stakeholders identified that current promotional materials used in healthcare services and research do not promote diversity. Promotional and research materials need to be designed to be more inclusive so that people feel seen and that they too can participate. Imagery used in leaflets and promotion materials often feature white people, which may lead to some people of ethnic minority groups to conclude that the health condition or disease may not affect them. Also informational materials used in research to the way participant information sheets are designed and tested with the populations of interest need to be adapted to be more inclusive. A community representative from the Arab community provided an example of being involved in a video to provide updates about COVID-19 in Arabic and how the community interacted with it. Having someone that looks like you, speaking your language can help people feel valued and included.

Workforce diversity

All stakeholders felt that more diversity within the workforce, both in health service and in research is needed to improve engagement from minority communities. Public stakeholders reflected that being able to see someone who looks like them, and who understands their culture would help them to open up and discuss their health. They would feel like their healthcare professional or researcher understands their culture and their experiences.

Representation of the messenger

In addition to representation within the workforce, public stakeholders also discussed the importance of the representation of the messenger providing communities with information about genes and health. Participants discussed how if the persons engaging within their community looked like them or understood their culture, they would know that their experiences were understood, and they would be more comfortable engaging in the discussion and sharing their thoughts. For communities to see themselves represented would help them feel more valued.

5.3.6 Specific barriers to genomic medicine services.

In addition to common barriers to services and research, we identified the following service-specific barriers that applied to precision medicine/ genomics services and healthcare services more generally (see Appendix 5.7 Service themes for supporting quotes):

Limited understanding of cultural and religious differences

All public and professional stakeholders highlighted that healthcare staff generally had little understanding of the cultural and religious norms for each community and the difference between communities. Stakeholders, especially public stakeholders across all community groups felt that lack of cultural awareness affected the quality of care they received.

Public stakeholders also felt that healthcare professionals' knowledge of differences within communities was poor. While minority communities such as African, Caribbean, South Asian, are acknowledged in the UK, public stakeholders highlighted that there are lots of different communities within these groups. Some participants who were African and from Uganda highlighted how within their country there were huge differences in dialect and cultural practices.

Participants from the Arab community also highlighted issues around lack of understanding of cultural differences, reflecting their experiences of being mislabelled as being Pakistani while discussing how these two communities differed. Additionally, participants from the Arab community stressed that there were many cultures and communities within the Arab community itself and this needs to be recognised and better understood by professionals.

Healthcare professionals' knowledge and confidence

Professional stakeholders discussed how healthcare professionals outside of genomics had limited knowledge and lacked the confidence to have conversations with patients about genomics services and testing. Public stakeholders also queried how well healthcare professionals they often saw such as GPs and nurses understood and could provide them with information about their genes and their health.

Healthcare professionals' biases (e.g. stereotyping)

Professional stakeholders with working experiences in clinical settings highlighted how healthcare professionals' and leaders' biases may affect how accessible services and testing are. One healthcare professional reflected on their experience with leadership when trying to develop a programme to engage with local ethnic minority communities and improve knowledge about genes and health and how leadership had preconceptions about the relevance to these communities.

Public stakeholders from Black African, Caribbean and Arab communities reflected on their experiences of encountering biases and stereotypes held by healthcare professionals when receiving care. One participant from the Arab community highlighted an example of how they felt when GPs see a woman wearing a hijab, they make assumptions about the patients' language, education and beliefs affecting how the individual is treated.

Gatekeeping by healthcare professionals

Public and professional stakeholders discussed how certain healthcare professionals may act as gatekeepers to services and make judgements about the patient's need without consulting the patient. Public stakeholders reflected in their challenges in getting a referral to specialist or tertiary care services from their GP, with some participants feeling that they would not be referred to services if they asked. This is because healthcare professionals would have made conclusions about the patient and not refer them for the appropriate service.

Negative feelings (stigma, shame, guilt, fear)

All stakeholders reflected on the potential for negative feelings as a barrier to genomics services and research. Professional and public stakeholders discussed how having a genetic condition, in some cultures, may impact the individual as well as their families and relatives. Professional stakeholders discussed how parents may experience shame or guilt if their child is diagnosed with a genetic condition and how often in minority communities these feelings of shame and guilt are largely the burden of the mother.

Participants from African communities discussed how feelings of fear may stop people from accessing genomics services and participating in genomics research. Four participants in one focus group highlighted how in some Black communities, the word cancer is associated with death and because of this consequently deterred people from undergoing screening and seeking treatment. Additionally, there is a fear of research within these communities; individuals were often sceptical about the destination and use of samples they were asked to provide.

The implications of testing outcomes

Professional stakeholders discussed the implications of testing outcomes for patients and their families. The result of a finding may impact future family planning for people from minority groups. Additionally, results may also bring about stigma and affect patients' social standing within their community, affecting prospects for members of their families (e.g. marriage). One healthcare professional reflected on anecdotal observations in cases where testing outcomes revealed genetic conditions within children from ethnic minority groups. They reported that the mothers often bore the burden of guilt and potential blame.

Professional stakeholders also pondered that possible ramifications of testing outcomes for people from already minoritised groups may further marginalise individuals and families within their own communities, which may lead to reluctance to access genomics services and undergo testing. Healthcare professionals also discussed the implications for findings of unknown significance or limited testing power for some communities.

A genetic counsellor reflected that there is a possibility that patients may receive a test result that is uninformative, which may mean they are more reluctant to undergo testing. Professional stakeholders also pondered that with improved datasets and more inclusive research, what we know about our genes and health will evolve, and although there may be no finding at present there may be a possibility of a future finding and wondered how this can be communicated to people from minority groups without causing confusion or affecting confidence in the service.

Waiting lists and a busy NHS

Most public stakeholders stressed how busy the NHS is and how long waiting lists are for other healthcare services. Public stakeholders were sceptical about how genomics services would be rolled out in an already struggling service and some public stakeholders felt there were more pressing issues the NHS needed to address like shortage of staff and long waiting lists to see a GP or specialist. Professional stakeholders also discussed how staff across the health service will be trained to support genomics services given most are already overstretched and work past their capacity. GMSA representatives anticipated this being a barrier to staff engagement and providing education and training to the workforce.

5.3.7 Specific barriers and facilitators for genomics medicine research

We identified the following research-specific barriers that applied to precision genomics research and are transferable to healthcare research more broadly (see Appendix 5.8 Research themes for supporting quotes):

Lack of diverse data

Professional stakeholders highlighted known issues about biased datasets and the barriers these present in precision medicine research. The impact of biased datasets on advances in research, development and utility of testing and treatments was also discussed. Professional stakeholders were clear there was a need for greater diversity in genomics databases to reflect the wider population. Current projects aimed at improving diversity in genomics data such as the diverse data and new-born screening programmes led by Genomics England and the Our Future Health programme were cited by professional stakeholders as examples of initiatives aiming to resolve the issue around the lack of diverse data.

Current recruitment methods

Current recruitment methods used in research generally and within genomics were also thought to be a barrier to the inclusion of ethnic minority groups. The need for tailored recruitment strategies was also highlighted by researchers.

Concerns around how data will be used and managed

Public stakeholders discussed concerns about how the samples and data they provide will be used and potentially misused. Some public stakeholders also referenced eugenics theory and associated practices and how genomics could use their samples and data for this purpose. Public stakeholders also raised questions about how their samples and data will be stored, and how they will be accessed and shared. Some participants were concerned that their data would be used for insurance purposes.

Increase awareness of research (talk to people)

All public and professional stakeholders discussed the need to improve awareness of research within communities. Public stakeholders highlighted that more awareness of research is needed within their communities. One participant from the Arab communities reflected on their experiences of being invited to participate in research but no one talked them through what was involved and so they ultimately declined to participate.

Another participant from Black Caribbean community also reflected on their family's personal experiences with research, recalling that it was impersonal, insensitive to their

loved ones circumstance and there was little opportunity to discuss the research with a healthcare or research professional. A researcher reflected on their experiences of recruiting participants and discussed how talking to people, being able to teach them the value of what is being done, why it is important and being able to have a discussion improved engagement with research.

Close the loop

Public stakeholders who had been involved in research previously said that they had not received any feedback on what happened with the study, what their results contributed to and what had happened since. Public stakeholders highlighted that getting feedback on what had happened with the research they had participated in would be helpful. It would enable them to understand the impact of getting involved and would motivate them to stay engaged with research. Professional stakeholders also acknowledged that more needs to be done to close the loop and communicate to participants the outcomes of their participation.

Inclusive research processes (go to the communities)

Healthcare and research professionals highlighted the need to develop and use more inclusive research processes. Participants discussed how research information is currently delivered using lengthy and wordy participant information sheets and how this could be made more accessible by adapting the language used and using different formats such as video clips to deliver study information. Additionally, all stakeholders felt that research and researchers need to go to the communities rather than expecting people to come to them. Engaging with community groups directly in their spaces will improve awareness of and participation in research. Public stakeholders also gave examples of how researchers and healthcare staff could reach communities through radio, social media, WhatsApp and by attending key events held by communities that are important to them such as cultural or religious festivals.

The value of being involved and the consequences of not participating

All public and professional stakeholders felt that to improve research participation, the value or benefit of participating for the individual, their families (specifically future generations), their communities and the potential future benefits needs to be communicated. Some stakeholders also suggested that researchers need to highlight the consequences of not participating, for their health and treatments available to them, their loved ones and the wider community.

5.3.8 Community engagement

The following themes pertaining to community engagement were noted (see Appendix 5.9 Community engagement themes for supporting quotes):

Sustained engagement

All stakeholders discussed the need for continued sustained engagement initiatives. Public stakeholders highlighted that researchers and healthcare professionals often approach them to collect information and ask questions, but afterwards, the communities do not receive feedback on how the provided information were used or its impact on effecting change. Researchers also highlighted how community engagement activities for patient recruitment and genetic studies can feel somewhat tokenistic leading to groups feeling used. Developing and sustaining long-term relationships with communities would help to address the communication gap communities experience and lead to a mutually beneficial relationship.

Strategies to improve community engagement

Both public and professional stakeholders discussed the need to acknowledge differences between diverse minority groups and factor these in when designing studies, recruitment strategies and informational materials. Stakeholders discussed different cultural and social norms that exist in minority communities that need to be appreciated, highlighting that a one-size-fits-all approach to community engagement is not appropriate and therefore more tailored approaches are needed to initiate and sustain community engagement.

Healthcare professionals from GMSAs discussed strategies they are considering to improve community engagement. One nurse discussed the importance of spreading knowledge and raising awareness of genomics among minority groups and discussed how they are considering a community ambassador approach, where key members of the community are trained to deliver information about genomics to people in their communities. Another nurse working in GMSA talked about how the organisation is exploring opportunities to collaborate with local schools, with a focus on learning about engagement strategies employed in computer science and STEM subjects, with the goal of adapting these strategies for genomics. Healthcare professionals and public stakeholders also discussed the role prominent community or public figures such as celebrities could play in engagement with minority communities. One public stakeholder cited the COVID-19 public campaign where celebrities from ethnic minority groups came forward to provide people with information about the COVID-19 vaccine and US politics where celebrities get involved to share information with the public.

5.3.9 Monitoring equity of access in genomics medicine services – the barriers and facilitators

Healthcare professionals working in clinical genetics and GMSA representatives discussed their challenges with ethnicity data recording, including issues around data quality. GMSA representatives also discussed challenges to being able to access the data and barriers with local data governance policies (see Appendix 5.10 GMSA equity of access themes for supporting quotes):

Current data recording practices

Each of the GMSA's representative we spoke with reported that data collection in each of the regions they represented is inconsistent and patchy, with some local areas they cover being more consistent than others. The clinical genetics services in the regions covered by the GMSA often do not use the same categories for ethnicity, meaning data is not always accurate or reliable. Additionally, some healthcare professionals and GMSA representatives discussed how ethnicity is not a mandatory field on genetic testing order forms and is often left blank by healthcare professionals ordering tests.

Current testing order process

The GMSA representatives we spoke to also highlighted that the format of referrals is either letters or forms and, in both cases, ethnicity is frequently not recorded. Each GMSA representative discussed the need to homogenise data collection by developing a standardised form that can be used to make referrals for clinical genetics services and testing. This could potentially improve data quality, monitoring and reporting of access to, and uptake of services by minority groups. One of the GMSA representatives recently developed a pro forma which is currently being piloted and another GMSA representative proposed collaborating with other GMSAs to develop a standardised form and process for data collection that is consistent at a local and national level. Professional stakeholders from the Genomics Unit at NHS England shared that there is work underway to develop an electronic testing ordering system that will standardise the process, and that a recently developed standardised dataset, will collect ethnicity, as part of the electronic testing order process.

Data sharing, access and governance

The GMSA representatives we spoke with discussed challenges around accessing data on ethnicity. The GMSAs are organisations covering regions with multiple different NHS trusts, however this also means due to individual information governance policies at each trust, access to data around ethnicity, service access and uptake is challenging. Professionals across each of the GMSAs highlighted that local policies means they are currently unable to access data and monitor access and uptake of services by different ethnic minority groups.

The needs to improve data sharing between GLHs and GMSAs was also stressed. The representatives we spoke with discussed the possible benefits of having a national level policy for data sharing and access which would be helpful for GMSAs to be able to negotiate and develop local level data sharing agreements.

GMSA EDI strategy development

GMSA representatives discussed their plans for developing local EDI strategies and are at different stages in the process. Each GMSA highlighted that developing and operationalising their local EDI strategies are key priorities for the year ahead. Each of GMSAs that participated in the informal focus groups also spoke about the need and potential benefits of having a national EDI framework in which each organisation can develop more localised strategies based on the populations they serve. Healthcare professionals including genetic nurses who work within and closely with GMSAs discussed in more detail their plans for their local EDI strategies and community engagement.

Collaboration

Each of the organisations we spoke with highlighted a need for collaborations amongst GMSAs to exchange ideas and discuss how GMSAs are tackling issues around data collection, accessing data and developing local data sharing agreements. Participants also discussed the need to collaborate with the other GMSAs to learn best practice from each other on how to improve equity of access and diversity in genomics medicine services.

Projects and initiatives aimed at addressing inequities

GMSA representatives and nurses working within the GMSAs also discussed projects they were aware of or involved in that were aimed at improving equity of access to genomics testing. A few examples cited by stakeholders were local transformation projects, for instance, one GMSA had been working with GPs in their local primary care network to provide patients concerned about family history of breast cancer with a direct pathway to genetic testing. Another GMSA did a data mapping exercise to explore the most common languages spoken in the area they serve, and the data were further used to develop information in those languages for patients.

The 'Genes and Health' project was another widely cited project in interviews and focus groups with participants. This project is a community-based genetic study running in East London, Bradford and Manchester. The study aims to improve knowledge, awareness, and inclusion of individuals from Pakistani and Bangladeshi communities, who often experience poorer health, by studying the genes and health of 100,000 people from these communities.

5.3.10 Workforce training

Both public and professional stakeholders identified training needs for the workforce. Professional stakeholders discussed how best to deliver these training. The following themes highlight several training needs for consideration (see Appendix 5.11 Workforce training themes for supporting quotes):

Genomics medicine training for healthcare and other professionals

Professional stakeholders discussed how training for healthcare professionals should provide knowledge and information on genomics services and available testing services so that they are better informed and able to support and signpost patients appropriately. All stakeholders highlighted that complex terminology relating to genomics medicine can be difficult to follow for patients and for healthcare professionals to communicate, especially for those working outside of genomics services. Additionally, professional stakeholders also discussed the need for training on how to best communicate complex information relating to genomics medicine services and research to patients and the public in an accessible way. Some professional stakeholders felt that there should be some level of training around genomics services, testing and research for other professionals such as social workers, and chaplains who may be involved in healthcare decision-making or providing support to patients. This would mean that other professionals are able to make informed decisions and direct patients to the most appropriate support.

Cultural and religion awareness

A need for training encompassing cultural and faith awareness across healthcare and research professionals was identified by stakeholders. One public stakeholder highlighted that healthcare professionals need to better understand the differences within communities citing the south Asian community as an example that is made of people of Indian, Pakistani, Bangladeshi communities to name a few. Research professionals also discussed the need for cultural awareness training for research professionals so that they can design studies, informational materials, study processes that are inclusive and sensitive to the needs of people from different backgrounds.

Community engagement training for researchers

Both professional and public stakeholders highlighted that researchers need training around community engagement. One research stakeholder highlighted that researchers involved in applied research with patients and basic science research within laboratories should be trained on how to engage with communities and communicate their complex research in an accessible way.

Importance of ethnicity data recording and having the conversations with patients

Issues around ethnicity data recording and being able to have a conversation about the purpose of collecting this data with patients were raised in discussion with professional stakeholders and GMSA representatives. These stakeholders felt that at present, the quality of the data available and recording of this data was poor. Some participants recalled conversations with healthcare professionals around the importance of ethnicity data recording and were advised by healthcare professionals they spoke with that they did not deem recording ethnicity as important to treating their patients. Other healthcare professional participants reflected on experiences when asking patients for this data and patients being unwilling to provide this information. Furthermore, some healthcare professionals enquired about the point at which this data should be collected and who should be asking the question, with mixed responses including healthcare professionals and administrators.

Professional stakeholders all recognised the importance of collecting ethnicity data and how it can be used to monitor and improve access to and uptake of services and research for minority groups. Professional stakeholders also stressed that the workforce needs to be trained on why this data collection is necessary and how to have the conversation with patients so that they understand the purpose of why this data is being collected and how it will be used.



5.4 SUMMARY

Knowledge and awareness of genes and health varied across ethnic minority groups. All professional and public stakeholders queried knowledge, awareness and confidence around discussing genomics with patients among healthcare professionals working in other areas of the health service. All professional and public stakeholders discussed strategies to improve knowledge and awareness among ethnic minority groups. The need for tailored engagement strategies to share knowledge and sustained messaging was suggested. Additionally, the role of healthcare professionals (mainly GPs) and community leaders being seen as reliable sources of information by ethnic minority groups was also emphasised.

Several barriers to genomics medicine services and research faced by ethnic minority groups were noted. Challenges accessing current healthcare services, a lack of information about what the services and testing are, what is available and how to access was highlighted by public stakeholders. Also lack of information about research, its purpose and the benefits of participating or consequences of not participating also need to be communicated to ethnic minority groups in an accessible way.

All stakeholders emphasised the impact of language barriers on the ability of healthcare professionals and researchers to effectively communicate complex information to ethnic minority groups. Stakeholders provided examples of how language barriers have affected the level of care, patient engagement and participation in research. There is a need for better access to interpreters who are qualified and able to communicate complex terminology between patients, healthcare staff and researchers.

Mistrust plays a major role in inequities of accessing services, testing and participating in research among ethnic minority groups. All stakeholders discussed the reasons for this, talking about historical examples of abuse that ethnic minority communities have experienced through the hands of healthcare systems, healthcare professionals and research. Public stakeholders also shared stories of personal past experiences of inadequate healthcare support that have led to feelings of mistrust, fear and suspicion. Additionally, there were concerns about providing samples for genomics testing, public stakeholders queried what the data would be used for and who would be accessing it. To build trust, stakeholders need to take time to understand the reasons for mistrust and create a space for people to share their stories to heal communities. Additionally, stakeholders need to acknowledge the past, and inform communities of what was learnt, what has changed and how it is different now.

All stakeholders highlighted the importance of community engagement to improve knowledge and awareness of genomics, precision medicine and research. Public stakeholders were highly aware of tokenistic engagement and discussed how regular engagement and feedback is required. Additionally, stakeholders discussed the need for meaningful, tailored and sustained engagement with ethnic minority groups and

the inclusion of these groups in developing and implement strategies. Community engagement is also key to building trust, and reassuring communities of current practices in place to keep them safe when accessing care or participating in research. Community engagement could also benefit from creating a space for ethnic minority groups to share their collective and individual stories.

Barriers to monitoring access to and uptake of testing, included problems with data recording of protected characteristics such as ethnicity. Recording of patient ethnicity data is inconsistent and quality of data available is poor. Healthcare professionals and GMSA representatives suggested reasons for this included not understanding the importance of collecting the data, not asking patients or healthcare staff feeling uncomfortable about having the conversation with patients. GMSAs who are tasked with addressing access inequity also stressed that systems used to record patient-level data are not compatible with one another making it difficult to access and make sense of the data available. Additionally, information governance policies and challenges with data sharing agreements are additional hurdles to monitoring of access and uptake of genomic testing.

Stakeholder interviews also highlighted a series of training needs for the workforce. These included training for all healthcare professionals across all levels to build their knowledge about what genomics and precision medicine is. Training around cultural awareness also needs to be developed and rolled out across the healthcare system more widely, not just in genomics and precision medicine.



6. DISCUSSION

6.1 OVERVIEW OF MAIN FINDINGS

Advances in genomic and precision medicine are encouraging and tailored approaches are being increasingly embedded within healthcare. However, our research brings attention to a variety of ethnic inequalities inherent within the field, that will further exacerbate health disparities, if these are not urgently addressed by research and policy.

Our findings highlighted that there is an ethnic bias in genetic datasets, with widespread underrepresentation of ethnic minority groups, where the focus has been on studying European origin populations. The implications of these data limitations are far-reaching, and run across genomic and precision medicine, from AI to PRS, where there is danger of misinterpreting the clinical relevance of research findings if diverse communities are not represented in research and service development. All three research studies conducted highlight that public and stakeholder engagement in the field of genomic and precision medicine is often piecemeal or not described in adequate detail. Some examples of public engagement with ethnic minority groups, such as Black African and African Caribbean communities were detailed, and this had led to recommendations for conducting equality impact assessments for clinical research (though this is not being done routinely either). Implementation plans arising from other engagement activity and more broadly, in relation to the genomic initiatives that are either being rolled out or planned are also lacking, often only aspirational statements are offered.

A concerted effort to reach out to and involve underrepresented ethnic minority groups using communication channels appropriate to the specific communities with tailored public engagement activities are clearly needed, which require investment in appropriate resources, to enable these groups to contribute to the field, optimally. Key to improving access to genomic medicine services is accurate monitoring of the ethnicity of those accessing the services, together with monitoring outcomes of care between different ethnic groups. Achieving this will also be dependent on the right infrastructure being in place and effective collaboration between relevant stakeholders. There is a drive to improve the genomic education of health professionals from undergraduate to advanced postgraduate training. Cultural awareness and implications of genetic diversity needs to be central to training modules.

The UK government has published a 10-year strategy to realise a genomic healthcare system to deliver better health outcomes. There are three areas of focus – diagnosis,

predictive and preventative care. To make advances in these areas, the strategy appears committed to investing in public engagement, workforce development, supporting industry growth, maintaining trust and co-ordinating efforts to data and analytics. Many of the findings of this research taps into the areas set out in this strategy and therefore the remainder of this section contextualises the findings from this research, accordingly.

6.2 ETHNICITY DATA IN GENOMIC DATASETS, GENOMICS RESEARCH AND PRECISION MEDICINE SERVICES

Repeatedly, it has been identified that ethnic minority populations are underrepresented across genomic and related large population databases, with suggestions that ethnicity data are missing altogether, for many patients ^(38, 39, 42, 282-284). In this review, we found that consideration of ethnicity was often limited to basic descriptive information about ethnic minorities in the databases, ^(92, 100, 104, 128, 132, 134, 151, 163, 188) and as most studies used the UK Biobank, this information was identical in several papers. Furthermore, in terms of analysis of genomic data on ethnic minority groups this also appeared superficial, where findings are rarely interpreted in results and discussion sections; though these studies do acknowledge that lack of ethnicity data is a limitation.

Unsurprisingly, GWAS were also dominated by European ancestry populations, which led to concerns about resulting PRS that lack predictive utility among ethnic minority populations. Ethnic bias in genetic datasets (i.e. towards European ancestry populations), GWAS and PRS were acknowledged widely across many of the policy and guidance documents included in our review. Additionally, our evidence synthesis revealed examples of ethnic diversity within existing datasets which were also mentioned in some of the policy documents. Findings from our evidence synthesis showed that when ethnicity data is used in data analysis, this is usually as a covariate in multivariate analysis rather than trying to identify clinically meaningful differences between ethnic minority groups and “European” groups ^(85, 86, 89, 92, 93, 96, 97, 100, 101, 113-117, 122-124, 126, 134, 135, 138, 140, 146-148, 150, 151, 154, 155, 159-162, 164, 165, 168, 173, 176, 177, 180, 181, 186, 188, 189, 191, 192, 194, 198, 200, 201, 204-206, 213, 215, 217, 221, 254, 264, 271).

Our evidence synthesis also brought attention to the many studies that are being published based on databases such as the UK Biobank that only include or attribute their findings to European ancestry populations, which is of immense concern given that policy and strategy documents set out that these sorts of initiatives were committed to recruiting ethnically diverse populations. Our qualitative interviews with health professionals, academics and focus groups with individuals representing the Genomic Medicine Service Alliances (GMSAs) do however suggest there are some encouraging pieces of work being done to improve ethnic representation in genomic medicine services. For instance, one GMSA conducted a data mapping exercise exploring languages spoken in their area from the Office for National Statistics Census 2021 and overlaid it with patient data to identify the most common languages spoken by

patients. This data is now being used to develop content in multiple languages relevant to the local area. Improving research practice among the research community is also warranted, for instance, research approval committees should appraise study proposals on how they will incorporate information on ethnicity in the overall sample.

Issues related to data recording on ethnicity need to be addressed. Challenges regarding the collection, recording and categorisation of race, ethnicity, and ancestry, and the interchangeable use of these terms have been previously reported ^(11, 285-287). Our document review and qualitative research supports the problems associated with ethnicity recording cited previously and offers some insights and possible resolutions. Our interviews with health professionals and GMSA representatives highlighted patient-level data around protected characteristics is not adequately captured and quality of the data available is poor – raising questions around the adequacy of systems in place, and the role(s) and perceptions regarding ethnicity recording among those responsible for collecting this information (individual, service and organisation); where our findings suggest that some health professionals find it difficult to ask questions about ethnicity.

Consultation with ethnic minority communities is also warranted because we found that Arab communities experienced being mislabelled as Pakistani. GMSA representatives also highlighted differences in ethnicity coding between different health services and electronic patient record systems. These sorts of shortcomings are likely to underestimate the true extent of ethnic inequities in genomic and precision medicine research and services. Moreover, these are unlikely to improve until a policy change or formalised guidance is introduced to improve patient-level data recording practices on ethnicity (and protected characteristics overall), ⁽²⁸⁸⁾, and that auditing and monitoring processes match ⁽²⁸⁸⁾, across services within the NHS that feed into genomics and precision medicine services.

Data monitoring and evaluation of datasets and genetic services (e.g. referrals and uptake of genetic testing, counselling etc) according to ethnicity are also not readily available for the UK context – and this was an aspect that our health professional participants and GMSA representatives discussed. This therefore suggests that routine monitoring and evaluation is currently not happening in the UK. Interviews and focus groups, particularly with GMSA representatives identified ‘stumbling blocks’ in terms of being able to access ethnicity data in relation to genomics testing uptake, research activity and databases, despite reports that such data should exist; though there were also admissions that data recording on ethnicity was of poor quality, often incomplete, inaccessible due to differences across electronic records systems or unavailable altogether.

Our qualitative research shows that standardised ordering processes for genetic testing, developing a core dataset relating to order testing and monitoring access and uptake of testing for marginalised groups is needed. Whilst we found that the development of an electronic order testing system is currently underway, the testing and roll out of the system needs to consider relevant stakeholders’ data and access requirements, ensuring this is factored into the core dataset and data sharing arrangements.

Another system change needed is around developing data governance and sharing policies that improve access to data so that GMSAs can access relevant patient-level monitoring data to assess current inequities in access and develop targeted community engagement strategies to improve knowledge, awareness and accessibility of services and genomic testing. These system changes require urgent attention to ensure that these weaknesses do not result in healthcare inequities, where at present, individuals of European ancestry would benefit most in terms of identifying risk of disease and any resulting advances in precision medicine healthcare. ⁽⁶⁷⁾

6.3 ENGAGING INDIVIDUALS REPRESENTING DIFFERENT ETHNIC MINORITY GROUPS

Engagement of different ethnic minority groups is lacking in both clinical research and genomic medicine services – this spans public engagement, research participation and access to resulting services, which is not a new phenomenon. Our findings along with those reported previously, show that ethnic inequities are the result of various factors, that include mistrust of the healthcare system, language barriers and discrimination, and being exposed to implicit bias, discrimination and stereotyping within the healthcare system. These facets map on to theories on structural and systemic racism, literacy theory, the resistance model and competency theory that have all been implicated when attempting to assess why ethnic inequities exist and persist within this field.

Despite the available evidence on the relevance of these constructs in relation to ethnic inequities in genomics and precision medicine, lack of commitment to understanding and thus addressing the underlying social, cultural, environmental and lifestyle factors that contribute to these disparities, is questionable ⁽²⁸⁹⁾. Improving the lack of genetic diversity in data, research participation and thus service access, requires the formulation of approaches that are relatable to the different groups concerned. For example, we found many references to the lived and historical experience of unethical research practices that will likely impact on ethnic minorities' response to the offer of study participation. These issues need to be addressed as part of the development of genomic research with these communities.

Our qualitative work showed that many of the individuals we spoke with appeared to have low levels of health and genomic literacy, lacking basic understanding of genetic concepts and the nature of precision medicine, altogether; suggesting that there is a lot of work to be done and that considering health and genomic literacy more widely i.e., among White populations is also warranted. As such, these and other barriers alluded to above, will lead to these underrepresented groups being unable to make informed decisions, use and interpret genomic information or benefit from genetic technologies – and that in the main, at present, these groups are unable to contribute to the genomic and precision medicine dialogue, until basic education on these concepts is cascaded in way that is meaningful.

Our document review suggested that efforts to reach out to and involve underrepresented ethnic minority groups using communication channels appropriate to the specific communities with tailored public engagement activities are needed. This requires clearly defined plans for implementation rather than the many examples of aspirational statements ('visions', 'ambitions'), that we reported in our results. A particular finding that stands out from our focus groups was that ethnic minority groups stressed that their "stories" needed to be acknowledged and "heard" and we posit that by doing this, we would foster greater understanding of the myriad of factors that are likely related to ethnic inequities in genomic and precision medicine.

Adopting a community-centred approach was advocated too, whereby barriers can be understood among communities and then knowledge can help inform development of approaches to reach underrepresented ethnic groups. For instance, in the US, Community Health Workers (lay health workers) serve as an intermediary between health professionals and patients and have a remit to improve this relationship through tailoring care according to the needs of the individual, ensuring it is conveyed in a culturally competent manner – which is an approach that has been found to facilitate greater engagement ⁽²⁹⁰⁾. Similar approaches are evident in the UK, such as the bilingual health researchers that led to the recruitment of Pakistani and Bangladeshi people as part of the Genes and Health Study ^(290, 291). During interviews with key stakeholders, one community engagement representative talked about Genomics England's community ambassador approach to improve genomic literacy in communities which involves connecting with community leaders and representatives who then share information and content within their own communities.

Our own work highlights the importance of gaining the support of gatekeepers who were pivotal in facilitating our engagement activity and subsequent research with the communities they represented. This is in line with utilising community champions who are from the same community, look like them, speak the language and share similar experiences. These sorts of approaches are also in line with co-production (or co-creation) techniques that were also advocated in some of the documents we reviewed, and in the recommendations sections of studies included in the evidence synthesis. For instance, some studies have utilised bilingual health researchers who recruited in community settings. Other examples that stress the importance of using such approaches is documented, but these need to be more commonplace.

6.4 THE HEALTHCARE WORKFORCE

Improving the provision of genomic medicine research and healthcare is also dependent on the practices of the healthcare workforce.

We found that both patients and health professionals have challenges accessing interpreters and translation services. This lack of language support has unfavourably

affected the quality of care and information available for ethnic groups. Hence, service provision to support all parties is warranted, as previous research cited not being able to speak English fluently or understand the healthcare system makes it difficult for people to interact constructively with health professionals e.g. about antenatal care,⁽⁵⁵⁾ and in, decisions about genetic screening.⁽⁵³⁾ This along with improving the provision of basic genomic education for health professionals (from undergraduate to postgraduate) and cultural awareness and implications of genetic diversity should be integral to these training modules – this was cited as a recommendation across all our research studies and has been highlighted in previous research ^(57, 290). The need for this is also emphasised by findings that referral rates of ethnic minority groups for genetic testing is lower when compared to White populations,⁽⁵⁴⁾ and evidence that some ethnic minority groups feel health professionals impose their own views and steer individuals' decisions ^(53, 54).

A separate but related point is that diversity among the healthcare workforce which would require diversifying entry routes into training may help break down many of the barriers impacting engagement of ethnic minority groups with genomic medicine services. In line with this, efforts to engage NICE to ensure clinical care guidelines (e.g. pharmacogenetic testing) incorporate and acknowledge the diversity among the population and how it relates to health conditions and disabilities, may also complement training, resulting in healthcare professionals being able to embed training and learning into their practice.



7. CONCLUSION

Genetic databases lack diversity and largely include populations of European ancestry. This has implications for the development of equitable genomics services, diagnostic tools and treatments for individuals from ethnic minority groups. Therefore, research studies need to develop inclusive approaches to recruit participants from under-represented communities. People from ethnic minority groups are uncomfortable with providing samples to be stored in large databases, such as UK Biobank. This reluctance is due to mistrust, discrimination and cultural insensitivity these communities continue to face within the healthcare setting, as well as the historical and lived experiences of unethical research practices.

Understanding and awareness of genomics services and research is limited among ethnic minority communities. The policy review found community engagement initiatives were not well described, though a few key successful projects were noted through stakeholder interviews. To improve awareness of genomics services and understanding of what biomedical research participation entails, policy, healthcare and research stakeholders must prioritise tailored and sustained programmes to engage with different communities. Community engagement is key to building trust between health services, research and ethnic minority communities. As part of this, healthcare providers and researchers need to demonstrate cultural awareness and competency, which requires the development of appropriate training. Also, for community engagement to be truly beneficial it requires better investment. There are also challenges with recording ethnicity data across the healthcare service which has limited the monitoring of access to genomics services and uptake of genomic testing. Stakeholders from the genomics medicine services and the health service more generally, must come together to improve data collection, data sharing and monitoring service access and testing uptake.

8. RECOMMENDATIONS

Recommendations for meaningful community engagement and building trust

1. There is a need for meaningful, sustained, and tailored community engagement activities across the healthcare systems with NHS England to ensure all benefit from new advances and with researchers/research councils to ensure all communities can engage in genomic/precision medicine research. Community engagement activities must:
 - Focus on improving knowledge and awareness of genomics services and research through tailored engagement approaches enabling communities to make informed contributions to the dialogue (including e.g. via the NHS GMS People and Communities Forum, and NHS GMS Alliances). Examples of community engagement activities such as 'Genetics in Communities' and Genomics England's initiative to improve genomics literacy across England provide a blueprint for how to do this at a local level using co-design and co-production (see Chapter 5).
 - Include the development of an engagement space or platform that brings together communities to inform them about genomics and its potential benefits. Further work is needed to tease out the format of this platform and how this could be built and implemented.
 - Inform the development and implementation of inclusive and accessible service provision for ethnic minority groups.
 - Build trust and overcome barriers relating to mistrust (discrimination, fear, suspicion, past trauma, lack of understanding of genomics and precision medicine). To do this, key stakeholders including policymakers, researchers and healthcare service providers must listen to and acknowledge the challenges ethnic minority groups continue to experience collectively as a community and as individuals.
 - Involve reaching out to and including underrepresented ethnic minority groups using tailored communication channels that are appropriate to different communities.
 - Include clearly defined plans for implementation and must include mechanisms for monitoring and evaluation. In tandem with evaluating engagement initiatives, the sustainability and scalability of these approaches need to be assessed.
 - Ensure public engagement is built into the development of future advances of health technologies within the fields of genomics and precision medicine. This will help to prevent further inequity and begin to build equity for ethnic minority groups.

- Are well supported with sustained financial, personnel and time investment.

Recommendations for policy and practice to ensure equitable access

2. All patients and healthcare professionals must have access to interpreters who are qualified and able to communicate complex medical terminology.
3. There is a need to develop a national Equality Diversity and Inclusion Framework with all relevant agencies in the NHS Genomics Medicine Services. The framework should also consider data governance and sharing policies that improve access to patient-level monitoring data for organisations key to implementing the NHS Genomics Medicine Services so that:
 - GMSAs can negotiate local data-sharing agreements with local trusts and Genomics Laboratory Hubs (GLHs) to obtain data on access and uptake of genomics services and testing for different communities.
 - GMSAs can evaluate current inequities in access by monitoring data relating to availability of tests, numbers and proportions of patients referred, ethnicity of patients accessing services and turnaround times for test results.
 - GMSAs and other relevant stakeholders can monitor care outcomes between different ethnic minority groups.
 - GMSAs can develop targeted community engagement strategies to improve knowledge, awareness and accessibility of services and genomic testing.
4. Regular monitoring, evaluation and publication of projects by NHS England Genomics Policy Unit which aim to address inequities in genetic medicine services and testing uptake must be routinely published and publicly accessible.

This should be routinely published, publicly available and shared across the NHS Genomics Medicine Services. Public authorities working in genomics, such as the NHS England Genomics Unit and NHS Genomic Medicine Service Alliances must hold key stakeholders to account through regular monitoring and evaluation of action and implementation plans.

5. NICE clinical care guidelines (e.g. implementation of pharmacogenetic testing) should acknowledge how population diversity relates to testing outcomes, health and disability. Where evidence shows ethnic differences, this should be included in NICE recommendations along with implementation tool to enable healthcare professionals to embed strategies to help facilitate equitable access into practice.
6. Better representation of ethnic minority groups within workforce across the genomics medicine services, precision medicine research and more generally across the healthcare service, including at leadership and decision-making levels. Further work is needed to explore how increasing diversity of the workforce can be achieved, perhaps through diversification of entry routes into medicine and applied healthcare training.

Recommendations for research: diversifying research participation

7. Governments, research bodies and funders should ensure research databases hold genetic information that is representative of our diverse population, with appropriate coding and recording of ethnicities. Work to increase representation of those that take part in research in genetic and precision medicine should be prioritised.
 - To improve understanding of genetic variation (according to ethnicity).
 - To improve subsequent development of genomic medicine services.
 - To ensure GWAS, PRS and other measures of risk are inclusive of different ethnic groups.
 - This should be underpinned by engaging with different ethnic minority communities (see recommendations on community engagement).
 - If ethnic bias is not addressed, ethnic inequities in genomic and precision medicine will be exacerbated. Oversampling of ethnic minority groups is recommended across genomic medicine research.
8. Ethnicity coding needs to be inclusive and consistent between different health services and electronic patient record systems:
 - Ethnicity coding should be developed in consultation with communities to ensure inclusivity and avoid mislabelling or arbitrary grouping.
 - There needs to be joint efforts across the health service to improve data recording practices for protected characteristics such as ethnicity.
9. Lived and historical experiences of unethical research practices need to be addressed and factored in when developing genomics research with ethnic minority groups. To ensure that this is done in a sensitive and meaningful way, researchers must engage with communities and ensure that research practices are sensitive to the needs of participants from different ethnic minority groups.
10. Research culture needs to change and develop more inclusive recruitment methods and research processes (e.g. informed consent, delivery of participant information):
 - In preparation for study recruitment, research teams need a clear plan to engage ethnic minority groups. This should include public engagement through tested communication channels and use of established community engagement models and support networks.
11. When researchers apply to use established databases (e.g. UK Biobank, Genomics England databases) or apply to funding bodies for research grants (e.g. Medical Research Council, Wellcome Trust, National Institute for Health Research), research approval committees should appraise the study proposals on how they will incorporate information on ethnic minority groups in the overall sample.

12. Legislation or official guidance for the UK pertaining to making research procedures and genomics research accessible for ethnic minority groups needs to be enacted. Lessons should be taken from the US where The National Institute for Health Clinical Diversity Act (2022) requires funding applications to provide clear plans for addressing accessibility and inclusion of diverse populations in clinical trials.
13. Genetic ancestry should not be used as a surrogate measure of race and ethnicity in genomic research; however, ancestry does provide insight into genetic predisposition.

Recommendations for workforce training and education

Several training needs for the workforce have been identified that should be considered as part of the national strategy to embed genomics medicine services across the NHS. The training needs include:

14. A drive to improve the genomic education of health professionals from undergraduate to advanced postgraduate training and for healthcare professionals currently working in the health service.
 - For health professionals currently working in the health service training needs to highlight how healthcare professionals may already be interacting with genomics and precision medicine and show the relevance to their practice and their patients.
 - Training for all healthcare staff must cover the implications of genetic diversity and cultural awareness. This training should address potential conscious and unconscious biases held by healthcare workers that may be affecting the quality-of-care patients receive. This will equip the workforce with an understanding of the needs of different groups, how to apply this knowledge to tailor conversations and inform interactions with patients. This will help to ensure that people from ethnic minority groups are receiving equitable care and support.
15. Providing general training around genomics services and precision medicine to non-healthcare workers involved in decision-making around healthcare such as social workers and other professionals (e.g. chaplains) involved in providing support to patients is key to ensure that minority groups can access accurate and reliable information.
16. Training around data collection for ethnicity and protected characteristics also needs to be developed and rolled out across the service. Healthcare workers across all levels who interact with patients as part of their role need to understand the importance of why this data needs to be collected and how to have a conversation with patients in a meaningful way.



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