

ETHNIC INEQUITIES IN GENOMICS AND PRECISION MEDICINE

LAY SUMMARY



OVERVIEW

Our health is influenced by the genes we inherit and the environments we live in. Changes in our DNA can be inherited or be a result of lifestyle or environmental factors, like smoking, sunlight and radiation. Some of these variations can protect against disease, others can increase our risk e.g. for certain cancers. Personalised medicine aims to understand more about how differences in our genes and other factors influence our health, and in doing so, aims to ensure the right individual, gets the right treatment at the right time – recognising that medicine isn't a 'one size' fits all approach.

Advances in technologies and more personalised healthcare are positive, but we know that people from many ethnic groups do not benefit from personalised medicine services in the same way as white people. For example, ethnic minority groups are hugely underrepresented in precision and genomic medicine research, which has negatively impacted health outcomes. The research and healthcare community has not engaged enough with ethnic groups in genomics research and the planning of precision medicine services either. We refer to these differences as ethnic inequalities, which need to be better understood and addressed, to ensure all communities are part of shaping healthcare in this area, and to ensure that these inequalities do not get worse. To do this, there is a need to understand the current conversation around ethnicity in relation to genomics and precision medicine within policy and guidance documents, academic literature and from the point of view of key stakeholders representing individuals from Black, Asian and minority ethnic groups, policymakers, researchers, healthcare professionals and those working within the genomics medicine service.

KEY TERMS

DNA	A chemical code containing instructions for how our body works.
Genes	A unit of DNA that carries instructions for a certain trait.
Genome	A complete set of DNA.
Genetics	Involves studying how certain traits are passed down from one generation to another (e.g. height, eye or hair colour).
Genomics	Studying all a person's genes (their genome) and how they interact with each other and the environment.
Precision medicine	Is interested in how the differences in our genes and other factors such as lifestyle influence our health. It aims to get the right person the right treatment at the right time.

WHAT WE DID

We wanted to understand how precision medicine services, genomic testing and genomics research are accessible to and involve people from different ethnic minority groups. To do this we used a mixed methods approach, by which we mean we collected data and information in a few different ways:

- We reviewed policy and guidance documents to understand the priorities of current approaches to ethnic health inequity in the development and rollout of genomics and precision medicine services.
- We searched for previously published studies in genomics medicine and patients' access to precision medicine services. For these studies, we considered the ethnic diversity among people who took part and how many people from different ethnicities are included in databases that help inform the development of genomic testing and risk prediction tools.
- We ran informal discussion groups and interviews with individuals from different ethnic minority groups to explore their attitudes, knowledge and awareness of genomics and precision medicine services. Through these discussions, we also explored what the barriers were for ethnic minority groups to access genomics services and participate in research and what needs to be done to improve access and participation.
- We also talked with healthcare professionals, researchers, patient and public involvement members, industry partners and community engagement coordinators to explore knowledge and awareness of current practices to address ethnic health inequalities in precision medicine and genomics services and to improve inclusion of ethnic minority groups in research.
- We also ran informal focus groups with representatives from the Genomic Medicine Service Alliances (GMSAs), to explore current challenges to improving equity of access for ethnic minority groups. We also explored current practices around ethnicity data recording. The NHS Genomic Medicine Service in England launched 7 Genetic Medicine Service Alliances or GMSAs. These alliances oversee and coordinate the roll-out of genomics and personalised medicine into mainstream clinical care within the geographical location that they cover.

WHAT WE FOUND

Policy review

We found and reviewed 70 policy and guidance documents. Fifty of these documents included information relating to precision medicine and/or genomics. The documents recognised that there is an underrepresentation of ethnic groups in genomic datasets and that this affects our understanding of genetic variation as well as the development of precision medicine services. These documents mentioned how poor ethnic representation in datasets limits the developments in artificial intelligence-based risk prediction tools which are used to predict a person's chances of diseases/conditions. Not having data from ethnic groups affects how well these tests work in different ethnic groups.

The documents mentioned public engagement efforts with diverse communities to inform genomics initiatives, but this appears to not be happening consistently enough. Also, there were limited details on how public engagement is happening on the ground, though working with individuals from ethnic groups to help co-design and co-produce approaches that would work for these groups, were suggested. We need to listen to and understand the needs of different ethnic minority groups. It is important that we take time to educate communities about genomics and precision medicine and empower them to contribute to how services are developed. To do this well though, financial and time investment is needed.

The documents also mentioned better diversity within the workforce would be helpful in breaking down historical barriers to service access among communities such as mistrust and suspicion. Some of the documents also suggested training to foster cultural awareness and understanding among healthcare staff would be a positive step towards improving ethnic inequities in precision and genomics medicine. Future work should look at the nature and content of such training and involve more public engagement activity with different ethnic minority groups.

Systematic review

A systematic review is an organised search to gather all the information available on a specific topic. In our case we conducted a systematic review to find out whether people from ethnic minority groups were included in genetic research or not. We found 143 studies in total. Most of these describe basic information about participants from ethnic minorities. However, they usually only considered ethnicity as part of the overall analysis, rather than to try to understand whether there are any important genetic differences between people of European heritage and ethnic minority populations.

91.1% of included studies mention ethnicity in their results sections. 78.5% analysed genetic data of ethnic minority individuals. 12.6% only mention basic background

information about ethnic minority participants and do not mention genetic data. Many studies explained that their research was limited by a lack of genetic data being available among ethnic minority participants.

Some ethnic minority participants who took part in the qualitative studies mentioned that ethnic minority groups were worried about their genetic samples being misused by researchers. Some participants also explained that they did not want to participate in genetic research due to historical violations against ethnic minorities. Others explained that they could not see how participation in genetic research would benefit ethnic minority populations.

To make healthcare fairer for all, it is important to address inequalities in access to health care - everyone deserves equal access to healthcare. Individuals from ethnic minority groups should be invited to join genetic research studies so that information gaps can be filled to ensure that everyone gets appropriate personalised care.

Stakeholder interviews

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 GMSAs 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 we found themes relating to knowledge and awareness of genomics and precision medicine, key barriers and facilitators to access and engagement and workforce training needs.

Current knowledge and awareness about the links between genes and our health, terms such as genomics and precision medicine varied across public stakeholders. All stakeholders felt that knowledge and awareness of genomics and precision medicine services among healthcare professionals working in other areas of healthcare was limited.

All stakeholders discussed strategies to improve knowledge and awareness of genomics, and precision medicine across ethnic minority groups. Healthcare professionals were seen as the first point of call for information. Faith or community leaders were thought to be key points of contact to share reliable information within communities. Stakeholders also talked about the need for tailored strategies for different communities and sustained messaging to improve knowledge, awareness and engagement.

Language was a common barrier to access, uptake and engagement with services, testing and research. This includes challenges around accessing translators and the accuracy of the translation. Public stakeholders shared how language barriers have impacted them being able to engage with healthcare services. Other barriers were mistrust, fear and suspicion of healthcare professionals, healthcare systems and research. Stakeholders advised acknowledging and understanding the reasons for mistrust were important 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. Participants felt that a tailored and sustained approach to engagement is likely to be most effective.

A series of workforce training needs were suggested by stakeholders. There is a need to improve genomics education across the healthcare workforce and for other professionals involved in health decision-making. Cultural awareness and competency training for healthcare professionals and researchers not just in genomics and precision but across the healthcare system is also needed.

WHAT NEEDS TO CHANGE?

To make sure that people from different ethnic backgrounds have fair access to genetic testing, precision medicine services and involvement in research, we suggest the following based on our research:

Recommendations for meaningful community engagement and building trust

1. We propose that different groups of people including – the public, patients, healthcare workers, government officials and the authorities should be involved in conversations about genetic medicine services. This is with the aim to help people understand genetic and precision medicine better. More conversation often leads to better engagement and therefore may improve access to genetic services for people from ethnic minorities.

When researchers engage with the public and patients, their ideas and concerns should be considered to make taking part in research more accessible. Involving people from ethnic minority groups in genetic research and services requires financial, resource and personnel investment. Clear plans which suit each community should be made and these should be assessed regularly. These plans need to be sustainable and scalable to improve engagement with ethnic minorities.

2. It is important to take time to reflect about past events to build trust with individuals and communities to break down barriers. These barriers usually arise from previous mistrust, discrimination, fear, and bad experiences. For these barriers to be addressed, decision-makers, researchers and healthcare professionals need to openly listen so that they can be more aware of the challenges experienced by ethnic minority communities.

Recommendations for policy and practice to ensure equitable access

3. All patients and healthcare professionals must have access to interpreters who understand and are able to communicate complex medical language. In our stakeholder interviews, we found that there is a lack of capable translators. Because of this, some individuals from ethnic minorities may not receive the information and care they need.
4. Rules and regulations about how and who can access genetic data need to be made. This is so that relevant organisations like GMSAs can monitor whether access to genetic testing is fair. This will allow for strategies to be developed to improve awareness of genetic services and how to access them among specific communities.

5. Working with the National Institute for Clinical Excellence (NICE) to ensure that healthcare guidelines consider differences between different populations about health and disability. This way, healthcare workers can use these guidelines to provide better and fairer care for everyone.
6. There needs to be regular monitoring and evaluation of projects which aim to address inequities in genetic medicine services. This is to ensure that plans to provide genetic services are fair between different communities. These plans need to be published and publicly available.
7. Further work is needed to see how to increase the diversity of healthcare workers involved in genetic services. This might mean increasing the number of routes towards becoming a doctor or other healthcare professional. This report found that more people from different ethnic backgrounds need to be involved in genetic services. Increasing the number of staff from ethnic backgrounds applies to healthcare, research, and leadership positions.

Recommendations for research: diversifying research participation

8. Databases which hold genetic information need to be more diverse. This will help us make sure that data from ethnic minority groups are included in the making of tools designed to reduce health risks. If data from ethnic minorities are not included, then genomic and precision medicine may become unfair towards ethnic minorities.
9. Coding and classifying ethnicities in genetic databases need to include all ethnicities and be better recorded across the healthcare system. Ethnic minority communities should be involved in developing the coding and classification of ethnicities. Our interviews revealed that some communities were often mislabelled or misrepresented. There are clear differences in terms of language and culture within communities that need to be better understood by healthcare professionals, researchers and decision makers. When we spoke with individuals from GMSAs they explained data on ethnicity is not well collected. Key players in genomics medicine services need to work together to improve the ways in which important data like an individual's ethnicity is recorded.
10. It is important that previous unethical research involving ethnic minorities is recognised. Researchers need to understand how these previous experiences discourage ethnic minorities from getting involved in research. Through understanding and recognition, ethnic minorities may feel more comfortable to take part in genomics research.
11. Researchers' attitudes and behaviours need to change to ensure that research participants are from diverse backgrounds. When inviting participants to join research studies, researchers should have clear plans to involve ethnic minorities. Researchers should use communication methods which are preferred by individuals and communities.

12. Groups which approve research should check how researchers plan to involve ethnic minority communities. This especially applies to research which involves existing information or databases. Official guidelines should be created to support the involvement of ethnic minority communities.

Recommendations for workforce training and education

13. Improve genetic medicine education for healthcare professionals - students and staff. Training needs to show why genetic medicine is important, how it can be a tool to provide good patient care with examples of how genetic medicine is currently being used in the health service.
14. Non-healthcare workers (like social workers and chaplains) who help make decisions about healthcare should receive training on genetic medicine. This is so that they can provide individuals from minority communities with accurate and trustworthy information.
15. Healthcare workers would benefit from 'cultural awareness' training so that they understand their patients better. 'Cultural awareness' means to be aware of and to respect how different people from different cultures think and behave. This would help to provide every patient with fair care.
16. Healthcare workers require training to improve the ways in which information about a person's characteristics is collected - including ethnicity. Healthcare workers need to understand why a person's characteristics is important and how best to have meaningful conversations with patients.



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