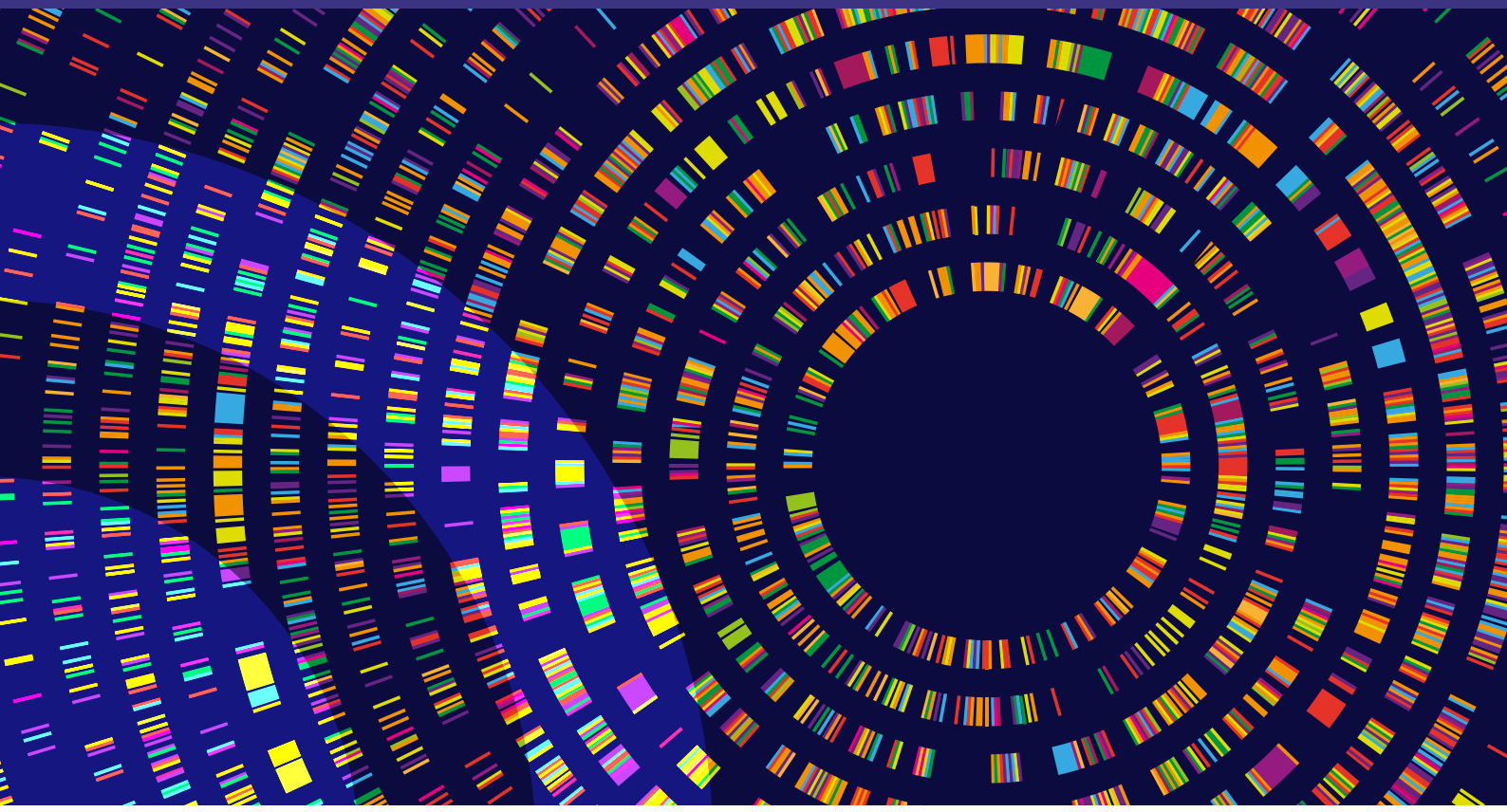
NHS RACE & HEALTH OBSERVATORY

ETHNIC INEQUITIES IN GENOMICS AND PRECISION MEDICINE

APPENDICES



The University of Nottingham

CONTENTS

Appendices

APPENDICES

APPENDIX 1

Search strategy

Ovid MEDLINE(R) ALL <1946 to October 19, 2022>

1. (Ethnic* or racial or race or racis* or “BME” or “BAME” or ancestr* or interethnic or multi-ethnic or multi*rac*).ti,kf,ab,hw. 384326
2. (Arab or Africa* or Afro* or Asian or Bangladesh* or Black or Caribbean or Chinese or India* or Irish or (Mixed adj other) or Pakistan* or Roma or traveller* or Gyps* or Gips* or Sikh* or Hindu* or Muslim* or Islam* or jew* or Hispanic or Latin or Caucasian or European).kf,hw,ti,ab. 1540199
3. (diversity or inclusion or under-represent* or under-served or equity or disparit* or equality or inequality or representation).ti,ab,kf,hw. 865642
4. 1 or 2 or 3 2488529
5. exp precision medicine/ 28186
6. exp genetic testing/ 52746
7. exp genetic services/ 63120
8. exp genetic counseling/ 15388
9. ((genetic or genom* or personal* or precision or pharmacogenetic* or pharmacogenomic* or bioinformatic*) adj3 (medicine or test* or counsel* or service* or intervention* or diagno* or treatment* or therap* or prevent* or screen* or drug* or prescri* or (risk adj3 (prediction or assessment or score)))).ti,ab,kf,hw. 301213
10. ((Target* adj2 screen*) or (tailor* adj3 therapy*) or ((polygen* adj2 risk) or PRS)).ti,ab,kf,hw. 21442
11. exp genomics/ or exp pharmacogenetics/ 157524

12. (pharmacogenetic* or pharmacogenomic* or bioinformatic* or genom* or Biobank or GWAS).ti,ab,kf,hw. 906745

13. 5 or 6 or 7 or 8 or 9 or 10 or 11 or 12 1231666

14. 4 and 13 172306

15. limit 14 to (editorial or letter or comment or newspaper article) 1864

16. 14 not 15170442

17. exp United Kingdom/ or exp Great Britain/ 386766

18. (gb or "g.b." or britain* or uk or "u.k." or "united kingdom*" or england* or "northern ireland*" or "northern irish*" or scotland* or scottish* or wales or "south wales" or welsh* or "national health service" or nhs).ti,ab,kf,hw. 553686

19. (london or "east midlands" or "west midlands" or yorkshire or "east anglia" or bedfordshire or hertfordshire or essex or peterborough or cambridgeshire or norfolk or suffolk or luton or bedford or "southend on sea" or thurrock or derbyshire or nottinghamshire or leicestershire or rutland or lincolnshire or derby or leicester or northamptonshire or nottingham or "tyne and wear" or "tees valley" or durham or darlington or hartlepool or "stockton on tees" or northumberland or teesside or sunderland or cumbria or cheshire or manchester or lancashire or merseyside or blackburn or darwen or blackpool or chester or liverpool or sefton or warrington or wirral or berkshire or buckinghamshire or oxfordshire or hampshire or "isle of wight" or kent or surrey or sussex or brighton or hove or "milton keynes" or portsmouth or southampton or devon or dorset or somerset or gloucestershire or wiltshire or bath or bournemouth or poole or bristol or plymouth or swindon or torbay or herefordshire or staffordshire or birmingham or coventry or dudley or sandwell or shropshire or solihull or "stoke on trent" or telford or wrekin or walsall or warwickshire or wolverhampton or worcestershire or barnsley or doncaster or rotherham or bradford or calderdale or kirklees or kingston or leeds or sheffield or wakefield or (york not new york) or antrim or ards or armagh or ballymena or ballymoney or banbridge or carrickfergus or castlereagh or coleraine or cookstown or craigavon or (down and (district or council)) or dungannon or fermanagh or larne or limavady or lisburn or magherafelt or moyle or "newry and mourne" or newtownabbey or omagh or strabane or londonderry or tyrone or belfast or aberdeen or aberdeenshire or angus or dundee or argyll or bute or clackmannanshire or fife or ayrshire or dunbartonshire or lothian or renfrewshire or edinburgh or falkirk or glasgow or highland* or inverclyde or midlothian or moray or lanarkshire or kinross* or stirling or "orkney islands" or "eileanan siaror shetland islands" or bridgend or "neath port talbot" or cardiff or "vale and glamorgan" or "central valleys" or conwy or denbighshire or flintshire or wrexham or "gwent valleys" or gwynedd or "isle and anglesey" or monmouthshire or newport or powys or swansea or ceredigion or carmarthenshire or pembrokeshire or "merthyr tydfil" or "rhondda cynon taff" or "blaenau gwent" or caerphilly or torfaen or caithness or

sutherland or cromarty or teeside or tyneside or wearside or “west mercia” or avon or ulster or derry or medway or “east riding” or “west riding” or “lake district” or “peak district” or cumberland or dartmoor or exmoor).mp. 237809

20. 17 or 18 or 19 737180

21. exp South Australia/ or exp Australia/ or exp Western Australia/ 165636

22. (“New South Wales” or Victoria or Queensland or “Western Australia” or “South Australia” or Tasmania or “Northern Territory” or “Jervis Bay Territory” or “Christmas Island” or “Australian Capital Territory” or “Norfolk Island” or “Cocos Islands”). ti,ab,kf,hw. 71524

23. 21 or 22 181163

24. 16 and (20 or 22) 5874

25. limit 24 to yr=”2021 - Current” 1674

Embase <1974 to 2022 October 19>

1. (Ethnic* or racial or race or racis* or “BME” or “BAME” or ancestr* or interethnic or multi-ethnic or multi*rac*).ti,kf,ab,hw. 593185

2. (Arab or Africa* or Afro* or Asian or Bangladesh* or Black or Caribbean or Chinese or India* or Irish or (Mixed adj other) or Pakistan* or Roma or traveller* or Gyps* or Gips* or Sikh* or Hindu* or Muslim* or Islam* or jew* or Hispanic or Latin or Caucasian or European).kf,hw,ti,ab. 2251961

3. (diversity or inclusion or under-represent* or under-served or equity or disparit* or equality or inequality or representation).ti,ab,kf,hw. 1108693

4. 1 or 2 or 3 3479765

5. exp precision medicine/ 62909

6. exp genetic testing/ 111974

7. exp genetic services/ 139899

8. exp genetic counseling/ 37229

9. ((genetic or genom* or personal* or precision or pharmacogenetic* or pharmacogenomic* or bioinformatic*) adj3 (medicine or test* or counsel* or service* or intervention* or diagno* or treatment* or therap* or prevent* or screen*

or drug* or prescri* or (risk adj3 (prediction or assessment or score)))).ti,ab,kf,hw. 335580

10. ((Target* adj2 screen*) or (tailor* adj3 therapy*) or ((polygen* adj2 risk) or PRS)).ti,ab,kf,hw. 33375

11. exp genomics/ or exp pharmacogenetics/ 152456

12. (pharmacogenetic* or pharmacogenomic* or bioinformatic* or genom* or Biobank or GWAS).ti,ab,kf,hw. 1204005

13. 5 or 6 or 7 or 8 or 9 or 10 or 11 or 12 1537343

14. 4 and 13 254748

15. limit 14 to (editorial or letter or comment or newspaper article) 3557

16. 14 not 15251191

17. exp United Kingdom/ or exp Great Britain/ 451098

18. (gb or "g.b." or britain* or uk or "u.k." or "united kingdom*" or england* or "northern ireland*" or "northern irish*" or scotland* or scottish* or wales or "south wales" or welsh* or "national health service" or nhs).ti,ab,kf,hw. 742835

19. (london or "east midlands" or "west midlands" or yorkshire or "east anglia" or bedfordshire or hertfordshire or essex or peterborough or cambridgeshire or norfolk or suffolk or luton or bedford or "southend on sea" or thurrock or derbyshire or nottinghamshire or leicestershire or rutland or lincolnshire or derby or leicester or northamptonshire or nottingham or "tyne and wear" or "tees valley" or durham or darlington or hartlepool or "stockton on tees" or northumberland or teesside or sunderland or cumbria or cheshire or manchester or lancashire or merseyside or blackburn or darwen or blackpool or chester or liverpool or sefton or warrington or wirral or berkshire or buckinghamshire or oxfordshire or hampshire or "isle of wight" or kent or surrey or sussex or brighton or hove or "milton keynes" or portsmouth or southampton or devon or dorset or somerset or gloucestershire or wiltshire or bath or bournemouth or poole or bristol or plymouth or swindon or torbay or herefordshire or staffordshire or birmingham or coventry or dudley or sandwell or shropshire or solihull or "stoke on trent" or telford or wrekin or walsall or warwickshire or wolverhampton or worcestershire or barnsley or doncaster or rotherham or bradford or calderdale or kirklees or kingston or leeds or sheffield or wakefield or (york not new york) or antrim or ards or armagh or ballymena or ballymoney or banbridge or carrickfergus or castlereagh or coleraine or cookstown or craigavon or (down and (district or council)) or dungannon or fermanagh or larne or limavady or lisburn or magherafelt or moyle or "newry and mourne" or newtownabbey or omagh or strabane or londonderry or tyrone or belfast or aberdeen or aberdeenshire or angus or dundee or argyll or bute or

clackmannanshire or fife or ayrshire or dunbartonshire or lothian or renfrewshire or edinburgh or falkirk or glasgow or highland* or inverclyde or midlothian or moray or lanarkshire or kinross* or stirling or “orkney islands” or “eileanan siaror shetland islands” or bridgend or “neath port talbot” or cardiff or “vale and glamorgan” or “central valleys” or conwy or denbighshire or flintshire or wrexham or “gwent valleys” or gwynedd or “isle and anglesey” or monmouthshire or newport or powys or swansea or ceredigion or carmarthenshire or pembrokeshire or “merthyr tydfil” or “rhondda cynon taff” or “blaenau gwent” or caerphilly or torfaen or caithness or sutherland or cromarty or teeside or tyneside or wearside or “west mercia” or avon or ulster or derry or medway or “east riding” or “west riding” or “lake district” or “peak district” or cumberland or dartmoor or exmoor).mp. 387497

20. 17 or 18 or 19 1040894

21. exp South Australia/ or exp Australia/ or exp Western Australia/ 188195

22. (“New South Wales” or Victoria or Queensland or “Western Australia” or “South Australia” or Tasmania or “Northern Territory” or “Jervis Bay Territory” or “Christmas Island” or “Australian Capital Territory” or “Norfolk Island” or “Cocos Islands”). ti,ab,kf,hw. 61939

23. 21 or 22 206180

24. 16 and (20 or 22) 10474

25. limit 24 to yr=”2021 - Current” 2860

APA PsycInfo <1806 to October Week 3 2022>

1. (Ethnic* or racial or race or racis* or “BME” or “BAME” or ancestr* or interethnic or multi-ethnic or multi*rac*).ti,ab,sh. 184965

2. (Arab or Africa* or Afro* or Asian or Bangladesh* or Black or Caribbean or Chinese or India* or Irish or (Mixed adj other) or Pakistan* or Roma or traveller* or Gyps* or Gips* or Sikh* or Hindu* or Muslim* or Islam* or jew* or Hispanic or Latin or Caucasian or European).ti,ab,sh. 351390

3. (diversity or inclusion or under-represent* or under-served or equity or disparit* or equality or inequality or representation).ti,ab,sh. 251657

4. 1 or 2 or 3 658070

5. exp precision medicine/ 573

6. exp genetic testing/ 2045

7. exp genetic counseling/ 2116

8. ((genetic or genom* or personal* or precision or pharmacogenetic* or pharmacogenomic* or bioinformatic*) adj3 (medicine or test* or counsel* or service* or intervention* or diagno* or treatment* or therap* or prevent* or screen* or drug* or prescri* or (risk adj3 (prediction or assessment or score)))).ti,ab,sh. 59038

9. ((Target* adj2 screen*) or (tailor* adj3 therapy*) or ((polygen* adj2 risk) or PRS)).ti,ab,sh. 2920

10. exp genomics/ or exp pharmacogenetics/ 6759

11. (pharmacogenetic* or pharmacogenomic* or bioinformatic* or genom* or Biobank or GWAS).ti,ab,sh. 21014

12. 5 or 6 or 7 or 8 or 9 or 10 or 11 80217

13. 4 and 12 9402

14. ("United Kingdom" or "Great Britain").ti,ab,sh. 14538

15. (gb or "g.b." or britain* or uk or "u.k." or "united kingdom*" or england* or "northern ireland*" or "northern irish*" or scotland* or scottish* or wales or "south wales" or welsh* or "national health service" or nhs).ti,ab,sh. 92202

16. (london or "east midlands" or "west midlands" or yorkshire or "east anglia" or bedfordshire or hertfordshire or essex or peterborough or cambridgeshire or norfolk or suffolk or luton or bedford or "southend on sea" or thurrock or derbyshire or nottinghamshire or leicestershire or rutland or lincolnshire or derby or leicester or northamptonshire or nottingham or "tyne and wear" or "tees valley" or durham or darlington or hartlepool or "stockton on tees" or northumberland or teesside or sunderland or cumbria or cheshire or manchester or lancashire or merseyside or blackburn or darwen or blackpool or chester or liverpool or sefton or warrington or wirral or berkshire or buckinghamshire or oxfordshire or hampshire or "isle of wight" or kent or surrey or sussex or brighton or hove or "milton keynes" or portsmouth or southampton or devon or dorset or somerset or gloucestershire or wiltshire or bath or bournemouth or poole or bristol or plymouth or swindon or torbay or herefordshire or staffordshire or birmingham or coventry or dudley or sandwell or shropshire or solihull or "stoke on trent" or telford or wrekin or walsall or warwickshire or wolverhampton or worcestershire or barnsley or doncaster or rotherham or bradford or calderdale or kirklees or kingston or leeds or sheffield or wakefield or (york not new york) or antrim or ards or armagh or ballymena or ballymoney or banbridge or carrickfergus or castlereagh or coleraine or cookstown or craigavon or (down and (district or council)) or dungannon or fermanagh or larne or limavady or lisburn or magherafelt or moyle or "newry and

mourne" or newtownabbey or omagh or strabane or londonderry or tyrone or belfast or aberdeen or aberdeenshire or angus or dundee or argyll or bute or clackmannanshire or fife or ayrshire or dunbartonshire or lothian or renfrewshire or edinburgh or falkirk or glasgow or highland* or inverclyde or midlothian or moray or lanarkshire or kinross* or stirling or "orkney islands" or "eileanan siaror shetland islands" or bridgend or "neath port talbot" or cardiff or "vale and glamorgan" or "central valleys" or conwy or denbighshire or flintshire or wrexham or "gwent valleys" or gwynedd or "isle and anglesey" or monmouthshire or newport or powys or swansea or ceredigion or carmarthenshire or pembrokeshire or "merthyr tydfil" or "rhondda cynon taff" or "blaenau gwent" or caerphilly or torfaen or caithness or sutherland or cromarty or teeside or tyneside or wearside or "west mercia" or avon or ulster or derry or medway or "east riding" or "west riding" or "lake district" or "peak district" or cumberland or dartmoor or exmoor).ti,ab,sh. 40865

17. 14 or 15 or 16 123352

18. "Australia".ti,ab,sh. 32698

19. ("New South Wales" or Victoria or Queensland or "Western Australia" or "South Australia" or Tasmania or "Northern Territory" or "Jervis Bay Territory" or "Christmas Island" or "Australian Capital Territory" or "Norfolk Island" or "Cocos Islands"). ti,ab,sh. 11271

20. 18 or 19 36364

21. 13 and (17 or 20) 455

22. limit 21 to yr="2021 - Current" 79

CINAHL (EBSCOhost) 20 October 2022

Interface
- EBSCOhost Research Databases

Search Screen
- Advanced Search

Database
- CINAHL Plus with Full Text

#	Query	Limiters/ Expanders	Results
S7	S1 AND S2 AND S5	Limiters - Published Date: 20210101-20231231 Expanders - Apply equivalent subjects Search modes - Boolean/ Phrase	6,371
S6	S1 AND S2 AND S5	Expanders - Apply equivalent subjects Search modes - Boolean/ Phrase	29,984
S5	S3 OR S4	Expanders - Apply equivalent subjects Search modes - Boolean/ Phrase	2,987,564
S4	MW (Australia) OR TX ("New South Wales" or Victoria or Queensland or "Western Australia" or "South Australia" or Tasmania or "Northern Territory" or "Jervis Bay Territory" or "Christmas Island" or "Australian Capital Territory" or "Norfolk Island" or "Cocos Islands")	Expanders - Apply equivalent subjects Search modes - Boolean/ Phrase	297,571

#	Query	Limiters/ Expanders	Results
S3	MW (United Kingdom or UK or Great Britain or Britain or Scotland or England or Wales or Northern Ireland) OR TX (gb or "g.b." or britain* or uk or "u.k." or "united kingdom*" or england* or "northern ireland*" or "northern irish*" or scotland* or scottish* or wales or "south wales" or welsh* or "national health service" or nhs) OR TX (london or "east midlands" or "west midlands" or yorkshire or "east anglia" or bedfordshire or hertfordshire or essex or peterborough or cambridgeshire or norfolk or suffolk or luton or bedford or "southend on sea" or thurrock or derbyshire or nottinghamshire or leicestershire or rutland or lincolnshire or derby or leicester or northamptonshire or nottingham or "tyne and wear" or "tees valley" or durham or darlington or hartlepool or "stockton on tees" or northumberland or teesside or sunderland or cumbria or cheshire or manchester or lancashire or merseyside or blackburn or darwen or blackpool or chester or liverpool or sefton or warrington or wirral or berkshire or buckinghamshire or oxfordshire or hampshire or "isle of wight" or kent or surrey or sussex or brighton or hove or "milton keynes" or portsmouth or southampton or devon or dorset or somerset or gloucestershire or wiltshire or bath or bournemouth or poole or bristol or plymouth or swindon or torbay or herefordshire or staffordshire or birmingham or coventry or dudley or sandwell or shropshire or solihull or "stoke on trent" or telford or wrekin or walsall or warwickshire or wolverhampton or worcestershire or barnsley or doncaster or rotherham or bradford or calderdale or kirklees or kingston or leeds or sheffield or wakefield or (york not new york) or antrim or ards or armagh or ballymena or ballymoney or banbridge or carrickfergus or castlereagh or coleraine or cookstown or craigavon or (down and (district or council)) or dungannon or fermanagh or larne or limavady or lisburn or magherafelt or moyle or "newry and mourne" or newtownabbey or omagh or strabane or londonderry or tyrone or belfast or aberdeen or aberdeenshire or angus or dundee or argyll or bute or clackmannanshire or fife or ayrshire or dunbartonshire or lothian or renfrewshire or edinburgh or falkirk or glasgow or highland* or inverclyde or midlothian or moray or lanarkshire or kinross* or stirling or "orkney islands" or "eileanan siaror shetland islands" or bridgend or "neath port talbot" or cardiff or "vale and glamorgan" or "central valleys" or conwy or denbighshire or flintshire or wrexham or "gwent valleys" or gwynedd or "isle and anglesey" or monmouthshire or newport or powys or swansea or ceredigion or carmarthenshire or pembrokeshire or "merthyr tydfil" or "rhondda cynon taff" or "blaenau gwent" or caerphilly or torfaen or caithness or sutherland or cromarty or teeside or tyneside or wearside or "west mercia" or avon or ulster or derry or medway or "east riding" or "west riding" or "lake district" or "peak district" or cumberland or dartmoor or exmoor)	Expanders - Apply equivalent subjects Search modes - Boolean/ Phrase	2,857,891

APPENDIX 2

Policy documents summary

Document title	Year published	Link	Source	Pages	Document type	Purpose	Includes information on health inequalities, equity, diversity or inclusion in healthcare or genomics?	Brief summary
Levelling up Health	2021	https://appg-longevity.org/levelling-up-health	All Party Parliamentary Group for Longevity	24	Report	This report highlights the impact of COVID-19 and health inequalities in the UK, discussing ways to level up health, at national and local levels.	Yes	Discusses widening health inequalities for those from more deprived areas and those from ethnic minority communities.
Fairness in precision medicine (name of project conducted) US	2018	https://datasociety.net/library/fairness-in-precision-medicine/	Data & Society (US)	54	Report	This report explores how bias, in datasets and outcomes, impacts precision medicine in the sphere of biomedical research.	Yes	Discusses the need for policy and regulation to prevent data insights being used surveil marginalised communities. Also highlights sources of bias in precision medicine through lack of diversity in datasets. Found current precision medicine research projects acknowledge the need to improve participant diversity. Qualitative study with a range of stakeholders highlights issues of bias in electronic health records, concerns around representation of gender, socioeconomic status, racial and ethnic backgrounds, researchers need to consider diversity in terms of ancestory and geography, importanace of engagement and represenation in research staff.

Document title	Year published	Link	Source	Pages	Document type	Purpose	Includes information on health inequalities, equity, diversity or inclusion in healthcare or genomics?	Brief summary
Women's Health Strategy for England	2022	https://www.gov.uk/government/publications/womens-health-strategy-for-england	Department of Health and Social Care	180	Policy paper	Provides details of the government's 10 year plan to improve health and wellbeing of women and girls in England.	Yes	Highlights racial disparity in mother and baby mortality. Discusses the need for improved data collection across the NHS and government around ethnicity. Discusses the use of screening and research to better understand genetic causes of cancer in ethnic minority women to develop better diagnostic tests and treatments.
England Rare Diseases Action Plan 2022	2022	https://www.gov.uk/government/publications/england-rare-diseases-action-plan-2022	Department of Health and Social Care	91	Policy paper	This document outlines Englands Rare Disease Action Plan, which has been developed to address the key priorities outlined in The UK Rare Diseases Framework. It highlights key action for the year 2021-2022 and outlines how progress will be monitored and evaluated.	Yes	Dicusses the community engagement work conducted as part of developing the action plan. Round table discussions and target online questionaire with key stakeholders to seek input on the action plan. Also partnered with Breaking Down Barriers organisation to run a workshop with various stakeholders to better understand health inequalities experiences by people from diverse and marginalised communities affected by rare dieases.
The UK Rare Diseases Framework	2021	https://www.gov.uk/government/publications/uk-rare-diseases-framework	Department of Health and Social Care	38	Policy paper	This document outlines key priorties and underpinning themes of the UK rare diseases framework.	Yes	Discusses the value of the knowledge and live experiences of people living with rare diseases, including represenation of ethnic minority communities for policy makers and services providers when designing relevant services. Propose that commitments will be developed in consultation with patient representatives, ensuring to include those from ethnic minority or disadvantaged background. Discusses issues with representation of the rare disease community in the national conversation on rare diseases survey in which a majority of the respondents were white females.

Document title	Year published	Link	Source	Pages	Document type	Purpose	Includes information on health inequalities, equity, diversity or inclusion in healthcare or genomics?	Brief summary
Data saves lives: reshaping health and social care with data	2022	https://www.gov.uk/government/publications/data-saves-lives-reshaping-health-and-social-care-with-data/data-saves-lives-reshaping-health-and-social-care-with-data	Department of Health and Social Care	n/a	Webpage/ Policy paper	This webpage details the strategy for using data to bring benefits to all parts of health and social care – from patients and care users to staff on the frontline and pioneers driving the most cutting-edge research.	Yes	Presents background around current health and care disparities in the UK exposed by the pandemic and the need to better understand where these disparties in health outcomes and experiences exist by group and how best to tackle them. Highlights the importance of potential of health service data to identify where these inequalties exist by population group (e.g. age, gender, ethnicity). Acknowledges the current challenges with varability in quality and quantity of available health service data. Highlights potential of health data held by the NHS to provde rich data insights, development of data drive models for early diagnosis and intervention and contributing to reducing health inequalities. Presents example of biobank study with a diverse participant pool enabling researchers to explore ethnic differenes in dementia risk. Discusses AI iniatives and research projects aiming to address health inequalties and improve health and care outcomes for minority ethnic groups.
10 year cancer plan - call for evidence	2022	https://www.gov.uk/government/consultations/10-year-cancer-plan-call-for-evidence/10-year-cancer-plan-call-for-evidence	Department of Health and Social Care		Closed consultation	This call for evidence will inform the development of the government's 10-Year Cancer Plan for England. Rather than a formal consultation on specific proposals, it constitutes a request for ideas and evidence on which we can build.	Yes	Discusses strategies to ensure recovery from COVID-1 9 is equitable such as, health inequalities analysis to target the national 'Help Us Help You' cancer awareness campaigns which is designed to identify people who may have cancer but did not present to health services due to the pandemic (e.g. older people, people from more deprived groups and ethnic minority audiences).

Document title	Year published	Link	Source	Pages	Document type	Purpose	Includes information on health inequalities, equity, diversity or inclusion in healthcare or genomics?	Brief summary
Genome UK: 2022 to 2025 implementation plan for England	2022	https://www.gov.uk/government/publications/genome-uk-2022-to-2025-implementation-plan-for-england/genome-uk-2022-to-2025-implementation-plan-for-england#cross-cutting-themes	Department of Health and Social Care	n/a	Policy Paper	Implementation play setting out actions for genomics delivery partners across England between2022 and 2025	Yes	Discusses commitments led by Genomics England to improve diversity of genomics data and address underpresentation of ethnic minority groups in these datasets and the resulting health inequalities. Discusses current research initiatives in England aimed at improving diversity and inclusion of under-represented groups in genomics reference datasets citing Genomics England's Diverse Data project and the Our Future Health Programme. Our Future Health Programme aims to recruit a cohort that is representative of the UK, so will be using the census 2021 data to inform recruitment by age, ethnicity nad socioeconomic status.

Document title	Year published	Link	Source	Pages	Document type	Purpose	Includes information on health inequalities, equity, diversity or inclusion in healthcare or genomics?	Brief summary
Newborn Genomes Programme	2021	https://www.genomicsengland.co.uk/initiatives/newborns	Genomics England	10	Report	This document outlines the joint NHS England and NHS Improvement (NHSE/I) and Genomics England vision for the Newborn Genomes Programme, developed following public and expert dialogues.The Newborn Genomes Programme will co-design and run a research pilot embedded in the NHS to explore the benefits, challenges, and practicalities of offering whole genome sequencing (WGS) to all newborns to accelerate diagnosis and access to treatments for rare genetic conditions.	Yes	Discusses engagement work with diverse groups conducted to date and highlights future plans to co-design pilot study.
Cancer 2.0 our initiatives section	Not available	https://www.genomicsengland.co.uk/initiatives/cancer	Genomics England	n/a	Webpage	Public facing information about long-read sequencing and multimodal data to improve cancer diagnosis.	No	
Cancer Genomics	Not available	https://www.genomicsengland.co.uk/genomic-medicine/understanding-genomics/cancer-genomics	Genomics England	n/a	Webpage	Public facing information describing cancer genomics and its application to deliver better patient care.	No	

Document title	Year published	Link	Source	Pages	Document type	Purpose	Includes information on health inequalities, equity, diversity or inclusion in healthcare or genomics?	Brief summary
Genomics England Covid-19 study	Not available	https://www.genomicsengland.co.uk/initiatives/covid-19	Genomics England	n/a	Webpage	Webpage provides and overview about the GenOMICC COVID-19 study, which involved analysis of whole genome sequences for 20,000 people who have been severely affected by COVID-19 and compared to 15,000 other genomes from people mildly affected by COVID-19	Yes	Explores why COVID-19 affects ages groups, genders and ethnic groups differently. Dicusses the important of including more volunteers from minority backgrounds as it is well established that minority groups were disproportionately affected by the coronavirus.
Genomics England Newborn Genomes Programme - our initiatives section	Not available	https://www.genomicsengland.co.uk/initiatives/newborns	Genomics England	n/a	Webpage	Webpage provides overview of the Newborn Genomes Programme which involves co-designing and running a study to explore benefits, challenges and practicalities of sequencing and analysing the genomes of newborns.	Yes	As part of the programme commissioned an in-depth national dialogue in 2021 involving a diverse group of 130 members of the public, including individuals with genetic conditions, new/expectant parents, people from BAME backgrounds and young adults. This programme will also look at how the study's design and delivery can be carried out equitably and fairly, to facilitate participation from a diverse range of communities.
Genomic England - Genomic Medicine Section	Not available	https://www.genomicsengland.co.uk/genomic-medicine	Genomics England	n/a	Webpage	Webpage provides an overview about genomic medicine and provides links to further information around the history of genomics, genomic sequencing, genomics in health, the NHS genomics service and the UK genomics industry.	No	

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Diverse Data Vision - Webpage info only	Not available	https://www.genomicsengland.co.uk/initiatives/diverse-data	Genomics England	n/a	Webpage	Webpage provides an overview of the Diverse data vision, presenting figures around issues with lack of diversity in genomics data, linking to further information around research, engagement and data sequencing.	Yes	Dicusses vision for patients to receive same quality of genomics based personalised medicine regardless of background. Highlights key statistics around representation in genetic studies, discusses statistics around accuracy of polgygenic risk scores based on ancestry. Presents strategy for diverse data initiative, highlighting the need for more diversity data, the current challenges to improving diversity in genomics research, plans for community engagement and sequencing of genomic data. Intended outcomes of this intiative are to earn and sustain trusts of diverse communities in genomics and to improve research, prognosis, diagnsos, treatment and care in genomics for diverse populations.
100,000 Genomes Project (including sections on rare disease and cancer related to the project)	Not available	https://www.genomicsengland.co.uk/initiatives/100000-genomes-project	Genomics England	n/a	Webpage	Webpage provides an overview of the 100,000 genomes project, with links to results, additional findings and participant resources.	Yes	Dicusses prevelence of cystic fibrosis based on ancestry.

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Diverse Data Strategy	2022	https://www.genomicsengland.co.uk/initiatives/diverse-data-#:~:text=The%20Diverse%20Data%20vision,and%20input%20into%20our%20design.	Genomics England	22	Strategy	This strategy aims to lay out the Diverse Data Initiative's focus areas for 2022-25 and how we will go about work on them.	Yes	Reseach priorities include address ethnic and ancestrol inequalities in cancer. Populations of interest include genetically diverse populations (people with ancestry from South Africa, Nigeria, Zimbabwe, Ghana, Kenya, Somalia, Uganda, Carribean), populations with high degressof consaguinity (people with ancetry from Pakistan, Arab nations, Traveller communities) Multiethnic cohorts (people with ancestry from India, Bangladesh, Philippines, Sri Lanka, Turkey, Iran, Iraq,Malaysia, Afghanistan, Brazil, Nepal).
Genome UK: Shared commitments for UK-wide implementation 2022 to 2025	2022	https://www.gov.uk/government/publications/genome-uk-shared-commitments-for-uk-wide-implementation-2022-to-2025/genome-uk-shared-commitments-for-uk-wide-implementation-2022-to-2025	Health and Social Care	n/a	Webpage/ policy paper	Building on the Genome UK- the future of healthcare stratergy, this webpage sets out shared commitments for UK wide implementation between 2022 and 2025.	No	

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Genome UK the future of healthcare	2020	https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare	HM Government	64	Policy paper	Sets out the UK strategy to maintain and extend the UK's leadership in genomic healthcare and research.	Yes	Presents research priority 'ensuring diverity and equity of access' in genomics research. Outlines the plans to achieve this through improving diversity in genomic data sets. Also sets out engagement plans with patients, public, workforce and diverse populations. Dicusses problems with under-representation of people from ethnic minority communities and non-european ancestry, the need to understand barriers to inclusion in genomic medicine and recruiting larger samples from minority communities compared than their overall population size (oversampling). The 10 year plan is to increase diversity in reference genomes and ensure future genomes studys reflect diverstiy within the UK.
Life Sciences Vision	2021	https://www.gov.uk/government/publications/life-sciences-vision	HM Government	63	Policy paper	This document sets out the UK's visions for applying the vaccine taskforce approach used to tackle the COVID-19 pandemic to search for life-changing breakthroughs against other diseases.	Yes	The life sciences vision includes more diverse clinical research that reflects the diversity of the UK population. Work with partners such as medical research charities to improve racial, age, gender, and geogrpahic diversity of clinical trials participants and real world data sets. Discusses the development of processes and guidance to increase uptake among traditionally underserved communities, including ethnic minority groups, women, children, the elderly and those in rural or small-town settings. Highlights the importance of data partnerships that can drive improvements in health outcomes and/or reduce health inequalities through clinical research and validating AI using the UK's uniquely diverse population.

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Genome UK: 2021 to 2022 implementation plan	2021	https://www.gov.uk/government/publications/genome-uk-2021-to-2022-implementation-plan	HM Government	n/a	Webpage/ Policy paper	This document sets out the UK's vision to create the most advanced genomic healthcare system in the world to deliver better healthcare at a lower cost. Additionally, this implementation plan details they key priority actions for 2021 to 2022.	Yes	The implementation plan includes a major drive 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. Also includes plans for widespread community engagement alongside sequencing and analytic tool development. This plan presents the UK's largest ever health research programme, Our Future Health, which will recruit up to 5 million diverse participants. This research programme will pilot participant recruitment processes and different recruitment routes achieve the cohort goals of scale and diversity. Also highlights the potential benefits of diverse genomic data for earlier interventions, better diagnostics, and innovative therapies for patients from all backgrounds from across the UK.
The report of the Commission on Race and Ethnic Disparities	2021	https://www.gov.uk/government/publications/the-report-of-the-commission-on-race-and-ethnic-disparities	HM Government	258	Report	Commissioned report on Race and Ethnic disparities in education, employment, crime, policing and health across the UK. The report makes 24 recommendations grouped into 4 themes; build trust, promote fairness, create agency and achieve inclusivity.	Yes	Identifies a need for more Black and Asian people to take part in medical research so data is represenative of the whole population. Highlights problems with the way public authorities collect and analyse data, reflect on the Census data as an example with limited ethnicity groups. Provides recommendations for research around health disparities in ethnici minority groups, looking at genetic and biological differences as well as cultural and social drivers.

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CORE20 PLUS5 (adults)	Not available	https://www.england.nhs.uk/about/equality/equality-hub/national-healthcare-inequalities-improvement-programme/core20plus5/	NHS England	n/a	Webpage	Core20PLUS5 is a national NHS England approach to inform action to reduce healthcare inequalities atboth national and system level. The approach defines a target population – the 'Core20PLUS' – and identifies '5' focus clinical areas requiring accelerated improvement.	Yes	Discusses NHS Englands approach to tackling inequalities across 5 key areas, namely maternity, severe mental illness, chronic respiratory disease, early cancer diagnosis and hypertension management for ethnic minority communities, people with learning disabilities, long term health conditions and other groups that share protected characteristics.
Improving outcomes through personalised medicine	2017	https://www.england.nhs.uk/publication/improving-outcomes-through-personalised-medicine/	NHS England	18	Guidance	The document outlines a vision for personalised medicine in the NHS.	Yes	Briefly mentions developing our understanding of how age, gender and ethnicity influence onset of diease.
Accelerating genomic medicine in the NHS	2022	https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/	NHS England		Strategy	This is an NHS strategy document detailing the genomic medicine service's approach to embedding genomics across the NHS over the next 5 years.	Yes	States over the next 1 to 3 years the NHS genomic medicine service will focus on improving equity of access to testing by exploring option to refer to testing through community and primary care with a focus on unmet need and undiagnosed populations. NHS GMSA's will have an explicit role in address inequalities in the regions they serve. Monitoring and evaluation data relating to testing access, turnaround times, inequalities will be used to drive service improvements and where possible made public. This strategy also refers how research initiatives such as the diverse data intiative being led by Genomics England will be used to inform future commissioning of services.

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NHS Long Term Plan	2019	https://www.longtermplan.nhs.uk/	NHS England	136	Strategy document	This plan states how to accelerate the redesign of patient care to future-proof the NHS for the decade ahead	Yes	Highlights differing risks and inequalities in various health conditions (obesity, diabetes, maternal and neonatal care, bowel cancer, mortality) in relation to ethnicity and socioeconomic status. Presents strategies on how to address health inequalities.
Technical support for the strategic system plan submission process	2019	https://www.longtermplan.nhs.uk/publication/technical-support-for-the-strategic-system-plan-submission-process/	NHS England and NHS Improvement	32	Technical guidance	The purpose of this document is to support systems to develop robust and high-quality five-year strategic plans.	No	
Science in Healthcare: Delivering the NHS Long Term Plan	2020	https://www.england.nhs.uk/publication/chief-scientific-officers-strategy/	NHS England and NHS Improvement	67	Strategy document	Outlines how healthcare science can help to deliver the NHS long term plan objectives.	No	
NHS Long Term Plan Implementation Framework: support offer	2019	https://www.longtermplan.nhs.uk/publication/implementation-framework-support-offer/	NHS England and NHS Improvement	31	Imple-mentation framework	signposts the national and regional support systems can draw ont o develop their five-year strategic plans.	Yes	Discusses ethnicity in the context of maternity and neonatal services and the implementation of direct local support, tools and resources, targeted and enhanced continuity of carer specifically for women whose ethnic background is Black or Asian and those living in the most deprived areas.

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Age-related macular degeneration and genomics - talks about the importance in ethnicity in risks for this disease	2021	https://www.phgfoundation.org/briefing/age-related-macular-degeneration-and-genomics	PHG Foundation	4	Policy briefing	Dicsusses potential applications of genetic testing, development of polygenic risk scores and genetic therapies for age related macular degeneration.	Yes	Discusses age-related macular degeneration prevelance based on ancestry. Highlights the need to larger scale genetic studies with different populations to uncover sources of heritability.
Vaccinomics	2021	https://www.phgfoundation.org/explainer/vaccinomics	PHG Foundation	n/a	Blog	Provides an overview of Vaccinomics.	Yes	Dicusses variations in vaccine response and differences in immunre response based on gender or ethnic background.
International lessons for personalised medicine	2021	https://www.phgfoundation.org/blog/international-lessons-for-personalised-medicine	PHG Foundation	n/a	Blog	Discusses personalised medicine in relation to pharmacogenomics.	Yes	Mention of ethnicity in relation to lack of population diversity in genomic datasets. Also suggests applying the 'one size does not fit all' approach to populations as well as individuals.
Policy and priorities for genomic medicine in Hong Kong	2020	https://www.phgfoundation.org/blog/policy-priorities-for-genomics-hong-kong	PHG Foundation	n/a	Blog	Discusses Hong Kong policy for genomic medicine and the Hong Kong Genome Project, while highlighting potential learning for genomics in the UK	Yes	Brief mention of ethnicity in relation to potential learning opportunities from the Hong Kong Genome Project around prevelenace of certain genetic diseases which vary between populations from different ethnic groups.
Further genetic clues to severe COVID-19	2020	https://www.phgfoundation.org/blog/further-genetic-clues-to-severe-covid-19	PHG Foundation	n/a	Blog	Explores findings from new research looking at genetic causes for most severe forms of COVID-19.	Yes	Dicusses genetic factors related to disease severity. Highlights interest in additional genetic factors that might account for increased risk of severe disease seen in certain ethnic groups.

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Genome UK: a new National Genomics Healthcare Strategy	2020	https://www.phgfoundation.org/blog/new-national-genomics-healthcare-strategy	PHG Foundation	n/a	Blog	Summaries the Genome UK: The future of Healthcare strategy.	No	
Identification and genomic data	2017	https://www.phgfoundation.org/report/identification-and-genomic-data	PHG Foundation	45	Report	Explores indentification, privacy and annoynymity in the context of genomic and genetic data.	Yes	Brief mention of terms diverse ancestry and ethnic heritage
Genomics in mainstream clinical pathways	2017	https://www.phgfoundation.org/report/genomics-mainstream-clinical-pathways	PHG Foundation	38	Report	Explores how genomics fits into current clinical pathways and how it will be used by clinicians.	No	
The personalised medicine technology landscape	2018	https://www.phgfoundation.org/report/personalised-medicine-technology-landscape	PHG Foundation	206	Report	Reviews developments in biomedical and digital technologies and how they can contribute to personalised medicine.	Yes	Discusses the need for better understanding of population diversity and for evidence of clinical effectiveness to be derived from populations that include all ethnicities,
Dementia risk prediction	2019	https://www.phgfoundation.org/report/dementia-risk-prediction-models	PHG Foundation	36	Report	Reviews the use of risk prediction for dementia as a tool for prevention	Yes	"Demographic models include ethnicity. Some models have been tested in different ethnic groups however reports highlight need for further work to test models in multiple populations including those of different ages and ethnicities. "

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Personalising prevention for breast cancer	2019	https://www.phgfoundation.org/report/personalising-prevention-for-breast-cancer-workshop	PHG Foundation	62	Report	Report on a multidisciplinary workshop held as part of the B-CAST project, exploring the science and technology advances in personalising prevention for breast cancer.	Yes	Confluence Project working to improve polygenic risk prediction across ethnic groups and breast cancer subtypes. Use of technologies could lead to increase in health inequalities but also reduce them by providing means of engagement with a more diverse range of communities.
	2019	https://www.phgfoundation.org/report/polygenic-scores-cardiovascular	PHG Foundation	83	Report	Report explores current evidence and readiness for clinical implementtion of polygenic risk scores from the perspectives of cardiovascular disease prevention.	Yes	Ethnicity is a major risk factor for CVD. Polygenic risk scores developed in European populations and do not perform as well in non-European populations
Regulating algorithms in healthcare: the GDPR and IVDR in practice	2019	https://www.phgfoundation.org/report/algorithms-workshop-report	PHG Foundation	33	Report	Report on a workshop exploring issues around regulating algorithms in healthcare	No	
Algorithms as medical devices	2019	https://www.phgfoundation.org/report/algorithms-as-medical-devices	PHG Foundation	60	Report	Report explores improving regulation of digital medical devices. This report makes recommendations for policy makers and regulators.	No	
The technologies of a healthy future	2019	https://www.phgfoundation.org/report/technologies-future-health	PHG Foundation	28	Report	The report highights the potential use of cutting edsge technology and biomedical advancements in healthcare	No	

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My healthy future: policy context	2019	https://www.phgfoundation.org/report/health-policy-context	PHG Foundation	34	Report	This report includes a detailed review of health policy in the UK.	No	
Health technologies and social impacts	2019	https://www.phgfoundation.org/report/health-technologies-social-impacts	PHG Foundation	27	Report	This report explores the risk of new technologies at aimed personalised prevention in widening health inequalities further and makes recommendations to mitigate this.	Yes	Explores need for health services to consider different perspectives, health needs, materials needed and cultural setting of diverse groups. Also highlights need to include people of non-european ancestry in genetic datasets. Discussed social factors contributing to health inequalities such as material circumstance, social cohesion, gender, ethnicity and race. Discusses 'digital divide' and the potential for technology to increase health inequalities.
Privacy and autonomy	2019	https://www.phgfoundation.org/report/privacy-autonomy	PHG Foundation	23	Report	This report explores the impact of new technologies in precision medicine on autonomy and privacy.	No	
My healthy future: over-diagnosis	2019	https://www.phgfoundation.org/report/overdiagnosis	PHG Foundation	21	Report	This report explores the role of technologies in health monitoring as part of a personalised medicine approach and discusses negative effects for users around increased anxiety and overdiagnosis.	Yes	Brief mention around offer of preconception carrier testing for certain ethnic groups.

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Person centred healthcare	2019	https://www.phgfoundation.org/reports/person-centred-care	PHG Foundation	23	Report	Report on a roundtable discussion on person centred healthcare. This report introduces relevant concepts, considers related policies and looks at future relevance and barriers of new biomedical and digital technologies to disease prevention.	Yes	General discusses strategies clinicians can use to adapt conversations to engage with particular groups. Also highlights importants of incorpating various factors including ethnicity in decision making. Identifies need to understand the barriers for certain groups. Suggests stratification should not be just by disea\se but also beliefs, values, and social characteristics to help promote equity.
Our healthy future	2019	https://www.phgfoundation.org/report/our-healthy-future	PHG Foundation	n/a	Report	This short report outlines the features of a future health system underpinned by science and technology and calls for alliances of stakeholders to support responsible design and effective delivery of health innovations.	Yes	General statement around ensuring that making sure wide range of people from different ethnic and socio-economic groups are involved in precision medicine.
Black box medicine and transparency	2020	https://www.phgfoundation.org/report/black-box-medicine-and-transparency	PHG Foundation	6	Report	A series of reports examining the human interpretability of machine learning in healthcare and research.	No	
Artificial intelligence for genomic medicine	2020	https://www.phgfoundation.org/report/artificial-intelligence-for-genomic-medicine	PHG Foundation	63	Report	This report examines the intersection between artificial intelligence and genomics medicine.	Yes	Highlights current machine learning algorithms are based on datasets of populations with european ancestry, not likely to be effective, potentially incorrect and harmful for populations of non-european ancestry.

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The GDPR and genomic data	2020	https://www.phgfoundation.org/report/the-gdpr-and-genomic-data	PHG Foundation	203	Report	This report includes a legal analysis of the impact of GDPR legislation in genomic medicine and research.	No	
Personalising breast cancer prevention – bridging the gap between research and policy	2020	https://www.phgfoundation.org/report/personalising-breast-cancer-prevention	PHG Foundation	41	Report	This report provides a comprehensive overview of the advancing science around breast cancer prevention.	Yes	General statement highligting that certain ethnic groups have higher risk of breast cancer.
Citizen generated data and health: predictive prevention of disease	2020	https://www.phgfoundation.org/report/cgd-predictive-prevention	PHG Foundation	37	Report	The report explores research and expert insight on using citizen generated data or health, disease prediction and prevention to offer more personalised assessments and interventions.	No	
Visual identifiers in the care of people with dementia	2021	https://www.phgfoundation.org/report/visual-identifiers-in-the-care-of-people-with-dementia	PHG Foundation	89	Report	The reports presents an ethical and legal analysis conducted as part of the DA VINCI (Developing a visual identification system for people with cognitive impairment in institutional settings) project.	No	

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Polygenic scores and clinical utility	2021	https://www.phgfoundation.org/report/polygenic-scores-and-clinical-utility	PHG Foundation	53	Report	This report provides a comprehensive background and exploration of dicussion around clinical readiness and clinical utility of polygenic risk scores.	Yes	General statement around ethnicity and model testing.
The ethical and legal framework for a Genomics England and Sano Genetics participant engagement platform	2021	https://www.phgfoundation.org/report/a-participant-engagement-platform	PHG Foundation	90	Report	This report, commissioned by Genomics England, explores legal and ethical implications of an patient engagement platform in genomics research.	Yes	Co-development of patient engagement platform includes potential freature to provide non-medical reports about ancestry and ethnicity. Highlights need to engage ethnic minority group in rare dieases research. Also highlightes potential impact for digital technologies to increase health inequalities. Also discusses ancestry in relation to genomic data highlighting the need for greater inclusion of populations of non-european ancestry.
Implementing polygenic scores for cardio-vascular disease into NHS Health Checks	2021	https://www.phgfoundation.org/report/prs-implementation-and-delivery	PHG Foundation	56	Report	This report explores implementation and delivery of polygenic risk scores analysis for cardiovascular disease as part of the NHS health checks programme.	Yes	Refers to need for diversity in ethnicity and ancestry data included in risk prediction tools to improve vaildity and utility in cardiovascular disease screening and NHS health check programme.
Control of patient information in the COVID-19 era	2021	https://www.phgfoundation.org/report/control-of-patient-information	PHG Foundation	20	Report	This report explores the impact of the extension of the processing of confidential patient information notice on genomic and medical research.	Yes	Refers to ethnicity in the context of participants who took part in a focus group which was explore patient and punlic views around the use of confidential patient information in research.

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Polygenic scores for cancer	2022	https://www.phgfoundation.org/report/polygenic-scores-for-cancer	PHG Foundation	33	Report	This report explores areas where implementation of polygenic risk scores for cancer is being considered in the health service and outlines current gaps in scientific evidence.	No	
Beyond the data: Understanding the impact of covid-19 on BAME groups	2020	https://www.gov.uk/government/publications/covid-19-understanding-the-impact-on-bame-communities	Public Health England	69	Report	This report, commisioned by the Chief Medical Officer for England, aimed to understand the extent that ethnicity impacts risk and outcomes. The Public Health England review of disparities in the risk and outcomes of COVID-19 shows that there is an association between belonging to some ethnic groups and the likelihood of testing positive and dying with COVID-19. Genetics were not included in the scope of the review.	Yes	Discusses the link between ethnicity and COVID-19 mortality. Also highlights the need for better recording of faith and ethnicity data.

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Equity of Access and Return in Global Genomics	2022	https://publicpolicyprojects.com/policy/	Public Policy Projects	31	Report	This report includes key themes discussed around equity of accesss and return in genomics at round table discussions with national and international researchers, cliniciians and industry represenatives.	Yes	This report discusses issues around equity of access to healthcare. It also discusses the strategy and conversation for engaging indigenous people and highlights how the current narrative only looks diversity in databases.
The Operationalisation of Precision Medicine	2020	https://publicpolicyprojects.com/policy/	Public Policy Projects	21	Report	This report includes recommendations for implementation and operationalisation of precision medicine for the NHS and other key partners.	Yes	This report includes a recommendation around genetic diversity in databases. It discusses the impact of the lack of diversity in databased on the progress of genomics medicine.
Genomics Revolution	2021	https://publicpolicyprojects.com/policy/	Public Policy Projects	53	Report	This report lays out current opportunities in genomics and key recommendations to enable to UK to develop a world leading genomics ecosystem.	Yes	The recommendations include and discuss the need to improve diversity in genomics data to prevent health data poverty and health inequalities. The report highlights the need for targeted community engagement, aswell as patient education.

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Bringing the Benefits of Genome Sequencing to the World	2022	https://publicpolicyprojects.com/policy/	Public Policy Projects	41	Report	This report discusses how the benefits of genome sequencing can be made global.	Yes	This report highlights the need to address existing global health inequalities prior to the roll out of genomics in healthcare, otherwise genomics may exacerbate these issues. The report also mentions the need to improve public trust and engagement. As well as this, the report discusses diverstiy in datasets, acknowledging that the global genomics dataset is not representative of the global population and provides examples of current projects such as Genomics England's 'Diverse Data Initative' and ELIXIR's 'Beyond 1 Million Genome Project'.
Socialising the Genome: Communications, Public Trust and Engagement	2021	https://publicpolicyprojects.com/policy/	Public Policy Projects	8	Report	This is a session report for a virtual webinar held which explored engagement, trust and improving public awareness of genomics in the general public and more specifically in minority communities.	Yes	The report highlights global lack of awareness of genomics as well as mistrust and disengagement in minority communities. It discusses the need to acknowledge and address communities history and reasons for mistrust, distrust and disengagement. The report presents more recent examples which may have impacted public confidence such as the NHS 'opt out' scheme. This report also explores how to better engage and communicate with the public about genomics, especially disengaged communities.
Build back fairer: the covid-19 Marmot review	2020	https://www.health.org.uk/publications/build-back-fairer-the-covid-19-marmot-review	The Health Foundation and Institute of Health Equity	222	Report	This report, commisioned by The Health Foundation, investigated how the pandemic affected health inequalities in England.	No	

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The Kings Fund	Not available	https://www.kingsfund.org.uk/publications/ethnicity-coding-health-records	The Kings Fund	n/a	Webpage/ Letter	The King's Fund in collaboration with UCL Institute of Health Equity submitted a letter to NHS England and NHS Improvement highlighting the issues around ethnicity related data recording and data quality, providing suggestions for improvement.	No	
Genomics for Precision Medicine Strategy (Wales)	2017	https://www.gov.wales/genomics-precision-medicine-strategy-0	Welsh Government	25	Policy and strategy document	This Strategy sets out the Welsh Government's plan to create a sustainable, internationally-competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales.	No	

APPENDIX 3

Descriptive overview of the methodology of the included studies

Other analytical techniques adopted include Model simulation analysis [102, 157, 160, 183, 185, 208, 223], mendelian randomisation analysis [98, 117, 130, 133, 150, 160, 167], phenome-wide association study analysis [129, 131, 187], allele frequency comparison [120, 146, 160, 205], Anova [120, 146, 160, 205], Wilcoxon rank sum test [94, 155, 193], Fisher's exact test [109, 160], Principal Component Analysis [172, 197], unspecified Association analysis [184], augmented variant analysis and reanalysis pipeline [184], batch screening iterative lasso (BASIL) algorithm [142], Cluster analysis [118], collapsing analysis [199], combined meta?analysis estimate (the GWAS, iCOGS and OncoArray study estimate) [176], Conditional analysis [207], gene-based enrichment analyses [182], genetic correlation analyses [104], Linkage analysis [184], Correlation analysis [111], Epigenome-Wide Association analysis [175], exposome-wide interaction study [211], Fine-mapping analysis [207], Pan-ancestry association analyses [245], Pathway analysis [207], polychoric correlations [263], Spearman correlation coefficient [179], T tests [146], whole-genome sequencing analysis [190, 270]. Another study also compared the PolyPred and PolyPred+ (and their summary statistics-based analogs) with state-of-the-art polygenic prediction methods via simulations and analyses [210]. However, analysis was not applicable to two studies – 1. Analysis was not done because restricting model development and validation to those of white ethnicity did not materially alter model performance for any of the models presented [128] 2. The study was a narrative synthesis of host genetic factors determining COVID-19 susceptibility and severity [90]. Finally, two studies of the included studies did not state the adopted analytical approach [136, 209]. [184], batch screening iterative lasso (BASIL) algorithm [142], Cluster analysis [118], collapsing analysis [199], combined meta?analysis estimate (the GWAS, iCOGS and OncoArray study estimate) [176], Conditional analysis [207], gene-based enrichment analyses [182], genetic correlation analyses [104], Linkage analysis [184], Correlation analysis [111], Epigenome-Wide Association analysis [175], exposome-wide interaction study [211], Fine-mapping analysis [207], Pan-ancestry association analyses [245], Pathway analysis [207], polychoric correlations [263], Spearman correlation coefficient [179], T tests [146], whole-genome sequencing analysis [190, 270]. Another study also compared the PolyPred and PolyPred+ (and their summary statistics-based analogs) with state-of-the-art polygenic prediction methods via simulations and analyses [210]. However, analysis was not applicable to two studies – 1. Analysis was not done because restricting model development and validation to those of white ethnicity did not materially alter model performance for any of the models presented [128] 2. The study was a narrative synthesis of host genetic factors determining COVID-19 susceptibility and severity [90]. Finally, two studies of the included studies did not state the adopted analytical approach [136, 209].

APPENDIX 4

Quantitative studies characteristics

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Adamson 2021	Observational study	Pulmonology/ Respiratory medicine (COVID-19)	9.433	9.433	Genetic & biomarker profile of BAME only	UKBB	Yes	Self-reported ancestry
Adjangba 2021	Observational study	Smoking/Tobacco	Not specified	Not specified	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry and genetic analysis
Aichang 2021	Observational study	Rheumatology	2.820	2820	Genetic & biomarker profile of BAME only	"1) Whole genome sequencing data of people with gout from clinical trials for urate-lowering therapy 2) A gout and related diseases study "	Yes	Self-reported ancestry
Alkhalfan 2022	Case control study	Neurology	11.155	346	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Apol 2022	Observational study	Public health	1541	19	Genetic & biomarker profile of White vs BAME	Primary data from recruited patients	Yes	Self-reported ancestry
Ashvetiya 2021	Case control study	Cardiovascular surgery	1.363	1.363	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Backman 2021	Observational study	Genomics	430.998	21.654	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Beaumont 2022	Observational study	Genomics	200.643	16.155	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Bell 2021	Observational study	Ophthalmology	54	15	Genetic & biomarker profile of White vs BAME	Patients' hospital records	Yes	Patient records
Blass 2022	Case control study	Gynecology	148.571	5.924	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Boulund 2022	Observational study	Gastroenterology, Cardiometabolic traits	4117	2.948	Genetic & biomarker profile of White vs BAME	1) UKBB 2) multi-ethnic Healthy Life in an Urban Setting (HELIUS) cohort	Yes	Self-reported ancestry
Bountziouka 2022	Observational study	Genomics	422.797	22.761	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Burnell 2022	Randomised controlled trials (two-arm)	Oncology, Psychiatry/ Psychological medicine	1034	1034	Genetic & biomarker profile of BAME only	Primary data from randomised controlled trial	Yes	Self-reported ancestry
Cai 2022	Observational study	Genomics	12.551	2.551	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Calvin 2022	Observational study	Psychiatry/ Psychological medicine	206.960	5.789	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Cavazos 2022	Case control study	Oncology	172.282	9.385	Genetic & biomarker profile of White vs BAME	1) Kaiser Permanente Research Bank (KPRB) 2) UKBB	Yes	Self-reported ancestry
Chan 2021	Cohort study	Ophthalmology	44	29	Genetic & biomarker profile of White vs BAME	Primary data from recruited patients	Yes	Self-reported ancestry
Chandrasekaran 2021	Interventional study	Oncology, Gynecology	303	107	Genetic & biomarker profile of White vs BAME	Primary data from recruited patients	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Charras 2022	Cohort Study	Rheumatology	348	107	Genetic & biomarker profile of White vs BAME	UK juvenile-onset systemic lupus erythematosus (JSLE) Cohort Study	Yes	Self-reported ancestry
Chiu 2022	Observational study	Demography	488.363	Not specified	Genetic & biomarker profile of White vs BAME	1) UKBB 2) The 1000 Genomes Project (TGP) 3) Human Origins (HO) dataset 4) Human Genome Diversity Project (HGDP)	Yes	Self-reported ancestry
Choquet 2021	Meta-analysis	Ophthalmology	487.321	Not specified	Genetic & biomarker profile of White vs BAME	1) UKBB 2) Genetic Epidemiology Research in Adult Health and Aging (GERA)	Yes	Self-reported ancestry
Choquet 2021	Meta-analysis	Neurology	465.435	27.257	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Chu 2022	Cohort study	Infectious diseases, Cardiology, Neurology	9689	510	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Clarke 2022	Cohort study	Rheumatology, Paediatrics, Epidemiology	Not specified	Not specified	Genetic& Biomarker Profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Clift 2021	Cohort study	Medical technology	420.560	21.298	Precision medicine approach to stratify care	UKBB	Yes	Self-reported ancestry
Constantinescu 2022	Observational study	Genomics	62.484	11.799	Methods to account for EM in genetic research	UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Crossfield 2022	Cohort study	Pulmonology/ Respiratory medicine (COVID-19), Epidemiology	9.560	865	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Cui 2021	Cohort Study	Oral oncology	539	57	Genetic & biomarker profile of White vs BAME	The Cancer Genomic Atlas (TCGA)	Yes	Self-reported ancestry
Curtis 2021	Observational study	Human heredity	49.953	6.303	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
David 2022	Case control study	Pulmonology/ Respiratory medicine (COVID-19)	2244	568	Genetic & biomarker profile of White vs BAME	Data from the Genetics Of Mortality In Critical Care (GenOMICC) study.	Yes	Self-reported ancestry
Derks 2022	Observational study	Genomics, Galactosemia	22	5	Country comparison	GalNet (an online patient registry including patients with galactosemia from several countries).	Yes	Self-reported ancestry
Dite 2021	Case control study	Pulmonology/ Respiratory medicine (COVID-19)	1582	564	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Elliott 2022	Observational study	Epigenetics	916	472	Genetic & biomarker profile of White vs BAME	UK-based cohorts: Southall and Brent Revisited (SABRE) and Born in Bradford (BiB).	Yes	Self-reported ancestry
Evans 2022	Observational study	Oncology	9475	578	Genetic & biomarker profile of BAME only	Predicting-Risk-of-Cancer-At-Screening (PROCAS) study	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Fatumo 2021	Case control study	Neurology, Endocrinology	1.874.463	6.614	Genetic & biomarker profile of White vs BAME	1) African Partnership for Chronic Disease Research study 2) UKBB 3)Million Veteran Program 4) Consortium of Minority Population Genome-Wide Association Studies of Stroke 5) MEGASTROKE study	Yes	Self-reported ancestry
Feng 2021	*Cohort study*	*Oncology, Nutrition*	*415.524*	*22.104*	*Genetic & biomarker profile of White vs BAME*	*UKBB*	*Yes*	*Self-reported ancestry*
Forrest 2022	*Observational study*	*Genomics*	*66.434*	*22.611*	*Genetic & biomarker profile of White vs BAME*	*1) UKBB 2) BioMe Biobank*	*Yes*	*Self-reported ancestry*
Fraszczyk 2022	*Meta-analysis*	*Endocrinology*	*3.200*	*2.659*	*Country comparison*	*five longitudinal cohorts were included: Doetinchem, ESTHER, KORA1, KORA2 and EPIC-Norfolk*	*Yes*	*Self-reported ancestry*
Fritsche 2021	*Observational study*	*Oncology*	485.434	16.453	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Gao 2022	Observational study	Oncology	Not specified	Not specified	Genetic & biomarker profile of White vs BAME	1) The Cancer Genome Atlas (TCGA) 2) UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Giannakopoulou 2021	Meta-analysis	Psychiatry/ Psychological medicine	15.771	Not specified	Country comparison	CKB, CONVERGE, Taiwan-MDD study, US- and UK-based cohorts with DNA samples of individuals of East Asian descent: 23andMe Inc, Women's Health Initiative (WHI), Mount Sinai BioMe Biobank, Intern Health Study (IHS), the Study to Assess Risk and Resilience in Servicemembers (Army-STARRS), and UKBB.	Yes	Self-reported ancestry
Gill 2021	Observational study	Endocrinology	598	262	Genetic & biomarker profile of White vs BAME	1) The Lipid Genetics Clinic (London, ON, Canada) 2) Cardiovascular Research Institute at the University of California, San Francisco.	Yes	Self-reported ancestry
Gratton 2022	Observational study	Endocrinology	367.264	14.098	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Gyftopoulos 2022	Case control study	Cardiology	363	31	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Haas 2022	Observational study	Gyne-endocrinology	229.966	5336	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Hale 2022	Case control study	Neurosurgery	155.154	Not specified	Country comparison	1) UKBB 2) FinnGen 3)Biobank Japan	Yes	Self-reported ancestry
Han 2021	Observational study	Ophthalmology	115.371	22.897	Country comparison	1) UKBB 2) The Canadian Longitudinal Study on Aging (CLSA).	Yes	Self-reported ancestry
He 2022	Observational study	Cardiology	495.534	76.631	Country comparison	1) Trans-Omics for Precision Medicine (TOPMed) 2) UKBB	Yes	Self-reported ancestry
Hodgson 2022	Cohort study	Endocrinology	22.490	22.490	Genetic & biomarker profile of BAME only	Genes and Health (G&H) study	Yes	Self-reported ancestry
Hujoel 2022	Observational study	Genomics	409.000	14.000	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Hulsizer 2022	Observational study	Haematology	8019	8019	Genetic & biomarker profile of BAME only	UKBB	Yes	Self-reported ancestry
Ingram 2021	Observational study	Pharmacogenetics	197509	32707	Genetic & biomarker profile of White vs BAME	1) University College Hospital (UCH) in London database 2) 1000 genome (1000G) study 3) Indian datasets (ie the Gujarati Indians in Houston and Indian Telugu in the UK)	Yes	Self-reported ancestry
Innes 2022	Observational study	Gastroenterology	197.509	27.267	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Jiang 2021	Case control study	Ageing & genetics	501.756	16.063	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Kappel 2022	Observational study	Psychiatry/ Psychological medicine	4.459	704	Genetic & biomarker profile of White vs BAME	1) CLOZUK2 2) CLOZUK3 3) Norwegian therapeutic drug monitoring (TDM) database	Yes	Self-reported ancestry
Kar 2022	Observational study	Haematology	200453	Not specified	Genetic & biomarker profile of White vs BAME	1) UK Biobank (UKBB)	Yes	Self-reported ancestry
Kelly 2022	Observational study	Cardiology	318.891	9.981	Country comparison	1) UK Biobank (UKBB), 2) Million Veteran Program (MVP), 3) Trans-Omics for Precision Medicine (TOPMed), 4) Centers for Common Disease Genomics (CCDG) program, 5) Reasons for Geographic and Racial Differences in Stroke (REGARDS) study	Yes	Self-reported ancestry
Kim 2022	Observational study	Gastroenterology, Oncology	414.209	24.757	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Knutson 2022	Cohort study	Molecular Genetics	9467	2757	Genetic & biomarker profile of White vs BAME	"1) UKBB 2) Alzheimer's Disease Sequencing Project (ADSP)"	Yes	Self-reported ancestry
Kum 2021	Cohort study	Oncology	458	103	Genetic & biomarker profile of White vs BAME	Electronic patients' records Active Surveillance (AS) cohort Guys and St Thomas' Hospital	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Laitman 2021	Cohort study	Oncology	2.503	2.503	Genetic & biomarker profile of White vs BAME	Patients records and Israeli National Cancer Registry.	Yes	Self-reported ancestry
Lamri 2022	Observational study	Obstetrics, Endocrinology	5.209	5.209	Genetic & biomarker profile of BAME only	1) SouTh Asian BiRth CohorT (START) cohort study 2) Born in Bradford (BiB) cohort study	Yes	Self-reported ancestry
Lewis 2021	Cross-sectional study	Endocrinology	459.503	6.990	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Liang 2022	Observational study	Genomics	370.694	14.218	Methods to account for EM in genetic research	UKBB	Yes	Self-reported ancestry
Madhvani 2022	Observational study	Gynecology	68.752	3.794	Methods to account for EM in genetic research	Hospital Episode Statistics (HES) of the English National Health Service (NHS)	Yes	Self-reported ancestry
Marino-Ramirez 2022	Observational study	Nephrology	35.590	25.590	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
McDonald 2022	Observational study	Rheumatology	163.015	40.220	Country comparison	Million Veteran Program (MVP) and UK Biobank (UKBB)	Yes	Self-reported ancestry
McInnes 2021	Observational study	Pharmacogenetics	487.409	Not specified	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry and principal component analysis of genetic data.
McManus 2022	Case control study	Pulmonology/ Respiratory medicine (COVID-19)	14.558	1.737	Genetic & biomarker profile of White vs BAME	1) UKBB 2)Patient hospital records	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Meijsen 2021	Case control study	Psychiatry, Gynecology	232.993	12.878	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Mone 2021	Cohort study	Obstetrics & Gynecology	425	260	Genetic & biomarker profile of BAME only	Birmingham Women and Children's NHS Foundation Trust, UK	Yes	Self-reported ancestry
Mukadam 2022	Cohort study	Neurology	294.162	6.356	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Muller 2022	Observational study	Ophthalmology	65.023	6.270	Precision medicine approach to stratify care	UKBB	Yes	Self-reported ancestry
Nag 2021	Observational study	Endocrinology	422.488	17.699	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Nag 2021	Observational study	Metabolomics	412.394	17.699	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Nagar 2021	Observational study	Chemical pathology	433.298	6.456	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Nagar 2021	Observational study	Endocrinology	474.184	16.902	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Namjou 2022	Case control study	Pulmonology/ Respiratory Medicine (Pediatrics)	428.725	30.479	Country comparison	1) Trans-National Asthma Genetic Consortium genome-wide association study summary statistics 2) Electronic Medical Records and Genomics (eMERGE) cohort 3) UK Biobank	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Naseri 2021	Observational study	Genomics	487.409	Not specified	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry and analysis of genetic data
Nongmaithem 2022	Observational study	Pediatrics	6.099	6.099	Genetic & biomarker profile of White vs BAME	1) UK BioBank 2) The Exeter Family Study of Childhood Health (EFSOCH) 3) The Indian cohort studies [Pune Maternal Nutrition Study (PMNS), Parthenon Study (PS), Mumbai Maternal Nutritional Project (MMNP) and Mysore Birth Records Cohort (MBRC)]. 4) Bangladeshi cohort study [GIFTS (Genomic and lIfestyle predictors of Fetal ouTcome relevant to diabetes and obesity and their relevance to prevention strategies in South Asian people)] 5) London UK Bangladeshi (UK-Bang) cohort study	Yes	Self-reported ancestry
Pagnamenta 2021	Observational study	Neurology	17	5	Genetic & biomarker profile of White vs BAME	100K Genomes Project	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Parker 2021	Case control study	Rheumatology/ Neurology	6.062	5.737	Genetic & biomarker profile of BAME only	UKBB, Penn Medicine Biobank and the Million Veteran Program	Yes	Self-reported ancestry and principal component analysis of genetic data.
Patel 2022	Observational study	Cardiology	59.411	4.776	Country comparison	1) Atherosclerosis in Risk Communities (ARIC) 2) UK Biobank	Yes	Self-reported ancestry
Patel 2021	Cohort study	Cardiology	457.473	8124	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Patel 2022	Observational study	Cardiology	519.432	23.643	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Pathak 2021	Case control study	Pulmonology/ Respiratory medicine	641331	20.800	Country comparison	1) UKBB 2) Vanderbilt Biobank 3) Biobank Japan	Yes	Self-reported ancestry
Piga 2021	Observational study	Smoking	10558	10558	Genetic & biomarker profile of BAME only	1) Africa Wits-INDEPTH Partnership for Genomic Studies (AWI-Gen) 2) Uganda Genome Resource (UGR) 3) UK Biobank (UKBB)	Yes	Self-reported ancestry
Prive 2022	Observational study	Bioinformatics	13737	7464	Methods to account for EM in genetic research	UKBB	Yes	Self-reported ancestry and genetic analysis
Puntis 2021	Cohort study	Psychiatry/ Psychological medicine	33710	2668	Genetic & biomarker profile of White vs BAME	Oxford Health NHS Foundation Trust electronic health records clinical register	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Reisel 2022	Cohort Study	Oncology	935	935	Methods to account for EM in genetic research	Self-referred participants attending pre-testing counselling/ recruitment clinics (primary data source)	Yes	Self-reported ancestry
Resurreccion 2021	Cohort Study	Haematology, Pulmonology	500822	8017	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Robbins 2022	Cohort study	Oncology	11006	857	Precision medicine approach to stratify care	National Lung Screening Trial (NLST)	Yes	Not reported
Rudnicka 2022	Cohort study	Ophthalmology, Cardiology	72281	4992	Precision medicine approach to stratify care	1) UKBB 2) European Prospective Investigation into Cancer (EPIC)-Norfolk	Yes	Self-reported ancestry
Runolfsdottir 2021	Cohort study	Nephrology	Not specified	Not specified	Country comparison	1) deCODE genetics database 2) UK Biobank 3) 100,000 Genomes Project 4) Genome Aggregation Database 5) Human Genetic Variation Database 6) Korean Variant Archive (KOVA)	Yes	Self-reported ancestry
Sarnowski 2021	Meta-analysis	Gynecology/ Reproductive health	57091	5148	Genetic & biomarker profile of BAME only	1) ReproGen Consortium 2) UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Satterfield 2021	Cohort study	Cardiology	908087	103580	Genetic & biomarker profile of White vs BAME	1) electronic Medical Records and Genomics (eMERGE) network 2) UK Biobank 3) Million Veteran Program (MVP)	Yes	Self-reported ancestry
Schachtl-Riess 2021	Cohort study	Cardiology	2504	2001	Genetic & biomarker profile of White vs BAME	1) UKBB 2) 1000 Genomes Project	Yes	Self-reported ancestry
Schneider 2022	Cohort study	Cellular biology, Internal medicine	Not specified	Not specified	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Sekimitsu 2022	Case control study	Ophthalmology	502506	10462	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Shah 2021	Cohort study	Endocrinology, Periodontics	Not specified	Not specified	Genetic & biomarker profile of White vs BAME	1) UK Biobank 2) MAGIC 3) Diabetes Meta-Analysis of Trans-Ethnic association studies (DIAMANTE) Consortium 4) FinnGen consortium 5) Biobank Japan	Yes	Self-reported ancestry
Shao 2022	Cohort study	Nutrition, Oncology	450482	42093	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Sharma-Oates 2022	Cohort study	Rheumatology, Immunology	3.902.748	622604	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Shears 2022	Cohort study	Immunology	40	27	Genetic & biomarker profile of White vs BAME	Patients' data provided by clinicians	Yes	Demographic data from clinical records
Sinkala 2022	Cohort study	Cardiology	389449	5978	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Sriram 2022	Cohort study	Pulmonology/ Respiratory medicine	487320	28070	Country comparison	UKBB	Yes	Self-reported ancestry
Stalbow 2022	Cohort study	Endocrinology	179412	Not specified	Country comparison	1) UKBB 2) BioMe	Yes	Self-reported ancestry
Sun 2022	Cohort study	Psychiatry/ Psychological medicine	2.269	139	Country comparison	1) UKBB 2) Qingdao Twins Registry	Yes	Self-reported ancestry
Sun 2022	Cohort study	Metabolomics	21745	21745	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry, k-means clustering of genetic principal components
Tanigawa 2021	Cohort study	Genomics	108362	40937	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Tcheandjieu 2022	Cohort study	Cardiovascular surgery	33031	836	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Thibord 2022	Cohort study	Pulmonology/ Respiratory medicine (COVID-19)	487320	28070	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Tideman 2021	Cohort study	Ophthalmology	54006	2165	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Tiller 2022	Cohort study	Oncology	2167	2167	Methods to account for EM in genetic research	2 BRCA-Jewish Founder Mutation (B-JFMs) screening programme databases in Australia-Melbourne and Sydney	Yes	Self-reported ancestry
Tong 2021	Cohort study	Nutrition	471593	5535	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Trinder 2021	Cohort study	Cardiology	725002	17838	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Vadgama 2022	Cohort study	Pulmonology/ Respiratory medicine (COVID-19)	1837	583	Genetic & biomarker profile of White vs BAME	100,000 Genomes Project	Yes	Self-reported ancestry
Vanent 2022	Cohort study	Nephrology, Neurology	6000	3897	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Velavan 2021	Systematic review (narrative synthesis)/ Literature review	Pulmonology/ Respiratory medicine (COVID-19)	Not specified	Not specified	Country comparison	PubMed, MEDLINE, EMBASE, GWAS catalogue, and preprints [medRxiv & bioRxiv- used case-control studies	Yes	Not reported
Wainberg 2021	Cross-sectional study	Psychiatry/ Psychological medicine	89205	2692	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Wainberg 2021	Cohort study	Psychiatry/ Psychological medicine	25916	655	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Wang 2021	Cohort study	Pulmonology/ Respiratory medicine (COVID-19)	914	53	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Ward 2021	observational study	Pulmonology/ Respiratory medicine (COVID-19)	2159742	Not specified	Genetic & biomarker profile of White vs BAME	Accident and emergency (A&E) data set from the Secondary Uses Service(SUS) suite of data sets.	Yes	Self-reported ancestry

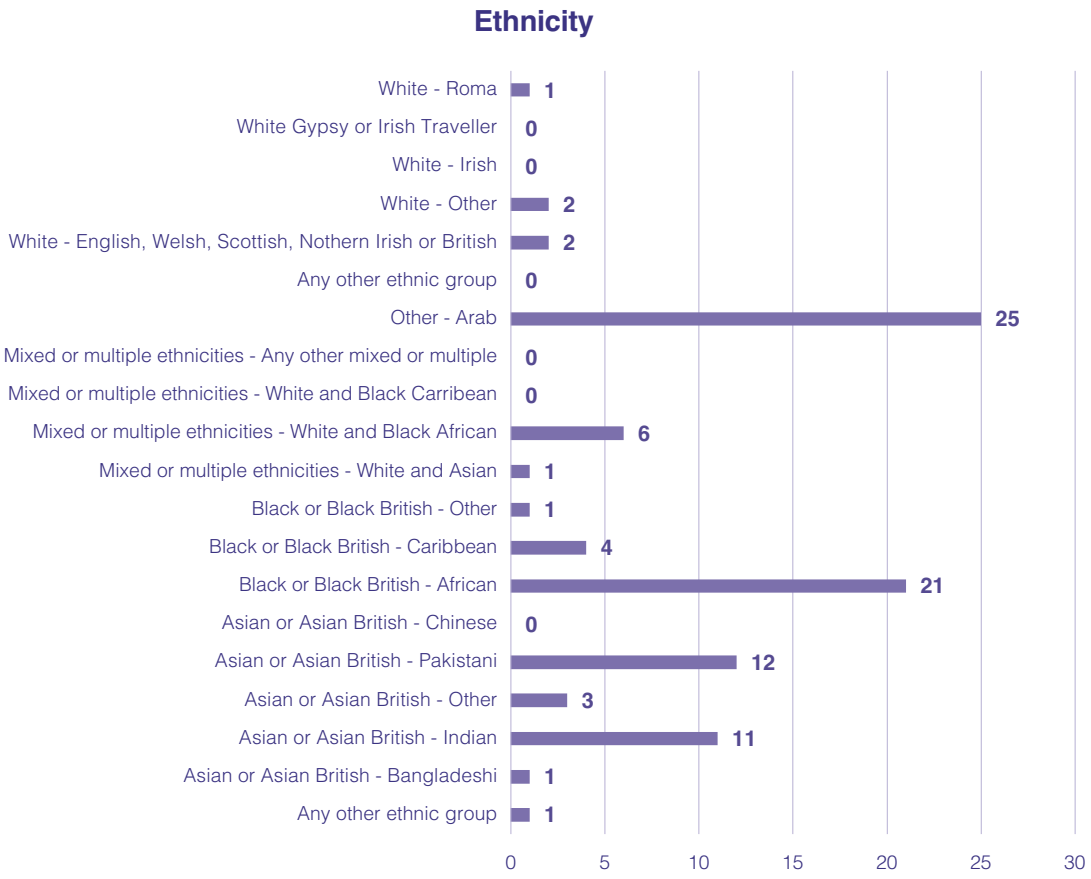
Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Watkins 2022	observational study	Genomics	4.338	439	Genetic & biomarker profile of White vs BAME	1) Accessible Resource for Integrated Epigenomic studies, ARIES [a sub study of the Avon Longitudinal Study of Parents and Children (ALSPAC)] 2) Born in Bradford (BiB) data.	Yes	Self-reported ancestry
Weissbrod 2022	observational study	Genomics	432.800	14.800	Country compariosn	1) UKBB 2) European Network for Genetic and Genomic Epidemiology 3) Biobank Japan 4) Uganda-APCDR	Yes	Self-reported ancestry
Welsh 2021	Cohort study	Cardiovascular surgery	401.820	18.129	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Westerman 2021	observational study	Cardiology, Genomics	350.016	13.718	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Westerman 2022	observational study	Nutrition, Endocrinology	28.824	13.086	Genetic & biomarker profile of White vs BAME	10 cohorts in the National Heart, Lung, and Blood Institute (NHLBI) Trans-Omics for Precision Medicine (TOPMed) program	Yes	Self-reported ancestry
Wright 2022	observational study	Genomics	13.610	2.166	Genetic & biomarker profile of White vs BAME	Deciphering Developmental Disorders (DDD) study	Yes	Self-reported ancestry

Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Xie 2022	Cohort study	Pulmonology/ Respiratory medicine (COVID-19), Haematology	336.790	22.605	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Yap 2021	Observational study	Neurology, Pediatrics,	2.477	303	Genetic & biomarker profile of White vs BAME	Australian Autism Biobank (AAB)	Yes	Analysis of genetic data
Yeap 2022	Cohort study	Cardiology, Reproductive Health	210.700	9.640	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Yngvadottir 2022	Case control study	Onconephrology	1336	132	Genetic & biomarker profile of White vs BAME	UK 100,000 Genomes Project	Yes	Self-reported ancestry and principal component analysis
Yuan 2022	Cohort study	Neurology	475.813	42.029	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Zhang 2021	Observational study	Pulmonology/ Respiratory medicine (COVID-19), Geriatrics	8847	451	Genetic & biomarker profile of White vs BAME	UKBB	Yes	Self-reported ancestry
Zhao 2022	Observational study	Genomics	18.453	18.453	Genetic & biomarker profile of BAME only	UKBB	Yes	Self-reported ancestry

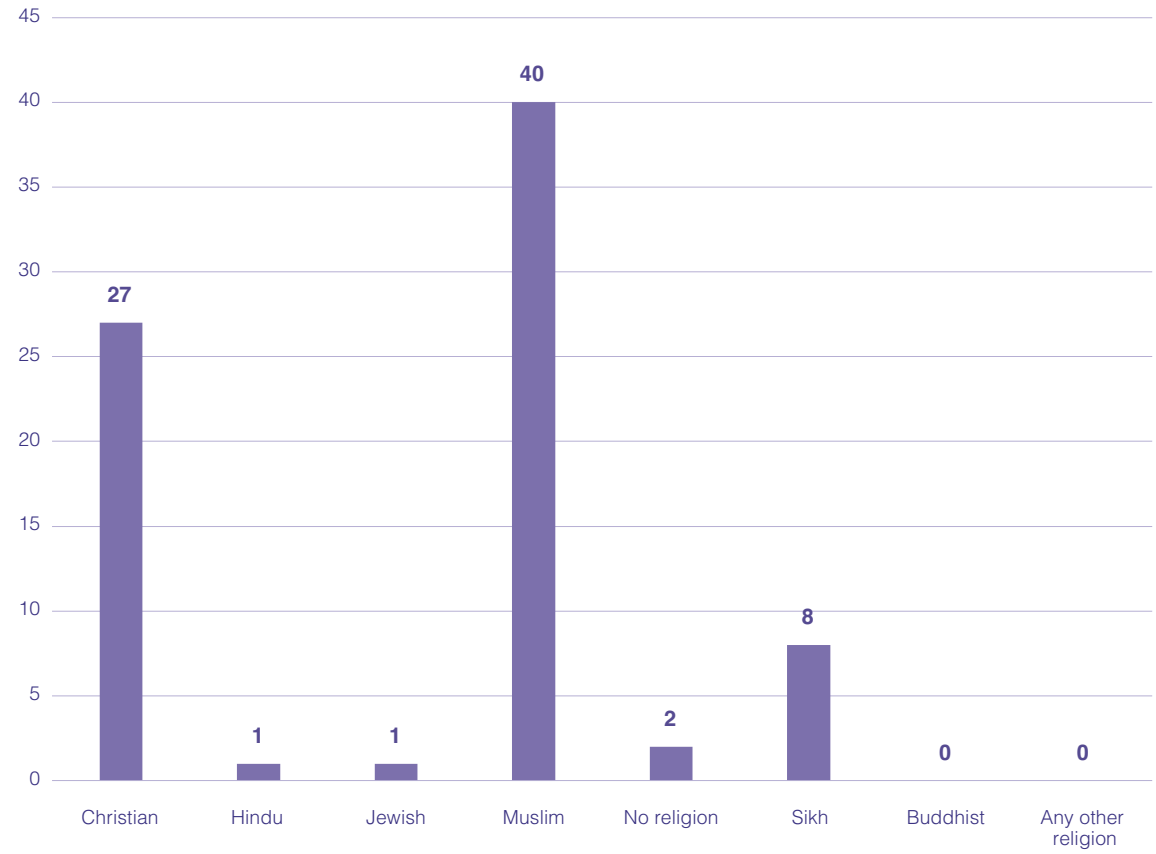
Study	Study design	Specialty/subject assessed	Total number of participants	Number of EM participants	Focus of study	Database/source of data used	Recognition of EM in database/ sample?	Means of EM identification
Zhixiu 2021	Case control study	Rheumatology	36.037	13.519	Country compariosn	1) UK Ankylosing Spondylosis (AS) cohort 2) Australo-Anglo-American Spondyloarthritis Consortium Cohort 3) Australian AS cohort 4) The Groupe Française d'Etude Génétique des Spondylarthrites (GFEGS) 5) The Chinese AS Cohort 6) The Turkish and Iranian AS cohort.	Yes	Self-reported ancestry
Zollner 2022	Observational study	Gastroenterology	5236	5236	Genetic & biomarker profile of BAME only	UK 5236 Genes & Health volunteers cohort	Yes	Self-reported ancestry

APPENDIX 4

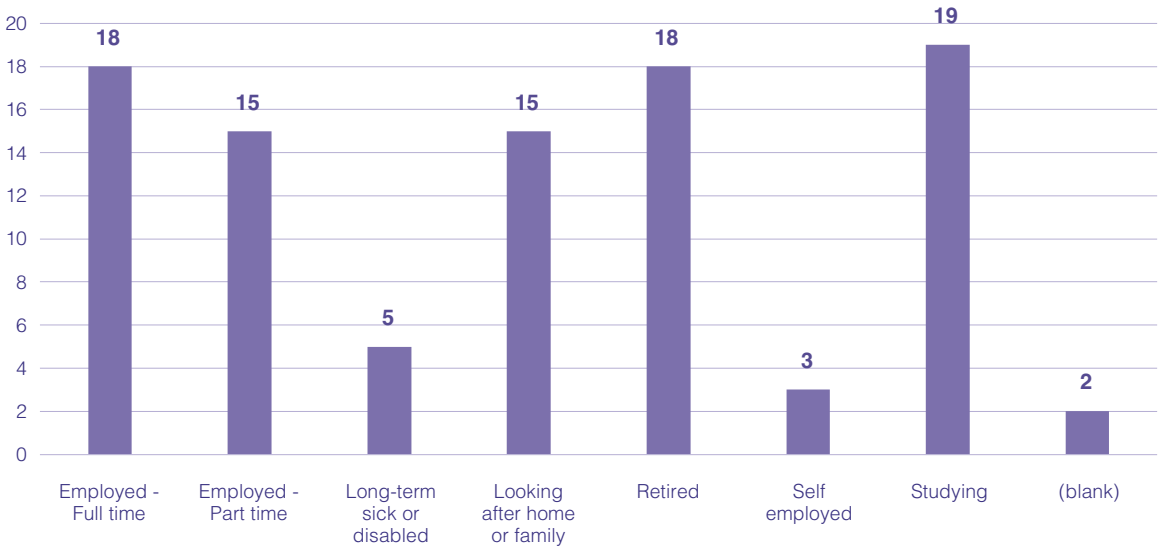
Public stakeholder demographics



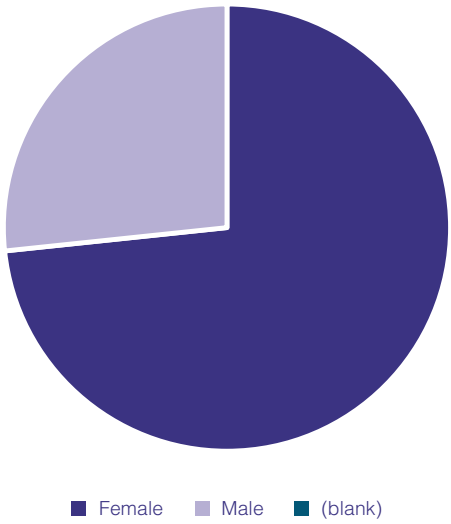
Religion



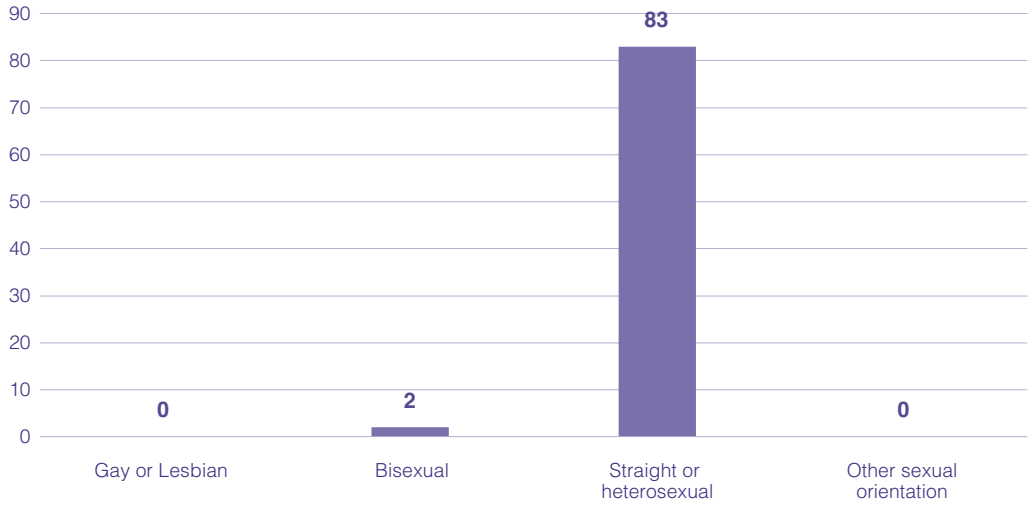
Employment status



Sex



Sexual orientation



Gender identity

APPENDIX 5

5.1 Knowledge and awareness themes

Theme	Sub theme	Quote
Current levels of knowledge and awareness	Genomics	"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
		"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
	Personalised medicine	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
		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
	Health inequalities	But I find it slightly confusing as to or are we supposed to, are the figures so low for the ethnic groups because they didn't take up the services or where they never offered them. - FG1
		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

5.2 Improving knowledge themes

Theme	Sub theme	Quote
Strategies for improving knowledge and awareness	Role of healthcare professionals	And so maybe it's through nursing that we can actually target that workforce, but I think that's where as well. If we're looking at those groups of those patients hard to reach patients within at some point, those patients are coming into hospital for one reason or the other. And so if we can start making sure that our our on the ground face to face teams understand and our way of genomics and can talk to their patients about it, then we might be a it it will slowly change. - C01
		I think you access this information for older generation and ethnic minorities probably you can start with the doctor's surgery, that you have leaflets in different languages, and even those surgeries, they should be incentivised they can give to those people that information which they think that needed. And similarly maybe in schools as well. - FG6
		I think GPs should have more information about this. Because everyone is going to go through the GP one day, one way or another. GPs, or within the outpatient clinics of hospitals, there is information, there is a person or pamphlets, I don't know. That they could inform us about all this. But I think it has to start with GPs, because here we don't listen to the radio, and if we do, we don't understand either. We don't read newspapers, because we don't understand, nor television. I think this is not the solution, I think it would have to be within the GPs that we have within our communities. Each of us is registered with a GP. I believe this is it. - FG8
	Role of faith and community leaders	Hey I think maybe try the the community groups, you know where the for the you know for Asian people, for minority groups, I think so like one place I think that is quite key. It would be quite good as maybe the mosques you know and the places of worship like mosques and temples and like churches as well and it will not church sorry and other places you know the space is worship or people place where they and they're like, and cultural events are held, perhaps, and things and things that would be good maybe and advertising as well in those areas. - FG4
		We've had in London, in South or particularly there's they do try and run the sort of like clinics and groups in the local Gurdwareh. Which they're quite popular actually, I think, because the people that speaking to them, they they can speak Punjabi. I think a lot of the. Sort of slightly and I don't know if if I should say older, but the ones that can't really wouldn't understand it in English basically. So I think those type of. Settings. They seem to be working well like around here, so maybe something like that in? -FG1
		I know that in the black community….. because you essentially listen to what …., so if we could get our leaders to be onboard, or if we should educate our leaders more, then the followers would definitely follow whatever they say. - FG9

Theme	Sub theme	Quote
	Presence in community spaces and events	Like Melton Rd. like where its Divali on Melton Road there was about 30-40 thousand people there. Have, have a little stand somewhere. Or have someone walking around like yourself or five, six other people and just chatting to families you turn the whole families then, then move on to the next family, cause a lot of people there, you know others four, 40,000 people. If 100, you know more. It's a lot of people. - FG4
		Like this, through focus groups, because in our communities I'm talking about the Arab community, it's the world of the mouth. So, I've attended this and I knew this. It's very important. - FG2
	Role of lived experience	I just feel like as if words are not enough, and most people would like it to be actions instead. If they see an example then they're more likely to go for it. So I feel like they should put more in actions than just words. I mean words can also work but that's just for a certain amount of people. A certain amount of people that are dependent, experience or like actions from like people they've seen go through it, and then they'll go for it. -FG9
		I think they really needs to be such a massive effort to say to, to know, to be out there, to give that information, to give experience. You know and and find patients that have been through a genetic journey it genetically over and start getting those patients as like that lived experience from those patients to the groups to the, to the groups that their communities as well. - C01
	Multigenerational approach	Secondary school. Because sex education is part of their curriculum. So if you can teach them about sex education, you can teach them about their genotypes. For example, I'm a carrier. I have known since I was a teenager the idea of not marrying someone who is also a carrier. And also we have a high risk of African essence. So I've known that since before I even started having emotional feelings towards the opposite sex, I've had it on the back of my mind. "OK, there are risks involved if I get married and have kids with somebody who is a carrier like me". So it's good to give the info to teenagers from that age, it's important. - FG8
		If you get the younger generation aware of what's going on, so like if we get the younger lot more where they can also like tell their parents about it, either on to their grandparents, because that college we do have that lesson. So we do learn about certain aspects of different stuff, but we don't retouch a lot on like cancers and diseases you can get due to your like ethnic might not like groups and stuff which it would be quite good to have that like. - FG1
		I think you access this information for older generation and ethnic minorities. Probably you can start with the doctor's surgery, that you have leaflets in different languages, and even those surgeries, they should be incentivised they can give to those people that information which they think that needed. And similarly maybe in schools as well. - FG6

Theme	Sub theme	Quote
	Multimedia approach	we get a lot of like a local monthly radio stations. So like like you say, you actually go there for an hour chat, show kind of thing and talk about it. Because I might not. Because it's Vaisakhi next month this month. And I believe that is a one month radio station at the same thing for Eid, same thing for what is divali same thing for other Asian festivals are coming up. They're one month radio shows and then now I'm sure if you get hold of who's promoting all presenters so I would get slot a week or every other week and make this aware to people that it's available . - FG4
		So, this type of zooms we are in right now and if you like, if you do zooms to other people which can tell them information about the thing we're knowing right now or you could either share your information online with other people, like on Google, YouTube and other media and stuff like that. - FG5
		I know the young ones will be different but nowadays things are intertwined, we tend to say that young ones, young ones but even the older ones are on social media 99% of our time and that is free, so I think while we can use technology to communicate.....- FG10
	Sustained messaging	And I think a lot of it is, is, is to keep going with the education and I people tend to forget a lot very quickly as well, unless like they could keep getting told again and again and again. - IPPIE03
		think its education and to making people aware. And I think first time people are listening is like a repetitive thing that you have to keep knocling the door right just to. And people are knowing what's going on. OK. Understanding first on the set that, you know, what is this? No interest. And obviously second, third, fourth time might be actually makes a bit more sense. And they know what to do. - FG4

5.3 Services and research themes

Theme	Sub theme	Quote
Shared barriers and facilitators for genomics services and research	Challenges accessing healthcare services	Even just to speak to a GP is a massive uphill struggle. ..... Every time you ring, first of all you wait for hours and hours and hours, and then you ring, you then get a turn, after three, four days you get an appointment, and then you get a telephone appointment next week.....When you see someone it's a different person all the time. it's always somebody else. They don't know you. They don't know anything about you. They're not interested. And when you get an appointment, they limit you to 10 minutes, so you're not able to tell them about, you know, you might have saved up quite a few symptoms because you haven't seen your doctor for a year nd you'll tell him about a couple and he'll say, "Right, that's it now. Ten minutes is up." ..... It's un-personalised now. And this is where you need to research
		So, in terms of my experience with the GP, I believe they got very little time and a lot of patients. So, let's say, for example, I am sitting there, and I have any like, for example, they are talking to me, this is what we are going to do, etc. I have a feeling that they are in a rush and they want me to finish and go also that they can move on to the next patient. Now, from coming from a medical background, I understand that in terms of having a lot of patients and fewer GPs in their practice, etc. So, I am also empathic about their time and needs as well. But sometimes, when I have a back-and-forth with my GP, I know that it is not necessarily they are not supporting me, or they are being racist, but I do feel like they can't, unwillingly or willingly, give me enough time ..... and I am on a waiting list to receive a call and about being able to see a specialist, and just that call has been rescheduled twice, and I have just been waiting for a month for the nurse to call me to give me an appointment with the specialist..... - FG6
		No, I don't think that we are that much aware of it, because we have a barrier where people don't know that they can access some services, that they are entitled to access some services, because if you are buying your prescription, you might not know that there are certain services you can access at the hospital .....these are some of the services that you can access. So since you don't have that information, you will not be able to engage that service, so you'll not get it. - FG9
	Limited knowledge and awareness	If people knew they could help, they would, but they don't because they don't have the knowledge. Our rulers are concerned with everything, except the main things, which are health and education. That's what we got. - FG8
		Sorry, to me, I don't think most people will be aware, especially those from the minority group. Something like this, if you are not into the health system or you work in the health sector, sometimes you might not even know something like this is going on, because if anything new come out, your GP might not even, they don't even have time to even tell you that this is what's going on. - FG9

Theme	Sub theme	Quote
	Literacy skills	Uh people don't have as much resource in terms of, you know, people's literacy skills are much lower, so they don't have the capacity necessarily to read up about things or full about things. - C07
		The second big problem is that sometimes a lot of people don't read in their language, so you sort of start translating things like surveys and participant information sheets, and sometimes that's not even helpful, because if someone not reading in their language then that is, you know, that's not helpful. - R03
	Socioeconomic factors	Um, I think in general, if you like for me, if your specialist centre is in central London trying to get in and paying for all of that, taking day off for work, that is a huge barrier to people who have, you know, who, who who, don't earn very much and I'm so that led us to actually doing what they call a what we call the hub and spoke model where we had spoke centres that sort of closer to where patients were, so that to to to kind of close that that gap . - C04
		One thing I've known that is critical is that an African, an average African who comes to this country and there are differences between the Caribbeans and Africans, even the Asian communities. It takes four times in terms of effort to survive in this country compared to a Caucasian counterpart, so most of the people are really preoccupied with survival in this country. You will find them having four jobs, three jobs or two jobs and the shifts and the language, the shift of booking it. The issue is that as much as there is research out there, even the information that goes back to them. "Because I'm from Amazon and I work at night and during the day I have another shift for four hours, then I have to sleep." It's a constant, the jobs and the tasks in there, heavy lifting things and people get tired, people are struggling with sleep, it's another mental health, so the issues around the survival, it preoccupies them much more than the Caucasian counterparts. Even if you sent a link to participate in research, engaging with that link alone and with this and they don't have that time. It's about the shift, it's about money, it's about the letters, the council tax dropping in there, things like that. - FG10
		Some of the genomic laboratory hubs cover really vast amount of space and they can be quite rural. So if you have to go to your nearest kind of tertiary hospital, it can be miles away. And if you don't have the financial means to to travel across the country to your nearest hospital that's gonna allow you to have this test that might allow you to have your treatment be tailored to you and you don't have the financial means to do that then you can't access that care. - R02
		I I will say sorry, I will say in terms of when we are asking patients during the 100,000 genomes project to come for an appointment because that was a face to face appointments initially there was the thing like about money for transport. So we did reimburse patients now, not everybody asked for it. So those that did have problems, we did reimburse their travel costs. - C01

5.4 Language themes

Theme	Sub theme	Quote
Language	Accessing interpreters	Even the fact that I guess most of genomics is obviously really complex to anyone anyway. And so when you have to convey really complex information to people, for instance, who don't have English as a first language. And I might need a translator. Access to translators is sometimes difficult. - R02
		Language is a huge problem, so I don't know about what's happening in other parts of the country. But since COVID in our trust, we haven't been allowed to have in person interpreters at all. So that's hugely difficult because you're trying to explain complex concepts to people without any language support. - C07
		You would quite often have couples come in with the woman didn't speak any English and you'd be saying to the man and he'll go. Yeah, yeah, it's fine. I'll tell her afterwards. And I'd say, well, no. Can you tell her every sentence as we go through? And I used to think I'm not conspiring with this. I I kind of. Need her to understand as much as you do as we go through. - C02
	Translation accuracy	If I'm thinking about language, I know that we use translator. The concern day for me is to those translators how much do they understand of what we asking them to translate to a patient? Umm so, so and and so. Is that why we're not getting so many? Because the information that's been translated is not being translated correctly and you know so. So there's people are still confused a lot. About what it might mean for their families. - C01
		the translators; some of them don't do a good job either, so it would be great if translators are also quick with knowledge. They're not just translating the direct conversation. - FG8
		Sometimes the translators don't actually have genomic understanding or literacy themselves. So then kind of having to explain these quite complicated. It's complicated information to parents. You wonder how much of that information is really getting through to help them make an informed decision - R02
	Language impacts engagement	Can I say something as well, regarding use of languages? You know in medicine and in the medical world, the language is a bit different, so on top of, as the participant has just mentioned about English, on top of the names and words being in English, they're in a medical English, so sometimes that is even more difficult. Even if you speak and know English, sometimes the medical terms used are quite difficult, so that's one point, so if there could be some help of translating the medical words into the layman's language that can help. - FG10
		For different communities and how they conceptualize them different disease definitions, how you conceptualize, and you know the language possibly isn't, terminology doesn't quite translate, there might not be the words that you can use and so you know, in that way you can see engagement falling off or not being encouraging engagement - R01

5.5 Mistrust themes

Theme	Sub theme	Quote
Mistrust	Fear and suspicion	And are we aware of people's fears? Trepidations pre-existing traumas, that they feel they've had, or historic trauma that they feel they've had? And I don't think that we know enough to do anything about that and there's maybe not been the permission to focus and I don't mean anyone to know you're not allowed to do that, but it's just been on the numbers to some extent. So with the 100,000 it was, we've got to get 100,000. So just the people who say yes. And there is no investigation into so why did some people say no, why were we not having some people present? So for me with precision medicine, it's great. But there's a bias as to. - C03
		Historic level of mistrust with healthcare professionals. I think that there might be an element of people wondering why do you want that information? What are you gonna do with it? Are you gonna not offer me something? Are you or are you gonna do something different to me? Because I'm from this background. So I guess what needs to be done is there needs to be some kind of. Counselling that goes with asking those questions and explaining exactly why you're asking those questions And so that people So you can kind of allay people's fears about why they might be having to disclose that information. - R02
		Maybe the Arabs are scared to share their genetic map with the BioBank and the health system - FG3
		"P013: We don't want to disclose information or share information. P014: Yes, exactly. P008: You know in movies? You might use it against us. - FG2 "
		I think one of the biggest problems is lack of trust, for me, I speak from the experience of coming to health, I've worked with Caribbean and African Health Network, the populations there begin from a point of suspicion, not a point of belief, it is a point of suspicion. "Why do you want to take my blood first of all, am I very sure that this sample is really meant for that ....?" - FG10
	Acknowledging the past	Don't pretend It didn't happen, you know, or the other one is like, ohh. But it wasn't us. It was you. It was a doctor's. It was the institutions, you know. Don't. No, no, it wasn't us. It it was, you know. So I think it is it has to be. Recognize not just spoken about, but understand. You need to explain what the differences now to. Then what's the difference? - Public stakeholder interview
		And we should address the past. People shy from what happened. So let's recognise what happened and we say, "That happened, but let's move on and build confidence that people actually participate, and make research fun, and have our voices there." . - FG9

Theme	Sub theme	Quote
	Government policies and politics	but thinking about my interactions with. Lots of people, and I don't know if this has come across in the interviews you've done so far, but for a lot of people, I feel that the starting point of genetics is a very, so they associate genetics with the Home Office, immigration, genetic testing, that sort of thing. So they have in their minds, they're very negative connotation of genetics. And it's all. Sort of very authoritarian. - C07
		Born from the overseas policies of western governments, how they are one-sided or the double-standards they employ. mass atrocities everywhere, artificial sort of wars are create.....Yeah they're suspicious.....That is closer to the psyche of the local community here, that we're not being treated as equal human beings, because people like us, we are treated in a bad way overseas, and perhaps they don't think much about us either. - FG6

5.6 Representation themes

Theme	Sub theme	Quote
Representation	Diversity in materials	Wow. Now for me, based on my experience of recently attending the XXXXX in XXXX, we, as I said I was the only non white in a room getting a presentation from a white presenter using information on leaflets or patients white patients. And or or white people. And I sat there. I thought, well, you're not really targeting. I I why would I listen to you? Because I don't relate to you. Don't look like me..... why would I, as a patient, look at all these leaflets about a condition or or or genetic condition or medical condition if it only has white people on those leaflets, so therefore it doesn't affect me as a non white person because it's only white people I'm seeing that are getting affected by this. It's targeted to white people, and I'm talking on a colored thing. - C01
		Then you've got things like, you know, the images that you might use so often you sort of look at promotion materials and things and uh, we sort of too focused on images that reflect sort of white populations, sort of. And what we might call, you know, a normal family, mum and dad and child. So I think you have to be really careful there as well that you know, if you're representing a white family, maybe people that aren't white don't necessarily feel that that research is for them at that, that it's sort of it's relevant to them. - R03
		So, when Coronavirus was around, so the council asked me to do a video and it was very well seen and it was very well shared as well. So, a video in Arabic, two minutes. Just giving the latest updatees. So, everyone really interacted with it. And when you see someone from your group speaking your language, they feel valued and they feel like it goes a long way - FG2

Theme	Sub theme	Quote
	Workforce diversity	The ethnic disparity of the genomic workforce is also like a really important part of the conversation. So being the only black genetic counselor in every service that I've ever worked in, it's a micro view of actually what genetics looks like in terms of workforce. And there are other workforces around the world that have done that work to map that out and we've never done that in the UK. So it's kind of just anecdotally that we're always like we know that this is not a representative workforce, but yeah, so there's that as well. - C08
		But we know that these people come in. I do not speak Indian, I do not speak Pakistani or any of those languages, but I find a way of connecting with those people, and a lot of times just showing kindness, just giving them, like here, making them know someone is listening makes them feel really relaxed and stuff. I had a patient once who told me that they look down on the coloured people. I'm sorry to say this, I haven't experienced that, but of course I don't think anybody's going to do that to me, but I've seen a lot of coloured people being treated like they don't matter. - FG9
		Yeah. And I think the other really important thing to say about this is. That we don't have enough researchers from minority ethnic groups actually doing research right. And I think there's, you know, I mean, I think there's something to be said about somebody approaching you if you looks like you. Right. You understand has the same background as you and saying, you know, this research is really important to me sort of, you know, a white person sort of trying to convince sort of a non white person to take part in research. So we need to be better at. And promoting and sort of recruiting researchers from different backgrounds cause if we also overlook the same, then I think we're less likely to to get that kind of... - R03
	Representation of the messenger	I also think the messenger is important. And if you honestly, if you were a white academic, I'll probably having a different conversation with you. Yeah, even whta happened in the Caribbean community with Windrush and everything people sort of rock up thinking on, yeah....., actually, we're not gonna talk to you. And I think that's what the problem is that the professionals, professionals don't have an understanding of who they're talking to and that we, you know, we don't have this. It's called colonized mindset we we do have our own history. We have our own conversations and this is all communities. Public stakeholder interview
		And maybe I think it depends. Maybe just depend perhaps if the person they have, they have the pressure is same doing the talking. I feel maybe from seeing my background then that might, that might encourage people and you know to learn more about don't know and it depends as well on the on the supposed depends as well on the pitch you know how they approach it as well maybe if they you know. - FG4
		he's talking about people coming from the same culture will be better for them, like, more comfortable and they can speak freely. XXXX is saying that people from same ethnic group will communicate better with each other rather than with an outsider - FG5

5.7 Service themes

Theme	Sub theme	Quote
Specific barriers and facilitators to genomics services	Awareness of cultural and religious differences	And the way that we describe what previous generations have been affected from or died of is different depending on how we talk about health and disease. So talking about, you know, having a broken heart or talking at, you know, I mean like, so there are other ways that we talk about pain and experience of kind of pain and disease that aren't recognized in a Western context. So that can be if you don't understand that, that can be a barrier to getting information. - C08
		And so I think there's that and then, you know, there were different stigmas for different groups around disease, around passing on of disease. So kind of the the guilt and blame and shame that goes along with disease and genetic disease. These are feelings that all cultural groups experience, but they do different things with those and are supported to manage those in different ways. So you kind of have to understand that really well. And that guilt in, you know, a Muslim family might look different. And guilt in a middle class white family and how you work through that so that people can access services is different. - C08
		And that's and so I and then also about this whole notion of, you know, this issue around cousin marriage it's a very sensitive subject and I thought why, why is it a sensitive subject? So we when you unpick that it's not sensitive to the communities that actually have cousin marriage, which is actually 1/5 of the world's population the fact that's not known very much. It's the sensitivity that comes in when you put a dominant culture that thinks it's incestuous next to a culture that thinks it's not. But it's the sensitivity is not there, the sensitivity is actually making the dominant culture because for them it's taboo, it's incestuous..... - R04
	Healthcare professionals knowledge and confidence	Healthcare professionals also are massively under confident in talking about genetics, #1 and then in terms of their cultural competence to talk about it with people from different backgrounds and people are so in this culture currently people are so risk adverse that they would rather not say the wrong thing rather than try and engage in in what they perceive as being a potentially difficult conversation. - C07
		And so now, looking at everything, it's it It's now that on reflection that I'm looking at it and I'm realizing, OK, theres major issues here for that is that for me to even just talk to a nurse about genomics and I mentioned the word genomics to any nurse, I say any to majority of the nurses I've spoken to It's like completely like ohh No Fear, or otherwise they haven't heard that term before. They don't understand it. So if medical people have that issue, then how will we broaching that with the Community and with the public. - C01
		I think they. I don't think they think it's necessarily relevant, and I think they think it's more work. - C05

Theme	Sub theme	Quote
		I've got so many friends and colleagues who are still in clinical practise and the responses are struggling to keep our head above water, just doing the bare minimum and even then sometimes not even getting the bare minimum done. I don't have the headspace to think about anything else.- C06
	Healthcare professionals biases (e.g. stereotyping)	I mean, there's a lot of issues, so if so, GP certainly can refer. But they're not that familiar with the referral pathways, and they also. Have it in their minds that people from certain backgrounds won't take up testing or won't attend appointments, and that's kind of true. So you can kind of understand how that's come about because actually they do find it difficult to attend appointments and they may not take up the offer of testing. So you can see how that's, you know, people think this is not that relevant to this group. So I think, you know GP's, big issue midwives, enormous issue, so you know midwives see women from these communities over and over and over again but they're not that confident in terms of talking about issues of genetics with them and they also. You know, they also very strongly are kind of set up to think that these people will decline this. So it's very unlikely that they'll take it up. So you know that also creates a barrier. - C07
	Gatekeeping by healthcare professionals	The GP always refuses to make an analysis or x-rays even if the doctor says so. There should be a referral if it is a critical or important issue. If you want to make an analysis but can't make it if you want to make it they always refuse to make an analysis and X-rays. - FG3
		I guess from a from a clinical role that you know genomics and genetics they're specialist services. So there were a lot of gates that you need to get through to access the services so that can have a very big impact on people who come from kind of different communities, marginalized communities, lower socioeconomic groups, people with language barriers, people with disability. So there are all of these things that actually make the system of getting into the system really quite difficult..... - C08
		And I think they were judgmental. They've got a criteria that if you're ABC and you're a certain age, NHS don't, you know, we won't give you this. We won't give you this chemo. Whether you're capable of tolerating it or not, you know, not, you know, your date of birth is this. That's why. And I see that as discrimination actually. But that's how they do things.- Public stakeholder interview
	Negative feelings (stigma, shame, fear, guilt)	I just wanted to add on something. I think conversations around cancer and issues that are terminal, we need to have a different conversation in the community. The two things people don't talk about easily within my community background is around sex and death. So we need to find a language where people – people associate that with fear..... And then there's this thing about cancer that there is association, cancer and death. So when people say, "When I know, I'm going to die." But I think the community need to know that cancer medicine has gone really ahead, and early diagnosis, actually many, maybe we need many live examples who say, "I diagnosed in this month, and look, I am still here," that people don't actually associate cancer and death. - FG9

Theme	Sub theme	Quote
		And there might be some sort of social and cultural factors. So for example around stigma, shame, in particular with some genetic conditions that are obviously inherited, say, passed on through families. So I've definitely encountered this situation sort of through my research, where you might be doing research on a condition like sickle cell disease and you might have less input from fathers because there's a sort of certain stigma with having particular mutation associated with the condition. And there's obviously a lot of work that's been done around sort of in Asian families, again, around guilt and shame. If a condition's been passed on and sort of blame on the mother for sort of having a child with a genetic condition. And that's obviously also particularly the case if it's an genetic condition where it's passed on from the mother to the to the son. So I guess sort of issues around sort of, shame, guilt, people maybe. - R03
		there is a lot of prejudice and stigma, so a lot of people, the number of people that I keep coming across that get told. And it's because you've married your cousin. If they're not married to the cousin to their cousin, it's because you live in the same area you live in the same village. And then one person just. She said I wasn't even married to somebody in the same village. It's bad luck. - PE03
	The implication of testing outcomes	Don't want to have genetic testing and access genetic services because they're worried about what they might find out in terms of Whether they passed the condition on to their child and that then in certain communities has issues around sort of you know whether you're, if you're if your child is known to have a genetic condition, will that child be able to like, find a partner and get married? And is there shame on the family because it's considered, you know, something that they did wrong or, you know, then it might be. And I'm guessing religious families, you know, whether it's punishment from God or something that they've done. So there's all kinds of these kind of social and cultural barriers, I think, in a lot of populations around accessing genetic services.- R03
		Sometimes they don't want to know [laughs]. So because my mum used to say, "If we doesn't know, it doesn't kill you," [laughs], so sometimes, yeah, yeah. - FG9
		Also other things that might be revealed From from the from, from the analysis you know, could there be a you like opening a Pandora's box? That might be the other thing? Yeah, I yeah. Yeah. And and I suppose You know, knowing more information is that how does that affect? Your work your sort of insurance. Um. Even people's ability to get married or be um matched. I think sometimes that that's also an issue. So like a bit of it, I think it's also the information that might come out of. - C04

Theme	Sub theme	Quote
		So it does affect your process clinically, particularly for genetic counselling, because I'm having a conversation with you about whether you want a test, whether you don't want a test about how you would feel if you get a result, if you get no result, if you get a result that is uninformative, and these things help you make a decision about whether you want the test at all. And that's different based on whether or not your ethnicity is going to have an effect or your ancestry is going to have an effect on whether you get an uninformed result or not. So it may be More negligible for some communities than others. - C08
	Waiting lists and a busy NHS	So it's one thing to be given access to the test, but it's also. People need to be given a realistic estimate of when, though, that testing result will come back through. - C05
		But it's, you know, like as I said, that conversation yesterday that I had with a senior leader that you know, that was absolutely their attitude was this is a complete waste of time we've got other things that we need to be doing we're really overwhelmed and I can't believe that you asked me for this half an hour meeting and that's really difficult - C07
		And that's the issue then, because a lot of tests that they're so expensive. So a lot of the genetic tests that are available are and like so routinely. If I wanted to have a genetic test for a condition that's in, my family would be 18 months or two years, I think. I'm sure last week she was saying it's gone up even further to have a routine test. - PE03
		Yes, and we need to check the analysis and check-up analysis, and they are still putting us on the waiting list, even the dentist. We didn't see any of them till now, and we have been here for 3 years now. - FG3

5.8 Research themes

Theme	Sub theme	Quote
Specific barriers and facilitators to genomics research	Lack of diverse data	And then I suppose in research there is that And you know, I think it's very relevant that you know, a lot of I the data isn't diverse so we don't have diverse data in genomics and so. So it's kind of hard to generalise or you know - C04
	Current recruitment methods	Yeah, I think just going via hospital clinicians, I think and relying on healthcare professionals to recruit on your behalf, I think is often quite difficult way. I know it's probably quite standard in, in the NHS. Umm, but I think again you've got that level of removal of you as the researcher from the person you want to speak to. And you also got that added bias of the person who you've asked to recruit on your behalf. Do they know fully the Inns and outs of what you're study is? You might. They might not explain it quite rright. And they also. To some extent, we'll select who they believe might say yes and might say no, and that might not be actually true.- R02
		It's just that it does work if you're well educated, if you're middle class, if you're from a Western context where you understand research in that way, then actually that process of being invited into research works for you. If that's not your cultural process of getting into research and it's not going to work. And I think that's, that's the only thing that makes it different is the process doesn't work for different cultures, yeah. - C08
	Concerns around how data will be used and managed	I think some of the reasons why is there some the reason why they don't come forward it's a matter of trust really and trust in the system and that you know because there's there's a lot of data involved in genomics, and I think people sometimes have that mistrust about what's gonna happen to all of that. And it's a bit like not just knowing what a person looks like, but it's like, you know, your your whole entity is becoming sort of analysed and and you know other things that might be discovered. - C04
		I know from speaking to other clinicians and interviewing other clinicians on their experiences there is a sense that certain communities feel that I was just talking about that, that lack of mistrust and that not necessarily understanding what their data is gonna be useful actually, for instance, an interview that I did with the parent and the other day when I asked about whether she would be willing to Keep her DNA samples on record for and you know to be used in ongoing research she just said absolutely don't. I like, I don't know what they're gonna do with with my data. And that was apparent from a particular minority group. And that was a very different reaction to the majority of the group of parents that I've spoken to who had come from predominantly white group. - R02
	Increase awareness of research (talk to people)	But if you told me you need information about the baby in this age or the respiratory system or this virus, actually, yes I will come. - FG2

Theme	Sub theme	Quote
		You know, but I know that I think it's the control who's doing it. Yeah, it is. Who's doing it? Who? What's their motivation for doing it? I don't like being under the microscope all the time. Somebody elses microscope all the time. I don't like being tinkered with all the time and being a subject of experimentation, you know? And I think that dehumanises and makes you feel very inhuman. Mum had that when they when she what really upset Mum they wanted her to have access to her tumor, you know. And she that really upset her because it it made. Well, are they treating me or not? They just want my my body parts. My, my tumor. That was so rare. But that really she didn't even fill it in. She just totally disengaged. And that's when I kind of took over. She completely disengaged and they didn't approach it just came in the post I'll you know your tumor is blah blah blah we'd like your tumour and La La La La la and it was nothing about her it was all about the tumour and I thought that was really upsetting so yeah she just completely disengaged. - Public stakeholder interview
		I mean if I sort of reflect back to sort of some of the other work I do, I think when I reach out and invite people and sort of, I'm able to sort of give that one to one Uh discussion around sort of you know why we're doing this research and why it's important. Then I think you actually get quite good engagement. I think maybe the issue is when you just sort of advertise things when you aren't able to have a more nuanced, tailored individual conversation with somebody it's much harder to recruit, but I think once you get to the point where you are able to sort of teach somebody about the value of what you're doing, then usually people are, you know are very positive - R03
	Close the loop	So it's continuous, but also you know, we're looking at research projects and we're asking people to take part in research. We ever actually give feedback to anybody about anything that we get from our research projects, so you know, so thinking about that as well. So we need I personally think we need to engage and work together or collaborate with strategy, whether it's a research proposal but also make sure that they are informed of the outcomes. And then we need to go back and evaluate and see how that worked. If it's not worked, then what needs to change? What? What's still going wrong? So that's what I think. - C01
		It's not like exactly today. Like incentives and the hiring, everything. This is what keeps us. If we have the right environment, we'll have the right interaction. And the most important thing that you keep us updated. So right now, after this, what will happen? You come back to us and tell us .....because we feel like this will encourage us to take part in further research because we feel like we're valued. And not only you want this for an hour and that's it.- FG2

Theme	Sub theme	Quote
		So sometimes with all this research, all this programme, when we've finished, if the outcome is being communicated to them, like we came to this group, this is the information that we had, this is what we've done with the information, and then when they see the results, maybe sometimes, from the next level, it might change their view. But if they come in, collect the information, and that's it, they don't see anything with the information that was given, and you come back again and ask them something else, they will not do it. They will not take part. - FG9
		Feedback is very important. It's very important and I think, but times, regrettably, you can't feed back as much as you would like to because they what happened when you search quite often is that you're funding runs out and you have to go on and work on another project and you can only feedback and disseminate at the end of the project. So you've always got to watch out for that, but you actually do do that at the end of the project. And if you do that or or other things that may come out of your project much later a couple of years later and you don't have the funding to send all that stuff, you know, all all all of them. So those can hinder it, but I think that is 1 aspect of umm research output which must be taken each. It's very important to take that seriously because you've built up a relationship with those people. You need to honor that relationship to the end and the way that research works doesn't always allow us as researchers to do that. But I have learned that that is a very important aspect. - R04
	Inclusive research processes (go to the communities)	Absolutely, yeah, that's exactly what I'm trying to say. It's not just about I translated this to Punjabi, just the just the process of doing that in a meaningful way makes people feel included. Yeah. Even if they don't even speak Punjabi, if they speak English very well. But you're from a Punjabi community and you've seen all of this stuff that even makes a difference to you, right. - R04
		Raising awareness is important, but like somebody mentioned earlier, if you want to get across to the average black man, if you can reach the hairdressers, the barbers, people who attend to them like on a regular basis, like average, normal people, it's going to be like really effective. So I think that's an approach that can be taken. - FG9
	The value of being involved and the consequences of not participating	Knowing the impact it has on your life. On our families as well. On our lifestyle, our health, our food, sport, work, etc. Our way of life. Yes, all of this would highly encourage us to involve ourselves - FG5

Theme	Sub theme	Quote
		I think the strategy is to engage with, with kind of ethnic minority groups relies on you just really understanding what do you what do they want from you? How can what they're doing for you as part of your study also benefit them? It's not about just going in sweeping in, taking all of their ideas and then not informing them about how the studies gone and not having any kind of longevity in that community after kind of just sweeping in and leaving. And I think it's important to make sure that you leave some sort of presence or some sort of some just leave some kind of mark where from where you've been and kind of I don't know. I don't know how to put it, but I guess just have some sort of Added value once you've left. - R02
		I think explain to people what the benefits are and also what the, what's the word, what what the consequences are if we don't do this. I think that's very important. It's alright to tick boxes. Our personalization, digital health, blah blah blah is wonderful. But what? What is your alternative if I say no? I don't want any of it. How would I be disadvantaged from that? Because if I see that well, there's no disadvantage to not having personalised care well, I will choose non personalized care because I feel safer that way. So I think that explain what you know why, what the benefits are, but also if you don't have it your health will suffer and I think that has to be quite strong in there to be more convincing.- Public stakeholder interview
		I I think it's like emphasising that if you, if you if you don't sound like if you it's like voting you know you don't have your say you're not counted. And so if you don't something around that I mean I'm sure they're more behavioural scientists, people who who know how to go about this but you know, how do you how do you motivate people to to believe in that that you know if if you don't participate if you're not part of it, that actually it's harder for us to harder to provide the right services - C04

5.9 Community engagement themes

Theme	Sub theme	Quote
Community engagement	Sustained engagement	I have a concern that there is a lot of focus and you know is concerned that that a lot of genetic studies, especially being done in European populations, white populations and now the focus is are we need to engage with minority populations and to some extent to me that comes across a little being a little bit exploitative as we don't have the data. Wait, you know you're interesting and different. -R01
		Umm. As we start seeing what's happening or if we, for instance, start realizing that in a particular area where we're expecting a certain population to participate, we're not getting many of them signing up many people from that particular group signing up. Then we have to be able to say We have to go back to the drawing board here. We have to go and figure out what's happening because we have, I think, a moral obligation if we if we're expecting people to participate and we're inviting people to participate, if we find they're not participate and we have to be able to react to that, if that means going back to the drawing board and replanning, even though we've already spent a year of planning, then I think that that's something that we need to be willing and prepared to do. - R05
		Yeah, I just wanted to add on these conversations around trust is a big thing, but I think we are coming every time when we want to collect information or data, then we start talking about trust. Let's keep on engaging the community; that they understand what we are doing, and using the voice that people really know that if we have so-and-so talking, I know that person, I can trust them. I'll listen. And that conversation goes on. Don't wait when there's surprises, an epidemic, they you say, "Okay, now you send that person to talk." That conversation should be on that people build it over time..... feeding back is really very, very important. - FG9
	Strategies to improve community engagement	So I am currently and I've just received some funding to do some focus groups that is looking specifically at the black and Asian community and my strategies to engage them will be entirely different, which I think for those communities, like I said because of I think because of that historic mistrust with healthcare professionals, it relies on you having to really build networks with the Community. I don't think you can just rely on sending out a letter sending out an e-mail And expecting people to come back to you when they don't know you at all and they don't know who you when. They don't know why you're studying what they're studying and they feel like they're just being used and they feel like it's very tokenistic. So I feel like if you want to engage with those communities, you need to get out there and actually be there and meet those communities and I think it's probably quite Often quite difficult, because in those research spaces a lot of researchers don't reflect how they look so. - R02

Theme	Sub theme	Quote
		Do we need community ambassadors where we don't have enough of us to go out and and raise awareness so we can train a kind of train, the trainer type thing, a community person to talk to their different communities about you nomics and to start raising awareness? So in in one way, I hope that means that that becomes sustainable self sustaining.- C01
		So one of the things we were looking at, XXX and I were really interested in the idea of doing like a hackathon health hackathon as opposed to an IT type one. But you could maybe join the two and get the kids who were there to talk about things like how they would like to be spoken to and engage with. And so to work in their own communities about engagement. And also potentially start looking at setting up some tools for genetics, genomics, family history, things, how they would work in different places. So we're talking about doing things like that. We're talking about, you know, having QR codes on post because they were doing it for the. Ohh, what's it called? The The Commonwealth Games? They've got QR codes and there was we were saying that they would be useful. You know, you scan a QR code and you can find out about. Such and such on your phone, so you don't even have to go and ask someone, but you find out where you go and it's maybe less invasive for people to do that And so that's something that we sort of got that we would like to maybe try and work on. - C03
		I think we learned that in the vaccine hesitancy issue that the messenger is very important and it's also the respect the messenger has in the vaccine hesitancy we had, like a campaign with I can't remember that this dreadlock academic, and that's done a lot of black history stuff on television like Lenny Henry. Some of the actors were also doing that with prostate cancer. You're getting people that are respected to speak to those communities. I'm sure that's happening in other communities that I'm not aware of, but you get people that are respected, that are cool and you know that deliver the message and you get that. Even in America, you get it. In politics you get uh Oprah Winfrey supporting Barack Obama. And you know what I mean you get. The messenger is important. - Public stakeholder interview

5.10 GMSA equity of access themes

Theme	Sub theme	Quote
Monitoring equity of access in genomics medicine services - the barriers and facilitators	Current data recording practices	The other thing is that when you are admitted into hospitals, you know you go via A and E and the and they'll ask you the question, will you take the paperwork that you get given the the collection of of the EDI information, there is actually a category on there that says not asked. So why is that a category? Do you know? - C01
		I I think the simple answer there is gonna be that that the current state of the recording ethnicity data within certainly within the GMs regionally it is is poor at the moment. It varies across the patch..... I would say we've got about 20 to 25% confidence that in the recording of ethnicity which which is poor..... - GMSA FG4
		Just as an example, we've had people and you know we, we we get them sometimes we used to get them to sign a form in which is your ethnicity and my ethnicity is not on here because it's none of these and there are people that, you know, we did some work on it about trying to increase the number of codes and things. So even if we all had the same codes and the same the same things are because there's a lot more than I think we capture. And there are a lot more ethnicities than we know. So I I don't know. Yeah, they're some of them are very broad. - GMSA FG2
	Current testing order process	There are issues of the numbers of staff and the type of staff at every single aspect of the whole of the system, which makes it clunky and because everyone is under time pressure and there's an increasing backlog of tests that need to be reported on from the clinical scientist at the GLH. Anywhere where they can shave two or three seconds off per request, they will. And of course, if that includes the EDI data, they're not going to look at it. - C06
		Yet just again to emphasize XXXX's point that you know most. So if we go back and look at the labs, most test request forms do not have ethnicity as a an item as a data item that you have to fill in. - GMSA FG4
		And I think the other really important works change that I haven't touched on yet with you. The genius got a massive IT team working on both interoperability and order comms because we recognise that we've got a what's recognised across the world has been a world beating service and yet we're in the dark ages having a paper system. So. There is and has been a lot of work ongoing around eliminating that paper requesting system so that it becomes electronic. - C06

Theme	Sub theme	Quote
	Data sharing, access and governance	just about to mention that actually one of the biggest barriers that we have is the information governance side of things and data sharing agreements. So as DS says, it feels like the data is being captured at multiple points. It's not readily available to those who need it, so we know from a PHP from population health management perspective there was a lot of work that was being done by geographies around local healthcare records, et cetera, et cetera. It's unclear the extent to which genomic data is incorporated in that particular data. So it, yeah, it's just a mine field. I mean even GMSA to GLH data sharing in itself is a nightmare, so we're having to do a significant amount of work arounds if if we're lucky to to have access to that data. - GMSA FG2
		Yeah. And I think the the main problem we've got, is from a data perspective, because we're because of the cause of the GMSA being seen as a specific entity and not a legal one. It it is a bit ridiculous that we can't actually as individuals access the GLH data without having some sort of data sharing agreement, which is something that we need to work on and and it's an issue nationally and I know I think it'sXXXX have done a bit of work on that themselves and they still haven't solved the problem, but they're trying to put in place kind of agreements where it is reasonable to get that sort of level, you know we can get very, very high level data in terms of you know a number of tests versus in terms of indications and where they might have been referred from both if we want to get deep into the ethnicity. - GMSA FG1
		we've been working to try and find a way of recording equity of access and and seeing where you know where maybe we have gaps in our data, data recording, data access or whether there's, you know, how can we kind of tie equity of access Into projects that we want to deliver in the future, so that essentially we can have a piece of work that is accessible to us as a GMSA that we can essentially use to Guide our decisions and also support decisions and helped us to kind of engage and they be groups that we haven't been able to. So we've been essentially trying to kind of find that data. - GMSA FG3
	GMSA EDI strategy development	Yeah so what we've done is we've meeting with the few people firstly in the first instance to see what information that we need to be aware of or what we need to understand about EDI now, because it's such a huge field we are, we've decided that we would use race and ethnicity as our focus because with that they are the issues of the socioeconomic issues, the disability issues, and so it will come in, but we that's where we started in at a first point I think. So we're trying to make contacts with different people. We've met with the Caribbean, African and African Health Network Group as well.....and we've also because of the work that we're doing with ovarian cancer, we have got information from the the ovarian cancer charity as well as a link with Macmillan Nurse that's doing a lot of EDI work. - C01

Theme	Sub theme	Quote
		we pioneered having an an EDI strategy implementing is another thing I think. And it's difficult because sometimes right because we're an entity, but we're also hosted by trust and then each trust has their own EDI, is it, you know, to try and type tie them all together and then I think we've had difficulties getting a chair or the actual EDI sort of team and strategy to implementation. So and then in XXXX there was also a there there there was also a Co creation, again the EDI the strategies are there, Implementation I'm not entirely sure how. - C04
		we are a point of trying to we're just about to agree our strategy for next year on EDI and in terms of how we're going to manage that we are we're creating a post so we're gonna implement a workforce development manager, but they they're they all have a responsibility for implementing the EDI strategy so. They will do that across everything, all the all the parts of work that we're doing and working in correlation with the PPIE panel that we have as well. - GMSA FG1
		So I've been in post for 15 months, one of the first things I I looked at was looking at an EDI strategy for the organization. So one of the ways of doing that was like what, what, what our aims and objectives or some of the services we provide. Looking at some of the national regional projects. And really, how would that strategy sit in terms of the GMSA and how do we then engage with organizations and partnerships? So I I looked at it from different perspectives, but it was to really develop a strategy which kind of sat to the heart of the organization in terms of the GMSA and then how then do we translate that? Because I wanted it not just to be a document, I wanted it translated it into a practical action.So we started to kind of roll that out. So the the strategy really it was It was really thinking about, uh, looking at the our aims and objectives are what's our vision for the GMSA? Some of our area work, what what, what do we want to achieve in the next three years of that EDI strategy? Who do we want to engage with what other some other areas that we need to concentrate on around equity of access. - GMSA FG5
	Collaboration	I think we've all articulated that we would all like a common framework to, work with and then we would like to be able to access access data iin if you like that would allow us to look at all of those particular groups that we want to be highlighting within that EDI framework to to make it obviously entirely relevant to the local populations and areas. I mean, we started to do this just dipping our toe into deprivation and we've managed to get absolutely nowhere over about 18 months simply because of IG issues and accessing data and data sharing. You can do it if everybody's literally sitting in the same department of the same trust, but if once you want to start sharing that out and so the the structure of the GMSA is because we work across 30 trusts immediately builds up an almost impenetrable barrier for data sharing and information governance. But we would like a dearly like a standardized sort of framework nationally, and perhaps a standardized data set where all of this could be generated. Big ask I'm afraid. - GMSA FG2

Theme	Sub theme	Quote
		I do work quite closely with the XXXX because on the on the, on the EDI perspective. And I think there's only two of us at the moment, so I worked quite closely with them. We do share it in information. We do share ideas around that, so. - GMSA FG5
	Projects and initiatives aimed at addressing inequity of access	So basically it's the primary care networks of in the GP they given an invitation, have they got concerned about breast cancer in their family? Would they like to fill in their family history questionnaire and then they, you know, they have an option straight to a pathway? And it's gone really well. Everything's good and bad, isn't it? So I can't say they haven't been problems cause there have. But I think the actual thinking behind it was brilliant. I think it could evolve.... - C05
		So that we work with we we've looked at it, it currently from the census information we we've drilled down to both ethnicity and also a spoken language at the local authority level across our region which is very interesting and very revealing because if you look at it from we we took like a top five..... So I mentioned before about spoken with the use of spoken languages, so that particularly has already started now to be used by the by our comms team and also particularly by the by the maternal work stream specifically to look to start producing both web and printed contents in in languages that are indicated as being, you know the best value for money if you like. Based upon this data - GMSA FG4
		It's around genes and health. So when we do each of the workshop, we do a presentation. So we give a background in terms of what the genes and health is because obviously Pakistani and the Bangladeshi Community, it's about raising awareness of what the genes and health project is and and also giving the participants that information to make that decision themselves. But it's also about we have colleagues from the trial and from genomic medicine as well. So we have a team that's there to from the trial department as well from the genomic medicine to to take those samples. But it's all done on voluntary basis. So there's no real pressure in terms of we leave it to them, but we do a presentation so they have that information and and then they can make that decision themselves. - GMSA FG5

5.11 Workforce training themes

Theme	Sub theme	Quote
Workforce training	Genomics and precision medicine training for healthcare and other professionals	So I think you know in terms of healthcare professionals, I think a short module which was incorporated into mandatory training for everybody. So whichever trust they joined in the country, they would have to do that I think would be really, really helpful. It's not rocket science doesn't have to be complicated, it just needs to be a couple of very basic you know very, very basic sort of foundational sort of points. So I think. From a healthcare professional point to you, I think that would be really helpful. - C07
		Yeah. And I think just the cross cutting nature of genomics, it is impacting on so many different services and so really supporting clinicians to become aware of genomics and what it means for their services and then being able to communicate that to the patients that they see in a culturally competent manner. I think is a challenge as well because it's almost like some of them are quite new to genomics and it has to be across every level I think of the clinical team, it can't just be the kind of oncologist it has to be. Anyone who might get asked a question, and so the work that the national Genomics education team, formerly Health Education, England are doing, is really to develop that just in time, resources for kind of through master's levels or through to kind of more bitesize learnings for healthcare professionals. - POL_01
		I think GPs should have more information about this. Because everyone is going to go through the GP one day, one way or another. GPs, or within the outpatient clinics of hospitals, there is information, there is a person or pamphlets, I don't know. That they could inform us about all this. But I think it has to start with GPs, because here we don't listen to the radio, and if we do, we don't understand either. We don't read newspapers, because we don't understand, nor television. I think this is not the solution, I think it would have to be within the GPs that we have within our communities. Each of us is registered with a GP. I believe this is it. - FG8
	Cultural and faith/ religion awareness	One thing, I don't know, maybe they have to start teaching them about cultural differences in nursing, in medicine, in the university, for them to know that we are from different backgrounds. Sometimes you don't even understand what is going on, why some people are being – you go to the hospital, you look at the nurse, and you feel like you want to go home, because the way she look at you, you feel like, I need to go home with whatever is disturbing me. You can't even open your mouth and talk. I'm a nurse myself. I've experienced so many things at the hospital, sometimes I don't feel like – I just want to stay home. But the way they will talk to you, open your mouth and you say you were a nurse, and the nurse midwife said, "Oh, are you sure?" And I was like, "What do you mean by am I sure? Apart from the nursing, I teach at XXXX University," and she was like, "Oh, okay." At that moment I just said, "No, I want to see another nurse, because I don't think I can get on well with you." - FG9

Theme	Sub theme	Quote
		Diversity, diversity. We are different and you need to respect that. If I am reacting like badly to what you're offering me, then find out, what can you do, what can you offer that would make me more at ease or help my condition better? They need to understand diversity, and that's where personal centred-ness comes in. It's not like one style fits all. We need to be a bit more compassionate as health workers and the likes, to know people are different. Yeah. - FG9
	Community engagement training for researchers	Yeah. So I'm so. What would that look like? So I think that the conversation around. And ethics and justice. Doesn't happen routinely, particularly if you're in biomedical sciences or in genetics, and there might be like a module. At some point, and then that's kind of it. And then whenever anyone talks about ethics, you're actually talking about like the ethics of your clinical study like. You're not talking about ethics like philosophy and ethics and. And so I think that therefore the, the, the complexity of how everything ties together and why what you do in a lab means something to what I do in a clinic doesn't actually get married together. So things get siloed. So they're like ohh EDI is something that Patients are doing around, clinicians are doing around their own countries, bias with patients like that like it sits over there. So I think practically bringing those conversations and into training across all bits of genetics I think is really important and we have opportunities to do that because we have things like scientist training programs which teach genomic counselors as well as teach lab people. - C08
		I don't know to what extent people. Well, what I would say is basic science within the genome, basic scientists within the genomics sort of research here to what extent they involve with or they use still familiar with sort of engaging with communities to do research and what comes across to me quite a lot is that not used to thinking in that kind of applied health research kind of way. - R01
	Importance of ethnicity data recording and having the conversation with patients	I mean, yes, but they're also needs to be a bit of an attitude shift. So as an example, we when patients arrive in clinic, the first thing that they do is go and check in at the reception desk. So they have to say what their name is and what their date of birth is. And then someone takes them off. And so there was a period two years ago where I was really pushing on this. And I I was. So there was two questions that I was keen for the receptionist to ask. One was around what do you consider to be your ethnicity? And the other one was around is there currently a family, a social worker involved with your family? So those were two questions that I said like, let's just ask them routinely to everyone and then that just makes all of our lives so much easier because I think it makes a big difference in terms of collecting data. And our reception staff absolutely freaked out like they just couldn't cope with the idea of asking these questions. They all got super anxious about it. They all, you know, they all went to their team leader and said under no circumstances can they do this. It was all absolutely they felt that it was completely impossible to do that and and so, you know, it's making people feel more comfortable that they're able to do more things. ..... That they think, is that they think is sensitive, but I think it needs to be normalized so that people do you feel comfortable talking about it because you know, there's no point, skirting around the subject. - CA06

Theme	Sub theme	Quote
		Well, I think one of the things that we've had a discussion about this within the GMSA as well. I think first of all is dp people understand why we're collecting that data. Is it important? Because how important it is somebody's ethnicity. If you're looking at a genome, you know it doesn't really give you that information. What it's giving us information, all those other factors, those socioeconomic factors that, that probably that can show a prove that there is a a, a barrier, umm. So it's understanding the importance of the data, but the other thing is and and this is partly based on personal experience and also partly based on on, on different discussions we've had is people are not comfortable to ask that question. .....I think what most of it is is like how do I ask that question without coming across as being?I don't know racist or didn't discriminatory in some sort of manner, so it's it's thinking about how do we I'm gonna say educate or or or or or explain why it's important for you to be able to also explain to the patient why you need to collect that information. - C01

APPENDIX 6

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