NHS England: Equality and Health Inequalities Impact Assessment (EHIA)

A completed copy of this form must be provided to the decision-makers in relation to your proposal. The decision-makers must consider the results of this assessment when they make their decision about your proposal.

1. Name of the proposal (policy, proposition, programme, proposal or initiative): Clinical Genomic Service Specification Revision

2. Brief summary of the proposal in a few sentences

Clinical Genomics is a rapidly evolving, vibrant clinical specialty providing clinical care through diagnosis, risk assessment, treatment and management to individuals and families with or at risk of genomic disorders due to changes in chromosomes or specific genes.

Since the publication of the last service specification in 2013, NHS England have formed an NHS Genomic Medicine Service (NHS GMS), building on the NHS contribution to the 100,000 Genomes Project. THE NHS GMS is comprised of seven NHS Genomic Laboratory Hubs (NHS GLHs) and seven NHS Genomic Medicine Service Alliances (NHS GMS Alliances) that were formed in 2021 and align to the NHS GLH geographical footprints. The new Service Specification reflects the new infrastructure of the NHS GMS and the most recent developmental aspirations.

Advances in technologies and increased availability of genomic tests will impact on the delivery of a Clinical Genomics Service (NHS CGS). The new service specification addresses the need to ensure a service that is equitable based on clinical need; highlights the need for education and training of the healthcare workforce in genomics; adoption of new ways to deliver the service, and the continued and expanded integration of high quality research to ensure a service that provides patients and their families with the best possible, evidence-based care.

It is recognised that individuals and families with different genomic/genetic disorders will have different levels of need based on the severity and range of their individual health problems. Their access to services will also vary due to the different levels of service provision. Therefore, in addition to considering equity of access based on need, this document also considers equity of provision of the services.

3. Main potential positive or adverse impact of the proposal for protected characteristic groups summarised

Please briefly summarise the main potential impact (positive or negative) on people with the nine protected characteristics (as listed below). Please state **N/A if your proposal will not impact adversely or positively on the protected characteristic groups listed below. Please note that these groups may also experience health inequalities.**

Protected characteristic groups	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
Age: older people; middle years; early years; children and young people.	The NHS CGS provides care to individuals and family members throughout the life course, including from the pre-natal to end of life settings.	All NHS CGSs will have a standardised operating procedure (SOP) to ensure the transition of care from paediatric to adult clinical services and acknowledging the autonomy of an individual as they reach adulthood, which may impact on their own decisions and consideration of their health. NHS CGSs are delivered in an age-appropriate manner, recognising that individuals in families may have different needs at different ages.
Disability: physical, sensory and learning impairment; mental health condition; long-term conditions.	It is well recognised that the physical and cognitive ability to access healthcare can be different for those who experience physical disabilities, mobility problems, sensory impairments or learning difficulties. NHS CGSs provide diagnostic and clinical management services for many patients and their families with severe physical, sensory and learning	Services should be provided which are sensitive to the needs of the individual e.g. individuals with hearing impairment should have access to British Sign Language interpreters or information should be provided in easy read formats for individuals with visual impairment. Appropriate means of consultation should be used e.g. video clinics if this reduces need for travel to the hospital. Provision should be made for those with learning difficulties or disabilities to be adequately supported

Protected characteristic groups	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
	impairments. Many of these conditions are lifelong and affect mental health.	during consultations and for the information provided to them during and after clinic to be in a format which is accessible to them. Facilities in which consultations take place should be accessible to those with physical disabilities, including wheelchair users, those of limited mobility and those of short stature with appropriate toilet facilities available on site.
Gender Reassignment and/or people who identify as Transgender	There is good evidence to demonstrate the barriers that transgender people face when accessing healthcare. These can be due to stigma, discrimination and lack of awareness.	Staff can discuss genomic testing and healthcare issues in a culturally competent manner which acknowledges the requirement for information about biological sex at birth that may inform the interpretation of genomic test results. Evidence that each NHS CGS acknowledges the health inequalities experienced by transgender and non-binary individuals and those considering or undergoing gender reassignment and provide a safe environment where gender identity is validated. Education and training for staff in ensuring competency in providing culturally sensitive services is provided.
Marriage & Civil Partnership: people married or in a civil partnership.	NHS CGSs should be available to all based on clinical need irrespective of marriage or civil partnership status.	Some individuals and couples will seek pre-conception counselling to consider reproductive risks and options, most usually when they are affected by a genomic condition or where there is a family history of relatives being affected by a

Protected characteristic groups	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
		genomic condition. Provision should be made for counselling to provide information and advice.
Pregnancy and Maternity: women before and after childbirth and who are breastfeeding.	NHS CGSs are relevant to pregnant women who may have a fetal abnormality identified on ultrasound scan, or non-invasive prenatal test indicating a risk of a genomic disorder in an unborn fetus. Due to personal health, that of a partner or family history, pregnant women may have concerns about the risks of a genomic disorder for their offspring. Genomic conditions can result in a higher risk of recurrent miscarriage (e.g. translocations), fetal loss in utero and stillbirth. Some genomic disorders result in a higher risk to health of the pregnant woman (e.g. Marfan syndrome)	NHS CGSs have established referral routes into fetal and maternal medicine services to support women with complex medical needs - relating to prenatal investigations, family history or personal history of a genomic disorder. Preconception and/or antenatal genetic counselling, provision of carrier/diagnostic/prenatal or preimplantation genomic testing and discussion of reproductive options should be provided to all women where there is a risk to their offspring a genomic condition. Appropriate genomic investigations and counselling about recurrence risk and options should be made available to women with recurrent miscarriage, fetal loss in utero and stillbirth. Advice and clinical support (e.g. referral to a specialist obstetric clinic) should be provided to all women with a genomic disorder which may have an adverse effect on their own health through pregnancy.

Protected characteristic groups	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
Race and ethnicity[1]	We acknowledge the sensitivities relating to genetic information that can have impacts on individuals and families of different races and ethnicity and that inappropriate use of genetic information has the potential to result in significant harm. Approximately 10% of the population worldwide are married to a biological relative or are the offspring of such a relationship. This is termed consanguinity. There are several social and economic advantages of consanguineous marriage. However, a significant association has been consistently demonstrated between consanguinity and the risks of severe inherited genomic condition. Consanguinity is more common in certain ethnic groups, increasing the risk of genomic conditions in these	All individuals working in NHS CGSs should undergo regular ethnicity and diversity awareness training in accordance with their local Trust policies. NHS CGSs are provided to all individuals based on clinical need, recognising that some conditions occur more commonly in certain ethnic groups due to the presence of specific genetic variants. Services should be delivered in a culturally competent way with input from patients and advocacy groups with experience and expertise in these conditions. Guidance has been produced in relation to addressing genetic risk associated with consanguineous marriage for inclusion within the published operational policy. Each NHS CGS should adhere to the guidance to ensure that the genomic needs of consanguineous families are met. Services should be provided that meet the language needs of the patients - use of interpreter services and translation of patient letters into the language of

[1] Addressing racial inequalities is about identifying any ethnic group that experiences inequalities. Race and ethnicity includes people from any ethnic group incl. BME communities, non-English speakers, Gypsies, Roma and Travelers, migrants etc. who experience inequalities so includes addressing the needs of BME communities but is not limited to addressing their needs, it is equally important to recognise the needs of White groups that experience inequalities. The Equality Act 2010 also prohibits discrimination on the basis of nationality and ethnic or national origins, issues related to national origin and nationality.

Protected characteristic groups	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
	communities. (Darr et al. J Community Genet. 2016; 7:65-79. Some inherited conditions are more common in certain ethnic groups due to the presence of specific genetic variants. Some individuals of different ethnicities do not use English as a first language potentially limiting their ability to access healthcare services. Lack of appropriate laboratory control data from individuals from different ethnic groups can result in misinterpretation of genomic test results (e.g. Manrai AK, et al. N Engl J Med. 2016;375:655-65)	choice to enhance communication and information provision. NHS CGSs should work with colleagues in the NHS GLHs to ensure that genomic tests meet the needs of all individuals, recognising that some assays have been designed based on information from specific ethnic groups and so may not be relevant to all populations. Professionals in clinical genomics should be aware of the potential to misinterpret genomic test results and take precautions to mitigate, due to the lack of control data in individuals from minority ethnic groups which have been under-represented in sequencing studies.
Religion and belief: people with different religions/faiths or beliefs, or none.	NHS CGSs are provided to all individuals based on clinical need and irrespective of religion and belief.	Staff delivering NHS CGSs are aware of the differences in how morally sensitive healthcare issues, including termination of pregnancy, contraception and end of life decisions are considered by individuals of different religions/faiths or beliefs. Staff are provided with education and training to provide a culturally competent service.
Sex: men; women	NHS CGSs are provided to men and women based on clinical need.	Each NHS CGS should make assessments if there are any barriers to males or females accessing the Service.

Protected characteristic groups	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
Sexual orientation: Lesbian; Gay; Bisexual; Heterosexual.	NHS CGSs are provided to all individuals irrespective of sexual orientation. There is evidence that barriers exist based on sexual orientation when accessing healthcare. These can be due to stigma, discrimination and lack of awareness.	Staff delivering NHS CGSs are aware of the health inequalities experienced by individuals due to sexual orientation and are provided education and training to provide care in a culturally sensitive manner.

4. Main potential positive or adverse impact for people who experience health inequalities summarised

Please briefly summarise the main potential impact (positive or negative) on people at particular risk of health inequalities (as listed below). Please state **N/A if your proposal will not impact on patients who experience health inequalities.**

Groups who face health inequalities[2]	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
Looked after children and young people	Looked after children (LAC) and young people may suffer health inequalities. The major reasons for referral into NHS CGSs are: 1. the child has a particular problem (diagnostic genomic testing) 2. The child is healthy, but the family history raises concern (carrier and predictive genomic testing)	Each NHS CGS should have, and comply with, their SOP for the provision of services to LAC. The role of potential genomic factors contributing to a looked after child's problems needs to be recognised, but there also needs to be an awareness of the purpose, principles and limitations of a genomic assessment. Consultation and genomic testing may be appropriate in the assessment of a child prior to

[2] Please note many groups who share protected characteristics have also been identified as facing health inequalities.

Groups who face health inequalities[2]	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
	3. There is a history of drugs or alcohol misuse or infections in pregnancy	adoption where this is considered in the child's best interest. LAC should not be excluded from some types of genomic testing simply because parental samples are not available for testing e.g. exome/genome trio analysis and alternative approaches considered. The child's best interests must ultimately remain of paramount importance.
Carers of patients: unpaid, family members.	Many individuals with genomic conditions require support from family members with care and activities of daily living.	Consideration that clinical appointments are convenient to allow carers to attend and support. Information and resources provided, with the patient's consent, to carers about the condition affecting their relative(s) to support their understanding of the condition. Ensure awareness of Healthcare Travel Costs Scheme (HTCS) to support attendance at hospital appointments.
Homeless people. People on the street; staying temporarily with friends /family; in hostels or B&Bs.	Homeless individuals are less likely to seek healthcare or be registered with a GP and more likely to have mental health problems. This reduces opportunities for referral into NHS CGSs.	Self-referrals will be accepted from this group of individuals. NHS CGSs will assess the patients clinical need, providing them with relevant guidance and/or offer them an appointment. Monitor failure to attend rates in this group and act on the findings in order to improve access.

Groups who face health inequalities[2]	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
People involved in the criminal justice system: offenders in prison/on probation, ex-offenders.	Some individuals in the criminal justice system have learning difficulties and behavioural problems which have a genomic basis.	NHS CGSs will be provided to individuals with genomic conditions where behavioural problems increase their likelihood of offending. Referrals to psychology and other support services will be made as appropriate. Monitor failure to attend rates in this group and act on the findings to improve access.
People with addictions and/or substance misuse issues	Individuals with some genomic conditions have an increased risk of addiction and substance misuse.	Where addiction and/or substance misuse is identified as part of routine clinical genomics assessment, referral to expert support services should be made or facilitated through communication to other healthcare professionals co-ordinating care where indicated.
People or families on a low income	It is recognised that many genomic conditions result in physical and learning disability which lead to many individuals and families with genomic disorders having a low income.	Recognition of the impact of the genomic conditions on the socio-economic well-being of the affected individual and their family. Ensure awareness of Healthcare Travel Costs Scheme (HTCS) to support attendance at hospital appointments.
People with poor literacy or health Literacy: (e.g. poor understanding of health services poor language skills).	Letters are provided to all patients attending NHS CGSs. Information leaflets are available to users of the service. Each NHS CGS has a website providing patients and family members with information.	Information leaflets should be written in plain easily accessible language. Letters provided to patients must be in language which is a at a level commensurate with user's understanding so that they can make maximal use of it.

Groups who face health inequalities[2]	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
		Websites must provide information in an accessible and readable format. Where English is not the user's first language special care should be taken to ensure that written material is accessible to them.
People living in deprived areas	NHS CGSs should be available to all individuals in deprived areas based on clinical need.	Face to face outreach clinics should be provided where possible. Video and telephone clinics should be provided where appropriate to increase access to individuals in deprived areas. Monitoring of geography distribution of referrals will increase awareness of areas with lack of provision/referrals and allow action to remedy. Monitor failure to attend rates in this group and act on the findings in order to improve access. Ensure awareness of Healthcare Travel Costs Scheme (HTCS) to support attendance at hospital appointments.
People living in remote, rural and island locations	NHS CGSs should be available to all individuals in remote, rural and island locations based on clinical need.	Face to face outreach clinics should be provided where possible. Video and telephone clinics should be provided where appropriate to increase access to individuals in living in remote, rural and island locations. Monitoring of geography of referrals will increase awareness of areas with lack of provision/referrals and allow action to remedy.

Groups who face health inequalities[2]	Summary explanation of the main potential positive or adverse impact of your proposal	Main recommendation from your proposal to reduce any key identified adverse impact or to increase the identified positive impact
		Monitor failure to attend rates in this group and act on the findings to improve access. Ensure awareness of Healthcare Travel Costs Scheme (HTCS) to support attendance at hospital appointments
Refugees, asylum seekers or those experiencing modern slavery	Refugees, asylum seekers or those experiencing modern slavery are less likely to seek healthcare or be registered with a GP and more likely to have mental health problems. This reduces opportunities for referral into NHS CGSs.	Self-referrals will be accepted from this group of individuals. NHS CGSs will assess the patients clinical need, providing them with relevant guidance and/or offer them an appointment. Ensure awareness of relevant funding schemes that may be available to support attendance at hospital appointments. Monitor failure to attend rates in this group and act on the findings to improve access.
Other groups experiencing health inequalities (please describe)	With the increasing availability of video clinics to provide clinical appointments it is noted that some groups do not have access to video-clinics or find this way of communication challenging (e.g. elderly, sensory impairment)	Alternatives to video-clinics will be provided - either as telephone or face to face consultation. Monitoring of video-clinics to ensure that they are used as a complimentary means of clinical engagement and that the outcomes of such clinics are acceptable to service users.

5. Engagement and consultation

a. Have any key engagement or consultative activities been undertaken that considered how to address equalities issues or reduce health inequalities? Please place an x in the appropriate box below.

Yes X	No	Do Not Know

b. If yes, please briefly list up the top 3 most important engagement or consultation activities undertaken, the main findings and when the engagement and consultative activities were undertaken.

Name of engagement and consultative activities undertaken		Summary note of the engagement or consultative activity undertaken	Month/Year
1	**Clinical Reference Group Stakeholder Workshops**	Three virtual workshops (by MS Teams) of ~75 individuals were held in the Summer of 2020 to address key issues relating to the update of the Service Specification in Clinical Genomics. Attendees included representatives from each NHS CGS in England, representatives from Clinical Genomics services in the devolved nations, Charities, patient representatives, the Royal Colleges and professional groups, including the British Society of Genetic Medicine, Clinical Genetics Society, Association of Clinical Genomic Science, Association of Genetic Nurses and Counsellors. Four specific themes were considered by the attendees, including workforce development, changes in clinical practice, new models of working and equity of access. The equity of access group considered how health inequalities can be recognised and addressed in Clinical Genomics.	**July/August 2020**
2	**Reducing disparities in access and the specification of the NHS Genomic Medicine Service (GMS)**	The Genomics England Disparities Subgroup set up to support the delivery of the 100,000 Genomes Project published recommendations to address health inequalities in the provision of Clinical Genomics.	**Nov 2018**

6. What key sources of evidence have informed your impact assessment and are there key gaps in the evidence?

Evidence Type	Key sources of available evidence	Key gaps in evidence
Published evidence	Bonvicini KA. LGBT healthcare disparities: What progress have we made? Patient Educ Couns. 2017; 100:2357-2361. Parker MJ, et al. The genetic assessment of looked after children: common reasons for referral and recent advances. Arch Dis Child 2016; 101:581–584. Manrai AK, et al. Genetic Misdiagnoses and the Potential for Health Disparities. N Engl J Med. 2016; 375:655-65 Burghel GJ, Khan U, Lin WY, Whittaker W, Banka S. Presence of pathogenic copy number variants (CNVs) is correlated with socioeconomic status. J Med Genet. 2020;57:70-72. Barnes H, Morris E, Austin J. Trans-inclusive genetic counseling services: Recommendations from members of the transgender and non-binary community. J Genet Couns. 2020 Jun;29(3):423-434. The care and treatment of young people in relation to their gender identity in the UK Nuffield Council Bioethics. September 2020. Fraiman YS, Wojcik MH. The influence of social determinants of health on the genetic	Need for further sequence data in different ethnic populations to allow interpretation of genomic results.

Evidence Type	Key sources of available evidence	Key gaps in evidence
	diagnostic odyssey: who remains undiagnosed, why, and to what effect? Pediatr Res. 2020 Sep 15. Epub ahead of print. PMID: 32932427. Salway S, et al. Responding to the increased genetic risk associated with customary consanguineous marriage among minority ethnic populations: lessons from local innovations in England. J Community Genet. 2016; 7:215-28. Graves KD, Tercyak KP. Introduction to the special series on health disparities in genomic medicine. Health Psychol. 2015; 34:97-100. Williams JK, et al. Advocacy and actions to address disparities in access to genomic health care: A report on a National Academies workshop. Nurs Outlook. 2019; 67:605-612.	
Consultation and involvement findings	Agreement by the CRG to adopt guidance to ensure that the NHS CGS meets the needs of all patients and families and addresses disparities in access. A document has been developed by Prof. Nadeem Qureshi, Chair of the Genomics England Disparities Subgroup & Tom Billins, Ethics manager, Genomics England to provide guidance to ensure that Clinical	

Evidence Type	Key sources of available evidence	Key gaps in evidence
	Genomics Services meet the needs of all patients and families and addresses disparities in access. This guidance will be included in published operational policy.	
Research	Adoption of guidance on provision of guidance to support NHS CGSs for consanguineous couples	Guidance has been produced in relation to addressing genetic risk associated with consanguineous relationships for inclusion within the published operational policy. Each NHS CGS should adhere to the guidance to ensure that the genomic needs of consanguineous families are met. We acknowledge that most research in disparities of access for different groups is based on studies from the USA and more work on the specific challenges in England is required.
Participant or expert knowledge For example, expertise within the team or expertise drawn on external to your team	Patient representatives on the CRG have contributed to the formation of the service specification. They also provide representation from NIHR patient representative groups, children charities (Christopher's Smile) and the cancer charity (Blood Cancer UK). Expertise on consanguinity and access to disadvantaged communities.	

7. Is your assessment that your proposal will support compliance with the Public Sector Equality Duty? Please add an x to the relevant box below.

	Tackling discrimination	Advancing equality of opportunity	Fostering good relations
The proposal will support?	X	X	X

The proposal may support?			
Uncertain whether the proposal will support?			

8. Is your assessment that your proposal will support reducing health inequalities faced by patients? Please add an x to the relevant box below.

	Reducing inequalities in access to health care	Reducing inequalities in health outcomes
The proposal will support?	X	X
The proposal may support?		
Uncertain if the proposal will support?		

9. Outstanding key issues/questions that may require further consultation, research or additional evidence. Please list your top 3 in order of priority or state N/A

Key issue or question to be answered		Type of consultation, research or other evidence that would address the issue and/or answer the question
1	What approaches will ensure increased uptake of genomics services by disadvantaged groups	There is an initial need to understand the scale and scope of the problem by data recording through the NHS CGSs and an approach to monitoring inequities in access to NHS CGSs will be established, sharing good practice and encourage adoption of practices to improve access to all based on clinical need.

10. Summary assessment of this EHIA findings

At each point in the development and writing of the Service Specification in Clinical Genomics we have been aware of the need to ensure the delivery of an equitable and high quality service. We have conducted specific stakeholder workshops to seek input and opinion about barriers to services and how these can be overcome. We also support the development of an approach to monitoring inequities in access to NHS CGSs - share good practice and encourage adoption of practices to improve access to all based on clinical need. We recognise the importance of having Clinical Genomics workforce and patient groups that broadly represent the communities which are served across all the protected characteristics.

11. Contact details re this EHIA

Team/Unit name:	Genomics Unit
Division name:	Strategy, Transformation and Quality
Directorate name:	Finance