North Thames
Genomic Medicine Service

Community Paediatric Genomics Pathway

Dr Ahmed Ahmed and Dr Veronica Govender

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Purpose of this Pathway

This pathway is intended to guide community paediatricians in North Thames in identifying, referring, and initiating WGS for children with suspected genetic conditions. It aligns with NHS Genomic Medicine Service (GMS) policies and aims to enable timely diagnosis and management.

We recognise that ordering WGS will be a change in practice for many community paediatricians. This document outlines key information and resources to support this transition. **Please note that genomic associates are available to support with consent and requesting relevant blood samples for health care teams in the North Thames region, and Clinical Geneticists / Genetic scientists are available for clinical support.**

Contents

Chapter 1 – Introduction to Whole Genome Sequencing

Whole Genome Sequencing

Whole genome sequencing (WGS) refers to DNA sequencing of the entire genome, including both coding and non-coding regions.

WGS is the most comprehensive form of genomic testing currently in clinical use. It enables a wide range of variant types in a large number of genes to be tested for simultaneously.

Trio whole genome sequencing (WGS), which involves sequencing the genomes of an individual and their biological parents, is recommended where possible because it significantly increases the diagnostic yield for genetic disorders, particularly rare ones, by helping to identify *de novo* mutations and inherited variants.

https://www.genomicseducation.hee.nhs.uk/blog/the-power-of-three-the-importance-of-trios-in-diagnosing-disease/

WGS is not the answer for everything and there may be other tests that could be more helpful.

Other genetics tests

FISH – Fluorescence in situ hybridization (FISH) is a technique that scientists use for some types of genetic testing. They mark DNA with fluorescent labels so they can see the areas they're interested in under a microscope.

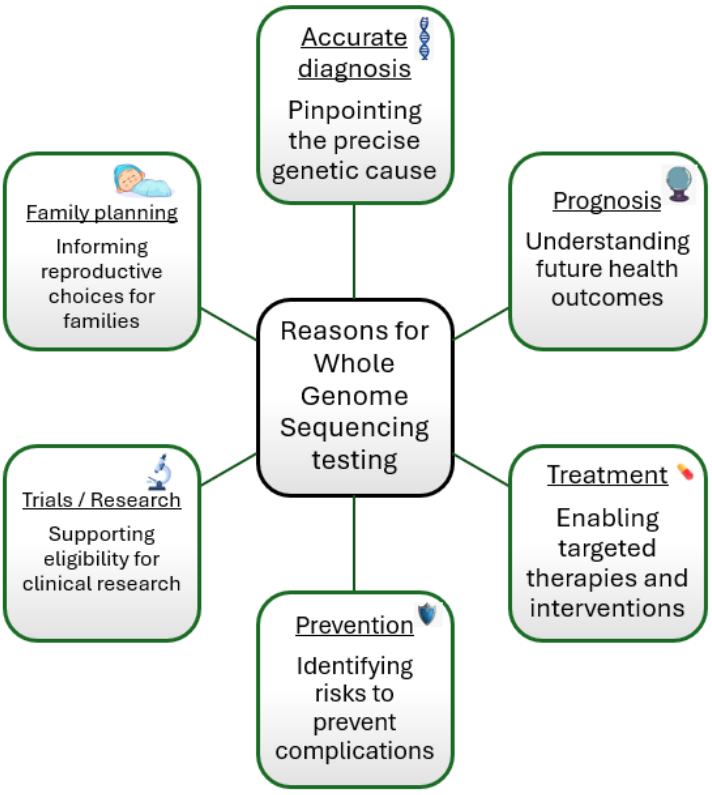
Microarray – A chromosomal microarray test is a genetic test that analyses a person's DNA to detect small missing or extra pieces of chromosomes, called copy number variations (CNVs). CNVs can be associated with various developmental, intellectual, or congenital abnormalities. Microarray was previously the first line test for children with intellectual disability and congenital abnormality. This is still a useful test in neonates with congenital anomalies but is being phased out as an investigation for intellectual disability as CNV analysis is included as part of WGS.

Karyotyping – Karyotyping is the microscopic examination of chromosomes and is used to confirm or exclude large structural changes e. g. Translocation, and for sex chromosome aneuploidies (X/XXX/XXY).

Chapter 2 – Overall Aims of a WGS pathway for Community Paediatrics

- Enable early, efficient, and equitable access to genetic testing for children with developmental concerns, learning disabilities, congenital anomalies, and related conditions.
- Facilitate timely intervention, appropriate management, and informed family planning.
- Reduce unnecessary investigations and testing.
- Ensure accurate and timely referrals to relevant specialists.
- Integrate whole-genome sequencing (WGS) responsibly within community paediatrics.

Why we test



Chapter 3 – Common panels for community paediatrics

When requesting WGS, you need to specify a 'clinical indication' (e.g. R27.3 paediatric disorders) for analysis. This determines which genes within the WGS data will be analysed.

https://norththamesgenomics.nhs.uk/request-a-genetic-test/test-request-guide/find-a-test-tool/

R27.3 Paediatric disorders

https://norththamesgenomics.nhs.uk/tool/congenital-malformation-and-dysmorphism-syndromes-microarray-and-sequencing/

Testing Criteria:

• **Congenital malformations and/or dysmorphism** highly suggestive of an underlying monogenic disorder where targeted genetic testing is not possible.

• **Unexplained moderate/severe/profound global developmental delay or unexplained**

moderate/severe/profound intellectual disability, and where clinical features are highly suggestive of an underlying monogenic disorder requiring sequencing and targeted genetic testing is not possible.

* Please note: **Intellectual disability panel is included in the Paediatric disorders panel**

R69.5 Hypotonic infant

https://norththamesgenomics.nhs.uk/tool/hypotonic-infant/

Testing Criteria:

Neonates or infants with unexplained hypotonia where the clinical picture is suggestive of a central cause i.e. particularly where the baby is not alert but lethargic or sleepy.

Clinically complex cases and additional panel analysis

Not all genetic panels can be requested directly by a community paediatrician. If you wish to request additional panels that require approval, or need advice on panel selection/ investigations, please attend Genomic Question Time (see below) or contact your clinical genetics team (contact details below) or relevant specialist. It is recommended to order all necessary panels together as part of a single WGS test. Adding panels after the initial analysis has been completed is not straightforward.

Please consider contacting/ referring directly to Clinical Genetics in cases of

- Multidomain regression
- Parental pregnancy
- Looked after child- if support is needed around consent
- Parental learning difficulty

Urgent WGS (R14)

Urgent WGS (R14) is available in some circumstances, usually for children on NICU/PICU, but may be considered in other exceptional circumstances. Please discuss directly with clinical genetics if you think that rapid testing is needed. There is an on-call consultant available for urgent advice.

R14 testing is carried out by the Exeter Clinical Laboratory, please refer to their website for details on this testing - R14 Rapid Genome Sequencing Service Exeter Clinical Laboratory International

Chapter 4 – What the WGS result may look like

Result Type	Description
Confirmed genetic diagnosis	A pathogenic or likely pathogenic variant matching the patient's condition is identified.
Incidental findings	An unrelated result was discovered during the genomic test. This may still have significant implications e.g. parentage, cancer risk gene etc.
Variant/s of uncertain significance	Insufficient information exists to determine if this variant is normal or relevant to the patient's condition. Sometimes additional testing may be advised.
No significant findings	No significant variants linked to the patient's condition have been found based on current knowledge. This does not exclude a genetic condition.

https://norththamesgenomics.nhs.uk/wp-content/uploads/2023/09/Types-of-Genomic-Test-Results.pdf

Trio whole genome sequencing (WGS), which involves sequencing the genomes of an individual and their parents, is recommended because it significantly increases the diagnostic yield for genetic disorders, particularly rare ones, by helping to identify de novo mutations and inherited variants. https://www.genomicseducation.hee.nhs.uk/blog/the-power-of-three-the-importance-of-trios-in-diagnosing-disease/

Chapter 5 – Genomic Associates

What is a Genomic Associate?

A Genomic Associate is a healthcare professional who works within the field of genetics and genomics, assisting clinical teams in providing care to patients and their families. They primarily focus on gathering and coordinating information, consent, and samples for genetic testing and counselling.

***For North Thames, they are based at GOSH, and they work with families virtually or via the phone.**

You can contact them by email: gos-tr.ntgenomicsassociate@nhs.net

Key responsibilities of a Genomic Associate for this pathway include:

- Ensuring patients understand and consent to genetic testing, and completing consent forms.
- Requesting blood tests for patients and parents and completing the sampling forms.
- Liaising with the genetic lab to ensure that the test and consent forms are received by the lab.
- Tracking the blood sample to the lab, and informing clinicians when samples have received to the lab within 3 – 4 months.

In the **North Thames region, the Genomic Associate team are based at GOSH but work virtually to support clinical teams across North Thames.**

Chapter 6 – Clinical Pathway for Whole Genome Sequencing

Identify child with suspected genetic condition

- 3-generation family history
- Thorough clinical exam
- Performs baseline investigations alongside genetics

Check eligibility criteria for WGS

- Moderate/severe/profound global developmental delay / intellectual disability, syndromic features, likely monogenic, relevant family history, congenital malformation
- https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-

Information to share with families

- Why the test is recommended
- That it is a blood test and trio test is recommended
- The laboratory will confirm biological relationships (including paternity/maternity) as part of the analysis.
- Possible outcomes of testing
- Mention briefly about NGRL (National Genomic Research Library)
- Process explanation and TAT (turnaround-time)
- Hand in Resources for parents / carers

Referring Clinician

- Doctor fills out the WGS test form
- Then sends the form and a referral to Genomics Associate

Genomic Associates

- Genomics Associates contact parent and fills out 3 x ROD forms
- Genomic Associates complete the sampling request forms X 3, and send them to parents / carers with information on how to arrange for the blood tests.
- Genomic Associate send a copy of ROD forms to clinician's service.
  Genomic Associates inform clinicians if the samples have not received by the lab in 3 – 4 months.

Results

- Results are returned to the referring doctors.
- The doctor is responsible for communicating the results to the family.
- If there are any queries, please discuss them with your local clinical geneticist or attend genomics question time meeting

Chapter 7 – Post-Test Considerations

Further information:

Turnaround time

- WGS Results turnaround-time – currently 12 months for routine WGS requests.
- Urgent WGS (R14) is available in some circumstances, usually for children on NICU/PICU. TAT is typically 2-3 weeks.

Genetic counselling

Ensure genetic counselling is available (pre- and post-test).

Pre-test discussion:

Prepare families for: Variants of uncertain significance (VUS) / incidental findings (e.g. BRCA, cardiomyopathy genes, non-paternity/maternity), and these should be discussed with Clinical Geneticists.

Post-test discussion:

- Families with a confirmed genetic diagnosis should be offered a referral to clinical genetics for counselling.
- Up to 50% still remain undiagnosed. In most cases no further routine genomic testing would be available after WGS. This could be revisited if there are new clinical concerns or after several years as testing is likely to improve.

Important links

- North Thames Whole Genome Sequencing - How To Guide https://norththamesgenomics.nhs.uk/wp-content/uploads/2025/08/Rare-Disease-WGS-How-To-Guide-April-2026-1.pdf

- Principles of Genetic testing: video by Dr Angela Brady: https://norththamesgenomics.nhs.uk/resources/session-1-principles-of-genomic-testing/

Appendix

i. Links for Whole Genome Sequencing, panels, etc

North Thames Genomic Medicine Service (NT GMS)

The North Thames GMS provides and coordinates all genomic tests, within agreed turnaround times, to bring genomic innovation to the people of North London, Hertfordshire and Mid and South Essex.

https://norththamesgenomics.nhs.uk/

National Genomic Test Directory Testing Criteria for Rare and Inherited Disease

This directory identifies the most appropriate test for each clinical indication and the testing methodology by which it should be delivered.

https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-and-inherited-disease-eligibility-criteria-v9.1.pdf

Genomics England PanelApp

Genomics England PanelApp is a publicly-available knowledgebase that allows virtual gene panels related to human disorders to be created, stored and queried. It includes a crowdsourcing tool that allows genes and genomic entities (short tandem repeats/STRs and copy number variants/CNVs) to be added or reviewed by experts throughout the worldwide scientific community, providing an opportunity for the standardisation of gene panels, and a consensus on which genes have sufficient evidence for disease association.

On the NHS GMS Panels Resource website, you can:

- Browse the approved/signed-off versions of panels in use in the NHS GMS
- Find which panel(s) a gene is on
- Find the content of a panel associated with a clinical indication

https://panelapp.genomicsengland.co.uk/

Human Phenotype Ontology

The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality e.g. microcephaly. The terms here are useful for filling out the WGS test form phenotype section.

https://hpo.jax.org/

GeneReviews

This is an international point-of-care resource for busy clinicians, provides clinically relevant and medically actionable information for inherited conditions in a standardized journal-style format, covering diagnosis, management, and genetic counselling for patients and their families. Each chapter is written by one or more experts on the specific condition or disease and goes through a rigorous editing and peer review process before being published online.

https://www.ncbi.nlm.nih.gov/books/NBK1116/

ii. Which conditions cannot be detected by Whole Genome Sequencing

Limitations: When WGS is used for the diagnosis of rare disease patients, it is important to be aware that virtual panels are sometimes used. This means that, although sequencing data are generated for the whole genome, only genes known to be associated with the patient's features are analysed. Clinical interpretation of the large number of identified variants is a significant challenge. Many more variants of uncertain significance are generated compared to more targeted testing. There is an increased risk of incidental findings compared to more targeted testing. Currently, WGS test results can take longer to come back than results of other genomic tests.

Whole genome sequencing (WGS) is a powerful tool, but it has limitations. It may **miss or poorly detect** certain types of genetic conditions or variants due to technical or biological reasons.

Here are key examples of what **WGS cannot reliably detect** or **may miss entirely**:

Repeat Expansion Disorders

WGS has been validated to detect unstable repeat expansions for some genetic conditions such as:

- **Fragile X syndrome** (FMR1 CGG repeat)
- **Myotonic dystrophy** (DMPK or CNBP repeats)
- **Friedreich ataxia** (FXN GAA repeat)

Epigenetic Conditions

WGS sequences DNA, but does **not assess methylation** or other epigenetic changes:

- **Prader-Willi / Angelman syndrome** (methylation status at 15q11-q13)
- **Beckwith-Wiedemann syndrome**
- **Silver-Russell syndrome**

→ These often need **methylation-specific testing** or **MS-MLPA**.

Mitochondrial DNA Disorders

WGS of **nuclear DNA** may not adequately cover:

- **Mitochondrial genome mutations**
- **Heteroplasmy levels** (variable proportions of mutated vs normal mtDNA)

→ These need **dedicated mtDNA sequencing**, often from muscle or other high-energy tissues

Low-Level Mosaicism

WGS may miss:

- **Somatic mosaicism**, especially at low allele frequencies
- **Postzygotic mutations** in conditions like segmental neurofibromatosis or some overgrowth syndromes

→ High-depth sequencing or **targeted methods** are better for detection.

Structural Variants

WGS may **miss or miscall**:

- **Balanced translocations**
- **Inversions**
- **Complex rearrangements** (like chromothripsis)

→ These are better detected by **karyotyping**, **FISH**, or **long-read sequencing** (e.g. PacBio, Oxford Nanopore).

Triploidy or Whole Genome Duplications

- WGS alignment and variant calling can struggle with **polyploidy** or **triploidy**
- Some copy number algorithms may not detect this reliably

→ **Karyotyping or SNP microarray** may be needed.

Interpretation Gaps

Even when a variant is detected, interpretation may be limited by:

- **Variants of uncertain significance (VUS)**
- **Lack of phenotype-genotype correlation**
- **Incomplete gene-disease annotation** (especially for rare or novel conditions)

iii. Education & Training

Several free online courses and resources are available for genomic medicine training in the UK. These resources offer basic training and advanced learning opportunities for healthcare professionals and other interested individuals.

🧬 North Thames Genomic Question Time

Please come and discuss your queries with the clinical genetic and wide MDT team.

- **Schedule:** First Thursday of each month, 12:30–1:00pm
- **Details:** Recurring live Q&A session with experts
- **How to Join:** Email nt-gms@gosh.nhs.uk for the meeting link
- **Link:** North Thames Genomic Question Time

🎓 Genomics Education Programme (HEE)

- **Overview:** Comprehensive education portal developed by NHS
- **Offerings:** Master's, Diploma, and Certificate programmes
- **Resources:** Guides, modules, and structured online learning
- **Link:** Genomics Education Programme

🌐 FutureLearn – *The Genomics Era*

- **Type:** Self-paced online course
- **Audience:** Beginners and intermediate learners
- **Developed With:** NHS organisations
- **Link:** The Genomics Era Course

💻 e-Learning for Healthcare

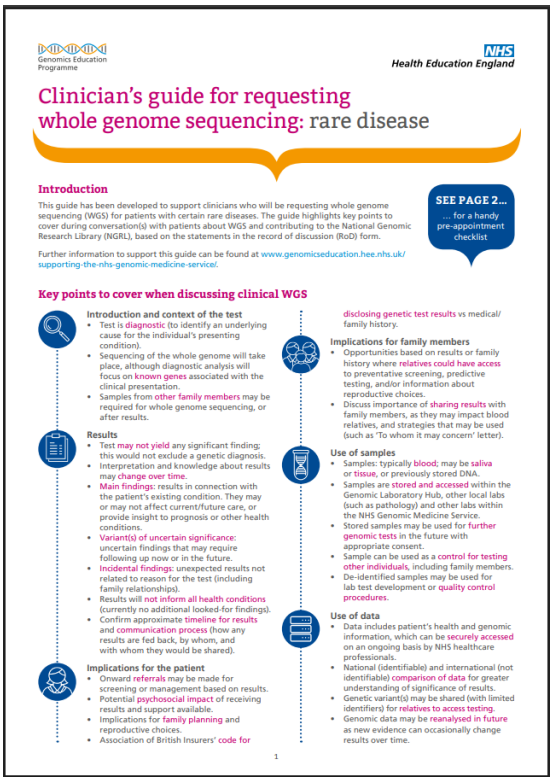
- **Platform:** NHS eLearning environment
- **Focus:** NHS Genomic Medicine Service and genomics topics
- **Audience:** Healthcare professionals and public learners
- **Link:** Genomics in the NHS

📚 East Genomics Learning Hub

- **Purpose:** Online training and resource portal
- **Audience:** Healthcare professionals
- **Link:** East Genomics Learning Hub

iv. Genomics Education Clinicians Guide

https://www.genomicseducation.hee.nhs.uk/wp-content/uploads/2019/11/Guide-to-requesting-WGS-RD-Nov-20.pdf



Genomics Education Programme

Health Education England

Clinician's guide for requesting whole genome sequencing: rare disease

Introduction

This guide has been developed to support clinicians who will be requesting whole genome sequencing (WGS) for patients with certain rare diseases. The guide highlights key points to cover during conversation(s) with patients about WGS and contributing to the National Genomic Research Library (NGRL), based on the statements in the record of discussion (RoD) form.

SEE PAGE 2... ... for a handy pre-appointment checklist

Further information to support this guide can be found at www.genomicseducation.hee.nhs.uk/supporting-the-nhs-genomic-medicine-service/.

Key points to cover when discussing clinical WGS

Introduction and context of the test
- Test is diagnostic (to identify an underlying cause for the individual's presenting condition).
- Sequencing of the whole genome will take place, although diagnostic analysis will focus on known genes associated with the clinical presentation.
- Samples from other family members may be required for whole genome sequencing, or after results.

Results
- Test may not yield any significant finding; this would not exclude a genetic diagnosis.
- Interpretation and knowledge about results may change over time.
- Main findings: results in connection with the patient's existing condition. They may or may not affect current/future care, or provide insight to prognosis or other health conditions.
- Variant(s) of uncertain significance: uncertain findings that may require following up now or in the future.
- Incidental findings: unexpected results not related to reason for the test (including family relationships).
- Results will not inform all health conditions (currently no additional looked-for findings).
- Confirm approximate timeline for results and communication process (how any results are fed back, by whom, and with whom they would be shared).

Implications for the patient
- Onward referrals may be made for screening or management based on results.
- Potential psychosocial impact of receiving results and support available.
- Implications for family planning and reproductive choices.
- Association of British Insurers' code for disclosing genetic test results vs medical/family history.

Implications for family members
- Opportunities based on results or family history where relatives could have access to preventative screening, predictive testing, and/or information about reproductive choices.
- Discuss importance of sharing results with family members, as they may impact blood relatives, and strategies that may be used (such as 'To whom it may concern' letter).

Use of samples
- Samples: typically blood; may be saliva or tissue, or previously stored DNA.
- Samples are stored and accessed within the Genomic Laboratory Hub, other local labs (such as pathology) and other labs within the NHS Genomic Medicine Service.
- Stored samples may be used for further genomic tests in the future with appropriate consent.
- Sample can be used as a control for testing other individuals, including family members.
- De-identified samples may be used for lab test development or quality control procedures.

Use of data
- Data includes patient's health and genomic information, which can be securely accessed on an ongoing basis by NHS healthcare professionals.
- National (identifiable) and international (not identifiable) comparison of data for greater understanding of significance of results.
- Genetic variant(s) may be shared (with limited identifiers) for relatives to access testing.
- Genomic data may be reanalysed in future as new evidence can occasionally change results over time.

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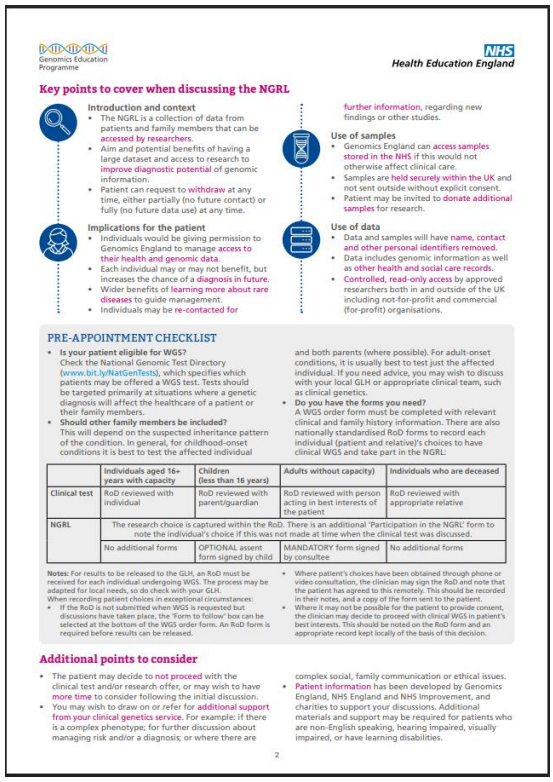


Genomics Education Programme

Health Education England

Key points to cover when discussing the NGRL

Introduction and context
- The NGRL is a collection of data from patients and family members that can be accessed by researchers.
- Aim and potential benefits of having a large dataset and access to research to improve diagnostic potential of genomic information.
- Patient can request to withdraw at any time, either partially (no future contact) or fully (no future data use) at any time.

Implications for the patient
- Individuals would be giving permission to Genomics England to manage access to their health and genomic data.
- Each individual may or may not benefit, but increases the chance of a diagnosis in future.
- Wider benefits of learning more about rare diseases to guide management.
- Individuals may be re-contacted for further information, regarding new findings or other studies.

Use of samples
- Genomics England can access samples stored in the NHS if this would not otherwise affect clinical care.
- Samples are held securely within the UK and not sent outside without explicit consent.
- Patient may be invited to donate additional samples for research.

Use of data
- Data and samples will have name, contact and other personal identifiers removed.
- Data includes genomic information as well as other health and social care records.
- Controlled, read-only access by approved researchers both in and outside of the UK including not-for-profit and commercial (for-profit) organisations.

PRE-APPOINTMENT CHECKLIST

- **Is your patient eligible for WGS?** Check the National Genomic Test Directory (www.bit.ly/NatGenTests), which specifies which patients may be offered a WGS test. Tests should be targeted primarily at situations where a genetic diagnosis will affect the healthcare of a patient or their family members.
- **Should other family members be included?** This will depend on the suspected inheritance pattern of the condition. In general, for childhood-onset conditions it is best to test the affected individual and both parents (where possible). For adult-onset conditions, it is usually best to test just the affected individual. If you need advice, you may wish to discuss with your local GLH or appropriate clinical team, such as clinical genetics.
- **Do you have the forms you need?** A WGS order form must be completed with relevant clinical and family history information. There are also nationally standardised RoD forms to record each individual (patient and relative)'s choices to have clinical WGS and take part in the NGRL:

	Individuals aged 16+ years with capacity	Children (less than 16 years)	Adults without capacity)	Individuals who are deceased
Clinical test	RoD reviewed with individual	RoD reviewed with parent/guardian	RoD reviewed with person acting in best interests of the patient	RoD reviewed with appropriate relative
NGRL	The research choice is captured within the RoD. There is an additional 'Participation in the NGRL' form to note the individual's choice if this was not made at time when the clinical test was discussed.			
	No additional forms	OPTIONAL assent form signed by child	MANDATORY form signed by consultee	No additional forms

Notes: For results to be released to the GLH, an RoD must be received for each individual undergoing WGS. The process may be adapted for local needs, so do check with your GLH.
When recording patient choices in exceptional circumstances:
- If the RoD is not submitted when WGS is requested but discussions have taken place, the 'Form to follow' box can be selected at the bottom of the WGS order form. An RoD form is required before results can be released.
- Where patient's choices have been obtained through phone or video consultation, the clinician may sign the RoD and note that the patient has agreed to this remotely. This should be recorded in their notes, and a copy of the form sent to the patient.
- Where it may not be possible for the patient to provide consent, the clinician may decide to proceed with clinical WGS in patient's best interests. This should be noted on the RoD form and an appropriate record kept locally of the basis of this decision.

Additional points to consider

- The patient may decide to not proceed with the clinical test and/or research offer, or may wish to have more time to consider following the initial discussion.
- You may wish to draw on or refer for additional support from your clinical genetics service. For example: if there is a complex phenotype; for further discussion about managing risk and/or a diagnosis; or where there are complex social, family communication or ethical issues.
- Patient information has been developed by Genomics England, NHS England and NHS Improvement, and charities to support your discussions. Additional materials and support may be required for patients who are non-English speaking, hearing impaired, visually impaired, or have learning disabilities.

2

The Approach to a Child with Dysmorphic Features: What the Paediatrician Should Know: https://pmc.ncbi.nlm.nih.gov/articles/PMC11120268/

dysmorphology article.pdf

v. Test request forms

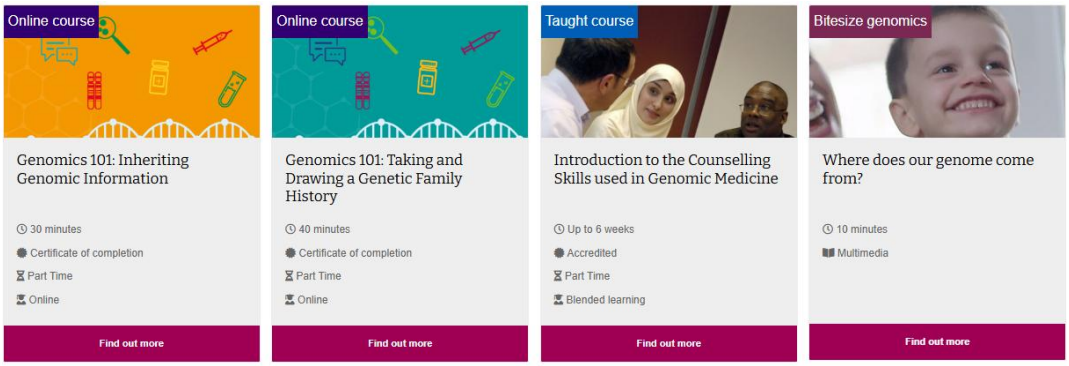
All test request forms, information about sending samples and consent can be found on the North Thames Genomic Medicine Service website:

Test request information - North Thames GMS : North Thames GMS

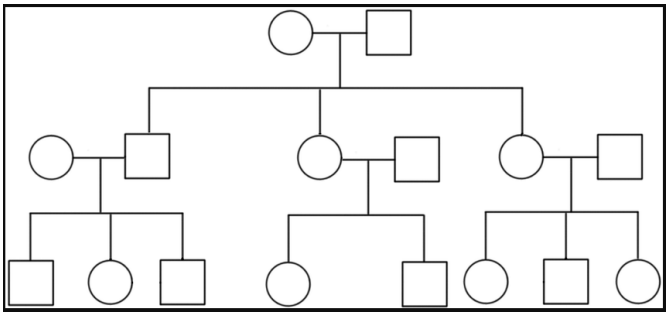
vi. Family Pedigree Template

Taking a family history tree – https://www.youtube.com/watch?v=7dgX9WiL5XQ

https://www.genomicseducation.hee.nhs.uk/taking-and-drawing-a-family-history/



Ideally should take a 3-generation pedigree:



	Male	Female	Sex Unknown
Individual			
Affected individual (symbol coloured in)			
Multiple individuals	5	5	5
Deceased			
Pregnancy	P	P	P
Miscarriage	male	female	
Person providing pedigree information			

Marriage/partnership	
Divorce/separation	
Where the partners are blood relatives (consanguineous relationship)	
Children/siblings	sibship line, line of descent, individual line
Identical twins (monozygotic)	
Non-identical twins (dizygotic)	

Reference: *Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counsellors; Bennet et al, Journal of Genetic counselling Sept 2008:* https://onlinelibrary.wiley.com/doi/10.1007/s10897-008-9169-9

vii. Referral to Genomic Associates – GOSH (Example)

Dear Genomic Associates Team – Great Ormond Street Hospital,

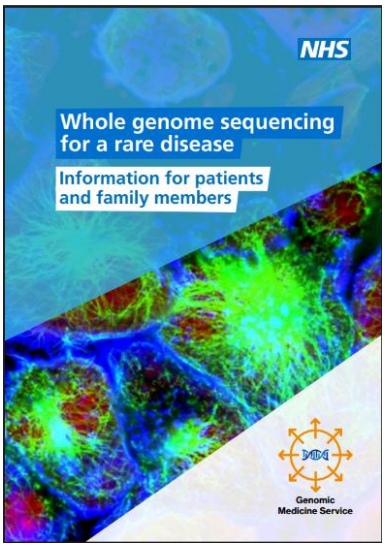
Re: BCYP details (Names, DOB, NHS Number)

I would be grateful if you could complete whole genome sequencing consent for the above child and their family and complete the blood sampling form for them. No sampling forms were completed locally. Please note the enclosed completed WGS test form.

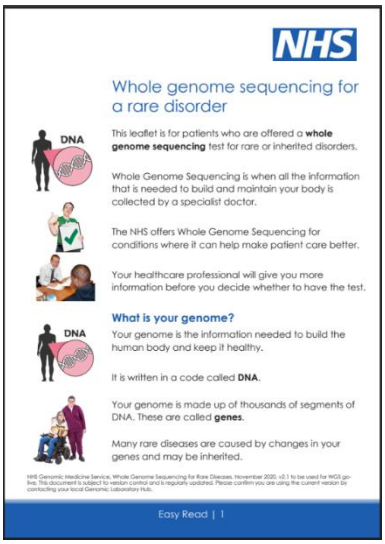
Test Name	R -
Test Type	Singleton / Duo / Trio
Mother	Name: Date of Birth: NHS Number: Address: Contact Number:
Father:	Name: Date of Birth: NHS Number: Address: Contact Number:
Method of contact	
Need of interpreter	
Urgency	
Safeguarding issues	
Others	
Have the blood tests arranged locally	Child: Yes / No Parents: Yes / No
How to arrange the blood test locally	Child: Parents:
Clinician's Name	
Department / Trust	
Date	
Service Email for GA to contact your service	

viii. Resources for parents / carers

https://www.england.nhs.uk/wp-content/uploads/2021/07/genome-sequencing-rare-disease-patient-information.pdf



https://www.england.nhs.uk/wp-content/uploads/2021/07/genome-sequencing-rare-disease-patient-information-easy-read.pdf

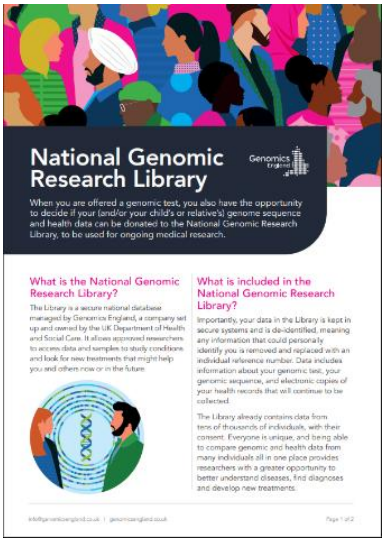


National Genomic Research Library – information for patients

The National Genomic Research Library, sometimes shortened to ‘the NGRL’, is a secure database that holds de-identified genomic and health data from participants.

All participants are patients or family members of patients, each of whom have consented for their data to be held by Genomics England and used for research.

The database is managed by Genomics England and serves as a resource for thousands of research projects.



https://www.genomicsengland.co.uk/assets/documents/One-page-NGRL-Patient-Information-Sheet.pdf

Principles of Genetic testing: Video by Dr Angela Brady

https://norththamesgenomics.nhs.uk/resources/session-1-principles-of-genomic-testing/

Patient Support Resources in Genomic Medicine

Resource	Link
NHS England Whole Genome Sequencing	https://www.england.nhs.uk/genomics/nhs-genomic-med-service/

Provides official NHS guidance for patients exploring genome sequencing—what it involves, potential outcomes, and support.	
Genetic Alliance UK Support for Genetic Conditions A national charity that empowers patients and families affected by genetic disorders through advocacy, information, and connections.	https://geneticalliance.org.uk/
SWAN UK Syndromes Without a Name Provides emotional and practical support for families living with a rare condition that remains undiagnosed.	https://geneticalliance.org.uk/support-and-information/swan-uk-syndromes-without-a-name/
Unique Rare Chromosome & Gene Disorders Offers condition-specific guides, community stories, and understanding for families affected by rare genomic changes.	https://rarechromo.org/
MedlinePlus Genetics Genetics and Health Conditions From the US National Library of Medicine, this site provides easy-to-understand explanations of genetic conditions, treatments, and inheritance.	https://medlineplus.gov/genetics/

viiii. Contact Information

1. North Thames GLH (Genomics Laboratory Hub)

- **Head of Laboratory Service:** Deborah Morrogh
- **Laboratory contact email:** gos-tr.norththamesgenomics@nhs.net
- **Phone:** 020 7762 6888
- **Samples should be sent to:** Great Ormond Street Hospital for Children NHS Foundation Trust, Levels 4-6 Barclay House, 37, Queen Square, London WC1N 3BH
- **https://norththamesgenomics.nhs.uk/about/our-glh/**

2. North West Thames Regional Genetics Service

The North West Thames Regional Genetics Service offers genetic counselling and genetic diagnosis to individuals and families living in north west London, parts of Hertfordshire and Bedfordshire.

- **Phone:** 020 8869 2795
- **Location:** North West Thames Regional Genetics Service, Level 8V, St. Mark's Hospital
- **https://www.lnwh.nhs.uk/genetics**

3. North East Thames Regional Genetics Service

- **Email:** gos-tr.clinicalgenetics@nhs.net
- **Phone:** 020 7762 6856
- **Location:** Level 4 Barclay House, Great Ormond Street, London. WC1N3JH
- **https://www.gosh.nhs.uk/wards-and-departments/departments/clinical-specialties/clinical-genetics-information-parents-and-visitors/**

4. Questions about Education and training:

If you have a query about education and training or cannot find the answer you are looking for on our site please contact **nt-gms@gosh.nhs.uk**.

References

- NHS England Rare Disease Guidelines
- Genomics Education Programme: Genomic Testing Framework
- Local Genomic Medicine Service Alliance (GMSA) pathways
- Genomics England: https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/whole-genome-sequencing/
- North Thames GLH: https://norththamesgenomics.nhs.uk/about/our-glh/