

# Referring Whole Genome Sequencing (WGS) for a Rare Disease *Clinician's How-To Guide*

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## A. WGS Checklist

### 1: Check WGS eligibility

On the [National Genomic Test Directory \(PDF\)](#), check “WGS” is indicated under “Methods” in Associated Tests.

### 2: Complete the Test Order Form

Complete the [Test Order form](#) (one per family)

### 3: Consent and complete the Record of Discussion form(s)

After consenting the patient, complete a [Record of Discussion](#) for each individual in the family being tested (+ a [Consultee form](#) if patient is an adult lacking capacity)

### 4: Organise WGS samples

Provide the patient and family members being tested with completed [Sample Forms](#) to take with them to their local hospital/phlebotomy

### 5: Send the Record of Discussion forms and Test Order Forms to the laboratory

Send to [gos-tr.wgsnorththamesglh@nhs.net](mailto:gos-tr.wgsnorththamesglh@nhs.net) save a copy to your records

## B. WGS Pathways

There are **two** suggested pathways for WGS referrals. One is by requesting and consenting for WGS yourself, and another is by requesting via a Genomic Practitioner/Associate.

#### **Genomic Practitioner/Associate:** [gos-tr.ntgenomicsassociate@nhs.net](mailto:gos-tr.ntgenomicsassociate@nhs.net)

role established to help meet the demands of genetic testing, with a specialised knowledge of WGS.

- Point of contact between consultants and the laboratory
- WGS request help – consenting, forms, sample collection, sample chasing
- Track WGS activation and dispatch

Non-WGS genetic testing, clinical details, decisions on clinical urgency and feeding back clinical information/WGS results to patients are **NOT** part of the role.

Key:

Referring Consultant

Genomic Practitioner

### Eligibility for Mainstream Genetic Testing

- Make sure patient fulfils [NGTD](#) criteria ([page4](#))
- Discuss family history
- Determine whether this is a singleton  
WGS (only the patient to be tested) or trio  
WGS (patient and both parents to be tested)

*Note: For other family combinations, please check possibility with laboratory*

### Patient Appointment

- Discuss family history
- Go through brief consent discussion to make sure the patient is happy with the genetic referral
- Complete [Test Order form](#) ([page 5 and 6](#))

NO

? Requesting via Genomic Practitioner

YES

### Patient Appointment [continued]

- Go through entire consent discussion
- Complete [Record of Discussion](#) ([page 7](#)) page can be virtually completed or with the patient face-to-face) (+ [Consultee form](#) if adult lacking capacity)
- Provide [Patient Information Leaflets](#) and [video](#) link
- Provide [Sample Form\(s\)](#) ([page 11](#)) to all family members being tested
- Save forms to Electronic Patient Record

### Following the Appointment

- Send Record of Discussion form(s) and Test Order form to [gos-tr.wgsnorththamesqlh@nhs.net](mailto:gos-tr.wgsnorththamesqlh@nhs.net)
- Confirm with laboratory that all samples have arrived and the test has been activated

- Send Test Order Form to Genomic Practitioner ([gos-tr.ntgenomicsassociate@nhs.net](mailto:gos-tr.ntgenomicsassociate@nhs.net))

### Genomic Practitioner Appointment

- Go through consent discussion
- Complete [Record of Discussion](#) (+ [Consultee form](#) if adult lacking capacity)
- Provide [Patient Information Leaflets](#) and [video](#) link
- Provide [Blood Test Request Form\(s\)](#) to all family members being tested (if not done by consultant)
- Send forms to department pathway coordinator to save to Electronic Patient Record

### Following the Appointment

- Send Record of Discussion form(s) and Test Order form to [gos-tr.wgsnorththamesqlh@nhs.net](mailto:gos-tr.wgsnorththamesqlh@nhs.net)
- Confirm with laboratory that all samples have arrived, and the test has been activated

## C. Eligibility for WGS

Patient eligibility for WGS clinical indications can be found on the [NHS England » National genomic test directory](#). If you need to determine which genes/panels are included in a clinical indication, please visit [PanelApp](#).

### R59 Early onset or syndromic epilepsy

### The Clinical Code/Indication

#### Testing Criteria

Unexplained epilepsy with clinical suspicion of a monogenic cause including:

1. Onset under 2 years, OR
2. Clinical features suggestive of specific genetic epilepsy, for example Dravet syndrome, OR
3. Additional clinical features: intellectual disability, autism spectrum disorder, structural abnormality (e.g. dysmorphism, congenital malformation), unexplained cognitive/memory decline

Testing may occasionally be appropriate where age of onset is between 2 and 3 years and following clinical agreement by a specialist MDT.

#### Overlapping indications

- R110 Segmental overgrowth disorders – Deep sequencing test should be used where megalencephaly is present to allow detection of somatic mosaic variants
- R14 Acutely unwell children with likely monogenic disorder should be used in acutely unwell children with epilepsy

**NOTE:** If a metabolic disorder is suspected, testing should be carried out either using R89 or R98 or under an alternative metabolic-related clinical indication

Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

#### Where in Pathway

At presentation

#### Requesting Specialties

- Clinical Genetics
- Metabolic Medicine
- Neurology

Check whether you are eligible for requesting this test

#### Specialist Service Group

- Neurology

#### Associated Tests

It is not a requirement to perform microarray testing in addition to WGS but microarray testing can be performed where appropriate

Code	Name	Optimal Family Structure	Scope(s)	Target Type	Target Name	Method
R59.2	Genomewide Microarray	Singleton	Genomewide CNVs	Genomewide	Genomewide	Microarray
R59.3	Epilepsy - early onset or syndromic WGS (phase 1)	Trio or singleton	Exon level CNVs, Small variants, STRs	Panel of genes or loci	Genetic epilepsy syndromes (402)	WGS

If trio is indicated, include parents if available

Make sure WGS is an associated test



Proband first name	Proband last name	Date of birth (dd/mm/yyyy)	NHS number
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\* **Mandatory**

HPO terms are important for the analysis and interpretation of WGS data.  
Please enter valid HPO terms present in the proband/family members being tested  
HPO terms can be copied from the lists below

Follow the link for a complete list of HPO terms

HPO Terms - Please ensure those given match those available at <a href="https://hpo.jax.org/spp/">https://hpo.jax.org/spp/</a>	Proband Name		Parent 1 Name		Parent 2 Name	
	Present	Absent	Present	Absent	Present	Absent
e.g. Epilepsy *	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Terms must be ticked as present or absent in all family members being tested.

- |  |  |  |
|--|--|--|
| <ul style="list-style-type: none"> <li>Intellectual disability, developmental and metabolic</li> <li>Intellectual disability - mild</li> <li>Intellectual disability - moderate</li> <li>Intellectual disability - profound</li> <li>Intellectual disability - severe</li> <li>Autistic behaviour</li> <li>Global developmental delay</li> <li>Delayed fine motor development</li> <li>Delayed gross motor development</li> <li>Delayed speech and language development</li> <li>Generalized hypotonia</li> <li>Feeding difficulties</li> <li>Failure to thrive</li> <li>Abnormal facial shape</li> <li>Abnormality of metabolism/homeostasis</li> <li>Microcephaly</li> <li>Macrocephaly</li> <li>Tall stature</li> </ul> | <ul style="list-style-type: none"> <li>Neurology</li> <li>Muscular dystrophy</li> <li>Myopathy</li> <li>Myotonia</li> <li>Fatigable weakness</li> <li>Peripheral neuropathy</li> <li>Distal arthroproposis</li> <li>Arthroproposis multiplex congenita</li> <li>Cognitive impairment</li> <li>Parkinsonism</li> <li>Spasticity</li> <li>Chorea</li> <li>Dystonia</li> <li>Ataxia</li> <li>Cerebellar atrophy</li> <li>Cerebellar hypoplasia</li> <li>Dandy-Walker malformation</li> <li>Olivopontocerebellar hypoplasia</li> <li>Diffuse white matter abnormalities</li> <li>Focal white matter lesions</li> <li>Leukoencephalopathy</li> <li>Cortical dysplasia</li> <li>Heterotopia</li> <li>Lissencephaly</li> <li>Pachygyria</li> <li>Polymicrogyria</li> <li>Schizencephaly</li> <li>Holoprosencephaly</li> <li>Hydrocephalus</li> <li>Neurodegeneration</li> <li>Dementia</li> </ul> | <ul style="list-style-type: none"> <li>Cardiology</li> <li>Hypertrophic cardiomyopathy</li> <li>Dilated cardiomyopathy</li> <li>Cardiomyopathy</li> </ul>  |
| <ul style="list-style-type: none"> <li>Craniosynostosis</li> <li>Bicoronal synostosis</li> <li>Unicoronal synostosis</li> <li>Metopic synostosis</li> <li>Sagittal craniosynostosis</li> <li>Lambdoidal craniosynostosis</li> <li>Multiple suture craniosynostosis</li> </ul>  | <ul style="list-style-type: none"> <li>Skeletal dysplasia</li> <li>Disproportionate short stature</li> <li>Proportionate short stature</li> <li>Short stature</li> <li>Skeletal dysplasia</li> </ul>   | <ul style="list-style-type: none"> <li>Eye Disorders</li> <li>Cataract</li> <li>Retinal dystrophy</li> <li>Macular dystrophy</li> <li>Microphthalmia</li> <li>Anophthalmia</li> <li>Coloboma</li> <li>Developmental glaucoma</li> <li>Aniridia</li> <li>Abnormal anterior eye segment morphology</li> <li>Nystagmus</li> </ul>   |
| <ul style="list-style-type: none"> <li>Diabetes</li> <li>Neonatal insulin-dependent diabetes mellitus</li> <li>Transient neonatal diabetes mellitus</li> </ul>   | <ul style="list-style-type: none"> <li>Epilepsy</li> <li>Seizures</li> <li>Generalized seizures</li> <li>Focal seizures</li> <li>Epileptic spasms</li> <li>Infantile encephalopathy</li> <li>Atonic seizures</li> <li>Generalized myoclonic seizures</li> <li>Generalized tonic seizures</li> <li>Generalized tonic-clonic seizures</li> <li>EEG with focal epileptiform discharges</li> <li>EEG with generalized epileptiform discharges</li> <li>Multifocal epileptiform discharges</li> </ul>   | <ul style="list-style-type: none"> <li>Immune Disorders</li> <li>Immunodeficiency</li> <li>Abnormal lymphocyte morphology</li> <li>Abnormal lymphocyte physiology</li> <li>Abnormal lymphocyte count</li> <li>Abnormality of neutrophils</li> <li>Abnormality of humoral immunity</li> <li>Abnormal inflammatory response</li> <li>Abnormality of complement system</li> </ul> |

Human Phenotype Ontology (HPO) terms are a standardised vocabulary of phenotypic abnormalities which are important for the analysis and interpretation of WGS data.

Please complete accurately for each family member being tested (present/absent boxes ticked).

A complete list can be found at [Human Phenotype Ontology \(jax.org\)](https://hpo.jax.org/).

Follow this [link](#) for more information on how to use HPO terms in a clinical context.

**HPO terms are very important for increasing the chances of finding a potential diagnosis via the panel agnostic exomiser variant prioritisation analysis that is carried out if the panel-tiering analysis is negative. If choosing HPO terms is done properly, this would also reduce the need for further analysis.**

## 2. The Record of Discussion - completed by the consultant or Genomic Practitioner

Page 3 (pictured) of the document to be completed after a full consent conversation – a summary of the conversation is listed on Page 1 and 2 (not pictured). This can be completed remotely (i.e. consent appointment over the phone/virtual). Information on what to include in the consent conversation is detailed page 8-10.

For adults lacking capacity, please also complete the [Consultee form](#).

NHS Genomic Medicine Service, Record of Discussion Form version 4.03.

First name *	NHS number (or postcode if not known) *
Last name *	Date of birth *



\* Mandatory

### Confirmation of Your Genomic Test and Research Choices

I confirm that I have had the opportunity to discuss information about genomic testing, I agree to the genomic test, and my research choice is indicated below.

- A. I have discussed taking part in the National Genomic Research Library  YES |  NO   
*If your answer to A is NO then please ignore B and sign directly below*
- B. I agree that my data and remainder sample may contribute to the National Genomic Research Library  YES |  NO

Patient name *	Signature	Date

If you are signing this form on behalf of someone else (children, adults without capacity or deceased patients) then please sign below.

Parent   Guardian   Consultee name* <i>please amend as appropriate</i>	Signature	Date

Mandatory signature for children, adults lacking capacity or deceased patients

- if patient is an adult lacking capacity, a separate Consultee form must be completed in addition to the Record of Discussion

### Healthcare professional use only

To be completed by the healthcare professional recording the patient's choices.

Patient category *	<input type="checkbox"/> Adult (made their own choices)	<input type="checkbox"/> Clinician has agreed to the test (in the patient's best interests)
	<input type="checkbox"/> Adult lacking capacity (choices advised by consultee)	<input type="checkbox"/> Deceased (choices made on behalf of deceased individual)
	<input type="checkbox"/> Child (parent or guardian choices)	
Test type *	<input type="checkbox"/> Rare and Inherited Diseases - WGS	<input type="checkbox"/> Cancer (paired tumour normal) - WGS
	<input type="checkbox"/> Patient would like to discuss at a later date	<input type="checkbox"/> Inappropriate to have discussion
If answer to research choice A is NO	<input type="checkbox"/> Patient lacks capacity and no consultee available	<input type="checkbox"/> Other
Remote consent	<input type="checkbox"/> Recorded remotely by clinician, no patient signature	
Responsible clinician *		
Hospital number		

A reason why the National Genomic Research library has not been discussed must be given (i.e. if A is ticked as NO)

The Record of Discussion form can be completed remotely if the patient appointment is virtual/over the phone

Healthcare professional name *	Signature	Date

## E. Consent Requirements:

### 1. WGS Consenting via the Genomic Practitioner

It is important to note that the consultant **ultimately has responsibility for patient consent**. Therefore, it is essential that you make sure the patient is happy to proceed with the genetic testing before referring them. The Genomic Practitioner would then speak to the patients and all family members to be tested and fill the Record of Discussion form.

#### a. Consultant Preliminary Discussion with the Patient:

1. What WGS is – reading through all the DNA and analysing specific areas (virtual panels) – NOT gene agnostic
2. Managing expectations - Turnaround time from the point that all blood samples/consent forms have arrived at the laboratory
  - a. 12 months (routine tests) *(as of August 2024)*
  - b. 12 weeks (urgent tests) *(as of August 2024)*
3. Understanding that the patient will be contacted by a Genomic Practitioner

The Genomic Practitioner will discuss the National Genomic Research Library with the patient.

If possible, please provide [Patient Information Leaflets](#) and the WGS [video](#) link

### 2. WGS Consenting yourself

#### a. Full Consent Conversation required:

	Individuals aged 16+ years with capacity	Children (less than 16 years)	Adults without capacity	Individuals who are deceased
Clinical test	RoD reviewed with each individual	RoD reviewed with parent/guardian	RoD reviewed with person acting in best interests of the patient	RoD reviewed with appropriate relative

1. What WGS is – reading through all the DNA and analysing specific areas (virtual panels)
2. Small blood sample required for each family member
3. Turnaround time from the point that all blood samples have arrived at the laboratory
  - a. 12 months (routine tests) *(as of August 2024)*
  - b. 12 weeks (urgent tests) *(as of August 2024)*
4. Family Implications
  - a. Implications on other family members or future pregnancies
  - b. Opportunities for relatives to have access to screening, predictive genetic testing and/or information about reproductive choices based on these results or family history
  - c. Importance of sharing results with family members if a pathogenic variant is found (it is helpful to start early conversations about this rather than only after the results are available)
5. Uncertainty



- a. Results may find a variant of uncertain/unknown significance (VUS) = a genetic change that may affect the way the gene is working, but there is not enough evidence available to confirm this as a disease-causing or likely disease-causing variant.
- b. May require a referral for further genetic testing via Clinical Genetics Service
- c. Variants of uncertain significance should not be used to make clinical decisions for the individual or family members
- d. This result may change over time as this can be re-analysed in future

#### 6. Unexpected Information/ Incidental Findings

- a. Pathogenic variants may be identified that are unrelated to the reason for the genetics referral, and may indicate an underlying predisposition to a different phenotype (e.g. risk of further cancer or diagnosis with other possible health problems)
- b. These are not routinely looked for and they are rare to come across as the laboratory focuses analysis on virtual panels relevant to the genetic referral
- c. The results will NOT inform all health conditions – currently, there are no additional looked-for findings, however these may still be found by chance
- d. Misattributed parentage is another example of incidental findings

#### 7. DNA storage

- a. The blood sample will be sent to the laboratory and DNA will be extracted
- b. This DNA will continue to be stored (approximately 30 years) unless the patient requests this to be destroyed
- c. This DNA can be accessed by other laboratories within the NHS Genomic Medicine Service
- d. The DNA will not be used for further genetic testing without consent – however, this may be used as a control sample for testing other family members
- e. DNA is not always of sufficient quality and another sample may be required to complete testing

#### 8. Data storage

- a. Data includes patient's health and genomic information, which can be securely access on an ongoing basis by NHS healthcare professionals
- b. Data is stored behind various NHS firewalls
- c. National (identifiable) and international (non-identifiable) comparison of data for greater understanding of significance of any results may be required
- d. Germline variants may be shared for relatives to access testing (limited identifiers to process the test) but medical information will not be shared with relatives
- e. Genomic data may be re-analysed in future as new evidence can occasionally change results
- f. The report will be available on the patient's clinical record

## b. [The National Genomic Research Library \(Genomics England\)](#)

	Individuals aged 16+ years with capacity	Children (less than 16 years)	Adults without capacity	Individuals who are deceased
<b>NGRL</b>	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 the time when the clinical test was discussed.			
	No additional forms	OPTIONAL assent form signed by child	MANDATORY form signed by consultee	No additional forms

For adults lacking capacity, a [Consultee form](#) is also required (will be completed by the Genomic Practitioner if using this pathway).

1. What it is - a comprehensive database that enables approved researchers to access *de-identified* genomic data, health data and samples
2. Research participation is an opt in process (they can choose to take part)
3. Who can access – national and international scientists, researchers, and healthcare companies
4. Data accessed
  - a. The Data is de-identified (pseudonymised) – each patient record is given a unique identification number instead of name, DOB and contact details
  - b. The data available included data about the sample, the raw data of the sample analysis, the patient clinical data (information about their condition that was submitted when ordering WGS) and secondary clinical data from NHS and GP records
5. Patients may be re-contacted for years to come by GE or clinical team
  - a. Certain approved staff within Genomics England will be able to see both identified and de-identified patient data to inform patients about any diagnosis found or to access a clinical trial
  - b. They will NOT be contacted for marketing purposes
6. They can withdraw from research and data sharing at any time
  - a. Partial withdrawal: the patient is happy for their data to continue to be stored but they do not wish to be contacted by Genomics England
  - b. Full withdrawal: all data will no longer be included in any future data releases for further research access

# F. WGS Sample Form

A completed NT GLH [Sample Form](#) with patient details **must** be attached to the labelled blood tube (1x EDTA) for WGS.

If the patient is unable to provide a blood test, a saliva kit can also be accepted. In this case, please provide the patient with a completed [Sample Form](#), a saliva kit (e.g. OG-600 or OG-500 kits) and a pre-paid envelope with the GLH address, for the saliva and form to reach the laboratory for extraction:

**North Thames GLH, Rare & Inherited Disease Genomic Laboratory**  
Specimen Reception, Level 5 Barclay House, 37 Queen Square,  
London WC1N 3BH

The form is double sided (page-2 not pictured) – please make sure to print both sides as information on the back is needed for phlebotomy and the processing of samples at the laboratory.

UKAS  
MEDICAL  
7883

North Thames Genomic Laboratory Hub  
Rare & Inherited Disease Genomic Laboratory  
Level 5 Barclay House  
37 Queen Square, London WC1N 3BH

University College  
London Hospitals  
NHS Foundation Trust  
UKAS  
MEDICAL  
8040

Please note that forms received with missing patient identifiers or no referring clinician/facility may not be tested

**\* Mandatory**

GENETIC TEST REQUEST FORM				Referring Clinician Details *	
Lab Ref <small>(lab use only)</small>	Date Received <small>(lab use only)</small>	Referring Clinician: (full name required)			
<b>Patient Details - use four patient identifiers *</b>		Contact Number:			
First name:	Surname:	NHS.net email: (mandatory)			
DOB:	Sex Assigned at birth:	Department:			
NHS Number: (mandatory)	Hospital No/Your Ref:	Hospital: (full hosp. name & address required)			
Ethnicity:	GOSH Family ID:	Submitter ID (Outreach):			
Patient Address:		Referring Consultant: (if different from referring clinician)			
Postcode:		Referring Consultant Email:			
Referring Clinician: I have discussed genomic testing with this patient and have retained a record of discussion (see page 2). Consent is not required for DNA storage.					
NHS Patient (England) <input type="checkbox"/>		*Billing Address (if organisation to be invoiced):		Purchase Order No.	
NHS Patient (Wales, Scotland, N.I.)* <input type="checkbox"/>					
Private/International Patient* <input type="checkbox"/>		*Patient Email Address (if Self Funding):			
<b>Specimen Details</b> If high risk please specify:					
High Risk Specimen? Yes <input type="checkbox"/> No <input type="checkbox"/>		Sample Type	Date / Time Collected	Collected By	
°Clinical Indication Code: R *		"WGS", or WGS Clinical R code		Urgent <input type="checkbox"/> Routine <input type="checkbox"/>	
Reason for referral: (please give clinical details & details of previous genetic investigations in the family, if known)				° For NHS England referrals, please refer to the National Genomic Test Directory for available tests and eligibility criteria - <a href="https://www.england.nhs.uk/publication/national-genomic-test-directories/">https://www.england.nhs.uk/publication/national-genomic-test-directories/</a>	

If only requesting a sample for WGS, this section is optional (as this information will be included in the Test Order Form).  
If requesting other non-WGS genetic testing (e.g. microarray, WES), please include clinical information

Molecular Genetic Testing <small>(EDTA, except NIPD, see below)</small>	Microarray (EDTA only)	Karyotype (Lithium Heparin)
DNA storage ONLY	If requesting urgent microarray (e.g. pregnancy, infants <3 months) please send a Lithium Heparin as well	To exclude Turner Syndrome (Short Stature/Amenorrhea ONLY)
<input checked="" type="checkbox"/> Diagnostic test *	Cytogenetic follow up (EDTA & Lithium Heparin) Please give the name & GOSH MRN of index patient above or include copy of index patient report	To exclude Ring 20 (Epilepsy)
Carrier test		Premature Ovarian Failure/IVF
Predictive test	Rapid testing for infants (Lithium Heparin & EDTA)	Azoospermia/Male Infertility/IVF
NIPD (PAXgene or Streck cell stabilising tube)	13/18 <input type="checkbox"/> 21 <input type="checkbox"/> Aneuploidy (please specify)	Sample requested by lab
Please provide relevant family history above	Presence of SRY (chromosomal sex)	Chromosome Breakage (not Fragile X) (Lithium Heparin)
		Fanconi Anaemia
		Bloom Syndrome
		Other—contact the lab

If requesting other non-WGS genetic testing, please tick the relevant boxes

## G. More resources:

### 1. Genomics Resources

[What is Genomics?](#)

[Genomics 101: Genomics in Healthcare](#)

[The Genomics Era: The Future of Genetics in Medicine](#)

[RCGP Genomics Toolkit](#)

[New Conditions Factsheet \(Genomics Education Programme\)](#)

### 2. WGS Resources

[Guide-to-requesting-WGS-RD-Nov-20.pdf \(hee.nhs.uk\)](#)

[Requesting whole genome sequencing: information for clinicians - Genomics Education Programme \(hee.nhs.uk\)](#)  
[Test order forms - North Thames GMS : North Thames GMS \(norththamesgenomics.nhs.uk\)](#)

[Whole Genome Sequencing - North Thames GMS : North Thames GMS \(norththamesgenomics.nhs.uk\)](#)

[Rare Diseases Test Order Forms and Clinician Packs](#) (scroll to Rare Disease- whole genome sequencing (WGS))

[Whole Genome Sequencing Animation – North Thames GMS](#)

[Genomic Question Time drop-in session](#) (Teams link)

- First Thursday of the month 12:30-13:00
- Passcode: aDYRNt
- Or contact us on: [nt-gmsa@gosh.nhs.uk](mailto:nt-gmsa@gosh.nhs.uk)

[Whole Genome Sequencing – Genetic Test Ordering](#)

How useful did you find this how-to guide? Please let us know how we can improve.