For full functionality of this form, use Adobe Reader or Acrobat. Web browser or Outlook views may be limited.

Genomic Medicine Service

Whole Genome Sequencing (WGS) Test Request
PLEASE DO NOT USE FOR NON-WGS TESTS

RARE AND INHERITED DISEASES

NHS

Requesting organisation:

GMS laboratory:

Proband's first name	Life status ☐ Alive ☐ Deceased; Ethnicity
Proband's last name	Family test ☐ Singleton ☐ Trio ☐ Other (provide number):
Date of birth *(dd/mm/yyyy)*; Hospital number	Relevant clinical information *Please include any previous molecular testing with date(s) and any other pertinent clinical information*
Sex assigned at birth ☐ Male ☐ Female	
Postcode	
NHS number	
Reason NHS Number not available: ☐ Patient not eligible for NHS number (e.g. foreign national) ☐ Other (please provide reason):	
Proband's age at onset of clinical features *years* *months*	

Test request

Test Directory Clinical Indication & code (reason for testing)

Additional panel(s) (if relevant; **mandatory for R89**)
(use panels with panel type 'GMS Rare Disease Virtual' - https://nhsgms-panelapp.genomicsengland.co.uk/)

State if specific rare disease is suspected or confirmed

Clinical Priority There is no urgent pathway, however prioritisation may be possible in exceptional circumstances. Please provide reason for urgency.

Reason for diagnostic test (Please provide additional information with other relevant clinical information above)

☐ Patient management: determining therapeutic decisions and/or clinical investigations and/or surveillance programme
☐ Patient, parents, or adult relative reproductive decision making
☐ Unaffected relatives are seeking predictive testing

Family members to be tested *(not required for proband only referrals)*

First name	Last name	Date of birth	NHS Number *(or postcode if not known)*	Sex assigned at birth	Deceased	Status	Ethnicity	Relationship to proband

Samples *being sent to GMS DNA extraction lab* ***(only required if also using this form for sample collection, please provide blood samples in EDTA)***

First name	Last name	Date of birth	Sample ID	Collection date / time	Sample type	Sample volume	Comments

☐ **I have attached a copy of the Record of Discussion form for all individuals**
☐ Patient conversation taken place; Record of Discussion form to follow

Also required - provide HPO terms on page 2

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

HPO terms are NECESSARY for the analysis and interpretation of WGS data and cannot commence until provided. Please ensure HPO terms match those available at https://hpo.jax.org/app/

HPO Terms At **least** one HPO term is required	Present	Absent	Unknown	Present	Absent	Unknown	Present	Absent	Unknown
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐	☐	☐	☐

The following list of HPO terms is provided as a guide but is not an exhaustive list. More terms are available at https://hpo.jax.org

Cardiology	**Developmental**	**Neurology**
Hypertrophic cardiomyopathy	Intellectual disability, moderate	Muscular dystrophy
Dilated cardiomyopathy	Intellectual disability, profound	Myopathy
Cardiomyopathy	Intellectual disability, severe	Myotonia
Immunology	Global developmental delay	Peripheral neuropathy
Immunodeficiency	Generalized hypotonia	Cognitive impairment
Abnormal lymphocyte count	Failure to thrive	Spasticity
Abnormality of humoral immunity	Abnormal facial shape	Chorea
Abnormal inflammatory response	Abnormality of metabolism/homeostasis	Dystonia
Opthalmology	Microcephaly	Ataxia
Cataract	Macrocephaly	Cerebellar atrophy
Retinal dystrophy	Tall stature	Cerebellar hypoplasia
Macular dystrophy	Short stature	Seizure
Renal	Skeletal dysplasia	
Multiple renal cysts	Hearing impairment	
Hepatic cysts		

Responsible clinician / consultant	**Main contact** (if different from responsible clinician/consultant)
Name:	Name:
Department address: Phone: Email:	Department address: Phone: Email:

For full functionality of this form, use Adobe Reader or Acrobat. Web browser or Outlook views may be limited.

First name	NHS number (or postcode if not known)
Last name	Date of birth (d d m m y y y y)

01-NGIS-ROD (v4.04)

Record of Discussion Regarding Genomic Testing

This form relates to the person being tested. One form is required for each person.

All of the statements below remain relevant even if the test relates to someone other than yourself, for example your child.

I have discussed genomic testing with my health professional and understand the following

Family and wider implications

1. The results of my test may have implications for me and members of my family. I understand that my results may also be used to help the healthcare of members of my family and others nationally and internationally. This could be done in discussion with me or through a process that will not personally identify me.

Uncertainty

2. The results of my test may have findings that are uncertain and not yet fully understood. To decide whether findings are significant for myself or others, my data may be compared to other patients' results across the country and internationally. I understand that this could change what my results mean for me and my treatment over time.

Unexpected information

3. The results of my test may also reveal unexpected results that are not related to why I am having this test. These may be found by chance and I may need further tests or investigations to understand their significance.

DNA storage

4. Normal NHS laboratory practice is to store the DNA extracted from my sample even after my current testing is complete. My DNA might be used for future analysis and/or to ensure that other testing (for example that of family members) is of high quality.

Data storage

5. The data from my genomic test will be securely stored so that it can be looked at again in the future if necessary.

Health records

6. Results from my genomic test will be part of my patient record, a copy of which is held in a national system only available to healthcare professionals.

Research

7. I understand that I have the opportunity to take part in research which may benefit myself or others, now or in the future. An offer to join a national research opportunity is available on the following page.

For any further questions, my healthcare professional can provide information. More information regarding genomic testing and how my data is protected can be found at www.nhs.uk/conditions/genetics

Please sign on page three to confirm your agreement to the genomic test.

First name	NHS number (or postcode if not known)
Last name	Date of birth
	d d m m y y y y

01-NGIS-ROD (v4.04)

The National Genomic Research Library

The NHS invites you to contribute to the National Genomic Research Library, managed by Genomics England.

Genomics England was set up in 2013 by the Department of Health and Social Care to work with the NHS to build a library of human genomes for researchers to study. Combining data from many different patients helps researchers to better understand disease and spot patterns in the data.

By agreeing to share your data you might get results which could lead to your own diagnosis, a new treatment, or offers to take part in clinical trials. Your taking part could enable diagnoses for people who don't have one.

Please read the following statements. Feel free to ask any questions before making a decision.

By saying 'yes' to research, I understand the following

The National Genomic Research Library

1. NHS England, on behalf of the Trusts that provided your genomic test, will allow Genomics England to access my personal data including my genomic record.

Security

2. Any samples and data stored by Genomics England and the NHS will always be stored securely. Genomics England will take all reasonable steps to ensure that I cannot be personally identified.

Re-Contact

3. My clinical team or Genomics England together with my clinical team, can contact me if the data or samples reveals any clinical trials or other research that I might benefit from.

4. If something is relevant to me or my family, there is a process by which this will be shared with my NHS clinical team.

Data and sample usage

5. Researchers may include national or international scientists, healthcare companies and NHS staff. To access the data,these researchers must all be approved by an independent committee of experts, including health professionals, clinical academics and patients. There will be no access to the data by personal insurers and marketing companies.

Data storage

6. Genomics England will collect different aspects of my health data from the NHS and other data from organisations listed at https://www.genomicsengland.co.uk/privacy-policy/. The collection and analysis of my health data for research will continue across my entire lifetime and beyond.

Withdrawal

7. I can change my mind about taking part at any time.

More information regarding research in the National Genomic Research Library can be found at www.genomicsengland.co.uk For any further questions, my healthcare professional can provide information.

Please use page three to indicate your research choices.

First name	NHS number (or postcode if not known)
Last name	Date of birth (d d m m y y y y)

01-NGIS-ROD (v4.04)

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
.....	.....	d d m m y y y y

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* * please amend as appropriate	Signature	Date
.....	.....	d d m m y y y y

Healthcare professional use only

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

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

Healthcare professional name	Signature	Date
.....	.....	d d m m y y y y

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For any further questions, my healthcare professional can provide information. More information regarding genomic testing and how my data is protected can be found at www.nhs.uk/conditions/genetics

Please sign on page three to confirm your agreement to the genomic test.

First name	NHS number (or postcode if not known)
Last name	Date of birth
	d d m m y y y y

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Re-Contact

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01-NGIS-ROD (v4.04)

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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
.....	.....	d d m m y y y y

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Hospital number		

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Last name	Date of birth
	d d m m y y y y

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First name	NHS number (or postcode if not known)
Last name	Date of birth (d d m m y y y y)

01-NGIS-ROD (v4.04)

Confirmation of Your Genomic Test and Research Choices

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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
.....	.....	d d m m y y y y

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* * please amend as appropriate	Signature	Date
.....	.....	d d m m y y y y

Healthcare professional use only

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

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

Healthcare professional name	Signature	Date
.....	.....	d d m m y y y y

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A SYNLAB pathology partnership

Guy's and St Thomas' NHS
NHS Foundation Trust

GENETICS SPECIMEN FORM

Genetics Laboratories, 5th Floor, Tower Wing, Guy's Hospital, Great Maze Pond, London, SE1 9RT

T: 020 7188 1696/1709
gstt.viapathgeneticsadmin@nhs.net
gstt.southeastglh@nhs.net

Surname:		**Sex assigned at birth:**	☐ Male ☐ Female
First Name:		**Ethnic origin:**	
Previous Name:		**Hospital number:**	
DOB:		**PRU Number:**	
Address:		**Post code:**	
NHS Number (Mandatory):	- -	**Private patient (please attach invoicing details):**	☐
GP name:		**GP Post code:**	
Consultant:		**Referring Hospital**:	

Signed: _____ **Date:** _____

Name: _____ **Email:** _____

Invoice address if different from referral address:

Samples please ensure specimens are dispatched to the laboratory promptly after sampling

Blood in potassium EDTA (DNA / MLPA / array CGH)	☐ 3-5 ml	Date of collection _____
Blood in lithium heparin (Chromosome rearrangements / Biochemical Genetics)	☐	Time of collection _____
Prenatal sample Please tick one:	CVS ☐ AF ☐ POC ☐	
Other – Please state		

Tests requested
NB For testing for chromosome imbalance (array CGH/chromosome analysis), please provide clinical details on the reverse of this form.

2x 1-3ml EDTA for Whole Genome Sequencing to be sent to Guys Genetics Laboratories

Samples for WGS - Proband

Clinical Details Please include full details of patient, with pedigree if relevant)
NB For testing for chromosome imbalance (array CGH/chromosome analysis), please provide clinical details on the reverse of this form.

In submitting this sample, the clinician confirms that **consent has been obtained**:
(a) for testing and possible storage
(b) for the use of this sample and the information generated from it to be shared with members of the donor's family and their health professionals (if appropriate).
(c) we assume that consent has been obtained for sensitive disposal of any fetal remains unless otherwise stated. **Please do NOT send the consent form**

Has this case been discussed with the Genetics Department? If so, with whom?

Is the patient pregnant? Yes ☐ / ☐ No

If YES: how many weeks gestation?

All fields above are mandatory. Samples supplied with inadequate or illegible information, will be subject to delay or rejection.

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GENETICS SPECIMEN FORM

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T: 020 7188 1696/1709
gstt.viapathgeneticsadmin@nhs.net
gstt.southeastglh@nhs.net

Surname:		**Sex assigned at birth:**	☐ Male ☐ Female
First Name:		**Ethnic origin:**	
Previous Name:		**Hospital number:**	
DOB:		**PRU Number:**	
Address:		**Post code:**	
NHS Number (Mandatory):		**Private patient (please attach invoicing details):**	☐
GP name:		**GP Post code:**	
Consultant:		**Referring Hospital**:	

Signed: _____ **Date:** _____

Name: _____ **Email:** _____

Invoice address if different from referral address:

Samples please ensure specimens are dispatched to the laboratory promptly after sampling

Blood in potassium EDTA (DNA / MLPA / array CGH)	☐ 3-5 ml	Date of collection _____
Blood in lithium heparin (Chromosome rearrangements / Biochemical Genetics)	☐	Time of collection _____
Prenatal sample Please tick one:	CVS ☐ AF ☐ POC ☐	
Other – Please state		

Tests requested
NB For testing for chromosome imbalance (array CGH/chromosome analysis), please provide clinical details on the reverse of this form.

2x 1-3ml EDTA for Whole Genome Sequencing to be sent to Guys Genetics Laboratories

Duo/Trio WGS samples

of

Clinical Details Please include full details of patient, with pedigree if relevant)
NB For testing for chromosome imbalance (array CGH/chromosome analysis), please provide clinical details on the reverse of this form.

In submitting this sample, the clinician confirms that **consent has been obtained**:
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Surname:		**Sex assigned at birth:**	☐ Male ☐ Female
First Name:		**Ethnic origin:**	
Previous Name:		**Hospital number:**	
DOB:		**PRU Number:**	
Address:		**Post code:**	
NHS Number (Mandatory):		**Private patient (please attach invoicing details):**	☐
GP name:		**GP Post code:**	
Consultant:		**Referring Hospital**:	

Signed: _____ **Date:** _____

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of

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