NHS East Genomic Laboratory Hub Clinician Guidance
Ordering Whole Genome Sequencing for Rare Disease

East Genomic Laboratory Hub webpage for Whole Genome Sequencing - Rare Disease:
Whole genome sequencing (rare disease) | East Genomics

Questions - email emee.glh@nhs.net (include "WGS Rare Disease" in the subject heading)

Sending Forms and Samples

- **WGS Test Order Forms and samples should be sent to the Leicester, Nottingham or Cambridge Genomic Laboratories:**
  Laboratory contact details | East Genomics

- **Test Order Forms and Record of Discussion forms should be emailed to** emee.glh@nhs.net

1) **Guidance:**
   See national Clinician Guidance for ordering WGS for Rare Disease here:
   Requesting whole genome sequencing: information for clinicians - Genomics Education Programme (hee.nhs.uk)

   Patient information leaflets are available here:
   NHS England » Whole genome sequencing patient information leaflets

2) **Clinical Indications:**
   Clinical Indications that use WGS for testing patient samples are listed within the National Genomic Rare Disease Test Directory (indicated by "WGS" in the Test Method).

3) **Eligibility:**
   Check the eligibility of your patient for the relevant Clinical Indication.
   This information can be found in the Rare and Inherited Disease eligibility criteria document on the National Genomic Test Directory webpage.

4) **Patient Choice Consent Framework Training:**
   Take the NHS East Genomic Laboratory Hub online Patient Choice training: https://elearning.cam-pgmc.ac.uk/

   We strongly suggest that you complete this training prior to submitting samples to the NHS East Genomic Laboratory Hub. If you have issues logging into this portal from your trust please contact gemma.chandratillake@nhs.net

5) **Family members:** The diagnostic yield of WGS is increased by the inclusion of parental samples to enable a trio analysis. Therefore, wherever possible a trio should be submitted for testing. In some circumstances parental samples may not be available for testing, therefore please submit duos or singletons. Please contact the team if you wish to submit multiple affected individuals per family or additional family samples.

6) **Ordering a Test**:
Download a WGS Test Order Form and save the document on your computer for electronic editing or print out for completion by hand.
One Test Order Form is required per proband.
The Test Order Form can be downloaded here:
NHS England » NHS Genomic Medicine Service test order forms

7) **Ordering a Test - CUH Clinicians**:
CUH Clinicians should use EPIC to place a WGS test for each family member.
CUH Epic Test codes:

LAB 9956 Rare Disease Genomic Testing
The EPIC form will take you through the questions required for a WGS test order.

LAB 7298 Genomic Test Request on Stored Sample
The EPIC form will take you through the questions required for a WGS test order.

8) **Record of Discussion forms:**
Download the Record of Discussion Forms (RoD) and save the document on your computer for electronic editing or print out for completion by hand.
One ROD should be completed for each family member tested.

The ROD form can be downloaded here:
https://www.england.nhs.uk/publication/nhs-genomic-medicine-service-record-of-discussion-form/

9) **National Genomic Research Library:**

The opportunity to participate in the National Genomic Research Library (NGRL) should be discussed with all family member tested using WGS and their choices recorded on the ROD form**.**

Information on the National Genomic Research Library for clinicians can be found here:
https://www.genomicseducation.hee.nhs.uk/supporting-the-nhs-genomic-medicine-service/national-genomic-research-library-information-for-clinicians/

Information on the National Genomic Research Library for patients can be found here:
https://www.genomicsengland.co.uk/patients-participants/taking-part/resources

10) **Remote appointments:**
If the appointment is conducted remotely and/or if you have a separate conversation by phone with a family member, you can record the individual's choices on the RoD form without the need for their signature. See example RoD form below.

11) Arranging samples:

Arrange for samples to be taken and sent to the Leicester, Nottingham or Cambridge East GLH Laboratories using our normal referral form (Rare disease tests - non WGS | East Genomics) or your local test request process.

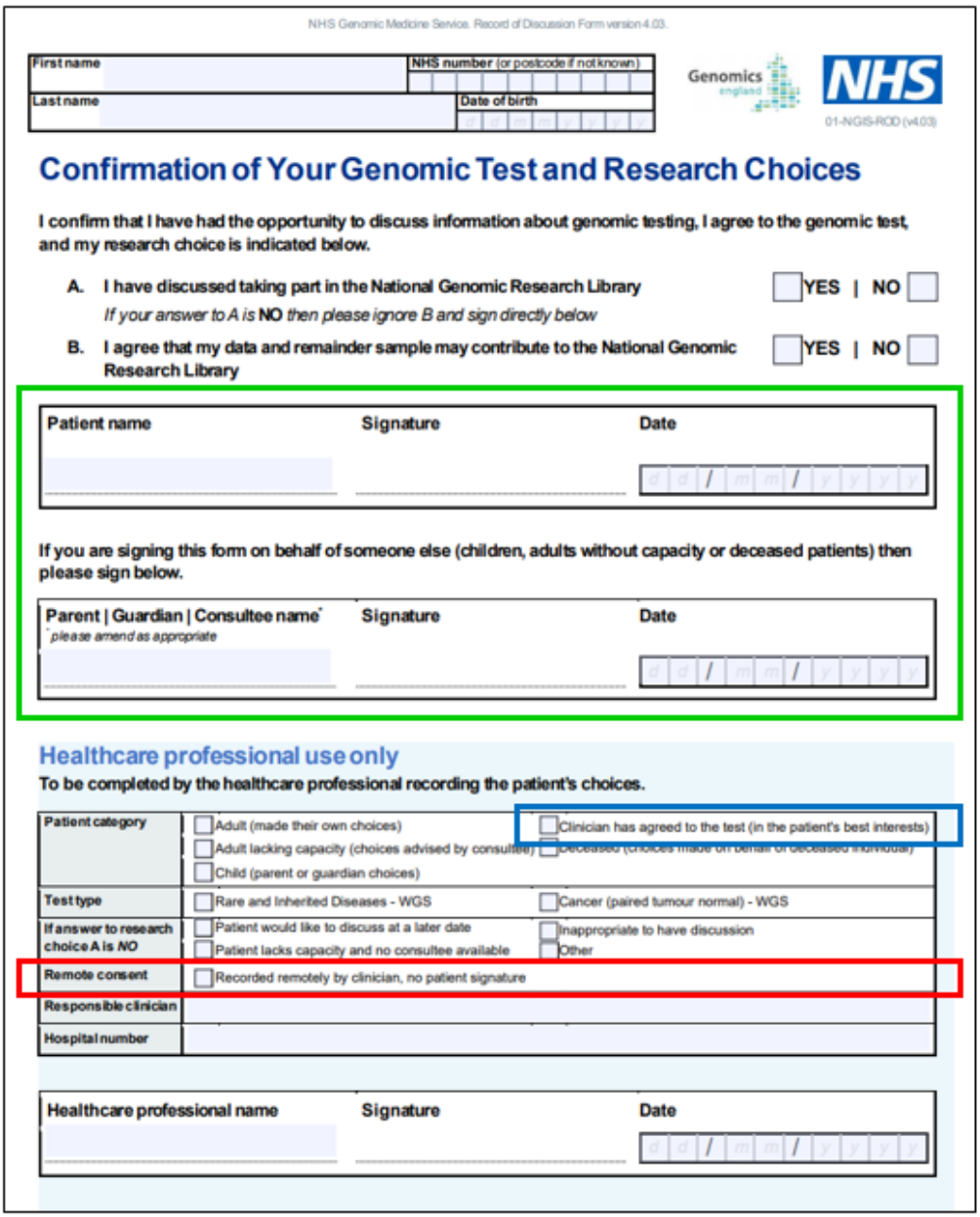
Clearly indicate "WGS for Rare Disease – East GLH" on the referral forms.

Samples should be **peripheral blood in EDTA**.

12) Arranging samples – stored DNA:

If DNA is already stored for an individual, please arrange for an aliquot to be transferred to the CUH Genomic Laboratory. You could do this by emailing the WGS Test order form to the laboratory holding the DNA sample. Please indicate that the DNA is required for WGS (a larger amount of DNA is required for WGS).

Example - Record of Discussion - Patient Choice Consent obtained remotely - annotated 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

Genomics england | NHS

01-NGIS-ROD (v4.03)

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)	☐ Clinician has agreed to the test (in the patient's best interests)
	☐ Adult lacking capacity (choices advised by consultee)	☐ Deceased (choices made on behalf of deceased individual)
	☐ Child (parent or guardian choices)	
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	☐ Inappropriate to have discussion
	☐ Patient lacks capacity and no consultee available	☐ 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

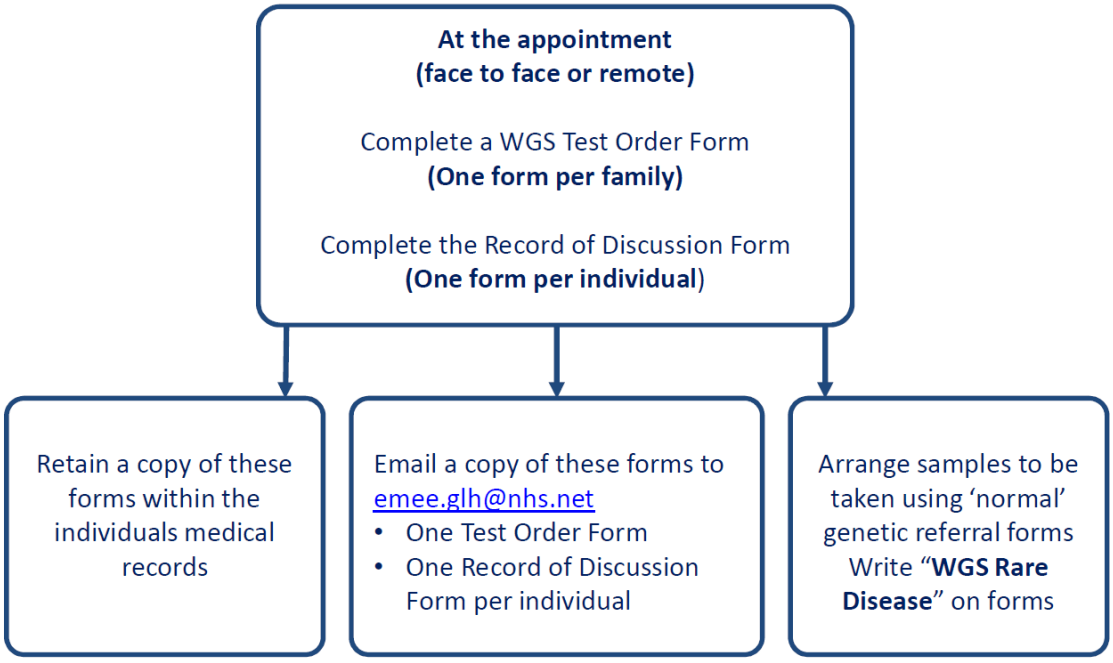
Leave blank if consent was obtained during a remote appointment

Only check this box for adults lacking capacity and where no consultee is available

Make a note here if consent was obtained during a remote appointment

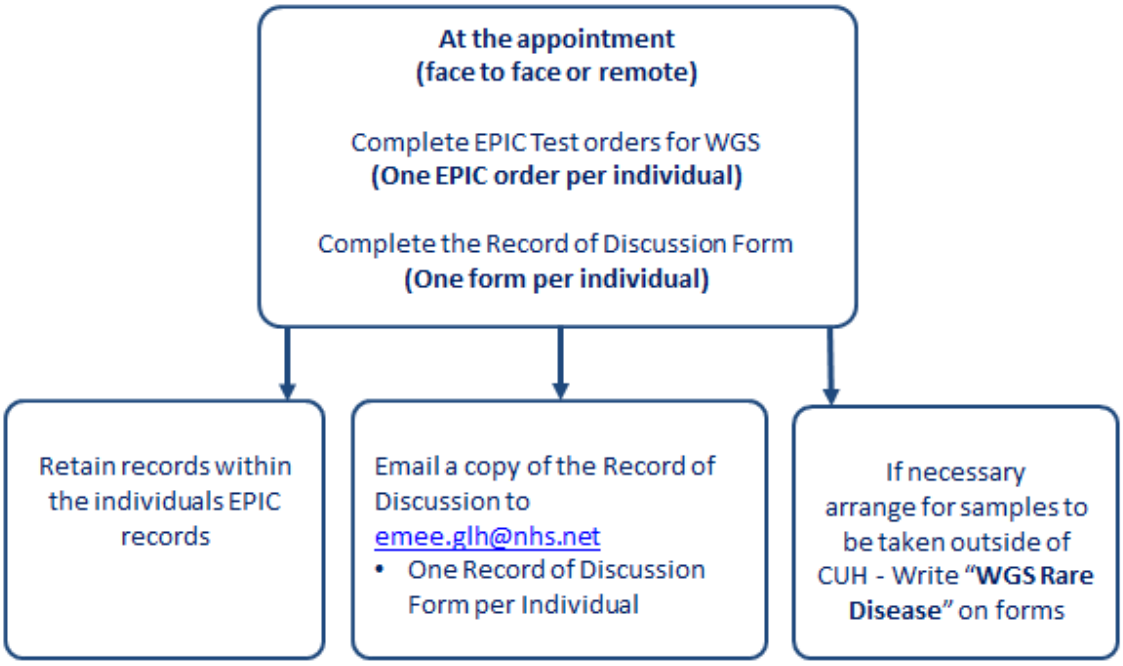
WGS Rare Disease – Flow Chart

East Genomic Laboratory Hub



CUH EPIC WGS Rare Disease – Flow Chart

East Genomic Laboratory Hub



CUH EPIC Test codes

LAB 9956	Rare Disease Genomic Testing
LAB 7298	Rare Disease Genomic Testing on stored sample