



North West
NHS Genomic Laboratory Hub

Genomic Testing Request Form Rare Disease

(DOC4900 Revision 1)

Lab use only
Lab No:

Patient Details – use sticker if available but please add any missing information

Referring Clinician/Healthcare Professional

NHS No:		D.O.B.:		Full Name:	
Surname:		Birth Gender:		Contact E-mail/Tel:	
Forename:		Ethnicity:		Hospital:	
Patient's Address:		Hospital No:		Department:	
	Postcode:	Clinical Genetics No:		Copy Report to (E-mail):	

Test Required – please refer to National Genomic Test Directory (<https://www.england.nhs.uk/publication/national-genomic-test-directories/>).
N.B. Samples will not be accepted for testing if the Clinical Indication Code and Test Code have not been provided. Please highlight any exceptions

Clinical Indication Code
(e.g. R53, Fragile X):

R

Test Code & Method
(e.g. R53.1, STR testing)

R

Clinical Details

Type of Test (please tick):

- Diagnostic Test
- Predictive/Pre-symptomatic Test
- Prenatal Test (please indicate fetal gestation below)
- Carrier Test (recessive disorder)
- Family studies
- Long term storage of cells (please state tissue type below)
- DNA STORAGE ONLY, NO TESTING (Tick this box ONLY)**

Consent Statement: Receipt of this form and sample(s) by the laboratory assumes that the clinician has obtained consent for genomic testing and for the use of the DNA sample(s) and/or test result(s) by healthcare professionals in the UK for family testing and quality control purposes.

Please tick if the patient does NOT want any remaining DNA, RNA or cells stored in the laboratory

- By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the: [National Genomic Test Directory](https://www.england.nhs.uk/publication/national-genomic-test-directories/)
- Please list how the patient meets the testing criteria and provide any additional pertinent clinical information and/or details of affected family members and familial variants.

N.B. WGS requests and certain specialist services require an additional proforma:
<https://mft.nhs.uk/nwglh/documents/test-request-forms/>

Specimen Details

High Infection Risk? Yes No

Sample Date:

Taken by:

Sample Type:

Further Details:

Fetal Gestation:

Once taken, samples should be sent to your local Genomics Laboratory



<https://mft.nhs.uk/nwglh/>

Laboratory Opening Hours:
09:00 – 17:00, Monday to Friday

Manchester

North West Genomic Laboratory Hub – Manchester Site
Manchester Centre for Genomic Medicine
Sample Reception (6th Floor)
St Mary's Hospital
Oxford Road
Manchester
M13 9WL

Tel: 0161 276 6122
Email: mft.genomics@nhs.net

Liverpool

North West Genomic Laboratory Hub – Liverpool Site
Manchester Centre for Genomic Medicine
Sample Reception (2nd Floor)
Liverpool Women's Hospital
Crown Street
Liverpool
L8 7SS

Tel: 0151 702 4228
Email: dna.liverpool@nhs.net

Guidance Notes – Genomic Testing Request Form – Rare Disease

Patient Details

The following details are mandatory, other details should be completed as fully as possible:

- **Surname & Forename**
- **D.O.B** – Date of Birth
- **NHS Number** (10 digits)
- Patient's **Birth Gender**
- Patient's **Postcode**

Please ensure a minimum of 3 matching identifiers on tubes and form.

Referring Clinician/Healthcare Professional

The following details are mandatory:

- **Name:** initials are not acceptable as the laboratory cannot identify the clinician/healthcare professional. A minimum of first initials and surname in full must be provided. For prenatal referrals, a **Midwife** name can be included if appropriate.
- **Hospital** should be clearly identifiable; initials are not acceptable as the laboratory cannot identify the hospital. Trusts with more than one hospital should clearly identify the referring hospital.
- **Department** should be clearly identifiable; initials are not acceptable as the laboratory cannot identify the department.

Other details should be completed as fully as possible:

- **E-mail/Tel;** without an email/telephone number, urgent results cannot be given. Reports will only be sent by first class post.

Copy Report to: if more space is required, use the Clinical Details box.

Clinical Indication Code and Test Code

Clinical Indication Code and Test Code are mandatory fields, however these codes may not be available, e.g. for a test that is in development. Please highlight these exceptions on the request form. More than one Clinical Indication Code and Test Code can be requested when relevant to the investigation, ensuring the appropriate sample type(s) are supplied for the requested test(s).

Full details of Test Required and Clinical Details must be supplied to ensure the correct analysis is performed. Illegible forms will result in delays for results. As much detail as possible should be provided, if required additional reports and letters can be attached to this referral form.

Specimen Details

High Infection Risk: In accordance with the Health & Safety at Work Act and COSHH Regulations, the laboratory must be informed of any infection risk associated with submitted samples. The sender has the responsibility for minimising the risk to laboratory staff by giving sufficient information to enable the laboratory to take appropriate safety precautions when testing a specimen.

Postnatal samples for DNA extraction (all genomic testing except FISH and Karyotyping) – Store sample at 4°C if required, send by courier or first class post. N.B. Blood samples for NF1 testing should be kept at room temperature.

- **Venous Blood:** use EDTA tube only:
 - 4ml for adults and children (BD Vacutainer preferred).
 - 1ml minimum for neonates (Sarstedt Micro Tube preferred).
- **Saliva Samples:** GeneFiX or Oragene collection kits only.
- **Other Sample Types:** by prior arrangement only.

Postnatal samples for Karyotyping/FISH – Store overnight at 4°C if required, DO NOT freeze or expose to heat. The sample must arrive in laboratory within 48 hours of being taken.

- **Venous Blood:** use Lithium Heparin (Li-Hep) tube only:
 - 4ml for adults and children.
 - 1ml minimum for neonates.
- **Solid Tissue:** DO NOT expose to formalin. Send in dry sterile plastic container (or if stored overnight in sterile saline).

Prenatal samples – Store overnight at 4°C if required, DO NOT freeze or expose to heat. The sample must arrive within 24 hours of being taken.

- **Amniotic Fluid:** 10-20ml in sterile leak proof plastic universal.
- **Chorionic Villi:** 10-30mg in sterile transport media. See guidance on website for further information.
- **Fetal Blood:** 1ml in a paediatric Li-Hep tube, mix well to prevent clotting.

Tissue Type: For solid tissue samples the tissue type should be specified. For fetal tissue samples the date of delivery and gestation must be included. Fetuses cannot be accepted under any circumstances.

Sample Packaging: The sample container should be sealed in a biohazard bag in case of a leakage. To prevent contamination of referral form and paperwork this should not be sealed with the sample. All packaging should conform to UN650 standards (as applied to UN3373 – Biological Samples, Category B).

Factors known to affect the performance of the examination/interpretation of the results: If this patient has had a bone marrow transplant/blood transfusion please contact the laboratory to discuss testing options prior to sending a sample.

This area is for Lab use only