NHS
Liverpool Women's
NHS Foundation Trust

The Test: Purpose and Process

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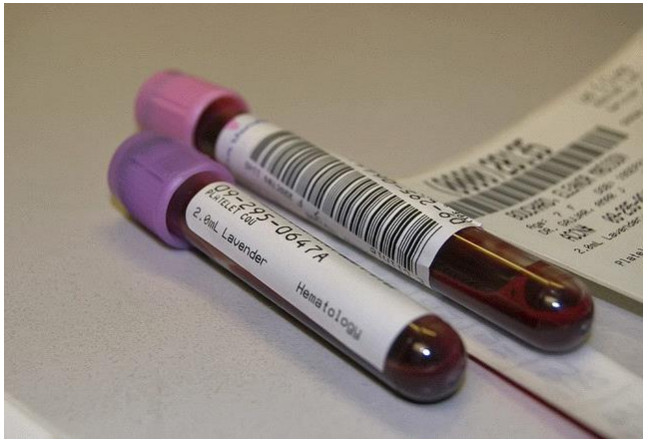
NHS
North West
NHS Genomic Laboratory Hub

NHS
North West Coast
NHS Genomic Medicine Centre

The Test: Purpose and Process

Liverpool Women's NHS Foundation Trust

- Sample requirements.
- Time frames
- Sample storage in lab
- Test request paperwork



North West NHS Genomic Laboratory Hub

North West Coast NHS Genomic Medicine Centre

The Test: Purpose and Process

Liverpool Women's NHS Foundation Trust

- Sample requirements:
- **Venous Blood**: use EDTA tube only:
- 4ml for adults (BD Vacutainer preferred).
- **Saliva Samples**: GeneFiX or Oragene collection kits only.
- **Other Sample Types:** by prior arrangement only.

The Test: Purpose and Process

Liverpool Women's NHS Foundation Trust

- Time Frame
- Results will take approximately 6 weeks

NHS North West NHS Genomic Laboratory Hub

NHS North West Coast NHS Genomic Medicine Centre

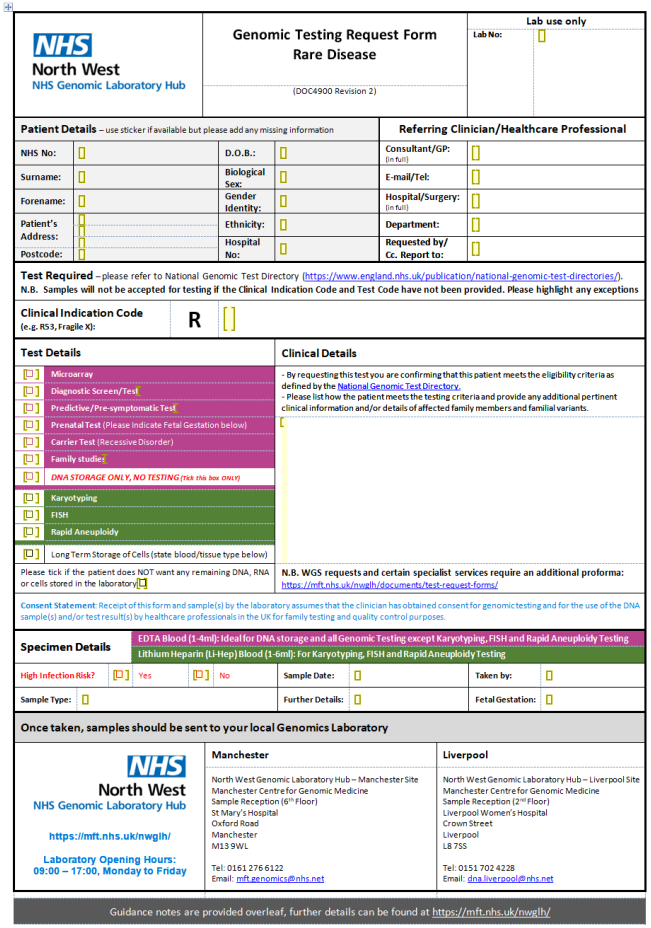
The Test: Purpose and Process

Liverpool Women's NHS Foundation Trust

- Sample storage in the lab
- DNA will be stored in lab in case further testing is required unless patient opts otherwise.

The Test request paperwork:

Liverpool Women's NHS Foundation Trust



NHS North West NHS Genomic Laboratory Hub

Genomic Testing Request Form
Rare Disease

(DOC4900 Revision 2)

Lab use only

Lab No:

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

NHS No:		D.O.B.:		Consultant/GP: (in full)	
Surname:		Biological Sex:		E-mail/Tel:	
Forename:		Gender Identity:		Hospital/Surgery: (in full)	
Patient's Address:		Ethnicity:		Department:	
Postcode:		Hospital No:		Requested by/ Cc. Report to:	

Referring Clinician/Healthcare Professional

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. RS3, Fragile X): R

Test Details

- Microarray
- Diagnostic Screen/Test
- Predictive/Pre-symptomatic Test
- Prenatal Test (Please Indicate Fetal Gestation below)
- Carrier Test (Recessive Disorder)
- Family studies
- *DNA STORAGE ONLY, NO TESTING (Tick this box ONLY)*
- Karyotyping
- FISH
- Rapid Aneuploidy
- Long Term Storage of Cells (state blood/tissue type below)

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

Clinical Details

- By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the National Genomic Test Directory.
- 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/

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.

Specimen Details

EDTA Blood (1-4ml): Ideal for DNA storage and all Genomic Testing except Karyotyping, FISH and Rapid Aneuploidy Testing
Lithium Heparin (Li-Hep) Blood (1-6ml): For Karyotyping, FISH and Rapid Aneuploidy Testing

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

NHS North West NHS Genomic Laboratory Hub

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

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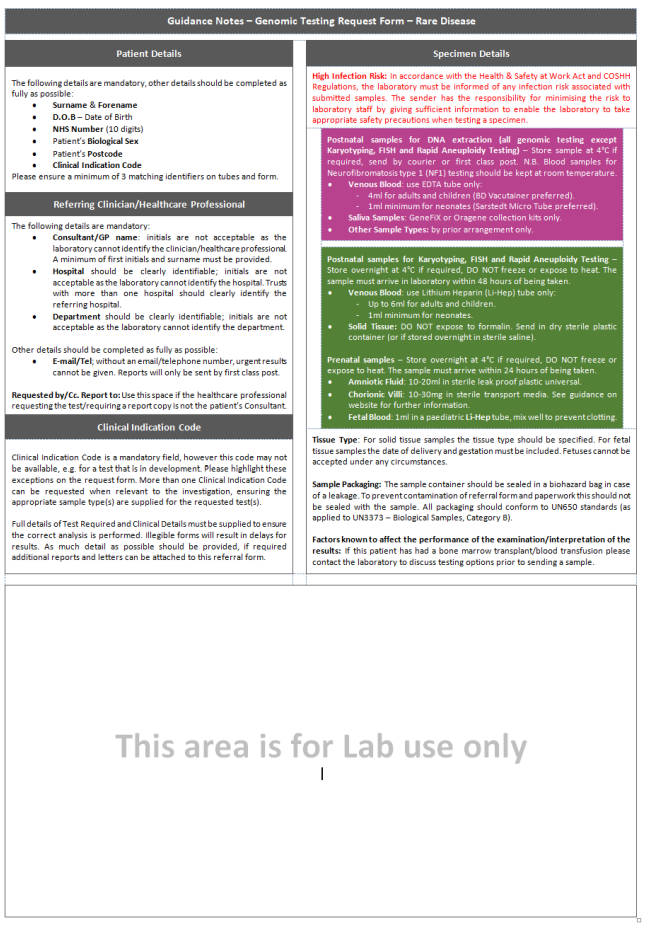
Guidance notes are provided overleaf, further details can be found at https://mft.nhs.uk/nwglh/



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 **Biological Sex**
- Patient's **Postcode**
- **Clinical Indication Code**

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

Referring Clinician/Healthcare Professional

The following details are mandatory:

- **Consultant/GP name**: initials are not acceptable as the laboratory cannot identify the clinician/healthcare professional. A minimum of first initials and surname must be provided.
- **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.

Requested by/Cc. Report to: Use this space if the healthcare professional requesting the test/requiring a report copy is not the patient's Consultant.

Clinical Indication Code

Clinical Indication Code is a mandatory field, however this code 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 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 Karyotyping, FISH and Rapid Aneuploidy Testing) – Store sample at 4°C if required, send by courier or first class post. N.B. Blood samples for Neurofibromatosis type 1 (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 and Rapid Aneuploidy Testing – 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:
  - Up to 6ml 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).

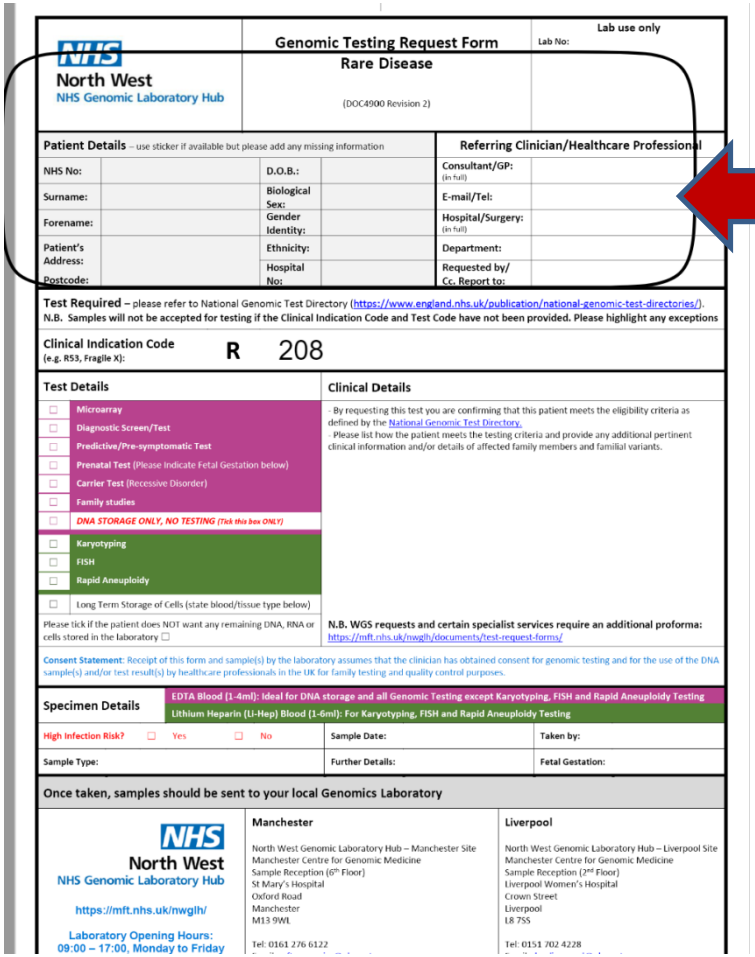
Factors known to affect the performance of the examination/interpretation of the results: If this patient has had a bone marrow transplant/blood transfusion please contact the laboratory to discuss testing options prior to sending a sample.

This area is for Lab use only

NHS North West NHS Genomic Laboratory Hub

NHS North West Coast NHS Genomic Medicine Centre

Patient demographics:



NHS North West NHS Genomic Laboratory Hub

Genomic Testing Request Form
Rare Disease
(DOC4900 Revision 2)

Lab use only
Lab No:

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

NHS No:		D.O.B.:		Consultant/GP: (in full)	
Surname:		Biological Sex:		E-mail/Tel:	
Forename:		Gender Identity:		Hospital/Surgery: (in full)	
Patient's Address:		Ethnicity:		Department:	
Postcode:		Hospital No:		Requested by/ Cc. Report to:	

Referring Clinician/Healthcare Professional

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 208**

Test Details

- ☐ Microarray
- ☐ Diagnostic Screen/Test
- ☐ Predictive/Pre-symptomatic Test
- ☐ Prenatal Test (Please Indicate Fetal Gestation below)
- ☐ Carrier Test (Recessive Disorder)
- ☐ Family studies
- ☐ *DNA STORAGE ONLY, NO TESTING (Tick this box ONLY)*
- ☐ Karyotyping
- ☐ FISH
- ☐ Rapid Aneuploidy
- ☐ Long Term Storage of Cells (state blood/tissue type below)

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

Clinical Details

- By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the National Genomic Test Directory.
- 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/

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.

Specimen Details

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Lithium Heparin (LI-Hep) Blood (1-6ml): For Karyotyping, FISH and Rapid Aneuploidy Testing

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

NHS North West NHS Genomic Laboratory Hub
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Laboratory Opening Hours: 09:00 – 17:00, Monday to Friday

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North West Genomic Laboratory Hub – Liverpool Site
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L8 7SS
Tel: 0151 702 4228

Patient details

Mandatory Fields. Can use sticker and fill in additional boxes by hand

NHS North West
NHS Genomic Laboratory Hub

Genomic Testing Request Form
Rare Disease
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Lab use only
Lab No:

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

NHS No:		D.O.B.:	
Surname:		Biological Sex:	
Forename:		Gender Identity:	
Patient's Address: Postcode:		Ethnicity:	
		Hospital No:	

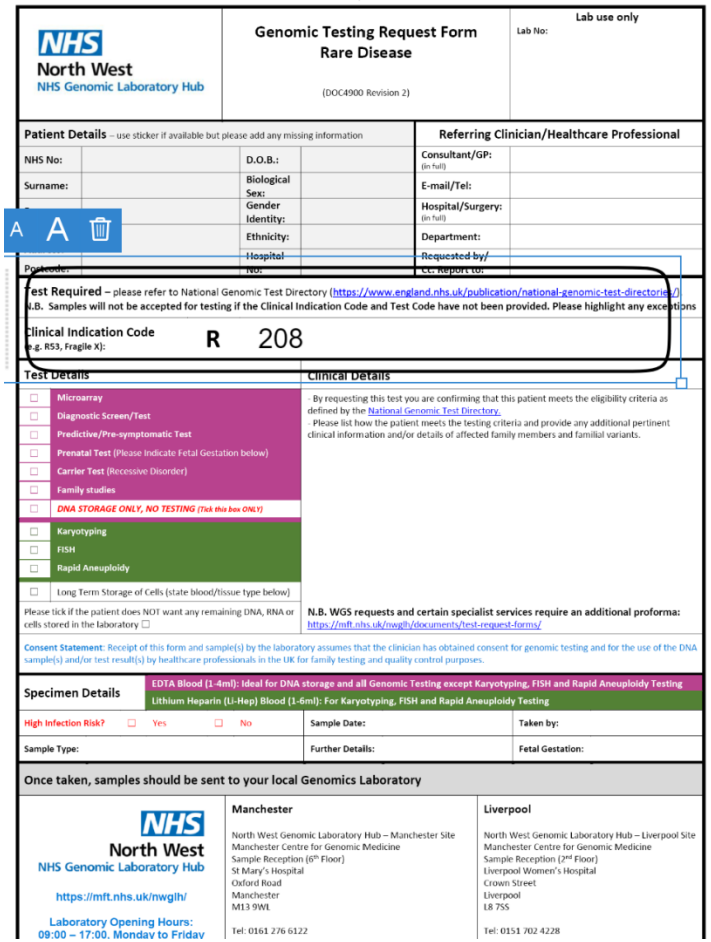
Referring Clinician/Healthcare Professional

Consultant/GP: (in full)	
E-mail/Tel:	
Hospital/Surgery: (in full)	
Department:	
Requested by/ Cc. Report to:	

Complete in full.

Use if clinician ordering not pts consultant

Test required:



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~~CC. Report to:~~	

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Test Details	Clinical Details
☐ Microarray ☐ Diagnostic Screen/Test ☐ Predictive/Pre-symptomatic Test ☐ Prenatal Test (Please Indicate Fetal Gestation below) ☐ Carrier Test (Recessive Disorder) ☐ Family studies ☐ *DNA STORAGE ONLY, NO TESTING (Tick this box ONLY)* ☐ Karyotyping ☐ FISH ☐ Rapid Aneuploidy ☐ Long Term Storage of Cells (state blood/tissue type below)	- By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the National Genomic Test Directory. - 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.
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High Infection Risk? ☐ Yes ☐ No		Sample Date:	Taken by:
Sample Type:		Further Details:	Fetal Gestation:

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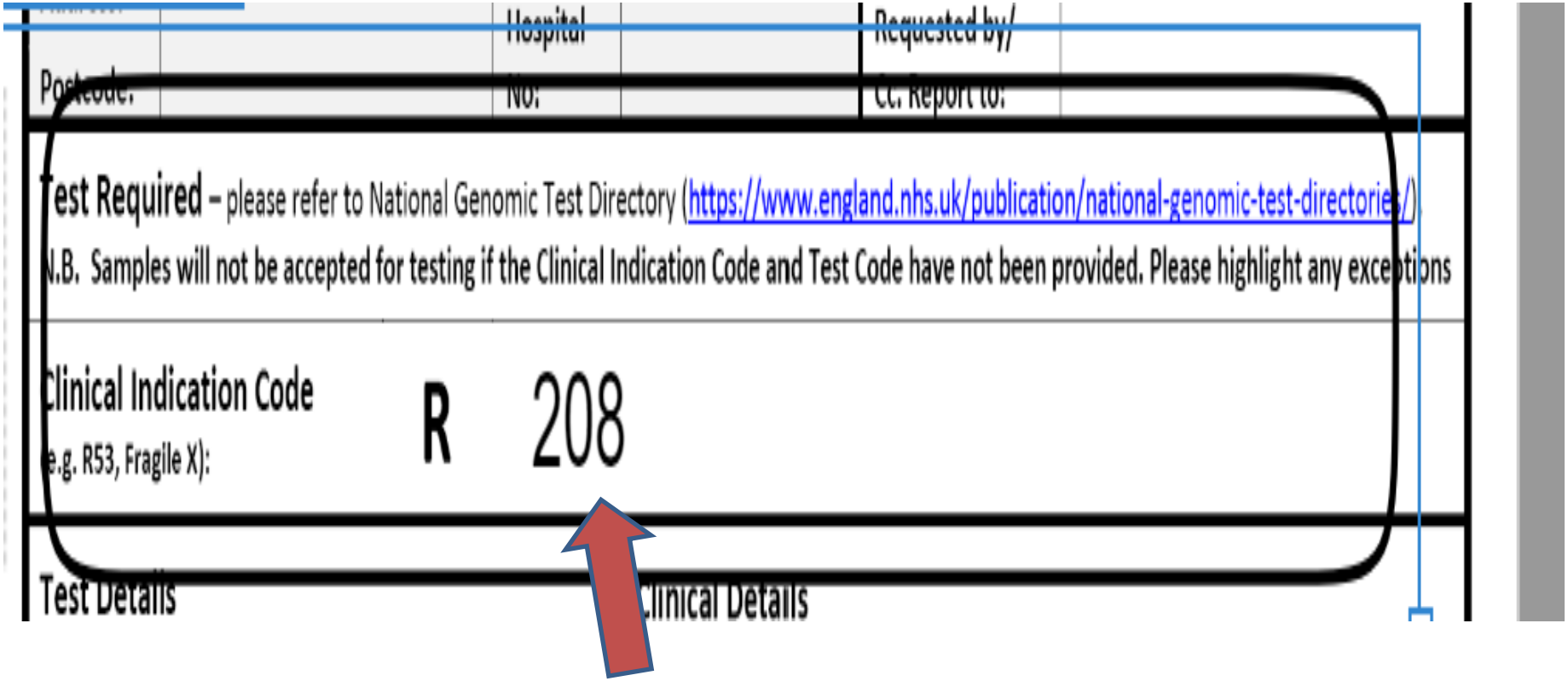
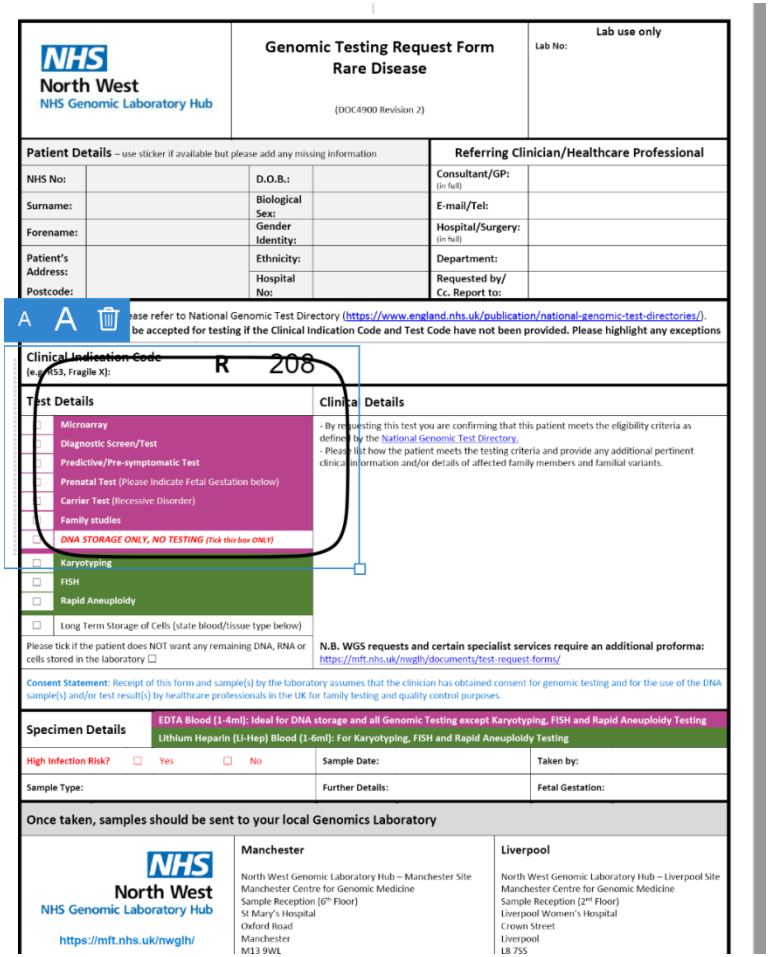


Pre-filled test code

Test Details



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Genomic Testing Request Form
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Surname:		Biological Sex:	
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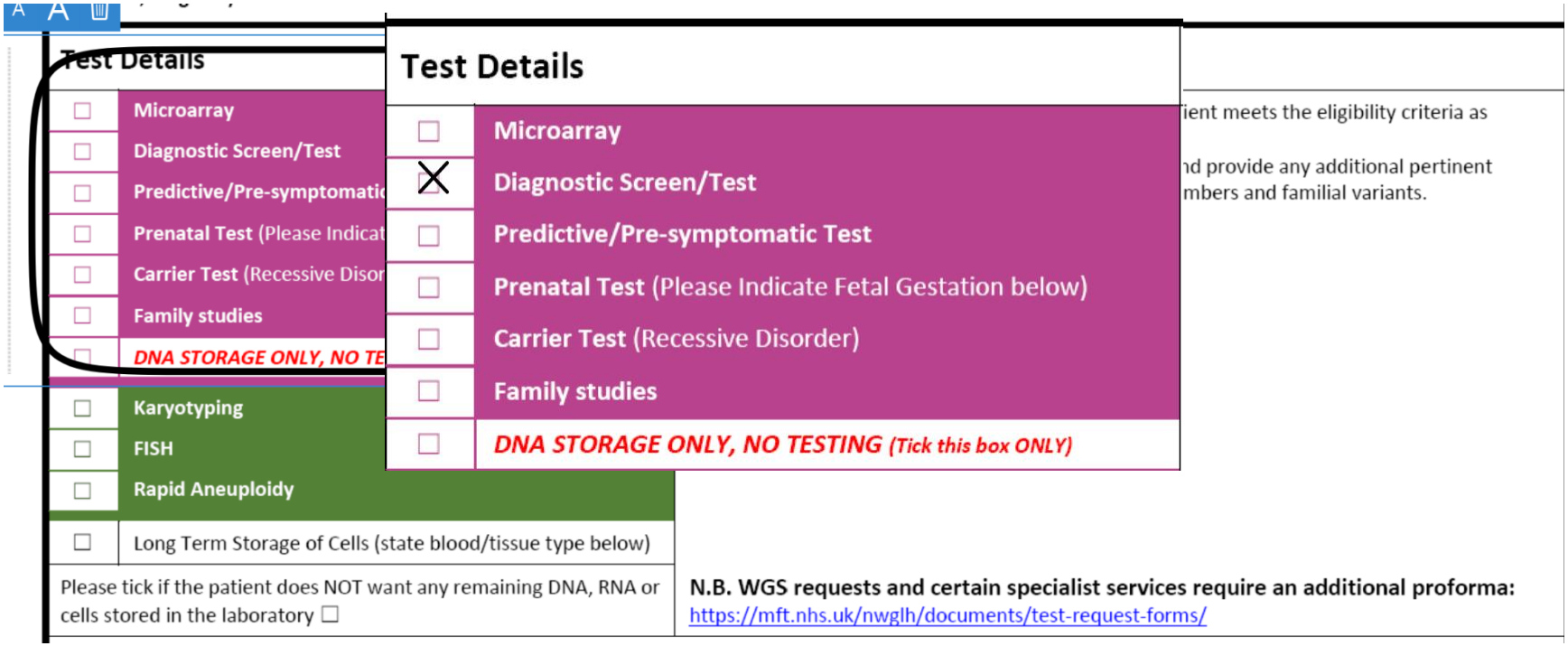

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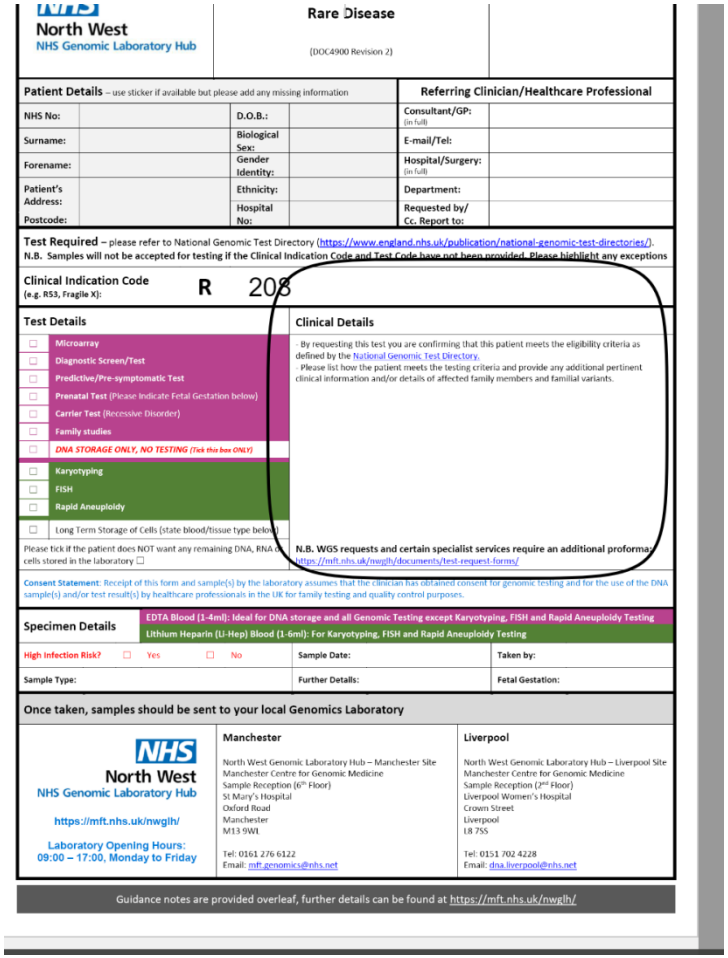

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Rare Disease

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Surname:		Biological Sex:		E-mail/Tel:	
Forename:		Gender Identity:		Hospital/Surgery: (in full)	
Patient's Address: Postcode:		Ethnicity:		Department:	
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High Infection Risk? ☐ Yes ☐ No		Sample Date:	Taken by:
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Guidance notes are provided overleaf, further details can be found at https://mft.nhs.uk/nwglh/

Clinical Details

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- 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.

Patient has a personal history of triple negative breast cancer aged 35 years and wider family history in her mother aged 54 and maternal Aunt aged 49 years.

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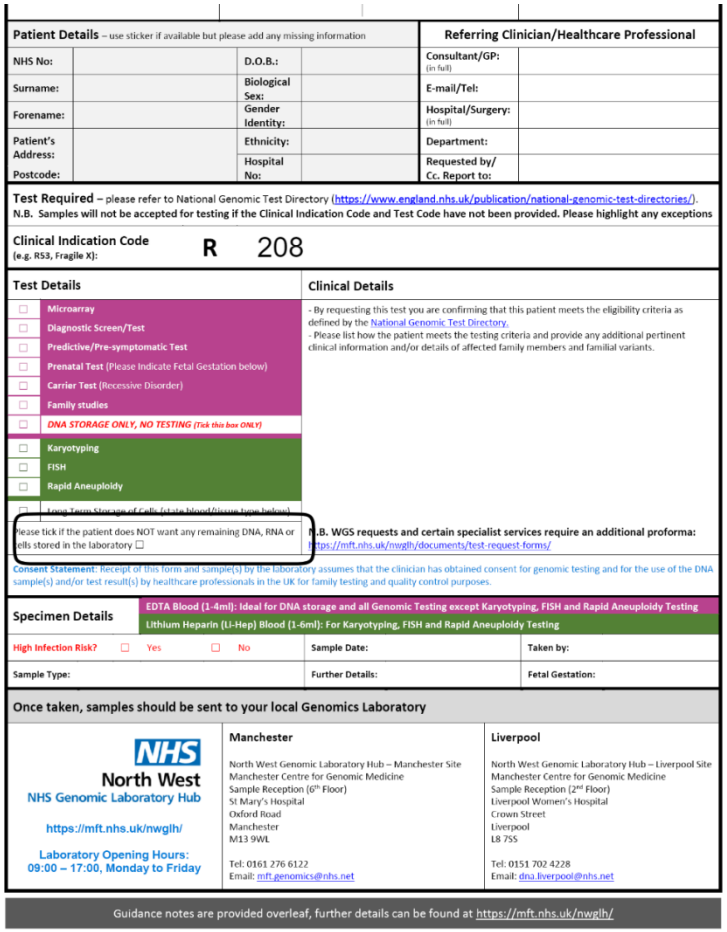
Option Not to store DNA



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NHS No:		D.O.B.:		Consultant/GP: (in full)	
Surname:		Biological Sex:		E-mail/Tel:	
Forename:		Gender Identity:		Hospital/Surgery: (in full)	
Patient's Address:		Ethnicity:		Department:	
Postcode:		Hospital No:		Requested by/ Cc. Report to:	

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 208**

Test Details	Clinical Details
☐ Microarray	- By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the National Genomic Test Directory.
☐ Diagnostic Screen/Test	- 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.
☐ Predictive/Pre-symptomatic Test	
☐ Prenatal Test (Please Indicate Fetal Gestation below)	
☐ Carrier Test (Recessive Disorder)	
☐ Family studies	
☐ *DNA STORAGE ONLY, NO TESTING (Tick this box ONLY)*	
☐ Karyotyping	
☐ FISH	
☐ Rapid Aneuploidy	
☐ Long Term Storage of Cells (state blood/tissue type below)	
Please tick if the patient does NOT want any remaining DNA, RNA or cells stored in the laboratory ☐	**N.B. WGS requests and certain specialist services require an additional proforma:** https://mft.nhs.uk/nwglh/documents/test-request-forms/

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.

Specimen Details	EDTA Blood (1-4ml): Ideal for DNA storage and all Genomic Testing except Karyotyping, FISH and Rapid Aneuploidy Testing Lithium Heparin (Li-Hep) Blood (1-6ml): For Karyotyping, FISH and Rapid Aneuploidy Testing	
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

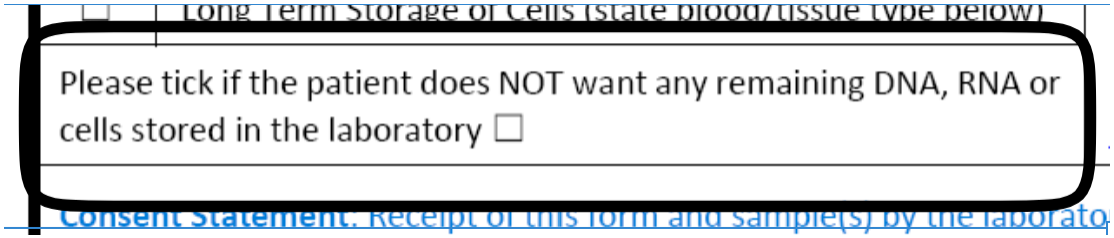
NHS North West
NHS Genomic Laboratory Hub
https://mft.nhs.uk/nwglh/
Laboratory Opening Hours:
09:00 – 17:00, Monday to Friday

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

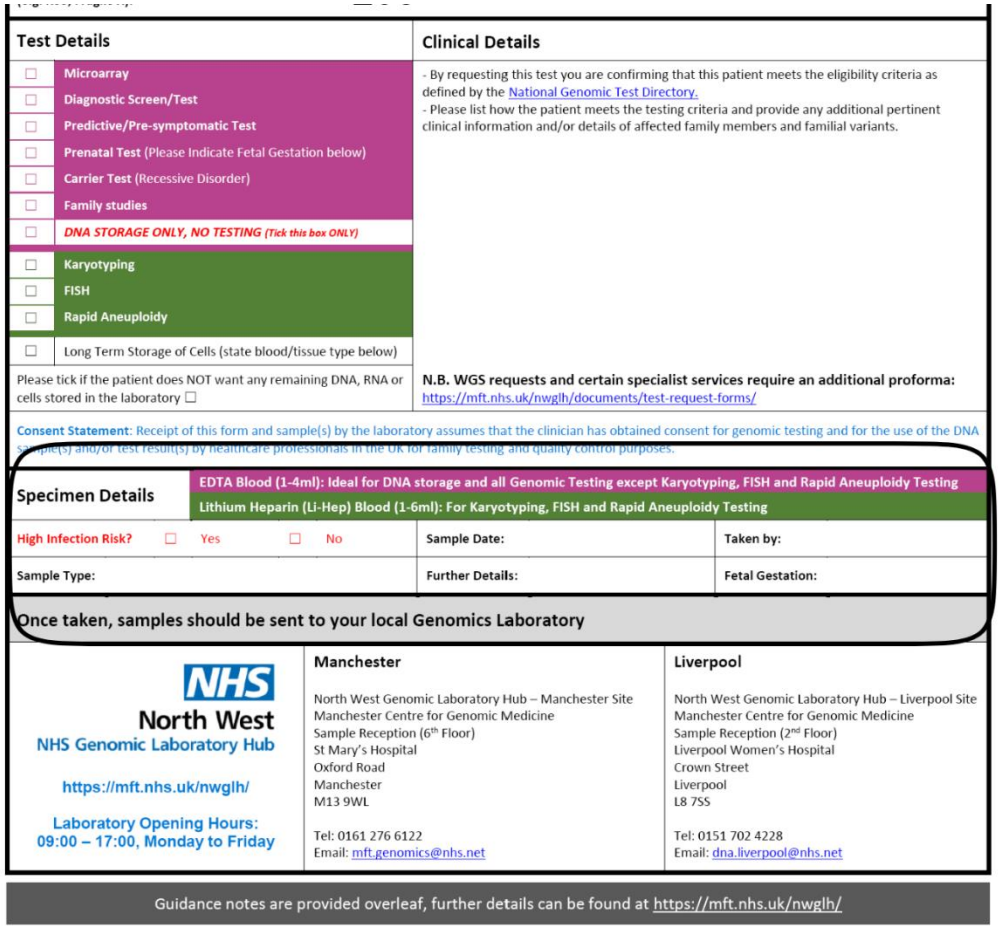
Liverpool
North West Genomic Laboratory Hub – Liverpool Site
Manchester Centre for Genomic Medicine
Sample Reception (2nd Floor)
Liverpool Women's Hospital
Crown Street
Liverpool
L8 7SS
Tel: 0151 702 4228
Email: dna.liverpool@nhs.net

Guidance notes are provided overleaf, further details can be found at https://mft.nhs.uk/nwglh/

Option Not to store DNA



Specimen details:



Test Details		Clinical Details
☐	Microarray	- By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the National Genomic Test Directory. - 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.
☐	Diagnostic Screen/Test	
☐	Predictive/Pre-symptomatic Test	
☐	Prenatal Test (Please Indicate Fetal Gestation below)	
☐	Carrier Test (Recessive Disorder)	
☐	Family studies	
☐	*DNA STORAGE ONLY, NO TESTING (Tick this box ONLY)*	
☐	Karyotyping	
☐	FISH	
☐	Rapid Aneuploidy	
☐	Long Term Storage of Cells (state blood/tissue type below)	
Please tick if the patient does NOT want any remaining DNA, RNA or cells stored in the laboratory ☐		**N.B. WGS requests and certain specialist services require an additional proforma:** https://mft.nhs.uk/nwglh/documents/test-request-forms/

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Specimen Details	EDTA Blood (1-4ml): Ideal for DNA storage and all Genomic Testing except Karyotyping, FISH and Rapid Aneuploidy Testing Lithium Heparin (Li-Hep) Blood (1-6ml): For Karyotyping, FISH and Rapid Aneuploidy Testing	
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

NHS North West
NHS Genomic Laboratory Hub

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

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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.

Specimen Details	EDTA Blood (1-4ml): Ideal for DNA storage and all Genomic Testing except Karyotyping, FISH and Rapid Aneuploidy Testing Lithium Heparin (Li-Hep) Blood (1-6ml): For Karyotyping, FISH and Rapid Aneuploidy Testing	
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

2nd page

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 **Biological Sex**
- Patient's **Postcode**
- **Clinical Indication Code**

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

Referring Clinician/Healthcare Professional

The following details are mandatory:

- **Consultant/GP name**: initials are not acceptable as the laboratory cannot identify the clinician/healthcare professional. A minimum of first initials and surname must be provided.
- **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.

Requested by/Cc. Report to: Use this space if the healthcare professional requesting the test/requiring a report copy is not the patient's Consultant.

Clinical Indication Code

Clinical Indication Code is a mandatory field, however this code 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 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 Karyotyping, FISH and Rapid Aneuploidy Testing) – Store sample at 4°C if required, send by courier or first class post. N.B. Blood samples for Neurofibromatosis type 1 (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 and Rapid Aneuploidy Testing – 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:
  - Up to 6ml 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