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| **9865 9322** | | | | | | | | | **Genomic Testing Request Form**  **Rare Disease** | | | | | | | | | | | | **Lab use only** | | | | |
| **Lab No:** | | Type Lab No. or Affix label | | |
| (DOC4900 Revision 5) | | | | | | | | | | | |
| **Patient Details** – use sticker if available but please add any missing information | | | | | | | | | | | | | | | | | | | **Referring Clinician/Healthcare Professional** | | | | | | |
| **NHS No:** | | Enter NHS No | | | | | | | | | | **D.O.B.:** | DD/MM/YYYY | | | | | | **Consultant/GP:**  (in full) | | Enter Consultant/GP name | | | | |
| **Surname:** | | Enter Surname | | | | | | | | | | **Biological Sex:** | Enter Biological Sex | | | | | | **E-mail/Tel:** | | Enter E-mail/Tel. | | | | |
| **Forename:** | | Enter Forename | | | | | | | | | | **Gender Identity:** | Enter Gender Identity | | | | | | **Hospital/Surgery:**  (in full) | | Enter Hospital/Surgery | | | | |
|  | | Address Line 1 | | | | | | | | | | **Ethnicity:** | Enter Ethnicity | | | | | | **Department:** | | Enter Department | | | | |
| Address Line 2 | | | | | | | | | |
| Address Line 3 | | | | | | | | | | **Hospital No:** | Enter Hospital No | | | | | | **Requested by/ Cc. Report to:** | | Enter Requested by/Cc. Report to | | | | |
| **Postcode:** | | Postcode | | | | | | | | | |
| **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** | | | | | **n/a - CYP2C19 genotyping to guide mavacamten dosing** | | | | | | | | | | | | | |
| **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](https://www.england.nhs.uk/wp-content/uploads/2018/08/Rare-and-Inherited-Disease-Eligibility-Criteria-November-2020-21.pdf).  - 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) | | | | | | | | | | | | **CYP2C19 genotyping to guide mavacamten dosing**  **SEND TO NWGLH – LIVERPOOL SITE (address below)** | | | | | | | | | | | | |
|  | **Carrier Test** (Recessive Disorder) | | | | | | | | | | | |
|  | **Family studies** | | | | | | | | | | | |
|  | **Rapid Aneuploidy** (for neonates send EDTA and Li-Hep) | | | | | | | | | | | |
|  | ***DNA STORAGE ONLY, NO TESTING (Tick this box ONLY)*** | | | | | | | | | | | |
|  |  | | | | | | | | | | | |
|  | **Karyotyping** | | | | | | | | | | | |
|  | **FISH** | | | | | | | | | | | |
|  |  | | | | | | | | | | | |
| Use alternative form (LF 160 001) for specialised cell culture service (cell lines/RNA) | | | | | | | | | | | | |
| 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: Ideal for DNA storage and all Genomic Testing except Karyotyping and FISH Testing** | | | | | | | | | | | | | | | | | | | | |
| **Lithium Heparin (Li-Hep) Blood: For Karyotyping and FISH Testing** | | | | | | | | | | | | | | | | | | | | |
| **High Infection Risk?** | | | |  | | Yes | |  | | | No | | | **Sample Date:** | | | | Select Date from Calendar | | | | **Taken by:** | | Enter Full Name | |
| **Sample Type:** | | | Select Sample Type | | | | | | | | | | | **Further Details:** | | | | Enter any relevant details | | | | **Fetal Gestation:** | | Enter Gestation | |
| **Once taken, samples should be sent to your local Genomics Laboratory** | | | | | | | | | | | | | | | | | | | | | | | | | |
| [**https://mft.nhs.uk/nwglh/**](https://mft.nhs.uk/nwglh/)  **Laboratory Opening Hours:**  **09:00 – 17:00, Monday to Friday** | | | | | | | | | | **Manchester** | | | | | | | | | | **Liverpool** | | | | | |
| 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](mailto:mft.genomics@nhs.net) | | | | | | | | | | 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](mailto:dna.liverpool@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** | | | | | | | | | | | | | | | | | | | | | | | | | |
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| **Patient Details** | | | | | | | | | | | | | | |  | **Specimen 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. | | | | | | | | | | | | | | | **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 and FISH)** – 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. | | | | | | | |  |
| **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. | | | | | | | | | | | | | | |
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| **Postnatal samples for Karyotyping and 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: * 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. | | | | | | | |
| **Clinical Indication Code** | | | | | | | | | | | | | | |
| **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. | | | | | | | | | |
| 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. | | | | | | | | | | | | | | |
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| **This area is for Lab use only** | | | | | | | | | | | | | | | | | | | | | | | | | |