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| **9865 9322** | | | | | | | [**https://mft.nhs.uk/nwglh/**](https://mft.nhs.uk/nwglh/)  **Laboratory Opening Hours:**  **09:00 – 17:00, Monday to Friday**  **mft.genomics@nhs.net** | | | | | | | | | | | | **Lab use only** | | |
| **Lab No:** |  | |
| **Genomic Testing Request Form (Rare Disease)**  DOC4900 (Revision 7) | | | | | | | | | | | | | | | | | | | | | |
| Please return this completed request form with all samples to the following NWGLH laboratory (tick as appropriate): | | | | | | | | | | | | | | | | | | | | | |
|  | NWGLH (Manchester), Sample Reception (6th Floor), St Mary’s Hospital, Oxford Road, Manchester, M13 9WL (Tel: 0161 276 6122) | | | | | | | | | | | | | | | | | | | | |
|  | NWGLH (Liverpool), Sample Reception (2nd Floor), Liverpool Women’s Hospital, Crown Street, Liverpool, L8 7SS (Tel: 0151 702 4228) | | | | | | | | | | | | | | | | | | | | |
| **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/**  **Clinician:** (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:** | | | Address Line 1 | | | | **Ethnicity:** | | | | Enter Ethnicity | | | | | **Clinical Specialty/ 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 Test Indication Code/Test Name and Clinical Utility have not been provided. Please highlight any exceptions.** * WGS requests and certain specialist services require an additional proforma: <https://mft.nhs.uk/nwglh/documents/test-request-forms/> * Use alternative form (LF 160 001) for specialised cell culture service (cell lines/RNA). | | | | | | | | | | | | | | | | | | | | | |
| **Test request details** | | | | | | **R code:**  **(e.g. R38.2)** | | |  | | | | | | **Test Name:**  **(e.g. Sporadic aniridia)** | | |  | | | |
| **Clinical Details** | | | | | Please provide the relevant eligibility criteria and clinical details for the patient, including their affected status.  If targeted familial testing is required, please provide details of the known familial variant and affected relative.  For DNA storage only, with no genomic testing required, please use test indication code **R346.1**. | | | | | | | | | | | | | | | | |
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| **Clinical Utility (please select as appropriate and provide any additional information in the section 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 | | | | | | | | | | | | | | | | | | | | |
| **Consent Statement:**  **Receipt of this form and sample(s) by the laboratory assumes that the clinician has obtained consent for genomic testing and storage to facilitate future testing, for quality control purposes and for use of the sample and/or test result(s) by healthcare professionals in the UK for family testing, as appropriate. If the patient does not consent for storage, please contact the laboratory once testing is completed to arrange disposal.** | | | | | | | | | | | | | | | | | | | | | |
| **Specimen Details** | | | | | **EDTA Blood: Ideal for DNA storage and all Genomic Testing except Karyotyping and FISH** | | | | | | | | | | | | | | | | |
| **Lithium Heparin (Li-Hep) Blood: For Karyotyping and FISH** | | | | | | | | | | | | | | | | |
|  | | **Urgent – please indicate reason in clinical details** (must satisfy one of the criteria listed overleaf) | | | | | | | | | | | | | | | | | | | |
| **Sample Type:** | | | | <select> | | | | | | | | | **Sample Date:** | | | Click or tap here to enter text. | | | **Taken by:** | Click or tap here to enter text. | |
| **High Infection Risk?** | | | | Yes |  | | No |  | |  | | | **Further Details:** | | |  | | | **Fetal Gestation:**  **(required for prenatal samples)** |  | |
| **Please state infection hazard, including whether blood-borne infection:** | | | | | | |  | | | | | |
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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** * **Test Request Details** (see further information below)   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. | | | | | | | | | |
|  | | **Blood 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 (also send ≥1ml in Li-Hep, see below). * **Fetal Blood**: ≥1ml in a paediatric EDTAtube. | | | | | | |  |
| **Referring Clinician/Healthcare Professional** | | | | | | | | | | |
| The following details are mandatory:   * **Consultant/Clinician**: initials are not acceptable as the laboratory cannot identify the clinician/healthcare professional. A minimum of first initials and surname must be provided. * **Hospital/Surgery** 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. * **Clinical Specialty/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 Consultant/Clinician. | | | | | | | | | | |
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| **Blood 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 (also send ≥1ml in EDTA, see above). | | | | | | |
| **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.   **Saliva Samples**: GeneFiX or Oragene collection kits only.  **Solid Tissue:** DO NOT expose to formalin. Send in dry sterile plastic container (or if stored overnight in sterile saline). 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.  **Other Sample Types:** by prior arrangement only. | | | | | | |
| **Test Request Details** | | | | | | | | | | |
| Test indication code (R 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 Test Indication Code can be requested when relevant to the investigation, ensuring the appropriate sample type(s) are supplied for the requested test(s).  Please ensure sufficient clinical information is provided to demonstrate eligibility for the test and complete the Clinical Utility section. The laboratory cannot proceed with testing if the form is incomplete and/or illegible. If required, additional reports and letters can be attached to this referral form. | | | | | | | | | | |
| **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. | | | | | | | | | |
| **Urgency** | | | | | | | | | | |
| Criteria for urgent genomic testing should include one of the following: neonate (<1 month), acutely/critical ill, pregnancy-related, prescribing, urgency defined in eligibility criteria from National Genomic Test Directory. | | | | | | | | | | |
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| **This area is for Lab use only** | | | | | | | | | | | | | | | | | | | | | |