

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Test Form** | **Specimen Type** | **Volume** | **Notes/Comments** | **Source** |
| **Rare Disease** | | | | |
| GLH Rare Disease Genetic Testing Form | | | | |
|  | EDTA Blood | 2-5ml | Molecular genetic analysis  2-5ml adults/children  1-2ml neonates | [411.027 V3.0](https://ney-genomics.org.uk/wp-content/uploads/2022/02/411.027-Rare-Disease-Referral-Form-v3.0web.pdf) |
| Heparin Blood | 2-5ml | Cytogenetic analysis |
| Skin |  | In transport medium |  |
| DNA | 3ug | Minimum concentration 50ug/ul |  |
| GLH Prenatal Form (Feto-maternal) | | | | |
|  | Maternal Sample for MCC | N/A | In EDTA | [411.030 V2.0](https://ney-genomics.org.uk/wp-content/uploads/2022/02/411.030-Prenatal-form-v2.0web.pdf) |
|  | Chorionic Villus | 10-30mg | In transport medium |
|  | Amniotic Fluid | 10-20ml | In universal container |
|  | Fetal Blood | 1-2ml | In EDTA |
|  | Maternal Blood for NIPD | 20ml | In Streck Tube |
|  | Pregnancy loss samples | N/A |  |
| Prenatal Genetic Testing for Haemoglobinopathies Referral Form | | | | |
|  | Chorionic Villus | 10-30mg | In transport medium |  |
|  | Amniotic Fluid | 10-20ml | In universal container |  |
| Genetic testing for Haemoglobinopathies Referral Form | | | | |
|  | Venous Blood | 4ml | EDTA tube. | [400.012 V2.0](https://ney-genomics.org.uk/wp-content/uploads/2022/05/NEY-Genetic-Testing-for-Haemoglobinopathies-Referral-Form-v2.pdf) |
|  |  |  | Lithium Heparin or Serum tubes are unsuitable for testing. Forward the completed referral form and EDTA blood sample to your local Genetics Laboratory |
|  |  |  | **Red cell indices and HPLC/IEF results must be supplied before the sample can be processed.** |
| **Cancer** | | | | |
| RNA Fusion Panel Request Form (Incorporating NTRK Fusion Testing) | | | | |
|  | FFPE curls | 5 x 10μM FFPE curls | Preferred | [411.029 V2](https://ney-genomics.org.uk/wp-content/uploads/2022/02/411.029-RNA-Fusion-Referral-Form-v2.0web.pdf) |
|  | FFPE sections | 10 x 5 μM FFPE sections | With labelled H&E slide for macro dissection |
|  | Fresh Frozen Tumour Tissue on DRY ICE | Minimum 1mm tissue | on DRY ICE |
|  | Fresh Frozen Tissue  Stored in RNA-later buffer | Minimum 1mm tissue | Do not freeze in RNA-later buffer - store/ transport at 4C |
| GLH DPYD Referral Form | | | | |
|  | Blood | 2.5mL tube | In EDTA | [411.028 V2.0](https://ney-genomics.org.uk/wp-content/uploads/2022/02/411.028-DYPD-Referral-Form-v2.0web.pdf) |
| tBRCA and HRD Request Form (Tumour HRD status and BRCA1&2 genomic testing) | | | | |
|  | Thick sections air dried on mounted slide with a corresponding marked H&E slide with regions of >30% neoplastic cells highlighted.  Sections should be cut under conditions that prevent cross contamination from other samples.  **FFPE curls NOT accepted** | 10 x 5 μM | Cytology samples can be accepted for HRD and tumour BRCA testing. It is essential that cells and tissue fragments from the cytology samples are processed into agar/cell blocks, formalin-fixed and paraffin embedded. They should then undergo a Pathology assessment process as per tissue samples | [411.032 V1.0](https://ney-genomics.org.uk/wp-content/uploads/2022/03/GLH-tBRCA-and-HRD-request-form-v1.0web.pdf) |
| Haematological Disorders Referral Card | | | | |
|  | Peripheral Blood (lithium heparin) | 5~10ml |  | [LGSR008 V1.4](http://leedspath.myeqms.com/Administrator/LoadDocADM.asp?ID=84328&Ext=True&CCID=1) |
|  | Bone Marrow Aspirate | 1ml marrow added to tranpsort medium | Transport medum tube provided by lab ((RPMI transport medium (10ml) + heparin) |
| Solid Tumour Specialist Diagnostic Services Referral Card | | | | |
|  | FFPE curls - DNA or RNA only | 5-10 x 10μm curls | If tumour sample >20%.  Double total if DNA & RNA required | [HPQA Form 62803 Dec 2021 K.Rankeillor](https://www.leedsth.nhs.uk/assets/Genetics-Laboratory/Referral-forms/aad39060e3/SMDS-referral-form-editable-v3.pdf) |
|  | FFPE sections - DNA or RNA only | 10 x 5μm sections with marked H & E | If tumour sample <20%.  Double total if DNA & RNA required |
|  | FFPE sections - FISH | 2-3 x 4μm FFPE sections on 'APES' or 'sticky' slides per test required with marked H&E slide |  |
|  | Fresh | Minimum 5mm3 | Fresh frozen tumour in dry ice or if refrigerated in stabilisation buffer (eg ‘RNA later’ or RLT buffer) |
|  |  |  |  |
|  |  |  |  |
|  |  |  |  |
|  |  |  |  |
|  |  |  |  |
| Circulating Lung Tumour Referral Card | | | | |
|  | Peripheral Blood | 2~10ml EDTA tube OR STRECK tube (fill with blood): | For Detection of EGFR Mutations Exons 19-20 in circulating tumour  Liase with Molecular Oncology 0113 206 4570 when a blood sample is taken to confirm transport arrangements.  If using EDTA tube, sample must arrive at lab within 4 hours of sampling  If using STRECK tube, tube must be completely filled where possible and then inverted ten times to mix well. | [LGSR009 V1.2](http://leedspath.myeqms.com/Administrator/LoadDocADM.asp?ID=84342&Ext=True&CCID=1) |