North West Genomic Laboratory Hub (Liverpool)
Manchester Centre for Genomic Medicine
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Q-Pulse Ref: DOC5463 User Leaflet; Revision: 2.

North West Genomics Laboratory Hub – Liverpool Site

Genetics Laboratory User Leaflet

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1 Laboratory Details

The North West Genomics Laboratory Hub – Liverpool Site – is situated on the 2nd Floor at Liverpool Women's Hospital and delivers a diagnostic service in both molecular and cytogenetics throughout the North West of England, the UK and worldwide. The site forms part of the North West Genomics Laboratory Hub in partnership with the Manchester site, based in St Mary's Hospital, Manchester. It aims to provide a clinical interpretative service which is responsive to its users' needs. It serves a large and diverse population and is a Genome Medicine Centre for the Northwest Coast contributing, together with our local delivery partners, towards patient recruitment and interpretation of variants for the 100,000 Genomes Project.

The Genomic Laboratory provides a comprehensive diagnostic service for prenatal samples (amniotic fluid, CVS), pre & postnatal blood samples, solid tissues, tumours and haematological malignancies, providing clinical interpretation for all cases. The Genetics service uses various molecular and cytogenetic analysis techniques to carry out testing for a wide range of genetic disorders.

For clinical advice please contact the department.

1.1 Quality Control & Accreditation

The laboratory is a UKAS accredited medical laboratory, UKAS No. 9322 accredited to ISO 15189:2012 for Medical Laboratories, for more info see: https://www.ukas.com/services/accreditation-services/medical-laboratory-accreditation-iso-15189/.

The laboratory participates in National External Quality Assurance Schemes for genomics run by Genomics Quality Assessment (GenQA), which is part of the UK NEQAS Consortium, and the European Molecular Genetics Quality Network (EMQN). From January 2018, the laboratory will take part in the combined GenQA EQA scheme.

The laboratory also conforms to the best practice guidelines of the Association of Clinical Genetic Sciences (ACGS).

For a full schedule of accredited tests please visit: https://www.ukas.com/wp-content/uploads/schedule_uploads/00007/9322%20Medical%20Single.pdf

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1.2 Postal Address for correspondence and samples

North West Genomics Laboratory Hub – Liverpool Site,
Liverpool Women's Hospital,
Crown Street,
Liverpool,
L8 7SS

Telephone: 0151 702 4228 / 4229 / 1425

Secure Fax: 0151 702 4230

Website: www.liverpoolwomens.nhs.uk

Genetic testing queries - email: dna.liverpool@nhs.net

Current Laboratory Working Hours 9.00 a.m. – 5.30 p.m. Monday - Friday

1.3 Contact Details:

Scientific Operational Director
Emma Howard, Consultant Clinical Scientist
Tel: 0151 702 4219
e-mail: emma.howard@mft.nhs.uk

Deputy Head of Laboratory
Victoria Stinton, Consultant Clinical Scientist
Tel: 0151 702 4231
e-mail: victoria.stinton@mft.nhs.uk

1.4 Feedback & Enquiries

Should you have any comments, suggestions, cause for concern or complaint about the service you receive from the laboratory, please contact the Head / Deputy Head of the laboratory using the contact details above.

2 Testing Provided

2.1 Reasons for referral:

The laboratory uses various techniques to carry out testing for a wide range of genetic disorders.

The types of investigation include:

- Confirmation / exclusion of a diagnosis for inherited and acquired disorders
- Carrier testing and risk assessment in families with a known genetic disorder
- Carrier testing and risk assessment in families with reproductive problems
- Pre-symptomatic / predictive testing in individuals at risk of a late-onset genetic disorder
- Prenatal diagnosis of genetic conditions, where appropriate

The laboratory offers testing for a range of 'core' disorders plus a set of more specialist services for which samples are received on a supra-regional or national basis.

The laboratory is also a member of the UK Genetic Testing Network (UKGTN) and can forward DNA samples to other UK genetics laboratories for testing of a large range of single gene disorders, where appropriate. Full details of services available through the UKGTN are available at www.ukgtn.nhs.uk or by contacting the laboratory.

Details of services for rare disorders not currently available in the UK can be found on the web sites www.orpha.net and www.genetests.org or by contacting the laboratory.

2.2 Request forms:

Requests for genetic testing should be accompanied by the laboratory referral card whenever possible (available for download from our website).

The following details regarding the correct patient identification are mandatory:

- Surname and Forename
- Date of Birth
- Full address, including postcode
- Patient's gender at birth
- NHS number, and where applicable local patient identifier (e.g. hospital number)

The following details regarding the referring clinician are mandatory:

- Name of referring Clinician

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- Address of referring Clinician, including Department

The referral card should also include:

- Sample type with date and time of collection
- Clear indication for why a test has been requested
- Consent for testing and possible storage of material

Failure to fill in the request form clearly and correctly may result in inappropriate testing, delays in reporting or even a sample not being processed.

Please note: this referral card forms a contract between the laboratory and the referring clinician.

2.3 Gate keeping & funding of UKGTN Samples

Access to certain tests provided by other UK laboratories is restricted to specific referring specialties or clinicians. There may also be specific referral criteria or forms required before these tests can be requested. Please contact the laboratory for details.

Please note that the costs of any tests referred to UKGTN centres or referred to non-UK laboratories will be recharged to the referring Trust.

Please refer to Appendix 1 for a full list of Molecular Tests offered by the laboratory

2.4 Prenatal Diagnosis (PND) Services

The hub offers genetic testing by microarray, conventional cytogenetic karyotyping, molecular cytogenetic Fluorescent In Situ Hybridization (FISH) and some single gene disorders on the following samples;

- Amniotic Fluid Samples
- Chorionic Villus Samples
- Fetal Blood Samples

The laboratory offers a rapid aneuploidy test via Quantitative Fluorescence-Polymerase Chain Reaction (QF-PCR) alongside a whole genome screen for all prenatal samples. A high resolution microarray test is carried out for any sample received following a fetal anomaly detected by detailed ultrasound scan (since October 2014) and only where suitable informed consent has been obtained. Conventional cytogenetic analysis via karyotyping is carried out for all other prenatal referral categories.

A range of testing for single gene disorders is available where there is appropriate clinical need or family history. For further details please contact the laboratory. The laboratory should be notified in advance for these samples.

Please note: It may not be possible to carry out rapid aneuploidy testing on heavily blood-stained amniotic fluid samples. Please send a maternal blood sample (in EDTA) with blood-stained samples. If rapid testing is not possible the laboratory will contact the referring clinician.

Prenatal Referral Policy

- Fetal Anomaly on Ultrasound Scan
- Serum Screen Positive
- Advanced Maternal Age/ maternal Anxiety
- Previous Pregnancy with Chromosome Rearrangement
- Familial Chromosome Rearrangement
- Single Gene Disorders (please contact the laboratory)

Please note: a maternal sample is also required for prenatal diagnosis of single gene disorders to rule out maternal cell contamination of the fetal DNA sample (5ml in EDTA).

For further information about prenatal testing, please contact the laboratory.

2.5 Fetal Blood Services

The laboratory offers a diagnostic service for fetal blood samples following appropriate consultation and agreement with a senior member of staff**.**

Fetal Blood Samples Referral Policy

- Abnormal Ultrasound Scan
- Abnormal Amniotic fluid /Chorionic Villus Sample result

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2.6 Solid Tissue Services

The laboratory also offers testing for solid tissue samples resulting from pregnancy loss. This testing should only be requested in the event of:

- A fetal abnormality
- Early stage pregnancy loss (after the 3rd pregnancy loss)

(https://www.rcog.org.uk/globalassets/documents/guidelines/gtg_17.pdf)

- IUD/Stillbirth

Samples not meeting one of these criteria may not be processed.

PLEASE NOTE: The laboratory will NOT accept the whole fetus or whole placenta. DO NOT EXPOSE TISSUE SAMPLES TO FORMALIN as this will render them unsuitable for culture.

2.7 Postnatal Services

The hub offers high resolution chromosome analysis by microarray, conventional cytogenetic analysis on cultured material and molecular cytogenetic (FISH) studies, detecting most structural anomalies, microdeletion syndromes and cryptic imbalances. We also carry out a range of molecular genetic tests for single gene disorders and gene panels, details of which can be found in **Appendix 1**.

2.7.1 Microarray Comparative Genomic Hybridisation Referral Policy

- Congenital Abnormality / Dysmorphism
- Developmental Delay / Learning Difficulties
- Epilepsy Screening

2.7.2 Conventional Cytogenetics (Karyotyping) Referral Policy

The laboratory will accept all appropriate referrals, major categories being:

- Delayed Puberty
- Sub Fertility / Recurrent Pregnancy Loss
- Breakage Syndromes (Please consult laboratory for details).

Samples MUST be sent in lithium heparin or they will not be suitable for culture and will be rejected.

For further information about microarray testing or karyotype testing, please contact the laboratory.

2.8 Oncology Services

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The laboratory offers a comprehensive conventional and molecular cytogenetic (FISH) diagnostic service for haematological and solid tumour malignancies, which include;

- Chronic / Acute Myeloid Leukaemias
- Acute Lymphoblastic Leukaemia
- Lymphomas
- Bone Marrow Transplant Monitoring
- Chronic Lymphocytic Leukaemia (FISH only)
- Plasma Cell Myeloma (PCM)
- Myelodysplastic syndrome (MDS) and Myeloproliferative neoplasms (MPN) (including *JAK2*, *CALR* and *MPL* testing)
- Pharmacogenomics: DYPD testing
- Molecular testing for solid tumours:
    - *KRAS/NRAS* codon 12, 13, 59 61, 117 & 146
    - *PIK3CA codon 542, 545 & 1042*
    - *BRAF* codon 599-600
    - *MLH1* hypermethylation analysis
    - *MGMT* promoter methylation analysis

For further information about oncology testing please contact the laboratory or email mft.genetics-oncology@nhs.net

3 Sending a sample to the laboratory

3.1 Consent

All genetic testing requires appropriate consent. Obtaining consent is the responsibility of the referring clinician. For further information and guidance, please refer to 'Consent and Confidentiality in Genetic Practice – A report of the Joint Committee on Medical Genetics', which is available as a download from the British Society of Human Genetics website at www.bsgm.org.uk.

3.2 DNA storage

Where DNA has been extracted, DNA from all referrals is currently retained for quality assurance purposes, unless the request card indicates that permission for storage is denied. Consent is not required for storage solely for quality assurance purposes but it is considered good practice for the referring clinician to inform the patient of this practice. The laboratory can also store DNA from patients on request (for example where no specific genetic test is currently available). Tests can be requested on

stored DNA samples without the need to take a fresh sample from the patient as long as the appropriate consent has been obtained and sufficient DNA is available.

3.3 Cell Storage

Fixed cell suspensions of cultured cells is routinely stored for 6 months following completion of the case. Exceptions include:

- Where no consent for storage has been obtained; these samples will be discarded after one month following successful completion of testing
- Where long term storage has specifically been requested. Please contact the laboratory for more details

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3.4 Suitable sample types

Test Type		Sample Types	Volume
DNA Testing	Single Gene Gene Panel	Blood (**EDTA**)	>4ml (Adult) 1-2ml (Young Children)
		Bone Marrow Aspirate	1-2ml in transport media
		Buccal Scrape	Swab
		Saliva sample	Single sample
		Tissue (fresh or frozen*)	N/A
		FFPE	Slides / Blocks/ Sections (+ H&E slide)
		Urine	15-20ml early morning sample
		Other samples by prior arrangement	
Microarray		Blood (**EDTA**)	3-5ml (Adult) 1-2ml (Young Children)
		Buccal Scrape	Swab
		Saliva	Single sample
		Tissue sample	In transport media
		Other samples by prior arrangement	
Conventional Cytogenetics	Karyotype FISH	Blood (**Lithium Heparin**)	3-5ml (Adult) 1-2ml (Young Children)
		Bone Marrow Aspirate	1-2ml in transport media Or in lithium heparin
		Tissue (cord insertion site) sample	1cm^2 in transport media
		Skin sample	In transport media
		Muscle biopsy	In transport media
		Tumour sample	In transport media
		Lymph node	In transport media
		FFPE (FISH only)	Slides / Sections (+ H&E slide)
		Other samples by prior arrangement	
Prenatal Diagnosis		Chorionic Villus	20-50mg
		Amniotic Fluid	10-20ml clear fluid (sterile universal)
		Fetal Blood Samples	1-2ml (Lithium Heparin)

*Frozen tissue samples should be transported on ice and be received frozen.

Molecular Genetic and Molecular Cytogenetic Testing:

3-5ml of blood **(EDTA)** from adults or 1-2ml from young children

DNA can be extracted from 2ml saliva (collected using the GeneFixTM DNA Saliva Collector or Oragene mouthwash sample), or using buccal swabs (collected using the IsoHelixTM system). Please note that buccal swabs may not necessarily provide sufficient DNA for all available tests. Please contact the laboratory before using these sampling methods to ensure that the test required can be carried out.

DNA can be extracted from fresh or frozen tissue samples, and it is also possible to obtain limited results for some assays from blood spots or paraffin embedded fixed tissue samples. However, please contact the laboratory before using these sampling methods to ensure that sufficient DNA of appropriate quality for the test required can be extracted.

Constitutional Karyotyping (Conventional Cytogenetics):

- 3-5ml of blood **(lithium heparin)** from adults or 1-2ml from young children.

Store overnight at 4°C if required. DO NOT freeze or expose to heat. The sample must arrive in the laboratory within 24 hours of being taken.

Prenatal Diagnosis:

Store overnight at 4°C if required. DO NOT freeze or expose to heat. The sample must arrive in the laboratory within 24 hours of being taken.

Chorionic Villus Samples – 12-15 weeks gestation:

- Conventional cytogenetic analysis only 7-10 mg
- Conventional & Molecular 10-15 mg
- Biochemical assays >20 mg

Quoted amounts refer to sorted material after biopsy dissection from maternal tissue and decidua.

Samples should be placed in sterile transport media (supplied by the laboratory upon request).

Amniotic Fluid Sample – 16+ weeks gestation

Prenatal diagnosis for single gene disorders is usually carried out on chorionic villus samples, but amniotic fluid or fetal blood samples can be used where necessary.

- 10-20ml in sterile leak-proof universal / plastic container

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Reproductive Loss:

- Sample of amnion cord and placenta from the cord insertion site
- Sample of fetal skin
- Sample of fetal blood (in Lithium Heparin or EDTA depending upon test required, see table above)

PLEASE NOTE: The laboratory will NOT accept the whole fetus or whole placenta

DO NOT EXPOSE TISSUE SAMPLES TO FORMALIN as this will render them unsuitable for culture. Transport media is available from the laboratory upon request.

Store overnight at 4°C if required. DO NOT freeze or expose to heat. The sample must arrive in the laboratory within 24 hours of being taken.

Acquired Haematological Disorders:

Sample Requirements (Conventional Cytogenetics and FISH):

- Bone Marrow Aspirate: 1-2 ml in transport medium (available from the laboratory)
- Peripheral Blood: 1-5 ml collected in a lithium heparin container
- Lymph Node: Biopsy placed in BM transport medium (available from the laboratory).
- Variant analysis 1.5 ml collected in EDTA or FFPE on slides: 4-5µm thick (with H& E slide, clearly highlighting area of interest)

Please note: Blood Oncology samples with a cell concentration <10^6/ml and heavily blood-stained bone marrow aspirates are less likely to yield a reportable result

Sample Requirements (molecular genetic tests – *JAK2* p.V617F, *JAK2* exon 12, *CALR* exon 9, *MPL* codon W515)

- Peripheral Blood: 1-5 ml collected in an EDTA container.

Please note: DNA extracted from bone marrow aspirate collected in BM transport medium or EDTA is also suitable for DNA analysis, however please note that DNA extracted from cultured and fixed bone marrow cells (stored following Cytogenetic analysis) is often of poor quality and may therefore be unsuitable for DNA testing.

For sample requirements for molecular testing of solid tumours (*KRAS*/*NRAS* codon 12, 13 & 61, *BRAF* codon 600 analysis, *MLH1* and *MGMT* promoter methylation analysis), please contact the laboratory for a copy of the appropriate specimen referral form.

Store overnight at 4°C if required. DO NOT freeze or expose to heat. The sample must arrive in the laboratory within 24 hours of being taken.

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3.5 Sample Handling and Storage

All samples should be sent to the laboratory as soon as possible after they have been taken; delayed transit can affect sample quality. If this is not possible, then they should be stored in a secure refrigerator at +4°C and sent to the laboratory as soon as possible.

3.6 Packaging & Transportation

All samples should be labelled with the Patient's Name, Date of Birth, Postcode, NHS number, Unit No. and the date of collection and be accompanied by a FULLY completed request card (Available from the laboratory or downloaded from the laboratory web site - see above). Details of the family history should also be included, where relevant.

The sample should be placed in a sealed specimen bag in such a way as to maintain patient confidentiality and to prevent spillage and contamination. Samples sent through the post should be packaged in accordance with PI 650 and current UN3373 regulations. (See Laboratory web site for further details).

Please note that clotted blood samples or samples that are inadequately labelled or packaged will not be accepted by the laboratory.

3.7 High-risk samples

If samples are known to present a high-risk to laboratory staff, then this should be clearly indicated on the referral card and sample tube – including full details of the risk carried

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3.8 Turnaround times

The laboratory aims to report results within the recommended reporting time targets set by NHS England.

Clinical Urgency	Reporting Target (Calendar Days)	Examples
Urgent	3 Days	• QF-PCR for rapid trisomy detection • Urgent haemato-oncology FISH/RT-PCR • PCR based tests where the result is needed urgently for prenatal diagnosis
Urgent	5 Days	• *DYPD* Pharmacogenomic analysis
Urgent	7 days	• NIPT (when available through the NHS)
Urgent	14 days	• Microarray for prenatal/urgent* postnatal referrals • Urgent haemato-oncology karyotyping • Variant Specific molecular pathology tests • Urgent Southern blot tests (eg for prenatal diagnosis) • PCR-based tests for predictive testing (asymptomatic patients) and confirmation of neonatal results
Urgent	21 days	• Urgent* panels and exomes • NIPD
Non-urgent	21 days	• Standard haemato-oncology karyotyping • NGS for haemato-oncology referrals • NGS for molecular pathology referrals
Non-urgent	42 days	• Standard microarray – postnatal and solid tissue • Standard single gene sequencing • Small NGS gene panels (less than 10 genes) • Known familial variant testing (symptomatic patients) • Postnatal karyotyping • Round STR based tests
Non-urgent	84 days	• Large gene panels (more than 10 genes) • Clinical exomes for standard referral indications • Whole genome sequencing (delivered centrally and validated by GLH)

*Urgent samples are defined as those received from a pregnant woman, or a man whose partner is currently pregnant, or a newborn baby

All reporting times are given in calendar days.

Day 0 is the day the sample is received into the laboratory, providing all appropriate information and supportive samples are also received.

Please see the website for notification of any significant delays.

4 Testing Locations Following Formation of the NW GLH

Following the formation of the NW GLH, a process of de-duplication of testing across the North West was commenced on 1st April 2019. This has resulted in some laboratory processes being split between the Liverpool and Manchester sites and some investigations that were previously performed on both sites now only being performed in one location.

The NW GLH will take responsibility for ensuring samples are transported between sites for testing and have a courier in place to facilitate this. For reference, samples which are split between sites prior to processing include:

- Manchester to Liverpool:
  - Pregnancy loss samples and any maternal blood samples for fetal identity testing
  - Tests for DMD/BMD, HD and CF (except newborn screening)
  - Blood sample karyotyping and/or FISH
- Liverpool to Manchester
  - Prenatal samples for QF-PCR and microarray/karyotype tests
  - Neonatal samples for QF-PCR and microarray
  - Postnatal samples for microarray, Fragile X and PWS/AS tests
  - Postnatal samples for hearing loss and neurology NGS panel tests
  - Whole genome sequencing samples
  - Tissue blocks from colorectal cancer patients for *KRAS*/*NRAS* testing

In some instances the sample is processed at the receiving laboratory and extracted DNA is then transferred between sites for testing. Samples which are currently sent to Manchester for initial processing and then the extracted DNA is sent to Liverpool includes:

- Prenatal samples for carrier testing (CF, DMD/BMD, HD) and accompanying maternal blood for fetal identity testing
- Connexin 26/30
- Myotonic dystrophy Type 1 (DM)
- Spinocerebellar Ataxis (SCA)
- Spinal muscular atrophy (SMA)
- Spinobulbar muscular atrophy (SBMA) aka Kennedy's disease
- Charcot Marie Tooth (CMT)
- Hereditary Neuropathy Pressure Palsy (HNPP)
- Leber hereditary optic neuropathy (LHON)
- Short tandem repeat (STR) tests for neurology

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Samples which are sent to Liverpool for initial processing and then the extracted DNA sent to Manchester include:

- Pregnancy loss tissue sample DNA for microarray testing and analysis
- BRCA testing
- Bone marrow samples for the investigation of Hairy Cell Leukaemia; DNA sent for *BRAF* V600E testing.

In these cases, the laboratory performing the DNA extraction stores the DNA on site and sends a proportion of the extracted DNA to the other laboratory for testing.

In all cases, either laboratory will be able to provide information on testing and sample requirements.

5 Advice Service

The Head of Laboratory and scientific staff will provide advice on scientific and technical issues. However, for advice on clinical and counselling issues, please contact the Clinical Genetics Service, based at Liverpool Women's Hospital (0151 802 5001).

6 Protection of Personal Information

The laboratory follows the Liverpool Women's Trust Information Governance Policy on the protection and use of personal information. In summary, the Trust regards all person identifiable information that it holds or processes as confidential, and implements and maintains policies to ensure compliance with all necessary mandatory obligations.

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Appendix 1 – Test directory for Molecular based Germline and Acquired Disorders

Germline Conditions (Single gene disorders)	
Adrenoleukodystrophy (X-linked)	
	Diagnostic testing for pathogenic variants in *ABCD1* gene
	Familial pathogenic variant studies
CADASIL	
	Diagnostic testing for pathogenic variants in *Notch3*
	Familial pathogenic variant studies
Cystic Fibrosis (CF)	
	Diagnostic testing and carrier detection for 61 'common' *CFTR* pathogenic variants
	Other rarer *CFTR* pathogenic variants tested by specific request - please enquire
	Cystic Fibrosis Newborn Screening (p.(Phe508del) and 3 other common pathogenic variants)
DRPLA (Dentatorubral-pallidoluysian atrophy)	
	Testing for CAG repeat expansion
DMD/BMD Duchenne / Becker Muscular Dystrophy	
	Diagnostic testing and carrier detection for pathogenic deletions / duplications in the dystrophin gene
DPD Deficiency	
	Dic testing for 4 'common' variants in DPYD gene that result in DPD deficiency; offered prior to treatment with fluorouracil based chemotherapy.
Torsion Dystonia	
	Diagnostic testing for *DYT1* pathogenic variants in Torsion Dystonia
	Familial pathogenic variant studies
Friedreich Ataxia	
	Diagnostic & carrier testing for the GAA repeat

	expansion
	Small pathogenic variant analysis in *FXN* gene
Gilbert syndrome	
	Testing for TATAA box *pathogenic variant* in *UGT1A1* gene
Haemochromatosis	
	Testing for p.C282Y pathogenic variant, and p.H63D pathogenic variant as a reflex test
Hearing Loss (Non-syndromic)	
	Diagnostic testing for Connexin-26 (*GJB2*) and Connexin-30 (*GJB6*) gene pathogenic variants
	Familial pathogenic variant studies for *GJB2* gene
	Diagnostic testing for Mitochondrial variant m.1555A>G
HMSN (Hereditary Motor and Sensory Neuropathy) & HNPP (Hereditary Neuropathy with Liability to Pressure Palsies)	
	Testing for common 17p (*PMP22*) duplication and deletion
	Testing for pathogenic variants in *PMP22*
	Familial pathogenic variant studies
Huntington disease (HD)	
	Testing for CAG repeat expansion. Pre-symptomatic referrals only accepted from Clinical Genetics Department
Infantile Neuroaxonal Dystrophy (INAD)	
	Screening for pathogenic variants in the *PLA2G6* gene
	Familial pathogenic variant studies in the *PLA2G6* gene
Kennedy Syndrome (SBMA)	
	Testing for CAG repeat expansion in the Androgen

	Receptor (*AR*) gene
Leber's Hereditary Optic Neuropathy (LHON)	
	Testing for 3 common mitochondrial DNA pathogenic variants (m.11778G>A, m.3460G>A & m.14484T>C)
Medium chain acyl dehydrogenase deficiency (MCADD)	
	Testing for common c.985A>G pathogenic variant only
	Newborn screening for c.985A>G pathogenic variant
Mitochondrial disease	
	Testing for common point pathogenic variants associated with MELAS (m.3243A>G), MERRF (m.8344A>G) and NARP (m.8993T>G/C)
	Testing for mitochondrial genomic rearrangements (muscle biopsy preferred)
Myoclonus Dystonia	
	Screening for pathogenic variants in the *SGCE* gene
	Familial pathogenic variant studies in the *SGCE* gene
Myotonic Dystrophy (type 1)	
	Testing for CTG repeat expansion
Myotonic Dystrophy (type 2)	
	Testing for CCTG repeat expansion
Myotubular Myopathy (X linked)	
	Diagnostic testing for pathogenic variants in *MTM1* gene
	Familial pathogenic variant studies in *MTM1* gene
Nail Patella Syndrome	
	Testing for pathogenic variants in *LMX1B* gene
	Familial pathogenic variant studies in *LMX1B* gene
Neuroferritinopathy	

	Testing for pathogenic variants in exon 4 of the FTL gene, including c.408insA pathogenic variant
Oculopharyngeal Muscular Dystrophy (OPMD)	
	Testing for GCN (Alanine) repeat expansion in *PABPN1*
Pancreatitis (Hereditary & Idiopathic)	
	Testing for *PRSS1* pathogenic variants
	Familial *PRSS1* pathogenic variant studies
	Testing for *CFTR* and *SPINK1* (N34S) pathogenic variants
Peutz-Jegher syndrome	
	Diagnostic testing for pathogenic variants in *STK11* gene
	Familial pathogenic variant studies in *STK11*
PKAN (Hallervorden-Spatz)	
	Diagnostic testing for pathogenic variants in *PANK2* gene
	Familial pathogenic variant studies in *PANK2*
Rett Syndrome	
	Diagnostic testing for pathogenic variants in *MECP2* gene
	Familial pathogenic variant studies for the *MECP2* gene
Spinal Muscular Atrophy (SMA)	
	Diagnostic and carrier testing for *SMN1* exon 7 deletion
Spinoerebellar Ataxia (SCA types 1, 2, 3, 6, 7, 17)	
	Testing for SCA1, SCA2, SCA3 and SCA6 CAG repeat expansions (SCA7 and SCA17 testing by specific request only)
Zygosity / Paternity analysis	

	Zygosity and Paternity testing is only available for clinical not social/legal cases
Next Generation Sequencing Panels	
Neuropathy, Epilepsy and Spastic Paraplegia Panel (NESPP)	
Orofacial Clefting Panel	
Acquired Disorders	
Haemato-oncology	
Myeloproliferative Neoplasms (MPNs): Polycythaemia rubra vera (PRV)	
	JAK2 V617F variant analysis
	JAK2 Exon 12 analysis
Myeloproliferative Neoplasms (MPNs): Essential thrombocythaemia (ET) and Myelofibrosis (MF)	
	JAK2 V617F variant analysis
	Calreticulin (*CALR*) exon 9 analysis
	Myeloproliferative Leukemia Oncogene (*MPL*) codon 515 analysis
Hairy cell leukaemia	
	BRAF V600E variant analysis
Molecular Pathology (oncogene analysis)	
***KRAS* & *NRAS* (codon 12, 13 and 61 pathogenic variants)**	*Please contact the laboratory for a copy of the appropriate referral form*
***BRAF* codon 600 pathogenic variant analysis**	
***MGMT* promoter methylation analysis**	
***MLH1* promoter hypermethylation analysis**	

Please note that turnaround times can be found in Section 3.8.