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University Hospitals of Leicester NHS Trust

Cytogenetics User Handbook

A user guide for UHL Cytogenetic Laboratory Pathology services

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1.0 Introduction

The information contained in this user handbook is designed to meet the needs and requirements of our service users. This handbook includes detailed information on the service available within the Cytogenetics Laboratory service at University Hospitals of Leicester (UHL).

If you require any further information on any aspect of the service or have a comment or suggestions on how we can improve please contact us via email at uhl-tr.cytogeneticsmailbox@nhs.net or telephone 0116 258 5637.

If you require clinical advice, please contact the laboratory and ask to speak with the Head of Service or one of the senior clinical scientist team.

2.0 Scope of the service

The cytogenetics laboratory is a Consultant Clinical Scientist led team that includes HCPC registered Clinical Scientists, Genetics Technologists and Biomedical Assistants

We offer an analysis and clinical interpretation service on a range of prenatal, postnatal, haematological and cancer samples using chromosome analysis, FISH, SNP microarray, single gene variant analysis and next generation sequencing (NGS).

The Laboratory is externally accredited by UKAS (ref 8069) to the ISO15189 (2022) Medical Laboratory standard and participates in the relevant Genetics External Quality Assessment Schemes (GenQA) and UK NEQAS Leucocyte Immunophenotyping Scheme to cover the full repertoire of our services and ensure the highest standards.

The laboratory is co-located and works in conjunction with the Clinical Genetics Service.

The Cytogenetic Laboratory Service is a subcontracted Local Genomic Laboratory (LGL) of the East Genomic Laboratory Hub (GLH) led by Cambridge University Hospital Foundation Trust. Nottingham University Hospitals NHS Trust are also an LGL for the East GLH.

The GLH provides testing in line with the NHS England genomic test directories for rare disease and cancer and associated eligibility criteria. Cytogenetic investigations, and some single gene and NGS cancer testing are undertaken within the UHL laboratory on behalf of the GLH.

There is no Molecular Genetics service within UHL for rare disease. Samples are sent to the required laboratory within the East GLH network for testing based on the referral reason in line with the GLH contract. Advice on sample requirements and the testing laboratory to be

used can be obtained from the cytogenetics laboratory or by contacting the GLH duty scientist by email.

Please note that we do not accept self-referrals from patients and that all results are strictly confidential.

3.0 General Information

3.1 Location of services

The Laboratory is located in the Genetics centre on the Leicester Royal Infirmary (LRI) site of UHL. Molecular pathology testing is carried out in a laboratory within the Robert Kilpatrick Clinical Sciences building (RKCSB) also on the LRI site.

3.2 Opening hours

The laboratory is open 9.00-17.30 Monday to Friday. There is no out of hours service

3.3 Contact information

Contact the laboratory if you require clinical advice or have any questions regarding the available service, results or sample requirements.

Enquiry line 0116 258 5637

Key staff contacts:

Head of service: Lara Cresswell FRCPath
Trainee Consultant Clinical Scientist &
Deputy head of service Paul Warman
Principal Clinical Scientists & Section Heads:
Karen Healey– Constitutional Section
Karen Marshall – Haematology and FISH
Claire Slater – Molecular pathology
Operational Manager: Rajesh Thakrar
Laboratory Co-ordinator: Anne-Marie Rocks
General enquiries mailbox – uhl-tr.cytogeneticsmailbox@nhs.net
East GLH duty scientist - cuh.geneticslaboratories@nhs.net

Please note that nhs.net email addresses conform to DCB 1596 accreditation.

3.4 Why is genetic testing undertaken in the laboratory?

Chromosome abnormalities usually involve large amounts of DNA that can be rearranged, deleted or duplicated. Chromosome analysis may be carried out to investigate fertility issues,

confirm some common chromosomal syndromes such as Down syndrome and to look for acquired chromosome changes associated with leukaemia.

Microarray investigation allows the identification of very small losses and gains of genetic material that are too small to be seen on chromosome analysis. Using a patient's DNA rather than their chromosomes very small loss and gain can be identified. In addition, where a particular type of microarray is used (a SNP array) uniparental disomy and absence of heterozygosity (AOH) may be identified. Microarray investigation is used in prenatal diagnosis and in the investigation of dysmorphism, congenital malformations and intellectual disability.

Molecular pathology testing is an important part of the diagnosis of cancer as acquired genetic changes can be of diagnostic, prognostic and therapeutic significance.

3.5 Protection of personal data

All laboratory staff undertake mandatory Information Governance training ensuring that patient information is handled legally, securely and confidentially.

UHL's privacy data protection notice can be found on the main website at https://www.uhleicester.nhs.uk/privacy/data-protection/
This Notice explains:

- Who we are and contact details for our Data Protection Officer (DPO)
- What kinds of personal information about you we collect and process, how we process it, and what the legal grounds for processing are
- How we keep the information safe and how long we keep it for
- What your rights are under Data Protection law
- What you should do if your information changes
- Who you can speak to for further information or to make a complaint

3.6 User feedback

The laboratory welcomes and encourages feedback and comments from our users regarding any aspect of our service. All sections within the laboratory can be contacted on 0116 258 5637 or extension 15637 or email; uhl-tr.cytogeneticsmailbox@nhs.net

3.7 Complaints and concerns

We aim to provide an excellent service however any complaints or concerns about our service, from patients or referring clinicians, will be dealt with according to the UHL Management of complaints policy. Complaints may be directed to the Head of Service in the first instance or direct to the Patient Advice and Liaison Service (PALS).

4.0 Requesting procedures

The laboratory **can only accept referrals that are eligible for testing** according to the NHS England genomic test directories. A separate document setting out eligibility for rare disease testing can be found alongside the test directory.

The laboratory provides specific cytogenetic referral forms for different referral types; sample bags can be attached. Please contact the laboratory to obtain a relevant supply. **For rare disease referrals the East GLH rare disease referral form should be used primarily (see section 4.3).** The forms give information on the sample types and volumes required. Cytogenetic tests cannot be requested electronically with the exception of JAK2.

Referrals for molecular testing to be undertaken at the GLH are sent to the cytogenetics laboratory at UHL where they are collated and sent to the relevant GLH lab via the daily courier service. The cytogenetics laboratory holds a record of the sample received and where they are sent but does not have access to the results for these investigations.

Please note that we do not accept self-referrals from patients and that all results are strictly confidential.

4.1 Sample collection and specimen types

All samples should be sent to the laboratory as soon as possible. Where samples are taken out of hours samples should be stored in a fridge and sent on the next working day.

Blood samples – please ensure samples are well mixed to prevent clotting
for chromosome analysis and/or FISH: lithium heparin tube; minimum 1ml
for microarray: EDTA tube
for JAK2 PCR : EDTA tube

Amniotic fluid samples – all investigations: minimum 15mls taken into a sterile universal container

Chorionic villus - all investigations: minimum 10mg villus sent in transport media (available from the laboratory)

Bone marrow - all investigations except NGS: minimum 1ml of marrow sent in transport media (available from the laboratory)

Skin biopsies: full dermal thickness sent in sterile physiological saline or transport media (available from the laboratory)

Solid tissue samples: sent in sterile physiological saline or transport media (available from the laboratory)

Products of conception (POC): POC samples must contain chorionic villus material or fetal parts. DO NOT send the entire evacuation products. Samples from UHL must be sent according to the UHL policy for sensitive disposal of fetal remains up to 16 weeks. Samples from other hospital trusts must comply with their own policy and provide evidence of disposal wishes with the sample.

Placental samples (from pregnancy losses): Placental samples will only be accepted if taken from the fetal side of the placenta. Cord samples should be taken from near the cord insertion site.

Formalin fixed paraffin embedded tissue (FFPE):
For FISH – 2µm sections on sticky slides sent de-paraffined with additional H&E slide with area of interest marked
For NGS - <20% tumour cellularity 8x4µm slides with additional marked H&E slides from either end
->20% tumour cellularity 2x10µm curls in an eppendorf
Where the tumour material is dispersed throughout the specimen and unsuitable for microdissection please send 2x10µm curls regardless of tumour cellularity %

Samples for Myeloid NGS
Myeloid NGS can be undertaken on bone marrow or blood in an EDTA tube or FFPE samples.
All samples should be sent via the HMDL lab and must be recieved with a completed HMDL referral form. If samples are sent directly to the genetics lab, this may delay sample processing while the appropriateness of the referral is investigated.

Where a suboptimal sample is received this may lead to a longer reporting time.

4.2 Specimen Labelling

To avoid errors in identification, samples must be clearly marked with the patient's full name and date of birth. This information should correspond exactly with the information on the request form.

If there is any discrepancy, the processing of the sample may be delayed until the information is checked. Ultimately the sample may be discarded if the patient cannot be positively identified. Unlabelled samples that cannot be reliably identified to a patient cannot be accepted for analysis.

4.3 Completion of request forms

Cytogenetic request forms should be used and be fully completed and signed by the requesting clinician. If available, addressograph labels should be used on request forms.

All forms must show patient details (at least first name and family name and date of birth), referring consultant, referring hospital, date of sample, full clinical details and where possible the appropriate test directory test code. Wherever possible the NHS number should be included on the form.

There are different coloured referral forms available from the laboratory however please contact our mailbox if you require a form urgently and we can email one to you.

The Genomic Laboratory Hub referral forms can also be used and are available from the East GLH website.

UHL referral forms – contact the laboratory for a supply:

White cytogenetics referral form should be used to request karyotyping/chromosome analysis and FISH.

Pale blue Molecular Haematology referral from should be used to request JAK2 V617F PCR testing and Myeloid NGS panel testing.

Orange molecular pathology referral form should be used to request NGS testing for solid cancer.

4.4 Consent for testing

All genetic testing requires consent. The responsibility for obtaining informed consent for genetic testing lies with the referring clinician. The laboratory assumes that by provision of a sample and completed referral from that consent has been obtained by the referring clinician. Documentation relating to this consent should be retained in the patient's clinical notes.

A record of discussion form is available on the East GLH website that can be helpful in recording consent.

4.5 Additional testing on a sample

There are some limited circumstances where additional tests may be undertaken on the original sample. Please contact the laboratory for advice before taking a further sample.

4.6 Verbal requests

Where a verbal request is made for testing the referring clinician must provide written confirmation of the verbal request. Laboratory staff will inform service users of this procedure if the verbal request is accepted.

4.7 Fibroblast cultures for onward referral

The laboratory will only grow fibroblasts cultures for onward referral outside of the GLH network by prior agreement, and an onward referral letter will be required upon receipt of the sample. The referring clinician will be responsible for selecting the testing laboratory.

5.0 Transport of specimens to the laboratory

Samples (especially bone marrow and prenatal samples) should be received in the laboratory as quickly as possible.

Samples must not be centrifuged or frozen or have been processed by another laboratory.

Out-of-hours samples should be stored in a refrigerator and sent the next working day. Significant delay in sending samples may result in the sample being unsuitable for processing.

Specimens must be placed in a sealed sample bag. In order to avoid accidental contamination, please do not place request forms in the same bag as the specimen pots.

5.1 Transport of samples within UHL

Samples can be sent to Pathology Specimen Reception (Sandringham level 2, LRI) using the regular LRI clinical distributor service for delivery to Cytogenetics or bought direct to the laboratory by portering services.

Samples from LGH or GH can be sent via the regular inter-hospital van transport and delivered to Pathology specimen reception for onward delivery by the clinical distributors.

5.2 Samples from external hospitals

Where samples are sent by post they must comply with current regulations (P650) on the transport and postage of biological materials and display the UN3373 symbol.

Samples should be placed in sealed specimen bags with the form attached to the outside and then placed in rigid outer packaging. It is recommended that samples are sent via Royal Mail special/tracked delivery or courier to ensure sample receipt can be confirmed.

5.3 Sending samples to the Genomic Laboratory Hub (GLH)

Samples from UHL and some other external hospitals or GPs that require molecular genetic/genomic testing are received by the cytogenetics laboratory and sent on to either the Nottingham Molecular Genetics laboratory or the GLH lab at Cambridge depending on the test directory code that is required. Transport is via a dedicated courier service provided by the GLH and staff in the cytogenetics laboratory will ensure samples are appropriately packaged for transport with the courier.

Samples will be logged for transit audit purposes, reports are not received back in the cytogenetics laboratory but sent direct to the referring clinician.

5.4 High risk specimens

The cytogenetic laboratory does NOT have a category 3 containment facility. This means any sample suspected of having or having a pathogen that requires containment level 3 (e.g. TB) cannot be accepted by the department for culturing.

It is the responsibility of the requesting clinician to ensure that all high risk or potentially high risk specimens are handled correctly and dealt with in the following manner:

Acceptable samples suspected or known to contain hazardous pathogens that require culturing are restricted to hepatitis B, hepatitis C, and HIV. Samples must be labelled with ‘Danger of Infection’ stickers. A sticker must also be attached to the request form, and both form and sample placed in a ‘bio-hazard’ bag.

The sample container must be appropriate for the purpose, must be properly closed and not externally contaminated by the contents.

The form must be placed in the separate pocket of the bag, NOT inside with the sample. Where bags are attached to the form the bag should be sealed shut after placing the sample inside.

The referral form must be fully completed including contact details of the clinician taking the sample. To avoid a delay in processing a high risk sample the laboratory must be able to contact the referring clinician.

Please inform the laboratory if the patient is subsequently shown not to carry the pathogen.

6.0 Constitutional investigations

The laboratory can undertake investigation of constitutional genetic conditions using karyotyping/chromosome analysis, microarray or fluorescent in situ hybridisation (FISH). The most appropriate method will be selected by the clinical scientists based on the clinical details provided and the relevant test directory code.

The constitutional section head is Karen Healey. If you have any questions regarding sample requirements or results please contact the laboratory on ext. 15637 or email

6.1 Postnatal microarray

Microarray investigation allows a genome wide screen at a high resolution for the detection of previously cryptic copy number imbalance (losses and gains of material).

The service offers microarray investigation using the Illumina Infinium GSA SNP array. This has a functional resolution across the genome of 200kb but in addition is able to identify uniparental disomy, absence of heterozygosity (AOH) and a lower level of mosaicism. The GSA array processing is undertaken by the GLH main lab at CUH with analysis undertaken by the team in cytogenetics. Whilst the GSA array has been fully validated this testing is not yet on the laboratory's ISO 15189 schedule of accreditation. An extension to scope assessment has been submitted and an assessment awaited.

A GLH rare disease referral form should be fully completed; see section 4.0 for further details of sample volumes and requirements.

Microarray analysis is undertaken using a patient's DNA. Therefore an EDTA blood sample is required. DNA is usually extracted in the CUH laboratory for the GSA array with remaining DNA stored long term by the CUH laboratory for any future testing needs. However, in some circumstances DNA may be extracted in the UHL laboratory and forwarded to the CUH laboratory for testing.

For neonatal microarray referrals a lithium heparin sample is also required if rapidFISH for a common aneuploidy is required e.g. Down syndrome is suspected; this will be undertaken prior to the array. Where a rapidFISH investigation is shown to be abnormal microarray investigation will not be undertaken, the FISH result will be confirmed by a targeted chromosome analysis.

6.2 Postnatal karyotyping/chromosome analysis

Microarray investigation is the first line test for the majority of postnatal blood referrals. Please see section 6.1.

Karyotyping/targeted chromosome analysis is used for specific referrals reasons such as mosaicism screening and looking for balanced structural chromosome rearrangements. Please contact the laboratory for advice on the appropriate test and which referral form to use.

6.3 Rapid aneuploidy testing

Rapid screening for common aneuploidies is undertaken by FISH for relevant urgent neonatal referrals and for some prenatal samples. See section 7 for further information on FISH testing.

The common aneuploidies that can be investigated are trisomies 13, 18 and 21 and sex chromosome aneuploidy.

Amniotic fluid samples which are heavily bloodstained may not be suitable for rapid aneuploidy testing. Microarray investigation may also be delayed as a direct extraction of DNA may not be suitable.

6.3.1 Rapid aneuploidy testing for the Fetal Anomaly Screening Programme (FASP)

Rapid aneuploidy testing for the FASP is undertaken by the QF-PCR method at the GLH main laboratory at CUH. The fetal medicine units at UHL send samples to the cytogenetics laboratory where DNA will be extracted and sent to CUH for QF-PCR. Samples are cultured at UHL and any follow up chromosome analysis undertaken in the cytogenetics laboratory. The prenatal microarray testing is undertaken at the CUH laboratory using the GSA SNP array with the analysis completed by the UHL laboratory.

Samples requiring rapid aneuploidy testing must reach the laboratory before 3.30pm to allow DNA to be sent to CUH for QF-PCR the next working day. QF-PCR results are usually available within 24-48 hours of receipt in the CUH laboratory however the turnaround time is 3 calendar days from receipt of the DNA.

Please note any samples outside of the FASP e.g. invasive samples for prenatal single gene disorder familial testing will continue to have rapid aneuploidy testing by FISH.

6.4 Prenatal microarray

The Cytogenetics service offers a prenatal microarray investigation for samples referred due to an increased nuchal translucency and/or abnormalities seen on an ultrasound scan. All prenatal samples are received from the Fetal Medicine service.

The sample requirements for prenatal microarray are the same as for prenatal cytogenetics and the microarray is undertaken on DNA extracted from the prenatal material received..

The service offers microarray investigation using the Illumina Infinium GSA SNP array. This has a functional resolution across the genome of 200kb but in addition is able to identify uniparental disomy, absence of heterozygosity (AOH) and a lower level of mosaicism. The GSA array processing is undertaken by the GLH main lab at CUH with analysis undertaken by the team in cytogenetics. Whilst the GSA array has been fully validated this testing is not yet on the laboratory's ISO 15189 schedule of accreditation. An extension to scope application has been submitted and an assessment awaited.

All samples will have a rapid aneuploidy test before microarray by QF-PCR. Where the QF-PCR result is abnormal it will be confirmed by a targeted chromosome analysis. Where the QF-PCR test is normal a GSA SNP microarray test will be undertaken. The QF-PCR test is able to indicate if there is any maternal cell contamination in the sample.

Long term storage of fetal DNA will be undertaken at the CUH laboratory where the QF-PCR and microarray investigations are carried out, appropriate consent must be obtained.

Where a suboptimal sample is received, or samples are taken late in the 3rd trimester, this may lead to a longer reporting time.

6.5 Prenatal karyotyping/chromosome analysis

Routine karyotyping is no longer carried out on samples received for a raised nuchal translucency or abnormalities found on ultrasound scan. Prenatal microarray investigation is the commissioned test.

Samples received due to a high risk Down Serum Screening Programme result will only have a rapid aneuploidy QF-PCR result which will be undertaken at the GLH main lab at CUH. Full karyotyping or microarray will not be undertaken however where the QF-PCR result is abnormal this will be confirmed by a targeted chromosome investigation.

Where a suboptimal sample is received, or samples are taken late in the 3rd trimester, this may lead to a longer reporting time.

6.6 Pregnancy loss investigation

Genetic investigation of pregnancy losses is undertaken using microarray. Referral reasons include recurrent miscarriage (3 or more), a termination of pregnancy for abnormalities found on ultrasound scan or any third trimester loss which cannot be otherwise explained. Referrals are accepted according to the referral criteria in the NHSE rare disease test directory. The relevant test directory codes are R22.2 and R318.2. Please note that samples are not accepted from isolated pregnancy losses in the second trimester where there are no anomalies on the ultrasound scan. A rejection report will be issued for any referrals that are not eligible for testing.

Pregnancy loss samples can be amniotic fluid, chorionic villus, products of conception (POC), cord or skin.

POC samples must contain chorionic villus material or fetal parts. Please do not send the entire evacuation products. Consent for disposal must be sent with the sample.

Amniotic fluid samples and chorionic villus samples are generally taken by fetal medicine consultants as for prenatal cytogenetic investigations.

Samples cannot be processed if they have been exposed to formalin.

7.0 Fluorescent in situ hybridisation (FISH)

The Cytogenetic Laboratory has a range of probes available for fluorescent in situ hybridisation (FISH) testing. These include probes for both constitutional and acquired abnormalities.

The head of the FISH section is Karen Marshall. If you have any questions regarding FISH testing please contact the laboratory on ext 15637 or email.

FISH testing is a useful tool for identifying rearrangements which may be missed by traditional karyotyping and is particularly useful in haematological malignancy investigation. It can also be used to confirm some microarray findings. FISH analysis can be undertaken on chromosomes or interphase nuclei and from paraffin embedded tissue samples .

As microarray investigation will assess all constitutional microdeletion and microduplication syndromes this is the best method of analysis for such postnatal and prenatal referrals. If you require FISH testing for a specific syndrome, please contact the laboratory to discuss the best method of investigation.

Rapid FISH screening is available for the common aneuploidies for all prenatal samples not part of the FASP and clinically relevant urgent neonatal referrals by prior agreement with the laboratory. The turnaround time for this test is within 3 calendar days.

Urgent FISH testing is a requirement for certain types of leukaemia and a preliminary FISH result should be available within 3 calendar days. These results are also available in the HMDL laboratory.

Where a routine FISH test is undertaken this is reported with the final report. **The turnaround time is based on the sample type received.**

Molecular genetic testing is the first line test for the specific investigation of Prader-Willi and Angelman syndrome. Microarray analysis is able to identify the common causes of these syndromes. Both tests require an EDTA blood sample. Please contact the laboratory for advice on the most appropriate test.

Some FISH probes may be used after the manufacturer's expiry date. These are not covered by our UKAS accreditation but have all been rigorously tested prior to reporting. A comment will be added to any reports where this has happened. Please contact the laboratory for any further information.

8.0 Acquired haemato-oncology cytogenetic investigation

The Haematology section performs chromosome analysis on haematological samples using traditional karyotyping and FISH analysis.

The Head of section is Karen Marshall. If you have any questions regarding sample requirements or results please contact the laboratory on ext 15637 or email.

Samples requiring cytogenetic testing should come to the laboratory via the HMDL laboratory (ext. 16518). The laboratory has specific criteria for its urgent classification.

Samples must be received as soon as possible after they have been taken. Old or clotted samples may not be successfully cultured.

Lithium heparin blood samples will generally only be accepted from CLL patients.

FISH studies will be undertaken as agreed with the HMDL Laboratory.

FISH studies on lymphoma referrals are undertaken for certain referrals as agreed with the HMDL laboratory. These studies are carried out using paraffin embedded tissue sections

(PETS) and are referred by histopathologists. FFPE slides are prepared by the cellular pathology lab for these tests. Turnaround times for these requests are agreed on a case by case basis.

Bone marrow samples from patient's known or suspected of having Myeloma should only be referred if the plasma cell count is >10%. Samples should be transported to the laboratory as quickly as possible and **should not be taken on Friday** where possible as these may not be able to be processed on receipt. Cytogenetic investigation for Myeloma is carried out on purified plasma cells. Delays in sample transport or processing can reduce the number of plasma cells available for purification and subsequent analysis.
See section 9.3 for details on NGS testing for myeloid disorders

9.0 Molecular pathology investigation

Molecular testing is an important part of the diagnosis of patients with cancer as acquired genetic changes can be of diagnostic, prognostic and therapeutic significance.

The head of section is Claire Slater. If you have any questions regarding sample requirements or results, please contact the laboratory on ext 15637 or email.

Molecular profiling of tumours can help provide information on the most effective therapy for a certain type of solid or haematological tumour.

Specific DNA variants (mutations) can be associated with sensitivity to certain therapeutic drugs; they can also indicate when a patient is becoming resistant to treatment allowing a change to an alternative therapy.

The service undertakes testing for certain gene variants associated with some solid and haematological cancers including myeloproliferative neoplasms (MPNs), myeloid leukaemias, lung cancer, colorectal cancer, melanoma, breast cancer, GIST and cholangiocarcinoma. Other tumour sites may be accepted if the genes of interest are included on the Thermofisher Oncomine Precision Assay (OPA) or Oncomine Myeloid Assay (OMA) gene panel. Please contact the laboratory to discuss.

DPYD testing:
Targeted variant testing of the DPYD gene for 5-fluorouracil toxicity is undertaken by the East GLH at Cambridge.

9.1 PCR testing

PCR testing is routinely undertaken for the JAK2 V617F variant associated with myeloproliferative neoplasm. A commercial quantitative PCR kit is used and a variant allele frequency >1% is reported. Monitoring requests will only be undertaken on an annual basis, any samples received out with of this time scale will be rejected.

Targeted PCR testing for KRAS, EGFR & BRAF is currently on the laboratory's schedule of accreditation and can be used where an urgent result is required and the NGS panel is not possible within the required timescale.

9.2 NGS testing for solid cancers

Molecular testing for lung cancer, colorectal cancer, melanoma, breast cancer, GIST and cholangiocarcinoma referrals is undertaken using the Thermofisher Oncomine precision Assay (OPA) gene panel on the Genexus NGS platform. Whilst the OPA panel covers single nucleotide variants, copy number variants and fusions across 50 key genes each cancer type will be analysed for the defined set of genes, covered by the OPA, in line with the multitarget NGS panel test described in the NHSE cancer test directory. In general, only actionable or driver variants are reported.

DNA and RNA extraction will be undertaken for all referred cases where possible to facilitate any downstream testing. This will be stored in the genetic laboratory.

Whilst the OPA assay has been fully validated this testing is not yet on the laboratory's ISO 15189 schedule of accreditation. An extension to scope application has been submitted and an assessment awaited.

The technical details for all molecular cancer tests are included in the report produced by the cytogenetics laboratory. Where this information is not visible on results transcribed into different systems e.g. iLab then this information is available from the laboratory.

9.3 NGS testing for myeloid disorders

The Oncomine Myeloid Assay (OMA) is used to test for SNVs and small indels as per the National Genomic Test Directory targets for AML, MDS and MPN across 45 genes.

Samples should only be received following review by the HMDL laboratory.

DNA is extracted from bone marrow, blood or FFPE samples as appropriate. This will be stored in the genetic laboratory.

Whilst the OMA assay has been fully validated this testing is not yet on the laboratory's ISO 15189 schedule of accreditation. An extension to scope application will be submitted in due course.

The technical details for all molecular cancer tests are included in the report produced by the cytogenetics laboratory. Where this information is not visible on results transcribed into different systems e.g. iLab then this information is available from the laboratory.

10.0 Molecular genetic and genomic testing

There is no molecular genetics service within UHL. Testing is undertaken at the relevant testing laboratory within the GLH network. Samples will be sent initially to either the GLH laboratory at Cambridge or the Nottingham Regional Molecular Genetics Service based at the Nottingham City Hospital.

The East Genomic Laboratory Hub (GLH) is based at Cambridge University Hospital Foundation Trust. The laboratory is accredited by UKAS to the ISO:15189 Medical Laboratories standard (ref 8840). The laboratory manager is Mark Dance.

The service manager for the Nottingham Regional Molecular Genetics Service is Amanda Corkill. The laboratory is accredited by UKAS to the ISO:15189 Medical Laboratories standard (ref 8044).

To find out which tests are undertaken at the Nottingham laboratory please see their website, all other tests will be arranged via the Cambridge laboratory.

For clinical advice, results or sample requirements please contact the GLH duty scientist on email or the Nottingham laboratory on 0115 9691169 ext 55207 or by email

Referral forms for the GLH laboratory can be found here which are accepted at both CUH and NUH.

Please note that results from molecular genetic investigations are not available in the cytogenetics laboratory. Results are emailed to the referring clinician by the testing laboratory.

If you require molecular genetic/genomic testing, samples should be sent to the Leicester cytogenetic laboratory for onward referral to either Cambridge or Nottingham. Samples are transported via the GLH courier each morning Monday to Friday.

11.0 Availability of results

Results will only be given to authorised clinical staff and we may refuse to disclose results if you are not from the referring clinician's team.

All authorised reports are emailed as pdfs direct to the referring clinician or their nominated mailbox. Valid reports will have an authorising signature.

Cytogenetic reports for prenatal and postnatal samples are not available on iLab.

Verbal requests for results can be made providing the clinician is involved in the care of the patient. Callers must be able to reliably identify themselves. Results are not given directly to patients.

Cytogenetic reports for haematology and solid cancer referrals including JAK2 are also available in iLab.

Uncertainty of Measurement

The laboratory considers the uncertainty of measurement for all its procedures. Please contact the laboratory if further details on our approach to uncertainty are required.

Molecular Genetic/Genomic test results

The cytogenetic laboratory does not have access to molecular genetic/genomic results. These results are only available from the molecular genetics laboratory who performed or organised the testing and will be routinely emailed to the referring clinician.

For referrals prior to December 2020 please contact the Nottingham Molecular Genetics Laboratory on 0115 969 1169 ext 55207 or secure email. Referrals received after this date may be available from Nottingham or the GLH laboratory who can be contacted by email in the first instance or on 01223 348866.

12.0 Turnaround times

The laboratory aims to meet the NHSE required turnaround time for all of our tests.

Turnaround times are monitored on a monthly basis and form part of our monthly assurance report.

Turnaround times are all in calendar days with the day of receipt being day 0.

Testing	subcategories	Turnaround time Calendar days	Examples
Postnatal microarray & chromosome analysis	Newborn <1 month Family testing where mother is pregnant	14 days	
	Routine	42 days	Intellectual disability Pregnancy loss
Rapid aneuploidy* FISH testing	Non-FASP prenatal requests and newborn babies	3 days	
Prenatal microarray & chromosome analysis		14 days	
Haemato-oncology karyotyping	New AML/ALL/CML & relapse samples	14 days	
	Non-acute requests	21 days	MDS/MPN
Haemato-oncology FISH	New AML/ALL/CML & relapse samples	3 days	Where treatment stratification is required
	Non-acute requests	14 days	CLL
		21 days	Myeloma
Molecular pathology PCR		14 days	JAK2 V617F
Solid cancer NGS testing		14 days	Lung cancer Colorectal cancer Melanoma
Myeloid NGS panel testing	AML IDH1 R132	7 days	
	AML panel	14 days	
	MPN/MDS	21 days	

- Please note that QF-PCR testing undertaken at CUH for FASP referrals has a turnaround time of 3 calendar days from receipt in the CUH laboratory

The laboratory has specific criteria for its urgent requests.

- Newborn babies under 1 month old
- Diagnostic testing on proband and or parental samples where the mother is pregnant
- New acute leukaemia or CML cases and where relapse is suspected

We will endeavour to meet appointment dates; please record this information on the referral form. If a result is required urgently and is not one of the above categories please record this on the referral form, including a statement of why this is urgent, or contact the laboratory.