

Haemato-oncology Genetic Analysis

Analysis of haematological malignancies samples using an appropriate investigation method (see below) depending upon the referral reason and type of sample sent.

Referrals / Indications

- Acute Myeloid Leukaemia (AML)
- Acute Lymphoblastic Leukaemia (ALL)
- Chronic Myeloid Leukaemia (CML)
- Chronic Lymphocytic Leukaemia (CLL)
- Myelodysplastic syndromes (MDS)
- Myeloproliferative Neoplasms (MPN)
- Myeloma
- Lymphoproliferative disorders (LPD)
- Lymphoma
- Waldenström's Macroglobulinaemia (WM)
- Systemic Mastocytosis (SM)

Referrals for the above conditions can be sent for a number of purposes including diagnosis, prognosis, relapse and treatment follow-up

Sample Requirements

Blood in lithium heparin
Child index case – >1ml
Adult index case – 5ml
Bone marrow – send in sterile transport medium supplied by laboratory or in a lithium heparin blood tube.
For molecular testing send in bone marrow in an EDTA tube
Please label samples with three identifiers and date of collection
All samples must be accompanied by a completed request form
Consent for testing and sample storage is assumed when the request is received – it is the responsibility of the referring

Analysis / Services

- Chromosome analysis by karyotyping
- Fluorescence in situ hybridisation (FISH)
- Molecular analysis (various methods)

Urgent Referrals

The following are processed as urgent – if there is a specific clinical need for sample to be processed urgently please indicate on the request form:

- New diagnosis for AML, ALL, CML
- Query relapse
- Query disease progression

Contact Details

All Wales Genomics Laboratory,
Institute of Medical Genetics,
University Hospital of Wales,
Heath Park,
Cardiff CF14 4XW
Tel: 02921 844023
Lab.genetics@wales.nhs.uk
<http://www.wales.nhs.uk/AWMGS/>
Accredited to ISO 15189:2012
(8988)

Turnaround Times (TAT)

Analysis	TAT
Urgent chromosome analysis	14 cd
Routine chromosome analysis	21 cd
FISH testing	varies
V617F (c.1849G>T) mutation analysis of exon 14 (<i>JAK2</i>) Gene fragment length analysis (<i>CALR</i> exon 9)	14 cd
ddPCR testing for MYD88 p. (Leu265Pro) Variant in Waldenström's Macroglobulinaemia ddPCR testing for C-KIT p.(D816V) Variant in Systemic Mastocytosis	14 cd
Molecular analysis of fusion transcripts found in CML, AML* & ALL *FLT3-ITD 10 cd	14 cd
NGS for AML, MDS, MPN, CMML, JMML and CLL	21 cd
Molecular monitoring of BCR/ABL1 transcript levels (quantitative testing – qPCR)	14 cd
Chimerism studies for post bone marrow transplant monitoring - microsatellite analysis - FISH	14 cd 14 cd
B cell clonality analysis	T cell clonality analysis 14 cd

Links

Orphanet - <http://www.orpha.net/>

OMIM - <http://www.omim.org/>

GeneTests - <http://www.genetests.org/>

Genetic Test Registry - <http://www.ncbi.nlm.nih.gov/gtr/>

Decipher - <http://decipher.sanger.ac.uk/>

