

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
 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 clinician to ensure that appropriate consent has been obtained

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

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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	14 cd
T cell clonality analysis	14 cd

Links

Orphanet - <http://www.orpha.net/> EDDNAL - <http://www.eddnal.com/> 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/>