Sample Requirements

HAEMATO-ONCOLOGY

Please make the suspected disease(s) clear in the 'Relevant Clinical Details' section of the HODS referral form.

If multiple disease types are suspected, please make this clear and send the samples required for all possible testing pathways (e.g. ?myeloma & ?MDS - please send 1 x marrow in transport medium and 1 x EDTA marrow for MDS testing, plus 1 x EDTA marrow for myeloma testing, on the same HODS episode).

Please note this is not an exhaustive list of available investigations. Testing on lower sample volumes can be attempted.

Category	HODS investigation names	Ideal specimen type, Container & Volume	Notes
Acute Myeloid Leukaemia (AML) or APML	Cytogenetics (Karyotyping)	1ml bone marrow in 5-10ml of transport medium	• For culturing/karyotyping and will also initiate the AML FISH panel in this context.
	APML Diagnostic FISH	1ml bone marrow in 5-10ml of transport medium OR 2ml bone marrow or blood EDTA / LiHep	
	FLT3 ITD & NPM1	2ml bone marrow EDTA	• RNA extraction on EDTA also initiated by default for all AMLs
	NGS - Myeloid		
Myelodysplastic syndrome (MDS)	Cytogenetics (Karyotyping)	1ml bone marrow in 5-10ml of transport medium	
	NGS - Myeloid	2ml blood or bone marrow EDTA	
Myeloproliferative neoplasms (MPN)	BCR/ABL Diagnostic FISH	2ml blood EDTA or LiHep	• *BCR::ABL1* FISH • Eosinophilia FISH - *PDGFRa* only
	Eosinophilia FISH FIP1L1/PDGFRA		
	Eosinophilia RNA Fusion Panel	2ml bone marrow EDTA	• Only done if *PDGFRa* FISH negative on blood; includes *PDGFRB, PDGFRA, FGFR1* and other rare fusions
	MPN NGS Panel	2ml bone marrow or blood EDTA	• EDTA blood is preferred • MPN NGS Panel includes JAK2, CALR and MPL
	Myeloid NGS		
	Cytogenetics (Karyotyping)	1ml bone marrow in 5-10ml of transport medium	• For karyotyping (and FISH)
Systemic mastocytosis	C-KIT	2ml bone marrow (or blood) EDTA	• ddPCR for cKIT hotspot analysis
Chronic Myeloid Leukaemia (CML)	BCR/ABL Diagnostic FISH	2ml blood or bone marrow EDTA or LiHep	• BCR::ABL1 FISH for diagnosis
	BCR/ABL Diagnostic FISH	1ml bone marrow in 5-10ml of transport medium	• BCR::ABL1 FISH for diagnosis • And for karyotyping
	Cytogenetics (Karyotyping)		
BCR::ABL monitoring	BCR/ABL1 Monitoring RT-qPCR	2ml blood or bone marrow EDTA	• BCR/ABL1 RQ-PCR Quantitation
Acute Lymphoblastic Leukaemia (ALL)	Cytogenetics (FISH)	1ml bone marrow in 5-10ml of transport medium	• For FISH (and culturing for karyotyping, if required) • Marrow in transport medium with Colcemid not required
	SNP Array for ALL	2ml bone marrow in EDTA	• RNA extraction also initiated by default for all ?ALL
	TPMT/NUDT15 genotyping		

Category	HODS investigation name	Ideal specimen type, Container & Volume	Notes
Chronic Lympho-proliferative Leukaemia (CLL)	SNP Array for CLL (TP53 deletion)	4ml blood (or bone marrow) EDTA	• 'SNParray for CLL' has replaced 'TP53 deletion (FISH)'. • 'NGS - Lymphoid' has replaced 'TP53 mutation NGS' • **Marrow in transport medium / blood in LiHep not required**
	NGS - Lymphoid		
	IgHVH mutational analysis		
Multiple myeloma / Plasma Cell Dyscrasia	CD138+ FISH for Myeloma	4ml bone marrow EDTA	• **Marrow in transport medium not required** • Do not need to separately request 'Cytogenetics'
Lymphoma / LPD / LPL	Cytogenetics (FISH)	2ml bone marrow (or blood) EDTA	• For FISH only • If known, please specify FISH probes or lymphoma subtype • **Karyotyping will not be done for lymphomas, as per GLH agreement.** • **Marrow in transport medium not required.**
	T Cell Gene Rearrangement	2ml bone marrow (or blood) EDTA	• Aka. T cell clonality/ TCR rearrangement • Aka. B cell clonality/ Ig gene rearrangement
	B Cell Gene rearrangement		
	MYD88 Hotspot - L265P	2ml bone marrow in EDTA	• Detects MYD88 L265P only
	NGS - Lymphoid	2ml bone marrow in EDTA	• NGS panel includes TP53, MYD88, CXCR4 and others.
	Cytogenetics (FISH)	Paraffin-embedded Tissue (PET)	• Not requested via HODS • Please specify the lymphoma subtype or FISH probes required
Chimerism	Chimerism (PCR) - Whole sample	2ml bone marrow or blood EDTA	• Post bone marrow / stem cell transplant • Analysis on whole sample
	Chimerism (PCR) - CD3 Lineage specific	5ml bone marrow or blood EDTA for each cell fraction required, **in addition to above**	• Analysis on CD3+ cell separated sample fraction
	Chimerism (PCR) - CD15 Lineage specific	5ml bone marrow or blood EDTA for each cell fraction required, **in addition to above**	• Analysis on CD15+ cell separated sample fraction

SOLID TUMOUR ONCOLOGY / PHARMACOGENOMICS

Please refer to sample information on referral forms:

https://ney-genomics.org.uk/wp-content/uploads/2024/03/NEY_GLH_solid_cancer_genomics_referral_form_mar24.pdf

https://ney-genomics.org.uk/wp-content/uploads/2023/02/411.028-DPYD-Referral-Form-v3.0.pdf

RARE DISEASE

Test	Specimen Type	Volume	Notes/Comments
Molecular-based assays (PCR, Sanger sequencing, Next Gen Seq, Whole Gen Seq)	Blood	0.5-5ml EDTA	For other sample types, contact the lab prior to sample dispatch
	Saliva / buccal swab	Samples only accepted in the following collection kits:- DNA Genotek Oragene OG-500 DNA Genotek Oragene OG-575.	Contact the lab prior to sample dispatch
Newborn screening - Cystic fibrosis - Sickle Cell Disease – transfused babies	Dried bloodspots	1x 10mm-diameter bloodspot	Referrals via Newborn Screening only
Aneuploidy Detection (QF-PCR)	Amniotic fluid / chorionic villus /	10-20ml sterile universal / 3-4 fronds sterile universal in CVS culture medium	
	Placental biopsy / skin biopsy	1-2mm cubed Sterile tissue culture medium pots	
	Neonatal blood	0.5-2ml EDTA	
Microarray	Amniotic fluid / chorionic villus	10-20ml sterile universal / 3-4 fronds sterile universal in CVS culture medium	
	Placental biopsy / skin biopsy	1-2mm cubed Sterile tissue culture medium pots	
	Blood	0.5-5ml EDTA	
Chromosome/karyotype (with or without FISH)	Blood	1--3ml LiHep	* smaller samples can be attempted but may reduce the likelihood of a successful result
	Amniotic Fluid sample	10-20ml sterile universal	
	CVS	3-4 fronds sterile universal in CVS culture medium	
	Fetal blood cordocentesis	0.5-1ml LiHep	
	Cord blood	1-3ml LiHep	
	Placental biopsy at cord insertion site, fetal membrane, villi and cord biopsies	<1cm cubed Sterile tissue culture medium pots	
	Skin biopsy	1-2mm cubed Sterile tissue culture medium pots	
	Solid Tumour Biopsy	<1cm cubed Universal with 5-10ml of transport medium	
Chromosome breakage testing (Fanconi anaemia, Blooms syndrome)	Blood	3-4ml LiHep	Inform lab prior to sample dispatch