

**Sample Requirements**

<b>Test</b>	<b>Specimen Type</b>	<b>Volume</b>	<b>Notes/Comments</b>
<b>ONCOLOGY</b>			
Leukaemia (AML, ALL, CML, CLL) +/- FISH ( <b>diagnosis/monitoring</b> ) Myelodysplastic syndromes (MDS) Myeloproliferative disease (MPD)	Bone marrow/ leukaemic blood	0.25-1ml BM in 5-10ml of transport medium <b>OR</b> 1ml BM/VB in Li Hep	
Chronic Lymphoproliferative Leukaemia (CLL) –FISH	Bone marrow/ leukaemic blood	Universal with 5-10ml of transport medium <b>OR</b> BM/VB Li Hep	
Myeloma – FISH	Bone marrow	0.5-5ml EDTA	
BCR-ABL1 Quantitation (RQ-PCR)	Blood / Bone marrow	0.5-5ml EDTA	Send to Lab immediately
Chimerism post bone marrow/stem cell transplant (Sex matched/ Powerplex)	Blood/Bone marrow	0.5-5ml EDTA (see below if lineage-specific analysis required)	
Chimerism post bone marrow/stem cell transplant (Sex mis-matched (FISH))	Blood/Bone marrow	2-3ml Li Hep (see below if lineage-specific analysis required)	
Chimerism if lineage-specific analysis required (MACS separated fraction e.g. CD3+)	Blood only	0.5-1ml EDTA (IN ADDITION TO ABOVE)	
MRD analysis by Ig/TCR gene rearrangements	Bone marrow	2.5-10ml ACD.	Peripheral blood in ACD is acceptable at diagnosis if WCC is > 20x10 <sup>9</sup> /l.
cfDNA testing for EGFR mutations (Relapse or diagnostic)	Peripheral blood	10ml PB in Streck/Paxgene preservative tubes	Preservative tubes can be supplied by the laboratory if required. Contact lab prior to sending
Solid tumour FISH	Paraffin-embedded Tissue (PET)	2x4µm sections on slides	
Molecular testing of solid tumours (BRAF, EGFR, KRAS, pharmacogenetics NGS panel)	Paraffin-embedded Tissue (PET)	8x10µm sections in universal	
Tumour Microsatellite Instability Analysis (MSI)	Blood and PETS 8x10µm sections in universal	0.5-5ml EDTA	

Date of issue: 16 Oct 2021	Version number: 2
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<b>RARE DISEASE</b>			
Routine molecular-based assays (PCR, sequencing, Next Gen Seq)	Blood	0.5-5ml EDTA	For other sample types, 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/Fish)	Amniotic fluid / chorionic villus	10-20ml sterile universal / 3-4 fronds sterile universal in CVS culture medium	
Postnatal Microarray	Blood	1-3ml EDTA	
Chromosome/karyotype (with or without FISH)	Blood	1–3ml Li Hep	* 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 Li Hep	
	Cord blood	1-3ml Li Hep	
	Placental biopsy at cord insertion site, fetal membrane, villi and cord biopsies	<1cm cubed Sterile tissue culture medium pots	<a href="#">See</a> our “additional information” page for information regarding sending samples
	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 Li Hep	Inform lab prior to sample dispatch

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