



## Haematology Probe List

Do you require a commercially available test *not* on this list?  
Please contact us on 0210075616 to discuss options

Probes can be ordered separately

<b>Billing codes:</b>
Tumour sections & Smears: 4757 + 4760 + (4761 x number of probes)
Blood or Bone Marrow: 4760 + (4761 x number of probes)

Mature B-cell Neoplasms (Probe sets can be ordered individually)	Chronic lymphocytic leukemia/small lymphocytic lymphoma (CLL) FISH Panel (Set 1)	11q22.3 17p13.1 (2 probes)	ATM Deletion TP53 Deletion	
	Chronic lymphocytic leukemia/small lymphocytic lymphoma (CLL) FISH Panel (Set 2)	CEP12 13q14 13q34 (3 probes)	D12Z3 Enumeration D13S319 Deletion LAMP1 Deletion	
	Multiple Myeloma Primary panel	1p32.3 1q21 (2 probes) 17p13.1 (2 probes) 14q32 (2 probes)	CDKN2C CKS1B Enumeration TP53 Deletion IGH Rearrangement	
	Multiple Myeloma Reflex panel – IGH rearranged	t(4;14)(p16;q32) (2 probes) t(14;16)(q32;q23) (2 probes) t(14;20)(q32;q12) (2 probes)	IGH/FGFR3 Translocation IGH/c-MAF Translocation IGH/MAF-B Translocation	
	Total: 6 probes			
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	Total: 6 probes			

Myelodysplastic Syndromes (MDS)	Myelodysplastic Disorder (MDS) Panel (Probes can be ordered individually)	5p15/5q31/5q33 (3 probes) 7q31 (2 probes) CEP8 (1 probe) 20q12 (1 probe)	CDC25C/RPS14/E GR1 Enumeration D7S486, CEP7 Deletion D8Z2 Enumeration D20S108 Deletion	
	Request additionally	11q23 (2 probes)	MLL Rearrangement	
	Total: 7 probes			
	Total: 7 probes			
	Total: 7 probes			

Micro-deletion	Williams Syndrome	7q11.23 7q31 (2 probes)	ELN Deletion	
	22q Deletion Syndrome	22q11.2 22q13.3 (2 probes)	TUPLE1 Deletion	

Sex Chromosomes	SRX	CEPX Yp11.3 (2 probes)	SRX Deletion	
	XY	CEPX CEPY (2 probes)	XY Enumeration	

### Requirements

Sample types are processed with sample related protocols.

#### Requirements for Blood and Bone Marrow samples:

Resuspend 2-4ml sample in EDTA or Lithium Heparin tube.

#### Requirements for Smears:

At least one slide per probe set (if possible an extra slide each as tests may need to be repeated)

Smear should not be thick as it is difficult for the probe to penetrate for analysable signals

Smear should not be older than 1 month (as sample quality deteriorates with time)

#### Requirements for Tumours:

Two slides per probe set (as tests may need to be repeated)

Tumour slice 2-3 µm thick

Placed on positively charged slide

**Invasive area marked on an accompanying H&E slide (time is wasted analysing unmarked tissue sections)**

Acute Myeloid Leukemia (AML)	Acute promyelocytic leukemia, FAB type M3	t(15;17)(q24;q21.1) and variants (2 probes) Request separately	PML/RARα Translocation	
	AML with deletion of 7q	7q31 (2 probes)	D7S486, CEP7 Deletion	
	AML with deletion of 5q	5p15/5q31/5q33 (3 probes)	CDC25C/RPS14/E GR1 Enumeration	
	AML, FAB type M2	t(8;21)(q22;q22) (2 probes)	RUNX1/RUNX1T1 Translocation	
	AML, FAB type M4Eo	Inv(16)(p13q22) or t(16;16)(p13q22) (2 probes)	CBFB Rearrangement	
	AML with 11q23 abnormalities	11q23 (2 probes)	KMT2A (MLL) Rearrangement	
	AML with deletion of 20q12	20q12 (1 probe)	D20S108 Deletion	

Chronic Myeloproliferative diseases	Chronic myelogenous leukemia	t(9;22)(q34;q11.2) (2 probes)	BCR/ABL Translocation	
	Hypereosinophilic syndrome	4q12 (3 probes) 5q33 (2 probes) 8q11 (2 probes) Total: 7 probes	FIP1L1-CHIC2-PDGFRα PDGFRβ FGFR1	
	Polycythemia vera	t(9;22)(q34;q11.2) (2 probes)	BCR/ABL Translocation	
	Essential thrombocythemia	t(9;22)(q34;q11.2) (2 probes)	BCR/ABL Translocation	

Precursor B-Lymphoblastic Leukemia	B-ALL	t(9;22)(q34;q11.2) (2 probes) 11q23 rearrangements (2 probes)	BCR/ABL Translocation KMT2A (MLL) Rearrangement	
		t(1;19)(q23;p13) (2 probes)	Rearrangement	
	Included for <25 yrs	t(12;21)(p13;q22) (2 probes)	ETV6/RUNX1 Translocation	

Other (Please discuss with lab first to determine if request is possible):

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## Pathology Probe List

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At least one slide per probe set (if possible an extra slide each as tests may need to be repeated)

Smear should not be thick as it is difficult for the probe to penetrate for analysable signals

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#### Requirements for Tumours:

Two slides per probe set (as tests may need to be repeated)

Tumour slice 2-3 µm thick

Placed on positively charged slide

### **Invasive area marked on an accompanying H&E slide (time is wasted analysing unmarked tissue sections)**

Indicate Tumour percentage and IHC score

Solid Tumour	Breast cancer	17p11.1-q11.1 17q11.2-q12 (2 probes)	Her-2/neu Amplification	
	Lung cancer	7p12 7p11.1-q11.1 (2 probes)	EGFR/CEP7 Amplification	
		2p23 (2 probes)	ALK Rearrangement	
	Diffuse Malignant Peritoneal Mesothelioma	9p21 (2 probes)	p16/CDKN2A Deletion	
	Neuroblastoma	2p24 2p11.1-q11 (2 probes)	NMYC Amplification	
	Uveal melanoma	CEP3 CEP8 8q24 (4 probes)	Monosomy 3 Gain of 8q	
	Gliomas	1p 19q Codeletion (4 probes)	1p36; 1q25; 19q13; 19p13	
		9p21 (2 probes)	CDKN2A/B Deletion	
		7p12 7p11.1-q11.1 (2 probes)	EGFR/ CEP7 Amplification	
		CEP7 CEP10 (2 probes)	Deletion of 7 Gain of 10	

Tumour %	IHC Score (HER2 only)
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Non-Hodgkins Lymphoma	Extranodal marginal zone B cell lymphoma (MALT lymphoma)	t(11;18)(q21;q21) (2 probes)	API2/MALT1 Translocation	
	Follicular lymphoma	t(14;18)(q32;q21) (2 probes)	IGH/BCL2 Translocation	
	Mantle cell lymphoma	t(11;14)(q13;q32) (2 probes)	CCND1/IGH Translocation	
	Diffuse large B- cell lymphoma	3q27 (2 probes)	BCL6 Rearrangement	
		8q24 (2 probes)	MYC Rearrangement	
		18q21 (2 probes)	BCL2 Rearrangement	
		6p25 (2 probes)	IRF4 Rearrangement	
Burkitt's lymphoma	t(8;14)(q24;q32) (3 probes)	MYC/IGH Translocation		
Anaplastic lymphoma	2p23 (2 probes)	ALK Rearrangement		

Sarcoma	Ewing sarcoma and others	22q12 (2 probes)	EWSR1 Rearrangement	
	Alveolar rhabdomyosarcoma	13q14 (2 probes)	FOXO1 Rearrangement	
	Soft tissue sarcoma	16p11 (2 probes)	FUS Rearrangement	
	Synovial sarcoma	18q11.2 (2 probes)	SS18 Rearrangement	
	Liposarcoma	12q15 CEP12 (2 probes)	MDM2/ CEP12 Amplification	
	Dermatofibrosarcoma protuberans (DFSP)	17q21.33 22q13.1 (2 probes)	COL1A1/ PDGFB Translocation	
	Myxoid lipo- sarcoma/Fibromyxoid sarcoma	12q13 (2 probes)	DDIT3 Rearrangement	
	Renal cell carcinoma	6p21.1 (2 probes)	TFEB Rearrangement	
		Xp11.23 (2 probes)	TFE3 Rearrangement	

Other (Please discuss with lab first to determine if request is possible):