FISH Test Menu

UCLA Pathology Outreach Services
David Geffen School of Medicine at UCLA

Hematologic

Diagnosis	Gene(s)/Unique Sequence	Probe Name	Cytogenetic Aberration Detected
Myelodysplastic Syndromes (MDS) Panel (Probes can be ordered individually)	5p15.2/5q31	LSI D5S721, D5S23/EGR1	deletion or loss of chromosome 5
	7q31/CEP7	LSI D7S486 / CEP 7	deletion or loss of chromosome 7
	8cen (chromosome 8)	CEP 8 (D8Z2)	gain of chromosome 8
	20q12	LSI D20S108	deletions in the long arm of chromosome 20
Myeloproliferative Neoplasm (MPN) Panel (Probes can be ordered individually)	5p15.2/5q31,7q31/CEP7, 8cen,20q12		
	t(9;22)(q34;q11.2)	LSI BCR-ABL1	BCR-ABL1 fusions
	4q12	LSI PDGFRa	deletion of CHIC2
	5q32	LSI PDGFRb	PDGFRb (5q32) gene rearrangement
	FGFR1	LSI FGFR1	FGFR1 (8q11) gene rearrangement
Chronic lymphocytic leukemia/small lymphocytic lymphoma (CLL/SLL) Panel (Probes can be ordered individually)	11q22.3	ATM/cep11	deletions of the ATM gene
	12cen	CEP12 (D12Z3)	gain of chromosome 12
	13q14, 13q34	LSI D13S319 (13q14)/13q34	deletions of the 13q14 region and loss of chromosome 13
	17p13.1	LSI p53 (17p13.1)/cep17	deletions of the TP53 gene
	6q23	LSI MYB/cep6	deletions of chromosome 6q
	14q32	LSI IGHα	translocation involving 14q32
	t(11;14)(q13;q32)	LSI CCND1-IGHα	CCND1-IGHα fusions
Lymphoproliferative Disorder (LPD) Panel (Probes can be ordered individually)	14q32	LSI IGHα	translocation involving 14q32
	12cen	CEP12 (D12Z3)	gain of chromosome 12
Multiple Myeloma (MM) Panel (Probes can be ordered individually)	14q32	LSI IGHα	translocation involving 14q32
	13q14, 13q34	LSI D13S319 (13q14)/13q34	deletions of 13q14 and loss of chromosome 13
	17p13.1	LSI p53 (17p13.1)/cep17	deletions of TP53 gene
	5p15.2/5q31	LSI D5S721, D5S23/EGR1	gain or loss of chromosome 5
	7q31/CEP7	LSI D7S486 / CEP 7	gain or loss of chromosome 7
	t(11;14)	LSI CCND1-IGHα	CCND1-IGHα fusions
	t(4;14)	LSI FGFR3-IGHα	FGFR3-IGHα fusions
	t(14;16)	LSI IGHα-MAF	IGHα-MAF fusions
B-Lymphoblastic Leukemia/ Lymphoma (B-ALL)	t(9;22)(q34;q11.2)	LSI BCR-ABL1 ES	BCR-ABL1 fusions
	11q23 rearrangement	LSI MLL	MLL gene rearrangement
	t(8;14)(q24;q32)	LSI MYC-IGHα	MYC-IGHα fusions
T-Lymphoblastic Leukemia (T-ALL)	14q11 rearrangement	LSI TCR A/D	TCR gene rearrangement
	11q23 rearrangement	LSI MLL	MLL gene rearrangement
Precursor B-Lymphoblastic Leukemia (B-ALL)	t(1;19)(q23;p13.3)	LSI TCF3-PBX1	TCF3-PBX1 fusions
	t(12;21)(p13;q22)	LSI Tel/AML1	TEl-AML1 (ETV6-RUNX1) fusions
	14q32	LSI IGHα	14q32α gene rearrangement
	11q23 rearrangement	LSI MLL	MLL gene rearrangement
	t(9;22)(q34;q11.2)	LSI BCR-ABL1 ES	BCR-ABL1 fusions
	4 cen (chromosome 4)	CEP 4	gain or loss of chromosome 4
	10 cen (chromosome 10)	CEP 10	gain or loss of chromosome 10
	17 cen (chromosome 17)	CEP 17	gain or loss of chromosome 17
Acute Myeloid Leukemia (AML Panel) (Probes can be ordered individually)	t(8;21)(q22;q22)	LSI ETO/AML1	ETO/AML1 (RUNX1T1,RUNX1) fusions
	t(15;17)(q24;q21.1) and variant translocation	LSI PML/RARALSI RARA	PML-RARA fusions, RARA rearrangement
	inv(16)(p13q22) or t(16;16)(p13;q22)	CBFB	CBFB gene rearrangement
	11q23	LSI MLL	MLL gene rearrangement
AML with translocation or inversion	3q26.2	LSI EVI1	EVI1gene rearrangement
Chronic Myelogenous Leukemia (CML)	t(9;22)(q34;q11.2)	LSI BCR/ABL1 DC DF	BCR-ABL1 fusions
	t(9;22)(q34;q11.2)	LSI BCR/ABL1/ASS1 TC DF	BCR-ABL1 fusions and deletion 9q34
	9q34	LSI 9q34 (ASS1)	loss of chromosome 9q34 (ASS1 gene)
CML Accelerated Phase	t(9;22)(q34;q11.2)	LSI BCR/ABL1	BCR-ABL1 fusions
	8cen (chromosome 8)	CEP 8 (D8Z2)	gain of chromosome 8
	17q	LSI RARA	gain of chromosome 17q (iso17q)
Bone Marrow Transplant	X/Y	CEP X/Y	Ratio of donor/recipient cells

09/01/11

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Constitutional Syndromes

Diagnosis	Locus	Probe Name	Cytogenetic Aberration Detected
Constitutional (numerical aberrations)	13q14,21q22.13-q22.2,Xp11.1-q11.1 Alpha Satellite DNA, Yp11.1-q11.1 Alpha Satelite DNA, 18p11.1-q11.1 Alpha Satellite DNA	AneuVysion	Trisomy 13, 21, 18. Sex chromosomes X numerical aberrations
Subtelomeric deletions	subtelomeric deletion (enumeration and integrity)	ToTelVysion	Identifies 41 of the 46 human subtelomeres with the exclusion of the p-arm telomeres of the acrocentric chromosomes.
Steroid Sulfatase Deficiency	Xp22.3	LSI STS/CEPX	Steroid Sulfatase gene deletion
Kallmann Syndrome	Xp22.3	LSI KAL/CEP X	Xp22.3 deletion
Turner Syndrome (45,X)	Xp11.1-q11.1 Alpha Satellite DNA Yp11.1-q11.1 Alpha Satelite DNA	CEP X/Y	Chromosome X and Y
Yp13 deletion	Yp11.3	LSI SRY/CEP X	SRY deletion, loss of Y chromosome
1p36 deletion	1p36	LSI p58 (1p36)/TelVysion1p/1q25	1p36 microdeletion
3q29 deletion	3q29	LSI TelVysion 3q	3q29 microdeletion/microduplication
Wolf-Hirschhorn Syndrome	4p16.3	LSI WHS/CEP 4	4p16.3 deletion
Cri-du-chat Syndrome	5p15.2	LSI EGR1/D5S23, D5S721	5p15.2 deletion
Sotos Region deletion	5q35	LSI NSD1/D5S23, D5S721	5q35 deletion/duplication
Williams Syndrome	7q11.23	LSI ELN/ D7S486, D7S522	7q11.23 deletion/duplication
Angelman/Prader-Willi Syndrome	15q11-q13	LSI D15S10/CEP 15 (D15Z1)/PML	15q11.2 deletion/duplication
Angelman/Prader-Willi Syndrome	15q11-q13	LSI SNRPN/CEP 15 (D15Z1)/PML	15q11.2 deletion/duplication
Miller-Dieker Syndrome	17p13.3	LSI LIS1/RARA	17p13.3 deletion
Smith-Magenis Syndrome	17p11.2	LSI SMS/RARA	17p11.2 deletion
DiGeorge/VCF Syndrome	22q11.2	LSI TUPLE1 (HIRA)/TelVysion 22q	22q11.2 deletion/duplication
Molar Pregnancy/aneuploidy	13q14,21q22.13-q22.2,Xp11.1-q11.1 Alpha Satellite DNA, Yp11.1-q11.1 Alpha Satelite DNA, 18p11.1-q11.1 Alpha Satellite DNA	AneuVysion	Chromosome 13, 21, 18, X and Y (numerical aberrations)

Lymphoma

Diagnosis	Gene(s)/Unique Sequence	Probe Name	Cytogenetic Aberration Detected
Follicular Lymphoma	t(14;18)(q32;q21)	LSI IGHα-BCL2 LSI BCL2/BCL2 breakapart	IGHα-BCL2 fusions
Mantle Cell Lymphoma	t(11;14)(q13;q32)	LSI CCND1-IGHα LSI CCND1/CCND1 breakapart	CCND1-IGHα fusions
Anaplastic Large Cell Lymphoma	2p23	LSI ALK rearrangements	ALK gene rearrangement
Burkitt Lymphoma	t(8;14)(q24;q32)/t(2;8)/t(8;22)	LSI MYC-IGHα and MYC breakapart	MYC-IGHα fusions
MALT Lymphoma	t(11;18)(q22.2;q21)	LSI API2-MATL1/LSI MALT1	API2-MALT1 fusions
DLBC Lymphoma	3q27	LSI BCL6	BCL6 gene rearrangement
T-cell Lymphoma	14q11.2	LSI TCR A/D	TCR gene rearrangement
Synovial sarcoma	18q11.2	LSI SYT	SYT gene rearrangement (proxy for X;18 translocation)

Solid Tumor

Diagnosis	Gene(s)/Unique Sequence	Probe Name	Cytogenetic Aberration Detected
Breast Cancer/Gastric Cancer	17q11.2-q12	LSI HER-2/neu	HER-2 gene amplification (ERBB2)
Bladder Cancer	chromosome 3, 7, 17 and 9p21	UroVysion	gain of chromosome 3,7,17 and loss of the 9p21
p16/CDKN2A 9p21	9p21	LSI p16/CDKN2A	loss of 9p21
Ewing sarcoma, PNET EWSR1	22q12	LSI EWSR1	EWSR1 gene rearrangement
Alveolar rhabdomyosarcoma FOXO1 (FKHR)	13q14	LSI FOXO1	FOXO1 gene rearrangement
Neuroblastoma	2p24.1	LSI MYCN	MYCN gene amplification
Myxoid/round cell liposarcoma DDIT3(CHOP) 12q13FUS1-16p11.2	12q13, 16p11.2	LSI DDIT3(CHOP) LSI FUS1	DDIT3 gene rearrangement FUS1 gene rearrangement
Malignant Liposarcoma/Angiomatoid fibrous histiocytoma FUS1	16p11.2	LSI FUS1	FUS1 gene rearrangement
Well-differentiated liposarcoma MDM2 amp	12q15	LSI MDM2/CEP2	MDM2 gene amplification
Retinoblastoma, various/RB1	13q14	LSI RB1	gain or loss of RB1 gene
Gliomas	1p36/19q13 centromere 7/7p12-EGFR centromere 10/10q23-PTEN	LSI 1p36/19q13 LSI EGFR LSI PTEN	co-deletion of 1p/19q EGFR gene amplification loss of PTEN
Uveal Melanoma	CEP3 (D3Z1)/6pter-6qter	CEP3/LSI 6pter/6qter	loss of chromosome 3 and 6pter and 6qter duplication

09/01/11