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PITTSBURGH CYTOGENETICS LABORATORY

ONCOLOGY FISH PROBE LIST (Hematological Disorders)

Break-apart Probe List				
Probes	**Locations**	**Cytogenetics Aberrations**	**Genes**	**Disorders**
ALK	2p23	t(2;5)(p23;q35) and variants	ALK	Anaplastic large cell lymphoma
AML1	21q22.12	t(12;21)(p13;q22)	*ETV6/RUNX1*	Precursor B-lymphoblastic leukemia/ALL
		t(8;21)(q22;q22)	*RUNX1T1/RUNX1*	Myeloid, AML FAB M2
BCL2	18q21.3	t(14;18)(q32;q21)	*IGH/BCL2*	Follicular lymphoma
BCL3	19q13.31q13.32	19q13 rearrangement	*BCL3*	Lymphoma
BCL6	3q27	3q27 rearrangement	*BCL6*	Diffuse large B-cell lymphoma
BCL10	1p22	t(1;14)(p22;q32)	*BCL10/IGH*	Lymphoid, MALT lymphoma, Diffuse large B-cell lymphoma
CBFB	16q22	inv(16)(p13q22) or t(16;16)(p13;q22)	*MYH11/CBFβ*	AML, FAB type M4eo
CCND1	11q13	t(11;14)(q13;q32)	*CCND1/IGH*	B-cell malignancies
CDK6	7q21.2	CDK6 gene rearrangement	*CDK6*	Myeloid malignancies.
CSF1R	5q33-34	5q rearrangement	*CSFR1*	Myeloid malignancies
FGFR1	8p11.23	t(8;13)(p11;q12) and variants	*ZMYM2-FGFR1*	myeloproliferative syndrome MPD
IGH	14q32.3	t(14;18)(q32;q21)	*IGH/BCL2*	Follicular lymphoma
		t(8;14)(q24.1;q32) and variants	*CMYC/IGH*	Burkitt lymphoma/leukemia
		t(11;14)(q13;q32)	*CCND1(BCL1)/IGH*	Mantle cell lymphoma
IGK	2p11.2	t(2;3;)(p12;q27)	*IGK/BCL6*	B-cell NHL, mainly diffuse large cell lymphoma
		t(2;8)(p12;q24)	*IGK/MYC*	B-cell ALL and NHL, especially in Burkitt
IGL	22q11	t(3;22)(q27;q11)	*BCL6/IGL*	B-cell NHL, mainly diffuse large cell lymphoma
		t(8;22)(q24;q11)	*MYC/IGL*	B-cell ALL and NHL, especially in Burkitt
IRF4/DUSP22	6p25	6p25 rearrangement	*IRF4/DUSP22*	Follicular Lymphoma or Diffuse Large B Cell Lymphoma or Anaplastic Large Cell Lymphoma
JAK2	9p24.1	9p24.1 rearrangement	*JAK2*	Myeloid and lymphoid malignancies
MALT1	18q21.3	t(11;18)(q21;q21) and variants	*BIRC3/MALT1*	B-cell NHL; MZBCL
MLL	11q23	11q23 rearrangement	*KMT2A*	MDS, AML with 11q23 abnormalities
				ALL, treatment related leukaemias
NUP98	11p15.5	11p15.5 rearrangement	*NUP98*	Myeloid and lymphoid malignancies
NUP214	9q34	9q34 rearrangement	*DEK/NUP214*	Myeloid and lymphoid malignancies
MYC	8q24.21	t(8;14)(q24;q32) and variants	*MYC/IGH*	Burkitt lymphoma/leukemia, other B cell lymphoma
PDGFRB	5q32	t(1;5)(q23;q33) and variants	*PDGFRB*	MDS/MPD/CMPD and AML
RARA	17q21.1-21.2	t(15;17)(q24;21) and variants	*PML/RARA*	APL
TCRB	7q34	t(1;7)(p34;q34) and variants	*LCK/TRB*	T cell lymphoid malignancy
TCRG	7p14	7p14 rearrangement	*TRG*	T cell lymphoid malignancy
TCRAD	14q11.2	14q11.2 rearrangement	*TRAC*	T cell lymphoid malignancy
TCF3	19p13.3	inv(19)(p13q13), t(19;19)(p13;q13) or t(1;19)(q23;p13)	*TCF3/TFPT PBX1/TCF3*	Precusor B cell lymphoblastic leukemia/lymphoma
ETV6	12p13	12p13 rearrangement	*ETV6*	MDS/AML,CML,CCML,B-ALL,T-ALL
TLX1	10q24	10q24 rearrangement	*TLX1*	T cell lymphoid malignancy
TLX3	5q35.1	t(5;14)(q35;q32) or t(5;14)(q35;q11)	*TLX3/BCL11B*	T cell lymphoid malignancy

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Translocation Probes	
Genes	Translocations
BCR/ABL	t(9;22)(q34;q11.2)
BIRC3/MALT1	t(11;18)(q21;q21)
CBFβ	t(16;16)(p13;q22) or inv(16)(p13q22)
ETV6/RUNX1	t(12;21)(p13;q22)
IGH/BCL2	t(14;18)(q32;q21)
IGH/CCND1	t(11;14)(q13;q32)
IGH/FGFR3	t(4;14)(p16;q32)
IGH/MAF	t(14;16)(q32;q23)
IGH/MALT1	t(14;18)(q32;q21)
IGH/MYC	t(8;14)(q24;q32)
PML/RARA	t(15;17)(q22;q21)
RUNX1T1/RUNX1	t(8;21)(q22;q22)
Chronic Lymphocytic Leukemia (CLL) Panel	
Aberration	**Probes**
13q-	*D13S319/LAMP1*
+12	*CEP 12*
11q-	*ATM/11CEP*
17p-	*p53/17CEP*
6q-	*CMYB/6CEP*
IGH rearrangement	*IGH*
*** If *IGH* is positive, we may recommend a few translocation probes involving 14q32**	
t(14;18)(q32;q21)	*IGH/BCL2*
t(11;14)(q13;q32)	*IGH/CCND1*
Multiple Myeloma (MM) Microarray + *IGH* FISH	
FISH testing for 14q32.3 (*IGH*) rearrangement *will be automatically performed with Microarray*	
*** If *IGH* is positive, FISH will be performed for**	
t(11;14)(q13;q32)	*IGH/CCND1*
t(4;14)(p16;q32)	*IGH/FGFR3*
t(14;16)(q32;q23)	*IGH/MAF*
t(6;14)(p21;q32)	*IGH/CCND3*
t(14;20)(q32;q12)	*IGH/MAFB*
Individual FISH testing for Plasma Cell Myeloma (MM)	
Aberration	**Probe(s)**
14q32.3 rearrangement	*IGH*
17p deletion	*TP53/CEP17*
del(13)(q14.3)	*D13S319/LAMP2*
1p del/1q dup	*CDKN2C /CKS1B*
Hyperdiploidy	*D5S23/D5S721,ASS1,CEP7*
8q24.21 rearrangement	*MYC*
*** If *IGH* is positive, FISH will be performed for**	
t(11;14)(q13;q32)	*IGH/CCND1*
t(4;14)(p16;q32)	*IGH/FGFR3*
t(14;16)(q32;q23)	*IGH/MAF*
t(6;14)(p21;q32)	*IGH/CCND3*
t(14;20)(q32;q12)	*IGH/MAFB*

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Myelodysplastic Syndrome (MDS) Panel	
Aberration	**Probes**
5q-	*EGR1* (5q31)
-7	*CEP 7* (7p11.1-q11.1)
7q-	*D7S486* (7q31)
+8	*CEP 8* (8p11.1-q11.1)
20q-	*D20S108* (20q12)

Myeloproliferative Disorder (MPD) Panel	
Aberration	**Probes**
+8	*CEP 8* (8p11.1-q11.1)
+9	*CEP 9* (9p11-q11)
t(9;22)	*BCR/ABL1* (9q34/22q11.2)
20q-	*D20S108* (20q12)

High Grade B-Cell Lymphoma Panel	
Testing for Rearrangements	**Chromosome Bands**
IGH	14q32.3
BCL6	3q27
BCL2	18q21
MYC	8q24
IGH/MYC	t(8;14)(q24;q32)

T-Cell Acute Lymphocytic Leukemia (T-ALL)	
Testing for Rearrangements	**Chromosome Bands**
TRAC	14q11
BCR/ABL1	t(9;22)(q34;q11.2)
TRG	7p14
TRB	7q34
STIL-TAL	1p33
CDKN2A/CEP9	9p21/9cen
MLLT10	10p12.31
If *TRG, TRB* or *TRAC* is positive then FISH will be performed for *TLX3* (5q35.1)	

Acute Lymphocytic Leukemia (ALL)	
Testing for Rearrangements	**Probes or Chromosome Bands**
Trisomies 4,10,17	*CEP4/CEP10/CEP17*
t(9;22)	*BCR/ABL1*
KMT2A	11q23
t(12;21)	*ETV6/RUNX1*
IKZF1 deletion	7p12.2
PAX5	9p13.2
CRLF2	Xp22.33/Yp11.32

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Acute Myelocytic Leukemia (AML)	
Aberration	**Probes**
5q-(5q31)	*EGR1*
monosomy 7	*CEP7/7q31*
t(8;21)	*RUNX1T1/RUNX1*
11q23 rearrangement	*KMT2A*
inv(16)	*CBFB*

Other probes	
1p32p36 deletion/1q21 amplification	*CDKN2C /CKS1B*
9p21 rearrangement	*P16(CDKN2A)(9p21)*

ONCOLOGY FISH PROBE LIST (Solid Tumors)

FISH Probes for Solid Tumors		
Tumors	**Abnormalities**	**Probes**
Alveolar Rhabdomyosarcoma	13q14 Rearrangement	FOXO1
Breast Cancer	*HER2* Amplification	ERBB2/CEP17
Ewing Sarcoma	22q12 Rearrangement	EWSR1
Germ Cell Tumor	Isochromosome 12p	CEP12/12p
Gliomas,	1p/19q co-deletion	1p36 /1q25 and 19q13/19p13
Inflammatory Myofibroblastic Tumors	2p23 Rearrangements	ALK
Low-grade Fibromyxoid Sarcoma	16p11.2 Rearrangement	FUS
Myxoid/Round Cell Liposarcoma	12q13 Rearrangement	CHOP
Neuroblastoma	*N-myc* Amplification	N-myc
Synovial Sarcoma (SS)	18q11.2 Rearrangement	SYT
Uveal Melanoma	Chromosome 3 Monosomy	CEP3
Alveolar Rhabdomyosarcoma	t(2;13)(q35;q14) *(PAX3/FOXO1)*	PAX3
Alveolar Rhabdomyosarcoma; embryonal rhabdomyosarcoma	t(1;13)(p36;q14) *(PAX7/FOXO1)*	PAX7

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