



A-Z Fluorescence *in situ* Hybridization assays

Gene Probe	Common name	Cytogenetic Aberration	Indication	Test Name	Test Code
NEOPLASTIC DISORDERS - Hematological Profiles					
BCR/ABL/ASS, MLL, TEL/AML1	Adult ALL profile	t(9;22)(q34;q11.2) 11q23 rearrangement t(12;21)(p13;q22)	Adult acute lymphoblastic leukemia	Acute Lymphoblastic Leukemia (ALL) Adult Profile	F AALL
BCL6/BCL2/C-MYC	Burkitt profile	3q27 rearrangement t(14;18)(q32;q21) 8q24 translocation	Burkitt Lymphoma, Large B-cell lymphomas	Burkitt/Large B-cell Lymphoma Profile	F BURKITT
13q14, CEP 12, ATM, p53, MYB	CLL profile	del(13)(q14.3), +12, del(11)(q22.3), del(17)(p13.1) del(6)(q23)	Chronic lymphocytic leukemia	Chronic Lymphocytic Leukemia (CLL) Profile	F CLL
EGR1, D7S486, CEP 8, D20S109	MDS profile	-5 /del(5)(q31) -7/del(7)(q13) +8, del(20)(q12)	Myelodysplastic syndrome	Myelodysplastic syndrome Profile	F MDS
BCR/ABL/ASS, CEP 8, 13q14, D20S108	MPD profile	t(9;22)(q34;q11.2) +8, del(13)(q14.3), del(20)(q12)	Myeloproliferative disorder	Myeloproliferative Profile	F MPD
D5S23/D5S721, CEP 9, CEP 15, 13q14, IGH/BCL1, p53	Myeloma profile	+5 , +9 , +15 del(13)(q14.3) IGH/BCL1 t(11;14) del(17)(p13.1),	Myeloma	Myeloma Profile	F MM
D5S23/D5S721, CEP 9, CEP 15, 13q14, IGH/BCL1, p53, IGH/MAF, IGH/FGFR3	Myeloma profile with auto reflex	+5 , +9 , +15 del(13)(q14.3) IGH/BCL1 t(11;14) del(17)(p13.1), t(14;16)(q32;q23) t(4;14)(p16;q32)	Myeloma	Myeloma Profile with autoreflex when IGH is rearranged with a locus other than BCL1	F MM
BCR/ABL/ASS, MLL, TEL/AML1, CEP 4, CEP 10, trisomy 17	Pediatric ALL profile	t(9;22)(q34;q11.2) 11q23 rearrangement t(12;21)(p13;q22) +4, +10,+17	Pediatric acute lymphoblastic leukemia	Acute Lymphoblastic Leukemia (ALL) Pediatric Profile	F PALL



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NEOPLASTIC DISORDERS - Individual Probes					
13q14	13q14.3	del(13)(q14.3)	CLL, NHL, MCL, MM	Deletion 13q14.3	F D1314
ALK	2p23	2p23 rearrangement	ALCL, NSCLC	ALK 2p23 Rearrangement	F ALK
AML1/ETO	t(8;21)	t(8;21)(q22;q22)	AML (M2)	AML1/ETO t(8;21)	F AML ETO
API2/MALT1	t(11;18)	t(11;18)(q22;q21)	NHL, MALT	API2 / MALT1 t(11;18)	F API MALT
ATM	11q22.3	del(11)(q22.3)	CLL	ATM deletion 11q22.3	F ATM
BCL6	3q27	3q27 rearrangement	NHL, DLCL, FCL, CLL, MALT, BL	BCL6 3q27 Rearrangement	F BCL6
BCR/ABL/ASS	t(9;22)	t(9;22)(q34;q11.2)	CML, ALL, AML,LL	BCR/ABL+9q34, t(9;22)	F BCR ABL
CBFB	Inv(16)	t(16;16),inv(16)	AML, AML (M4Eo)	CBFB t(16;16), inv(16)	F CBFB
CEP 3	+3	trisomy 3	MM, MZL, PBCL	trisomy 3	F T3
CEP 4	+4	trisomy 4	Pediatric ALL	trisomy 4	F T4
D20S108	20q-	del(20)(q12)	AML, MDS, MPD	Deletion 20q12	F D20
D5S23/D5S721, CEP 9, CEP 15	+5p/+9/+15	trisomies 5, 9, and 15	MM	trisomy 5, 9 and 15	F T5T9T15
CEP 8	+8	trisomy 8	AML, CML, MDS, BL	trisomy 8	F T8
CEP 10	+10	trisomy 10	Pediatric ALL	trisomy 10	F T10
CEP 12	+12	trisomy 12	CLL	trisomy 12	F T12
CHIC2 / F1P1L1/PDGFRA	4q12	4q12 rearrangement	Hypereosinophilic syndrome	FIP1L1/PDGFRA 4q12 Rearrangement	F 4Q12
D7S486, CEP 7	-7/7q-	-7/del(7)(q31)	AML, MDS	CEP 7 -7/7q31-	F CEP7
EGR1, D5S23/D5S721	5q-	-5/del(5)(q31)	5q- syndrome; MDS, MPD, AML	EGR1 -5/5q-	F EGR1
IGH	14q32	14q32 rearrangement	NHL, MM	IGH 14q32 Rearrangement	F IGH
IGH/BCL2	t(14;18)	t(14;18)(q32;q21)	NHL, DLCL, FCL, MCL, BL	BCL2, IGH/BCL2 t(14;18)	F BCL2
IGH/CCND1 (BCL1)	t(11;14)	t(11;14)(q13;q32)	MCL, MM, MALT	BCL1 IGH/CCND1 t(11;14)	F BCL1
IGH/FGFR3	t(4;14)	t(4;14)(p16;q32)	MM, MG	IGH/FGFR3 t(4;14)	F FGFR3
IGH/MAF	t(14;16)	t(14;16)(q32;q23)	MM, MG	IGH/MAF t(14;16)	F IGH MAF
IGH/MALT1	t(14;18)	t(14;18)(q32;q21)	NHL, MALT	IGH/MALT1 t(14;18)	F IGH MALT



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Gene Probe	Common name	Cytogenetic Aberration	Indication	Test Name	Test Code
IGH/MYC	t(8;14) or MYC	t(8;14)(q32;q24)	NHL, FCL, DLCL, LL, MM	IGH/MYC, CEP 8, t(8; 14)	F IGH MYC
MALT1	18q21	t(11;18)(q21;q21)	NHL, MALT	MALT1 18q21	F MALT
MLL	11q23	11q23	AML, ALL	MLL 11q23	F MLL
MYB	6q23	del(6)(q23)	CLL, B cell SLL, MM	MYB deletion 6	F MYBD6
MYC	8q24	8q24 translocation	ALL, NHL, LL, BL, MM solid tumors: breast, prostate	C MYC 8q24 Rearrangement	F CMYC
p53	p53 or 17p13	del(17)(p13.1)	CLL, B-cell APL, B-cell LPD, MM	p53 deletion 17p13	F P53
PML/RARA	t(15;17)	t(15;17)(q22;q21.1)	APL (AML M3)	PML/RARA t(15,17)	F PML-RARA
SRY/CEP X	SRY region	Yp11.3/Xp11.1q11.1	Post sex-mismatched transplantation	X/Y Post Sex-Mismatch Transplant SRY/CEP X deletion Yp11.3	F SRY
TEL/AML1 aka ETV6/RUNX1	t(12;21)	t(12;21)(p13;q22)	ALL	TEL/AML1 t(12;21)	F TEL AML1

ALL acute lymphoblastic leukemia
 ALCL anaplastic large cell lymphoma
 AML acute myeloid leukemia
 APL acute promyelocytic leukemia
 BL Burkitt lymphoma / leukemia,
 CLL chronic lymphocytic leukemia

CML chronic myelogenous leukemia
 DLCL diffuse large cell lymphoma
 FCL follicular cell lymphoma
 LL lymphoblastic lymphoma
 LPD lymphoproliferative disorder
 MALT MALT lymphoma
 MCL Mantle cell lymphoma

MDS myelodysplastic syndrome
 MG monoclonal gammopathy
 MM multiple myeloma
 MPD myeloproliferative disorder
 MZL marginal zone lymphoma
 NHL non-Hodgkin lymphoma
 PBCL polyclonal B-cell lymphocytosis



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NEOPLASTIC DISORDERS – Solid Tumor

Gene Probe	Common name	Cytogenetic Aberration	Indication	Test Name	Test Code
ALK	2p23	2p23 rearrangement	ALCL, NSCLC	ALK 2p23 Rearrangement	F ALK
1p36/1q25, 19q13/19p13	1p36/19q13	del(1)(p36), del(19)(q13)	Oligodendroglioma	Oligodendroglioma 1p36-/19q13- Glioma	F GLI
CEP 3, CEP 7, CEP 17, p16	UroVysion®	+3, +7, +17, del(9)(p21)	Bladder cancer	UroVysion® : +3/+7/+17/deletion 9p21	F URO
EGFR, CEP 7	7p12	7p12 amplification	Lung, colon, breast	EGFR 7p12 Amplification	F EGFR
EWSR1	22q12	22q12 rearrangement	Ewing sarcoma	EWSR1 22q12 Rearrangement Ewing Sarcoma	F EWSR1
HER2/ <i>neu</i> , CEP 17	HER2/ <i>neu</i>	17q11.2-q12 amplification	Breast cancer	HER2/ <i>neu</i> PathVysion®	F HER2
HER2/ <i>neu</i> , CEP 17	HER2/ <i>neu</i>	17q11.2-q12 amplification	Gastric & GEJC Cancer	HER2 Gastric	F HER2 GA
N-MYC	N-MYC or 2p24	2p24.1 amplification	Neuroblastoma	N-MYC 2p24.1	F NMYC
PTEN, CEP10	10q23	del(10)(q23.3)	Breast cancer, endometrial carcinomas, glioblastoma	PTEN/CEP10 deletion 10q23.3	F PTEN
SYT	18q11	18q11.2 rearrangement	Synovial sarcoma	SYT 18q11.2 Synovial Sarcoma	F SYT



CONGENITAL DISORDERS					
Common name	Gene Probe	Cytogenetic Aberration	Indication	Test Name	Test Code
AneuVysion	13q14, CEP 18, CEP X, CEP Y, 21q22.13-q22.2	+13,+18,+21 ,X, Y	AneuVysion® Prenatal Test	AneuVysion®: +13/+18/+21/X/Y	F ANEU
Angelman	SNRPN, CEP 15, PML	del(15)(q11.2q13)	Angelman syndrome	Angelman syndrome	F AS
Cri-du-Chat	D5S23, D5S721	del(5)(p15.2)	Cri-du-Chat syndrome	Cri-du-Chat Syndrome	F CAT
DiGeorge	N25, ARSA	del(22)(q11.2)	DiGeorge/Velo-Cardio-Facial syndrome	DiGeorge/Velo-Cardio-Facial Syndrome	F VCF
Kallmann	KAL, CEP X	del(X)(p22.3)	Kallmann syndrome	Kallmann Syndrome	F KAL
Lissencephaly	LIS1, RARA	del(17)(p13.3)	Miller-Dieker syndrome / Isolated Lissencephaly	Miller-Dieker Syndrome / Lissencephaly	F LIS
Phelan-McDermid	ARSA, N25	del(22)(q13)	Phelan-McDermid syndrome	Phelan-McDermid Syndrome deletion 22q13	F PMS
Prader-Willi	SNRPN, CEP 15, PML	del(15)(q11.2q13)	Prader-Willi syndrome	Prader-Willi Syndrome	F PWS
Smith-Magenis	SMS, RARA	del(17)(p11.2)	Smith-Magenis syndrome	Smith-Magenis Syndrome	F SMS
X-linked ichthyosis	STS, CEP X	del(X)(p22.32)	Steroid Sulfatase Deficiency	Steroid Sulfatase Deficiency	F STS
SRY region	SRY/CEP X	Yp11.3/Xp11.1q11.1	Sex determination/ sex chromosome aneuploidy	SRY/CEP X	F SRY
Subtelomeres	ToTelVysion	subtelomeric rearrangements, or deletion	Subtelomeric rearrangements, or deletion	ToTelVysion™: Subtelomeres	F SUB
Trisomy 21	AML1	Trisomy 21	Down syndrome / trisomy 21	Trisomy 21	F T21
Williams	ELN, D7S486/D7S522	del(7)(q11.23)	Williams syndrome	Williams Syndrome	F WIL
Wolf-Hirschhorn	WHS, CEP 4	del(4)(p16.3)	Wolf-Hirschhorn syndrome	Wolf-Hirschhorn Syndrome	F WH