MAYO CLINIC

Hematologic Cytogenetics Reference Sheet

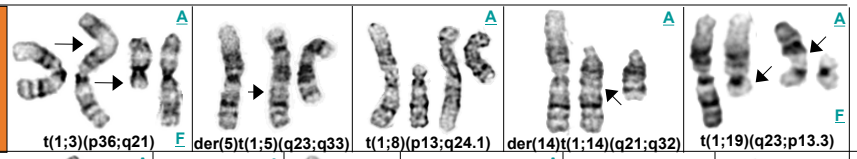
A/Atlas – (Links provided with permission) Atlas of Genetics and Cytogenetics in Oncology and Haematology. Ed. Jean-Loup Huret. University Hospital of Poitiers. 28Nov.2012
http://atlasgeneticsoncology.org

F/FISH- Mayo Medical Laboratories Test Catalog

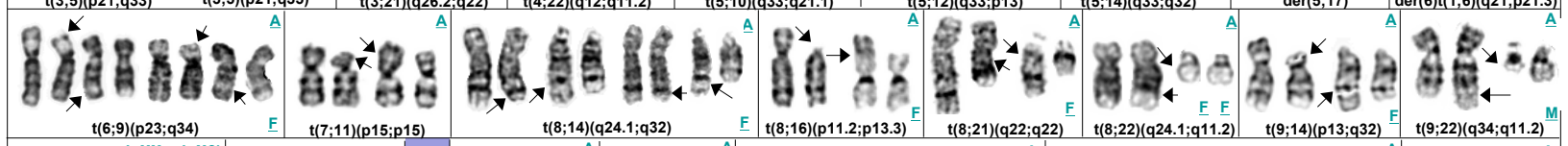
•Abbreviations and disease testing categories

January 2018

trans

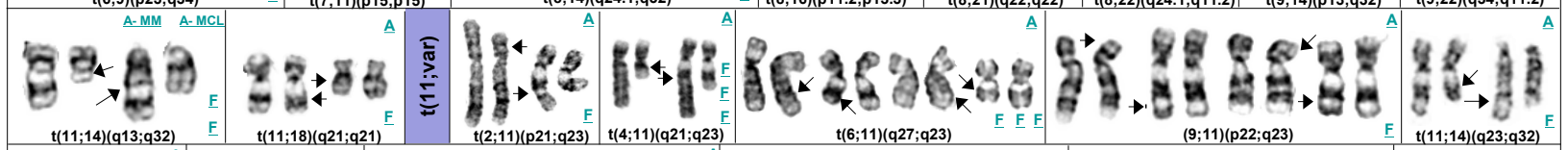


der(1;var)

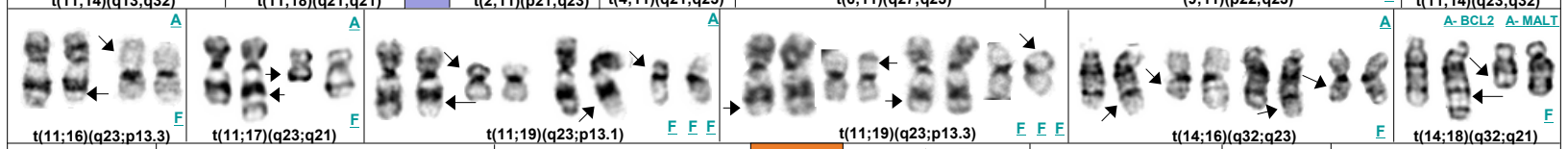


A- MM A- MCL

t(11;var)



A- BCL2 A- MALT



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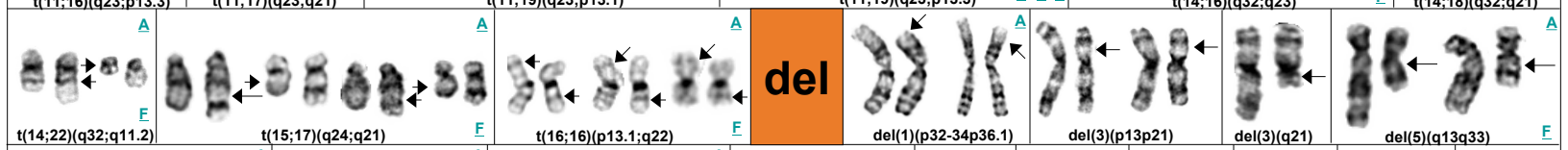

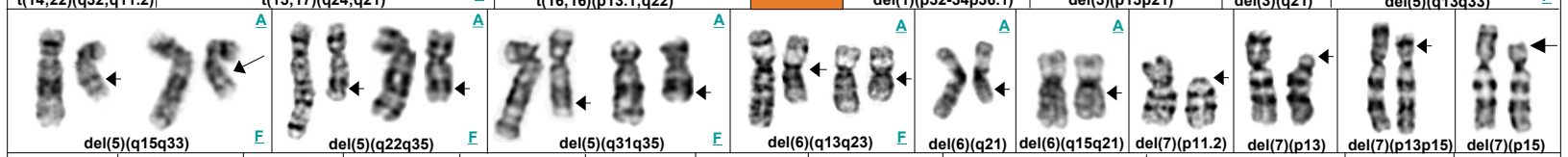

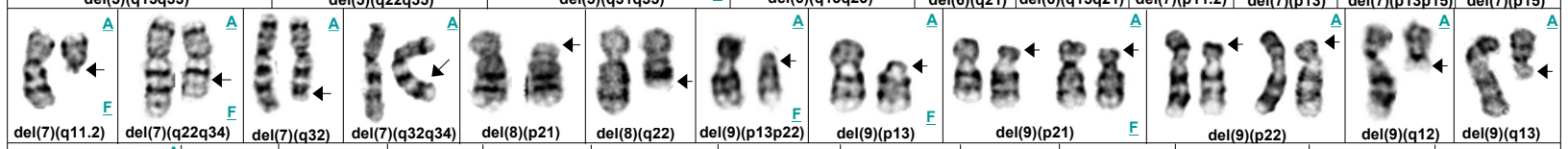

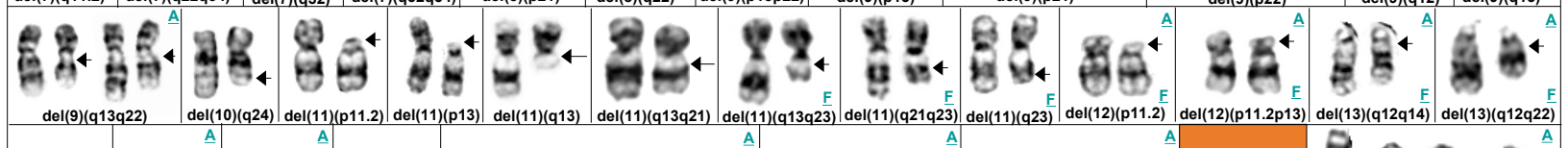

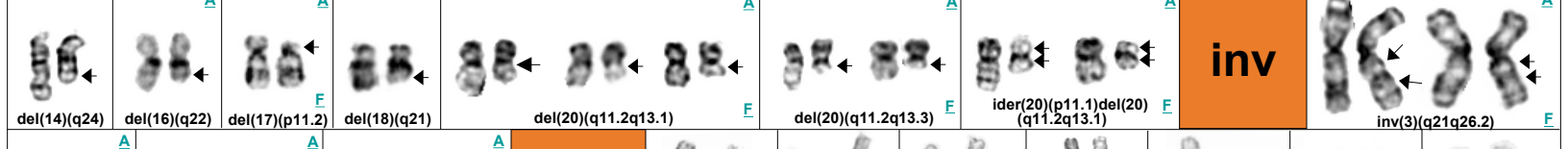

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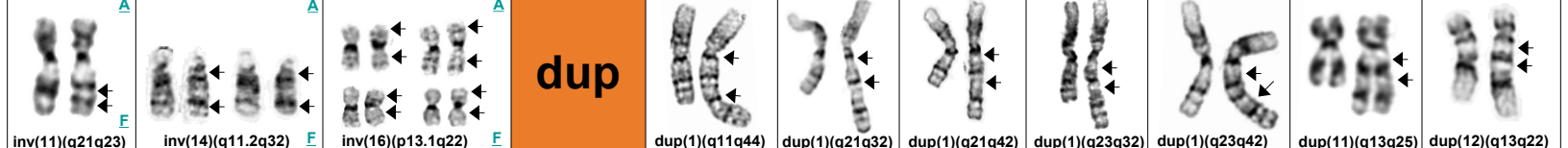

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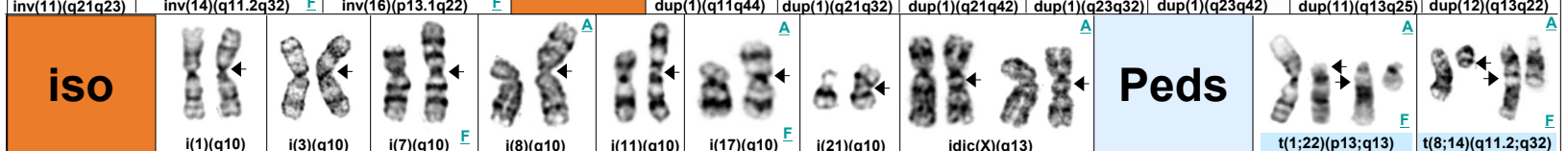
Peds



•*Abbreviations:* B-ALL, B-cell acute lymphoblastic leukemia; AML, acute myeloid leukemia; ALCL, anaplastic large cell lymphoma; ATL, adult T-cell lymphoma; BL, Burkitt lymphoma; CEL, chronic eosinophilic leukemia; CLL, chronic lymphocytic leukemia; CML Ph+, chronic myeloid leukemia with Philadelphia chromosome; CMML, chronic myelomonocytic leukemia; DLBCL, diffuse large cell lymphoma; FL, follicular lymphoma; HCL Hairy Cell leukemia; HSγδTL, hepatosplenic gamma delta T-cell lymphoma; MCL, mantle cell lymphoma; MDS, myelodysplastic syndrome; MM, multiple myeloma; MPD, myeloproliferative disorder; MGUS, monoclonal gammopathy of undetermined significance; MALT, mucosa associated lymphoid tissue; NHL, Non-Hodgkin lymphoma; MZL marginal zone lymphoma; PLL, prolymphocytic leukemia; PMF, primary myelofibrosis; PV, polycythemia vera; RARS, refractory anemia with ringed sideroblasts; SLVL, splenic lymphoma with villous lymphocytes; SMCD-eos, systemic mast cell disease with eosinophilia; T-ALL, T-cell acute lymphoblastic leukemia; t-AML, therapy-related acute myeloid leukemia; t-MDS, therapy-related myelodysplastic syndrome; WM/LPL, Waldenstrom macroglobulinemia / lymphoplasmacytic lymphoma

ANOMALY	ASSOCIATED DISORDERS with FISH test	GENES
TRANSLOCATIONS		
der(1)t(1;1)(p36.1;q21)	NHL	
t(1;3)(p36.1;q21)	NHL	
t(1;3;5)(p36.1;p21;q33)	CEL	WDR48-PDGFRB
t(1;5)(q21;q33)	MPD	TPM3-PDGFRB
t(1;5)(q22;q33)	MPD with eosinophilia, CEL, B-ALL	PDE4DIP-PDGFRB
der(1;7)(q10;p10)	MDS, AML, MPD	
t(1;8)(p13;q24.1)		MYC(C-MYC)
t(1;14)(q21;q32)/ t(1;22)(q21;q11.2)	B-ALL, NHL	BCL9-IGH, IGL-BCL9
t(1;19)(q23;p13.3)	B-ALL	PBX1-TCF3(E2A)
t(2;5)(p23;q35)	ALCL, NHL	ALK-NPM1
t(2;8)(p12;q24.1)	BL, B-ALL, NHL	IGK-MYC(C-MYC)
t(2;11)(p21;q23)	AML, MDS, B-ALL	KMT2A(MLL)
t(2;14)(p15;q32)	CLL, NHL, B-ALL	IGH-BCL11A
t(3;3)(q21;q26.2)	MDS, AML	RPN1-MECOM(EVI1)
t(3;5)(q25;q34)	AML	MLF1-NPM1
t(3;17)(q26.3;q21)	APL	TBL1XR1-RARA
t(3;21)(q26.2;q22)	t-AML, t-MDS	RUNX1(AML1)-MECOM
t(3;22)(q27;q11.2)	NHL	IGL-BCL6
t(4;14)(p16;q32)	MM, cryptic abnormality	FGFR3-IGH
t(4;22)(q12;q11.2)	CML, MPN	PDGFRA -BCR
t(4;5;5)(q23;q31;q33)	chronic basophilic leukemia	PRKG2-PDGFRB
t(5;7)(q33;q11.2)	CMML	HIP1-PDGFRB
t(5;10)(q33;q21)	atypical MPD	CCDC6-PDGFRB
t(5;12)(q31;p13)	CEL, MPD	ETV6(TEL)-ACSL6
t(5;12)(q33;q24)	CMPD with eosinophilia	GIT2-PDGFRB
t(5;12)(q33;p13)	CMML, MDS, MPD	ETV6(TEL)-PDGFRB
t(5;14)(q31;q32)	B-ALL with hypereosinophilia	IGH-IL3
t(5;14)(q33;q24)	MPD, atypical MPD	NIN-PDGFRB
t(5;14)(q33;q32)	CML, MPN, MDS	PDGFRB-CCDC88C
t(5;15)(q33;q22)	CMML atypical MPD	TP53BP1-PDGFRB
dic(5;17)(q11.1;p11.1)	MDS, AML, t-MDS, t-AML	TP53
t(5;17)(q33;p11.2-p13)	JMML, CMML, atypical MPD	RABEP1-PDGFRB
t(5;17)(q35;q21)	APL	NPM1-RARA
der(6)t(1;6)(q21;p21.3)	AML, PMF, PV	
t(6;9)(p23;q34)	MDS, AML	DEK-NUP214(CAN)
t(6;11)(q27;q23)	AML, t-AML, T-ALL	AFDN(MLLT4)-KMT2A(MLL)
t(6;14)(p21.3;q32)	MM	IGH-CCND3
t(7;11)(p15;p15)	AML	NUP98-HOXA9
t(8;var)(p11.2;var)	MPN	FGFR1
t(8;9)(p22;p24)	AML, B-ALL, MDS	PCM1-JAK2
t(8;14)(q24.1;q32)	BL, MM, NHL	MYC(c-MYC)-IGH
t(8;16)(p11.2;p13.3)	AML, t-AML, t-MDS	MYST3(KAT6A)-CREBBP(CBP)
t(8;21)(q22;q22)	AML	RUNX1(AML)-RUNX1T1(ETO)
t(8;22)(q24.1;q11.2)	BL, B-ALL, NHL, MM	MYC(C-MYC)-IGL
add(9)(p24)		JAK2
t(9;11)(p22;q23)	AML, B-ALL	MLLT3(AF9)-KMT2A(MLL)
t(9;14)(p13;q32)	MM, CLL, DLBCL, FL, MCL splenic MZL	PAX5-IGH
t(9;22)(q34;q11.2)	CML (BCR/ABL by PCR) , B-ALL, T-ALL, AML	ABL1(ABL)-BCR
t(11;14)(q13;q32)	MCL, MM	CCND1(BCL1)-IGH
t(11;14)(q23;q32)	t-AML	KMT2A/KIAA1524
t(11;16)(q23;p13.3)	t-MDS, t-AML	KMT2A-CREBBP(CBP)
t(11;17)(q23;q21)	APL	ZBTB16(PLZF)-RARA
t(11;18)(q21;q21.3)	MALT	BIRC3(API2)-MALT1
t(11;19)(q23;p13.1)	AML, B-ALL, T-ALL	KMT2A(MLL)-ELL
t(11;19)(q23;p13.3)	AML, B-ALL, T-ALL	KMT2A(MLL)-MLLT1(ENL)
t(14;14)(q11.2;q32.1)	T-ALL	TRA-TCL1A
t(14;16)(q32;q23)	MM	IGH-MAF
t(14;18)(q32;q21.3)	BL, DLBCL, FL, CLL	IGH-BCL2
t(14;19)(q32;q13.3)	CLL, DLBCL, MCL, MZL, SLVL	IGH-BCL3
t(14;20)(q32;q11.2)	MM	IGH-MAFB
t(14;22)(q32;q11.2)	B-ALL, CLL, HCL, AML, NHL, DLBCL	IGH-IGL
t(15;17)(q24;q21)	APL (PML/RARA by PCR)	PML-RARA
t(16;16)(p13.1;q22)	AML	MYH11-CBFB
t(16;21)(q24;q22)	AML	CBFA2T3-RUNX1-(AML1)
der(Y)t(Y;1)(q12;q12)	MDS	
INVERSIONS		
inv(3)(q21q26.2)	MDS, AML	RPN1-MECOM(EVI1)
inv(11)(q21q23)	MDS, AML, T-ALL	MAML2-KMT2A(MLL)
inv(14)(q11.2q32)	T-PLL	TRAD,TCL1A(TCL1)
inv(16)(p13.1q22)	AML	MYH11-CBFB
DUPLICATIONS		
dup(1)(q-var)	CDR q11q44	
dup(1)(q21q32)	B-ALL, NHL	
dup(11)(q13q25)	NHL	KMT2A(MLL)
dup(12)(q13q22)		
ISOCHROMOSOMES		
i(1)(q10)	NHL	
i(3)(q10)	NHL	
i(7)(q10)	T-cell lymphoma	
i(8)(q10)	T-PLL, AML, B-ALL	
i(11)(q10)	AML, MDS	
i(17)(q10)	MDS, MPD, AML, CML, B-ALL, CLL, Hodgkin and NHL	TP53
i(21)(q10)	MDS, MPD, AML, CML, B-ALL, CLL, Hodgkin and NHL	
idic(X)(q13)	AML, MDS, RARS	Unknown gene within XIST
DELETIONS		
del(1)(p32-34.1p36.1)	AML, MDS, MM, ALL	
del(3)(q21)	B-ALL, AML, NHL	RPN1
del(5q)(var)	AML, MDS, MPD	EGR1
del(6q)(var)	In lymphoid malignancies, WM/LPL, CLL, MM, NK lymphoma, DLBCL, uncommonly myeloid	
del(7)(p-var)	CDR p15	
del(7)(p11.2)	B-ALL, AML, MDS	
del(7)(q11.2)	B-ALL, AML, MDS	
del(7)(q-var)	CDR q22-q34 AML, MDS, NHL	
del(9p)(var)	AML, B-ALL, NHL, pediatric B-ALL	
del(9q)(var)	AML, MDS	
del(11)(p11.2)	NHL	
del(11)(p12p13)	B-ALL	LMO2
del(11q22-q23)	in myeloid AML, MDS, in lymphoid CLL, NHL	KMT2A(MLL) or ATM
add(12)(p)/ del(12)(p11.2p13)	AML, MDS, B-ALL	ETV6(TEL)
del(13)(q12q14)	AML, PMF, MDS, NHL, CLL	
del(13)(q12q22)	AML, PMF, CLL, MM, MDS, NHL	
del(14)(q24)	NHL	
del(16)(q22)	AML, NHL	CBFB
del(17)(p11.2)	B-ALL, AML, CLL, MDS, NHL	TP53(P53)
del(18)(q21)	AML, NHL	
del(20)(q11.2q13.1) del(20)(q11.2q13.3)	AML, MDS, MPD	
ider(20)(p11.1) del(20)(q11.2q13.1-13.3)	AML, MDS, MPD	
del(X)(q24)	NHL	
NUMERIC ANOMALIES		
+3	SLVL, T-cell lymphoma, MZL, MCL, MALT	
+4	AML, t-AML	
+8	B-ALL, AML, MPD, MDS, PV	
+9	AML, MDS, MPD, PV, B-ALL, T-ALL	
+10	AML, B-ALL, MDS	
+11	AML, MDS	KMT2A(MLL)
+12	AML, CLL, NHL, MALT, HCL, SLVL, WM/LPL, FL, MCL and DLBCL	
+13 / +13,+13	AML	
+14	MDS RA, RAEB±T mainly, AML, atypical MPD	
+15	Age related when sole anomaly or with -Y	
+19	AML, MDS	
+21	AML, B-ALL, MDS, MPD	
+22	AML	
-7	MDS, MDS-RS, CMML, J-CML, AML, NHL	
-Y	Age related	
PEDIATRIC (primarily)		
Ph-like ALL loci for 3' kinase		
1q25.2	B-ALL	ABL2
4q12	B-ALL	PDGFRA
5q22.1	B-ALL	TSLP
5q32	B-ALL	CSF1R or PDGFRB
8p21.2	B-ALL	PTK2B
8q12.1	B-ALL	LYN
9p24.1	B-ALL	JAK2
9q34.12	B-ALL	ABL1
10q24.1	B-ALL	BLNK
13q12.2	B-ALL	FLT3
15q25.3	B-ALL	NTRK3
19p13.2	B-ALL	TYK2 or EPOR
22q12.3	B-ALL	IL2RB
t(X/Y;14)	B-ALL, cryptic abnormality	CRLF2-IGH-P2RY8
Pediatric B-ALL		
t(1;19)(q23;p13.3)	B-ALL	PBX1-TCF3(E2A)
del(5)(q32q33)	B-ALL, cryptic abnormality	EBF1-PDGFRB
t(8;14)(q11.2;q32)	B-ALL	CEBPD-IGH
i(9)(q10)/idic(9)(q11)	B-ALL	
dic(9;12)(p11;p11.2)	B-ALL	
dic(9;20)(p11-13;q11.1)	B-ALL	PAX5
t(11:var)(q23;var)	B-ALL	KMT2A(MLL)
	t(4;11), t(9;11), t(6;11), t(10;11), t(11;16), t(11;19), t(11;19)	AFF1, MLLT3, AFDN, MLLT10, CREBBP, ELL, MLLT1
t(12;21)(p13;q22)	B-ALL, cryptic abnormality	ETV6-RUNX1
t(17;19)(q22;p13.3)	B-ALL	TCF3(E2A)-HLF
der(19)t(1;19)(q23;p13.3)	B-ALL	PBX1-TCF3(E2A)
Hyperdiploid	B-ALL	
Pediatric AML		
t(1;11)(q21;q23)	AML	KMT2A(MLL)-MLLT11
t(1;22)(p13;q13)	AML M7	RBM15-MKL1
t(5;11)(q35;p15)	AML	NUP98-NSD1
t(7;12)(q36;p13)	AML	MNX1-ETV6
t(8;21)(q22;q22)	AML	RUNX1T1-RUNX1
del(9)(q34)	B-ALL, AUL	NUP214
t(9;9)(q34;q34)	AML, cryptic abnormality	SET-NUP214
t(10;11)(p12;q23)	AML	MLLT10 (AF10)/ KMT2A(MLL)
t(11;12)(p15;p13)	AML	NUP98-KDM5A
t(11:var)(q23;var)	AML	KMT2A(MLL)
	t(9;11), t(10;11), t(4;11), t(6;11), t(11;16), t(11;19), t(11;19)	MLLT3, MLLT10, AFF1, AFDN, CREBBP, ELL, MLLT1
t(16;21)(p11.2;q22)	AML	FUS-ERG
Pediatric T-ALL		
del(1)(p32p32)	T-ALL, cryptic abnormality	TAL1-STIL
t(5;14)(q35;q32)	T-ALL, cryptic abnormality	TLX3(HOX11L2)-BCL11B
del(9)(q34)	B-ALL, AUL	NUP214
t(10;11)(p13;q14)	T-ALL	MLLT10(AF10)/PICALM
t(10;14)(q24;q11.1)	T-ALL	TLX1-TRAD
t(11;14)(p13;q11.2)	T-ALL	TRAD-LMO2
t(11:var)(q23;var)	T-ALL	KMT2A(MLL)
t(11;19), t(6;11), t(4;11), t(9;11), t(10;11), t(11;16), t(11;19)		MLLT1, AFDN, AFF1, MLLT3, MLLT10, CREBBP, ELL

Blue background = common pediatric anomalies
Dark blue font = links to Mayo Medical Laboratories test catalog
Green font = (links provided with permission) Atlas of Genetics and Cytogenetics in Oncology and Haematology Ed. Jean-Loup Huret. University Hospital of Poitiers. 28Nov.2012 http://atlasgeneticsoncology.org.
CDR = Common Deleted Region

Helpful Mayo Clinic Algorithm links

B-Lymphoblastic Leukemia/Lymphoma Algorithm
Myeloproliferative Neoplasm: A Diagnostic Approach to Bone Marrow Evaluation
Myelodysplastic Syndrome: Guideline to Diagnosis and Follow-up

Abbreviations: **AUL, acute undifferentiated leukemia: AML, acute myeloid leukemia; AMM, Agnogenic myeloid metaplasia; ALCL, anaplastic large cell lymphoma; APL, acute promyelocytic leukemia; ATL, adult T-cell lymphoma; B-ALL, B-cell acute lymphoblastic leukemia; BL, Burkitt lymphoma; CEL, chronic eosinophilic leukemia; CLL, chronic lymphocytic leukemia; CML Ph+, chronic myeloid leukemia with Philadelphia chromosome; CMML, chronic myelomonocytic leukemia; DLBCL, diffuse large B-cell lymphoma; FL, follicular lymphoma; HSγδTL, hepatosplenic gamma delta T-cell lymphoma; LPL, lymphoplasmacytic lymphoma; MALT, mucosa associated lymphoid tissue; MCL, mantle cell lymphoma; MDS, myelodysplastic syndrome; MM, multiple myeloma; MPD, myeloproliferative disorder; MGUS, monoclonal gammopathy of undetermined significance; MZL marginal zone lymphoma; NHL, Non-Hodgkin lymphoma; PLL, prolymphocytic leukemia; PMF, primary myelofibrosis; PV, polycythemia vera; RARS, refractory anemia with ringed sideroblasts; SLVL, splenic lymphoma with villous lymphocytes; SMCD-eos, systemic mast cell disease with eosinophilia; T-ALL, T-cell acute lymphoblastic leukemia; t-AML, therapy-related acute myeloid leukemia; t-MDS, therapy-related myelodysplastic syndrome; WM/LPL, Waldenstrom macroglobulinemia / lymphoplasmacytic lymphoma**