

Clinical Molecular Diagnostic Tests
Human Molecular Genetics Laboratories, UConn Health Center
(updated 04/25/2007)

DNA tests:

Cystic fibrosis carrier screening
CF mutation 3199del6 detection
Connexin 26 (*GJB2*) mutation detection
by sequencing
Methylation PCR for Prader Willi and
Angelman syndromes
Factor V Leiden
Prothrombin 20210G>A (factor II)
MTHFR 677C>T
Tay Sachs mutations
JAK2 mutation in MPD
B-cell clonality
T-cell clonality
BCL2/IGH rearrangement

RNA tests:

BCR/ABL
AML1/ETO
PML/RARA
CBFB/MYH11
MLL/AF4
E2A/PBX1
TEL/AML
Real-time quantitative PCR (Q-PCR) for
BCR/ABL

Oncology FISH tests

METHODS DISEASES	FISH	RT-PCR
CML	t(9;22)(q34;q11.2)	BCR/ABL
AML	t(8;21)(q22;q22) t(15;17)(q22;q12) Inv(16)/t(16;16) 11q23 (MLL) rearrangements	AML1/ETO PML/RARA CBFB/MYH11
ALL	t(9;22)(q34;q11.2) 11q23 rearrangement for t(4;11) +4, +10, +17	BCR/ABL MLL/AF4 E2A/PBX1 [t(1;19)]
MDS/MPD	-5 or del(5q) -7 or del(7q) del(20q) +8	
lymphoma -Burkitt's -Mantle cell -follicular	8q24 (MYC) rearrangement t(11;14)(q13;q32) t(14;18)(q32;q21)	B cell clonality T cell clonality Bcl2/IgH
CLL	Trisomy 12 Rb deletion [-13 or del(13q)] P53 deletion [del(17p)]	
CLL panel Multiple myeloma panel MDS panel	+12, 11q, 13q14, 14q32, 17p13 t(11;14), 11q, 13q14, 17p13 -5/del(5q), -7/del(7q), +8, del(20q)	
Post-bone marrow transpla	X/Y ratio	
Breast Cancer	Her2/neu amplification	