

Fluorescence in situ hybridization (FISH)
Uses

Disease	Cytogenetic Abnormalities/Fluorescence in situ hybridization (FISH) Probe	Significance
CML	t(9;22) (BCR/ABL) (q34;q11): LSI BCR/ABL single fusion probe t(9;22) (BCR/ABL) (q34;q11): LSI BCR/ABL double fusion probe t(9;22) (BCR/ABL) (q34;q11): LSI BCR/ABL extra signal probe Trisomy 8: CEP 8 probe	Diagnosis Prognosis Monitoring of therapy Detection of minimal residual disease Identification of major and minor break points(ALL de novo /on top of CML)
ALL	t(12;21): LSI TEL/AML1 single fusion probe	Favorable Outcome and good therapeutic response (Positive in about 30% of pediatric precursor B-ALL)
	t(9;22) (BCR/ABL) (q34;q11): LSI BCR/ABL single fusion probe t(9;22) (BCR/ABL) (q34;q11): LSI BCR/ABL double fusion probe t(9;22) (BCR/ABL) (q34;q11): LSI BCR/ABL extra signal probe 11q23 rearrangements: LSI MLL break apart rearrangement probe	Poor prognosis
AML	t(8;21): LSI RUNX1/RUNX1T1 double fusion probe t(15;17): LSI PML/RARA single fusion probe inv(16): LSI CBFB break apart rearrangement probe 11q23 rearrangements: LSI MLL break apart rearrangement probe del(7q)/monosomy 7: LSI D7S522 7q31 probe del(9q) : LSI BCR/ABL extra signal probe Trisomy 8: CEP 8 probe	Favorable prognosis, common in AML-M2 Favorable prognosis, common in AML-M3 Favorable prognosis, common in AML-M4 Poor prognosis, common in AML-M5 Poor prognosis Intermediate prognosis

Disease	Cytogenetic Abnormalities/Fluorescence in situ hybridization (FISH) Probe	Significance
NHL	14q32 Rearrangements: LSI IGH break apart rearrangement probe 11q13 rearrangements: LSI CCND1 probe BCL6 gene rearrangement	Clonality analysis in B-NHL Diagnosis of mantle cell lymphoma Most common abnormality in DLBCL
Multiple Myeloma (MM)	del(13q): LSI 13q del p53: LSI 17P13.1 Probe 14q32 Rearrangements: LSI IGH break apart rearrangement probe t(11;14): LSI IGH break apart rearrangement probe, LSI cyclin D1 break apart rearrangement probe	Favorable Outcome and good therapeutic response Poor prognosis
Myelodysplastic syndrome (MDS)	del(5q): LSI del 5q31 probe	Favorable Outcome and good therapeutic response
	Trisomy 8: CEP 8 probe	Intermediate prognosis
	Chromosome 7 aberrations (Monosomy 7/del 7q): LSI 7q31 probe	Poor prognosis
CLL	del p53: LSI 17P13.1 Probe	Poor prognosis
	del(13q): LSI 13q	Good prognosis
	del ATM: LSI 11q22 rearrangements (deletion, translocations/ trisomy 11)	Poor prognosis
	Trisomy 12: LSI Centromeric 12 probe	Poor prognosis

-Fluorescence in situ hybridization (FISH) is also used for assessment of bone marrow engraftment by LSI Y probe

Real Time PCR analysis common mutations in Thrombophilia or hypercoagulability:

- Factor V-Leiden
- Prothrombin-Factor II G20210A
- MTHFR C677T

Real Time PCR analysis for BCR/ABL fusion transcripts