

CG-203 Attachment B: FISH PROBE LIST

Probe	Description	Abnormality Detection	Type of Probe
Centromere Probes	CEP probes consist of highly repeated human α-satellite DNA sequences, usually located at the centromere region of the chromosome.	Chromosome specific aneuploidy (gains/ losses)	Single Color Can be mixed for multi-color assay
All probes listed are manufactured by Abbott Molecular or CytoCell, listed next to probe name.			
List of available aneuploidy tests: All Abbott Molecular probes (Note: For chromosomes not listed below, and appropriate locus specific probe may be useful for detecting gain/loss)			
Chromosome 4 (orange) Chromosome 8 (orange)	Chromosome 10 (green) Chromosome 17 (aqua)	Chromosome X (orange) / Yq12 (green) (For sex-mismatched BMT chimerism)	
Locus Specific or Unique Sequence Probes	LSI probes consist of specific gene sequences mapped to a specific chromosome locus.	Targeted abnormality (translocation, deletion, etc.)	Single or multi-color
CRLF2 (Xp22.33/Yp11.32) CYTOCELL PROBE	The CRLF2 probe consists of a proximal red labeled 243kb portion centromeric (5') to the CRLF2 gene and a distal green labeled telomeric (3') probe consisting of two probes (71kb and 131kb).	CRLF2 gene rearrangements including a cryptic t(X;14) or t(Y;14) translocation	Dual Color Break apart
CRLF2 (Xp22.33/Yp11.32)/ IGH (14q32.33) CYTOCELL PROBE	The CRLF2/IGH fusion probe set consists of a 436kb probe, labeled in red, covering a region within Xp22.22/Yp11.32 including the CRLF2 gene and a 976kb probe, labeled in green, covering 14q32.33 region including the IGH gene.	CRLF2-IGH gene rearrangement/ fusion; t(X;14) or t(Y;14) translocation	Dual Color Dual Fusion
1p36/1q25 Abbott Molecular	The 1p36 / 1q25 Dual Color Probes include a 620 kb green-labeled 1q25 probe and a 400 kb orange-labeled 1p36 probe.	Copy number detection of 1p36 deletions or 1q25 region in neuroblastoma and gliomas	Dual Color
ABL2 (1q25.2) CYTOCELL PROBE	The ABL2 dual color break-apart probe consists of 318 kb portion centromeric/ proximal (3') region labeled in red and 351kb (5') distal/telomeric region labeled in green.	ABL2 gene rearrangements in high risk ALL	Dual Color Break apart
ALK (2p23.2-p23.1) CYTOCELL PROBE	The dual color ALK break-apart rearrangement probe consists of a 486kb probe proximal to the ALK gene, labeled in red (5'), covering the telomeric end of the ALK gene, as well as a 420kb probe distal to the ALK gene, labeled in green (3'), covering the centromeric end of the ALK gene..	The ALK (Anaplastic Lymphoma Kinase) probe is designed to detect the known 2p23 rearrangements that occur in t(2;5) and its variants.	Dual Color Break apart
MYCN (2p24.1) / 2 centromere Abbott Molecular	The MYCN (2p24) green-labeled probe is a 200kb probe that hybridizes to chrom. 2p24 region and contains sequences that flank both 5' and 3' ends of the MYCN gene. The orange-labeled probe hybridizes to alpha satellite sequences on chromosome 2.	MYCN amplification and chromosome 2 copy number in neuroblastoma	Dual Color
RPN1 (3q21.3)/ MECOM (3q26.2) CYTOCELL PROBE	The RPN1/MECOM fusion probe set consists of a 589kb probe, labeled in green, covering a region within 3q21.3 including the RPN1 gene and a 1207kb probe, labeled in red, covering 3q26.2 region including the MECOM gene.	Inversion 3 and t(3;3) in AML	Dual Color Dual Fusion

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D5S23 (5p15) / EGR1 (5q31) Abbott Molecular	The EGR1/D5S23, D5S721 probe set targets 5q31 containing the EGR1 locus. It is a mixture of 200kb orange-labeled EGR1 probe and a 450kb green-labeled LSI D5S23, D5S721 (5p15) probe.	Deletions of 5q31 (5q-) and chromosome 5 (-5) copy number in AML/MDS	Dual Color
PDGFRB (5q32) CYTOCELL PROBE	The PDGFRB probe set consists of a 107kb red-labeled centromeric (3') probe and a 154kb green-labeled telomeric (5') probe.	Rearrangement of PDGFRB with partner gene	Dual Color Break apart
DEK (6p22.3)/ NUP214 (9q34.12-q34.13) CYTOCELL PROBE	The DEK/NUP214 fusion probe set consists of a 387kb probe, labeled in green, covering a region within 6p22.3 including the DEK gene and a 413kb probe, labeled in red, covering 9q34.12-q34.13 region including the NUP214 gene.	Translocation (6;9)(p22.3;q34.13) in AML	Dual Color Dual Fusion
EGFR/CEP 7 (7p11.2-7p12/7p11.1-q11.1) CYTOCELL PROBE	The dual color deletion probe EGFR/CEP 7 consists of an EGFR 295kb probe labeled in red and a centromere 7 probe (7p11.1-q11.1) labeled in green.	Amplification of EGFR seen in solid tumors such as, lung adenocarcinoma, conventional glioblastoma multiform, glioblastoma, breast invasive ductal carcinoma, and colon adenocarcinoma	Dual Color
D7S522 (7q31) / 7 centromere Abbott Molecular	The D7S522/CEP 7 probe set includes a 200 kb orange-labeled D7S522 (7q31) probe and the green-labeled probe hybridizes to alpha satellite sequences on chromosome 7.	Deletions of 7q31 and chromosome 7 copy number in AML/MDS	Dual Color
TCRB (7q34) CYTOCELL	The dual color TCRB break-apart rearrangement probe consists of a 177kb probe proximal to the TCRB gene, labeled in red (5'), covering the centromeric end of the TCRB gene, as well as a 133kb probe distal to the TCRB gene, labeled in green (3'), covering the telomeric end of the TCRB gene.	Rearrangements of TCRB gene	Dual Color Break apart
MYST3 (8p11.21)/ CREBBP (16p13.3) CYTOCELL PROBE	The MYST3/CREBBP fusion probe set consists of a 651kb probe, labeled in green, covering a region within 8p11.21 including the MYST3 gene and a 686kb probe, labeled in red, covering 16p13.3 region including the CREBBP gene.	Translocation (8;16)(p11.21;p13.3) in AML	Dual Color Dual Fusion
PLAG1 (8q12.2) CYTOCELL PROBE	The dual color PLAG1 break-apart rearrangement probe consists of a 399.9kb (118, 170 and 153 kb overlapping) probe proximal to the PLAG1 gene, labeled in red (3'), covering the centromeric end of the PLAG1 gene, as well as a 349.4kb (172 + 170kb) probe distal to the PLAG1 gene, labeled in green (5'), covering the telomeric end of the PLAG1 gene.	Rearrangements of the PLAG1 gene in pleomorphic adenomas and lipoblastomas	Dual Color Break apart
RUNX1 (21q22) / RUNX1T1 (8q24) Abbott Molecular	The RUNX1/RUNX1T1 probe set contains a 1.3 Mb green-labeled probe hybridizing to the 21q22 band containing the RUNX1 gene and a 480 kb orange-labeled probe hybridizing to the 8q22 band containing the RUNX1T1 (ETO) gene.	Translocation (8;21)(q22;q22) in AML-M2	Dual Color Dual Fusion

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MYC (8q24) / IgH (14q32) / CEP 8 Abbott Molecular	The IGH/MYC, CEP 8 Tri-color probe set contains a 1.5 Mb green-labeled probe containing sequences homologous to essentially the entire IGH locus as well as sequences extending about 300kb beyond the 3' end of the IGH locus. An 821 kb orange-labeled CMYC probe extends 385 kb upstream of CMYC and 431 kb 3' beyond CMYC. An aqua labeled probe hybridizes to alpha satellite sequences on chromosome 8.	Translocation (8;14)(q24;q32) in Burkitt's lymphoma with 8 centromere control	Tri-Color Dual Fusion
MYC (8q24.1) Abbott Molecular	The MYC probe set flanks MYC gene consisting of a 260kb orange-labeled probe centromeric (5') to the MYC gene and a 400kb green-labeled probe telomeric (3') to the MYC gene.	Rearrangement of MYC with unknown partner gene and amplification of MYC	Dual Color Break apart
JAK2 (9p24.1) CYTOCELL PROBE	The dual color JAK2 break-apart rearrangement probe consists of a 159kb probe proximal to the JAK2 gene, labeled in red (5'), covering the telomeric end of the JAK2 gene, as well as a 293kb (185 + 108kb) probe distal to the JAK2 gene, labeled in green (3'), covering the centromeric end of the JAK2 gene.	Rearrangements of the JAK2 gene usually seen in myeloproliferative disorders	Dual Color Break apart
PAX5 (9p13.2) CYTOCELL PROBE	The dual color PAX5 break-apart rearrangement probe consists of a 318kb (175+168 kb overlapping) probe proximal to the PAX5 gene, labeled in red (5'), covering the centromeric end of the PAX5 gene, as well as a 292kb (129 + 158kb) probe distal to the PAX5 gene, labeled in green (3'), covering the telomeric end of the PAX5 gene.	Rearrangements of the PAX5 gene in approximately 30% of pediatric patients affected by BCP-ALL	Dual Color Break apart
ABL1 (9q34.11-q34.13) CYTOCELL PROBE	The ABL1 dual color break-apart probe consists of 238 kb portion centromeric/proximal (5') region labeled in red and 390 kb (3') distal/telomeric region labeled in green.	Rearrangements of ABL1 (other than BCR-ABL1 fusion) in high risk ALL	Dual Color Break apart
BCR (22q11.2) / ABL1 (9q34) Abbott Molecular	The BCR/ABL1 probe set consists of a 650kb orange-labeled probe spanning the ABL1 breakpoint. A 1.5 MB green-labeled probe spans the BCR gene breakpoint cluster region.	Translocation (9;22)(q34;q11.2)	Dual Color Dual Fusion
LSI CDKN2A / 9 centromere Abbott Molecular	The CDKN2A/CEP 9 probe set consists of an 190kb orange-labeled probe spanning multiple genetic loci including D9S1749, D9S1747, p16 (INK4A), p14 (ARF), D9S1748, p15 (INK4B), and D9S1752. The green-labeled probe hybridizes to alpha satellite sequences on chromosome 9.	Deletions of 9p21 in ALL and other tumors	Dual Color
NUP98 (11p15.4) CYTOCELL PROBE	The NUP98 probe consists of a 132 kb probe proximal to the NUP98 gene, labeled in red (5'), covering the telomeric end of the STIM1 gene including markers D11S2344 and D11S2347, as well as a 151kb probe distal to the NUP98 gene, labeled in green (3'), covering the ZNF195 gene and the D11S4731 marker.	AML with rearrangement of NUP98 with partner gene	Dual Color Break apart

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KMT2A (MLL) (11q23) Abbott Molecular	The MLL probe set consists of a 350kb green-labeled probe targeting the centromeric (5') half of the MLL gene breakpoint cluster region (bcr) and a 190kb orange-labeled probe targeting the telomeric (3') half of MLL bcr. In approximately 25% of 11q23 translocations, a region beginning at the MLL breakpoint and extending distally is deleted resulting in a deleted orange signal.	Rearrangement of MLL gene Ex: Translocations (4;11), (9;11), (10;11), and (11;19) in ALL/AML	Dual Color Break apart
ETV6 (12p13.2) / RUNX1 (21q22.12) CYTOCELL PROBE	The dual color, dual fusion ETV6-RUNX1 probe consists of an ETV6 (ETO) 348kb probe labeled in red and a 315kb probe labeled in green covering the RUNX1 (AML1) gene.	Translocation (12;21) in ALL, changes in ETV6 and RUNX1 copy numbers or iAMP21 (RUNX1 amplification) in ALL	Dual Color Dual Fusion
ETV6 (12p13) BAP Abbott Molecular	The ETV6 BAP probe set flanks ETV6 gene consisting of a 490kb orange-labeled probe telomeric (5') to the ETV6 gene and a 630kb green-labeled probe centromeric (3') to the ETV6 gene.	Rearrangement of ETV6 with partner gene	Dual Color Break apart
13q14.3 / 13q telomere CYTOCELL PROBE	The LSI D13S319 probe set consists of a 400kb red-labeled probe spanning D13S319 telomeric to D13S262 on 13q14.3. The entire probe lies telomeric to the RB1 gene. A green-labeled probe hybridizes to 13q telomere sequences.	Deletions of 13q14.3 in B-cell neoplasms and chromosome 13 copy number	Dual Color Deletion 13qter control
FOXO1 (13q14) Abbott Molecular	The FOXO1 probe set flanks the FOXO1 gene consisting of a 720kb green-labeled probe lying centromeric (3') to the FOXO1 gene and a 650kb orange-labeled probe telomeric (5') of the FOXO1 gene. 3' amplification may be seen in some cases of A-RMS.	FOXO1 rearrangement (1;13 and 2;13) in alveolar rhabdomyosarcoma	Dual Color Break apart
TCR A/D (14q11) Abbott Molecular	The TCR alpha/delta probe set flanks the TCR constant gene segments and consists of a 660kb orange-labeled flanking probe completely centromeric (5') to TCR and a 710kb green-labeled probe targeting the telomeric (3') TCR variable region.	Rearrangements of TCR A/D gene	Dual Color Break apart
IGH (14q32) CYTOCELL PROBE	The IgH probe set flanks the IgH constant gene segments and consists of a 250kb orange-labeled flanking probe completely centromeric (3') to IgH and a 900kb green-labeled probe targeting the telomeric (5') IgH variable region. As a result of this probe design, any translocation with a breakpoint at the J segments or within switch sequences should produce separate orange and green signals.	Rearrangement of IgH with partner gene other than CMYC	Dual Color Break apart
PML (15q22) / RAR α (17q21) Abbott Molecular	The PML/RAR α probe set consists of a 500kb orange-labeled probe spanning PML gene. A 700kb green-labeled probe spans the RAR α gene.	Translocation (15;17) in AML-M3	Dual Color Dual Fusion
NTRK3 (15q25.3) CYTOCELL PROBE	The dual color NTRK3 break-apart rearrangement probe consists of a 222.3kb probe proximal to the NTRK3 gene, labeled in green (3'), covering the centromeric end of the NTRK3 gene, as well as a 155.5kb probe distal to the NTRK3 gene, labeled in red (5').	Rearrangements of the NTRK3 gene in Fibrosarcoma	Dual Color Break apart

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	covering the telomeric end of the NTRK3 gene.		
FUS (16p11.2) CYTOCELL PROBE	The dual color FUS break-apart rearrangement probe consists of three probes (189kb, 160kb and 96kb) probe proximal to the FUS gene, labeled in red (3'), covering the centromeric end of the FUS gene including markers SHGC-110391 and D16S2933E, as well as a 502kb probe distal to the FUS gene, labeled in green (5'), covering the telomeric end of the FUS gene including markers SHGC-58827 and D16S2660.	Rearrangements of the FUS gene, primary translocation t(16;21)(p11;q22) in Ewing-like sarcomas and in rare cases of AML, results in the fusion of the FUS gene with the ERG gene on 22q12	Dual Color Break apart
GLIS2 (16p13.3) CYTOCELL PROBE	The GLIS2 probe consists of a 232 kb probe proximal to the NUP98 gene, labeled in green (5'), covering the telomeric end of GLIS2, as well as a 347kb probe distal to the GLIS2 gene, labeled in red (3'), covering the centromeric side of GLIS2.	Rearrangements of the GLIS2 gene in AML	Dual Color Break apart
CBFβ-MYH11 inv(16)/ t(16;16) CYTOCELL PROBE	The CBFβ (CBFB) /MYH11 fusion probe set consists of a 617kb probe, labeled in red, covering 16q22 region including the CBFB gene and a 621kb probe, labeled in green, covering a region within 16p13.1 including the MYH11 gene.	Inversion 16 and t(16;16) in AML-M4	Dual Color Dual Fusion
TP53 (17p13.1) / 17 centromere Abbott Molecular	The TP53 probe is a 172kb orange-labeled probe targeting the TP53 gene (previously designated as p53). The probe mix also contains a control probe for the 17 centromere (D17Z1) in green.	TP53 deletion / amplification in a variety of hematological and solid tumor neoplasms	Dual Color
SS18 (18q11.2) (SYT- Synovial Sarcoma) Abbott Molecular	The SS18 probe set flanks the SS18 gene consisting of a 1044kb green-labeled centromeric (3') probe and a 650kb orange-labeled telomeric (5') probe.	Translocation (X;18) in synovial sarcoma	Dual Color Break apart
TCF3 (19p13.3) CYTOCELL PROBE	The TCF3 probe set flanks the E2A gene and consists of a 164kb green-labeled telomeric (3') probe and a 191kb red labeled centromeric (5') probe.	Translocation (1;19) in ALL	Dual Color Break apart
LSI D20S108 (20q12) Abbott Molecular	The D20S108 Probe is a 170kb orange-labeled probe targeting 20q12 region.	Deletions of 20q12 in MDS/AML	Single Color
EWSR1 (22q12) Abbott Molecular	The EWSR1 probe set flanks the EWSR1 gene consisting of a 500kb orange-labeled probe flanking the centromeric (5') side of the EWSR1 gene and 1100 kb green-labeled probe flanking the telomeric (3') side of the EWSR1 gene.	Rearrangement of EWSR1 gene Ex: Translocations (11;22), t(7;11), t(2;11), t(11;17), and other partners	Dual Color Break apart