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
List of available aneuploidy tests: (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**
1p36/1q25	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
ALK (2p23)	The ALK probe contains an orange-labeled 250 kb probe on the telomeric (3') side of the ALK breakpoint and a green-labeled 300kb on the centromeric (5') side.	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
MYC-N (2p24.1) / 2 centromere	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
5p15 / 5q31 (EGR1)	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)	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
7q31 / 7 centromere	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
RUNX1 (21q22) / RUNX1T1 (8q24)	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 (control)

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MYC (8q24) / IgH (14q32) / CEP 8	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 (Control)
CMYC (8q24.1)	The CMYC probe set flanks CMYC gene consisting of a 260kb orange-labeled probe centromeric (5') to the CMYC gene and a 400kb green-labeled probe telomeric (3') to the CMYC gene.	Rearrangement of CMYC with unknown partner gene and amplification of CMYC	Dual Color Break apart
BCR (22q11.2) / ABL1 (9q34)	The BCR/ABL1 probe set consists of a 650kb orange-labeled probe spanning the ABL1 breakpoint. A 1.5mb green-labeled probe spans the BCR gene breakpoint cluster region.	Translocation (9;22)(q34;q11.2)	Dual Color Dual Fusion (Control)
LSI CDKN2A / 9 centromere	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
MLL (11q23)	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) / RUNX1 (21q22)	The ETV6/RUNX1 ES probe set consists of a 350kb green-labeled ETV6 probe entirely telomeric to the ETV6 breakpoint and a 500kb orange-labeled RUNX1 probe which spans the RUNX1 gene. This strategy typically yields a residual RUNX1 signal.	Translocation (12;21) in ALL, changes in ETV6 and RUNX1 copy number in ALL	Dual Color Extra Signal (ES) Fusion (Control)
ETV6 (12p13) BAP	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	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

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FOXO1 (13q14)	The FOX01 probe sets flanks the FOX01 gene consisting of a 720kb green-labeled probe lying centromeric (3') to the FOX01 gene and an 650kb orange-labeled probe telomeric (5') of the FOX01 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)	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)	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) / RARα (17q21)	The PML/RARα probe set consists of a 500kb orange-labeled probe spanning PML gene. A 700kb green-labeled probe spans the RARa gene.	Translocation (15;17) in AML-M3	Dual Color Dual Fusion (Control)
CBFβ (16q22)	The CBFβ probe set flanks inversion 16 breakpoint region and consists of 150kb red labeled centromeric (5') probe and a 170kb green-labeled telomeric (3') probe.	Inversion 16 and t(16;16) in AML-M4	Dual Color Break apart
TP53 (17p13.1) / 17 centromere	The TP53 probe is a 172 kb 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)	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)	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)	The D20S108 Probe is a 170kb orange-labeled probe targeting 20q12 region.	Deletions of 20q12 in MDS/AML	Single Color
EWSR1 (22q12)	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