

**Cancer Cytogenetic Studies**

**Primary specimen containers must be labeled with two patient identifiers**

**Lab location: F buidling 511; T: 212-7464148**

Test Name	Specimen Type	Specimen Collection	Specimen Container	Specimen Preparation	Unacceptable Conditions	Storage/Transport Temperature	Stability (from collection to initiation)	TAT
Chromosome Analysis - Neoplastic (lymphoma)	Lymph node/spleen/mass tissue	3~4 mm <sup>2</sup> lymph node/spleen/mass tissue	Sterile container with sterile RPMI	Collect sample using aseptic/sterile technique.	Frozen specimens Excessive delay in transport, >4 days Fixed specimen	Room temperature or refrigerated	Ambient: 48 hours; Refrigerated: 48 hours	5~28 days
Chromosome Analysis - Neoplastic (Hematological malignancies, leukemia)	Bone Marrow Leukemic Peripheral blood with blasts Peripheral blood for CLL/SLL/MPN	Adult: 1~3mL bone marrow 5~7 mL blood Pediatric: 0.3~0.5mL bone marrow 2 ~ 5 mL blood	Sodium Heparin (green top) tube	Collect sample using aseptic technique. Agitate sample by inverting several times to prevent coagulation.	Clotted specimen Frozen specimens Excessive delay in transport, >4 days Fixed specimens	Room temperature or refrigerated	Ambient: 48 hours, Refrigerated: 48 hours	5~21 days
Fluorescence in situ hybridization (FISH), Interphase - Neoplastic/Hematological Malignancies	Bone marrow aspirate or peripheral blood Fresh lymph node/solid tumor/mass Other specimens such as cerebrospinal fluid (CSF), ascites fluids may be acceptable.	Adults: 2~3mL bone marrow (Minimum: 1mL) 5mL whole blood (Minimum: 2mL) Pediatric: 1~2mL bone marrow (Minimum: 0.5mL) 3~4mL whole blood (Minimum: 1mL) 2~3mm <sup>2</sup> Lymph node/solid tumor pieces	Bone marrow/Peripheral blood: Green top sodium heparin  Lymph node/tumor: sterile container with RPMI	Collect sample using aseptic technique.  For sodium heparin containers, agitate sample by inverting several times to prevent coagulation.	Frozen specimens Clotted specimens	Room tempertaure or refrigerated	Ambient: 48 hours; Refrigerated: 72 hours	3~14 days  NOTE: Time required for testing can vary depending on specimen type and FISH tests ordered.

Fluorescence in situ hybridization (FISH) - HER2/neu Gene Amplification	Tumor tissue		Slides	Formalin fixed (10% neutral buffered formalin) paraffin embedded tissue. Fixative duration: 6-72 hours. Submit 1 H&E and 2 consecutively cut, 4-5 micron thick, mounted on positively charged glass slides	Specimens fixed or processed in alternative fixatives (alcohol, prefer) or heavy metal fixatives (B-4 or B-5) No or insufficient tumor in tissue Decalcified specimens Fixation times less than six hours and more then 72 hours	Room tempertaure	Ambient: indefinitely	5~14 days
Flourescent in situ hybridization (FISH) - Brain Tumors	Brain Tissue		Slides	Formalin fixed (10% neutral buffered formalin) paraffin embedded tumor tissue Provide H&E and unstained slides, consecutively cut, 4-5 micron thick sections, mounted on positively charged glass slides	Specimens fixed or processed in alternative fixatives (alcohol, prefer) or heavy metal fixatives (B-4 or B-5) No or insufficient tumor in tissue Decalcified specimens Fixation times less than six hours and more then 72 hours	Room tempertaure	Ambient: indefinitely	5~14 days
Fluorescence in situ hybridization (FISH) - ALK Gene Rearrangements, Lung Cancer	Tumor tissue		Slides	Formalin fixed (10% neutral buffered formalin) paraffin embedded tumor tissue Provide 1 H&E and 2 unstained, consecutively cut, 4-5-micron thick sections, mounted on positively charged glass slides	Specimens fixed or processed in alternative fixatives (alcohol, prefer) or heavy metal fixatives (B-4 or B-5) No or insufficient tumor in tissue Decalcified specimens Fixation times less than six hours and more then 72 hours	Room temperature	Ambient: indefinitely	5~14 days

**Constitutional Cytogenetics Studies**

**All constitutional studies must be accompanied by a Cytogenetics Consent Form**

**Primary specimen containers must be labeled with two patient identifiers      Lab location: F buidling 511; T: 212-7464148**

Test Name	Specimen Type	Specimen Collection	Specimen Container	Specimen Preparation	Unacceptable Conditions	Storage/Transport Temperature	Stability (from collection to)	TAT
Chromosome Analysis - Peripheral Blood for Constitutional studies	Peripheral blood	Adults: 5~7 mL of blood Percutaneous Umbilical Blood Sampling (PUBS): 1~3 mL Pediatric: 1~3 mL	Sodium Heparin (green top) tube	Collect sample using aseptic technique.  Agitate by inverting the tube several times to prevent clotting.	Excessively clotted specimen Excessive delay in transport, > 5days Frozen specimen	Room temperature or refrigerated	Ambient: 48 hours Refrigerated: 72 hours	5~28 days
Chromosome Analysis - Products of Conception	Products of Conception: Abortus material, placental, fetal and D&C tissue		Sterile container with sterile isotonic saline or sterile culture media	Collect specimen using sterile technique	Visible signs of bacterial contamination Fixed tissue Frozen tissue Lack of villi or fetal parts	Room temperature or refrigerated	Ambient: 48 hours Refrigerated: 72 hours	8~28 days
Fluorescence in situ hybridization (FISH), Peripheral Blood (constitutional)	Peripheral blood	Adults: 5~7 mL of blood Percutaneous Umbilical Blood Sampling (PUBS): 1~3 mL Pediatric: 1~3 mL	Sodium Heparin (green top) tube	Collect sample using aseptic technique.  Agitate by inverting the tube several times to prevent clotting.	Excessively clotted specimen Excessive delay in transport, > 5days Frozen specimen	Room temperature or refrigerated	Ambient: 48 hours, Refrigerated: 72 hours	3~10 days

**FISH Test Menu**  
Cytogenetic Laboratory  
New York Presbyterian/Weill Cornell

Oncology FISH Panels			
Disease Panels	Genes/Loci		FFPE
Acute Myelogenous Leukemia (AML)	PML::RARA, CFBF, RUNX1::RUNX1T1		
Acute Promyelocytic Leukemia (APL)	PML::RARA		
Acute Lymphoblastic Leukemia (adult)	ETV6::RUNX1, CDKN2A, BCR::ABL1, KMT2A (MLL), CRLF2, TCF3(E2A)::PBX1		
Acute Lymphoblastic Leukemia (pediatrics)	ETV6::RUNX1, CDKN2A, BCR::ABL1, KMT2A (MLL), CRLF2, TCF3(E2A)::PBX1, MYC		
Burkitt Lymphoma	MYC, IGH::MYC		
Chronic Lymphocytic Leukemia (CLL)	ATM, CEP12, RB1, TP53, IGH::CCND1, CEP2/N-MYC, MYB		
Chronic myeloid leukemia	BCR::ABL1/ASS1		
DLBCL/High-grade B-cell lymphoma	MYC, IGH::MYC, BCL2, BCL6		√
Multiple Myeloma (MM)	CDKN2C/CKS1B/erg, RB1/13q34, TP53, CEP5/CEP9/CEP15, IGH, MYC		
Multiple Myeloma Reflex Panel (MM)	IGH::FGFR3, IGH::CCND1, IGH::MAF, IGH::MAFB, IGH::CCND3		
Myeloproliferative Neoplasm (MPN)	CDKN2C/CKS1B, CDKN2A/CEP9, BCR::ABL1, RB1, D20S108, CEP8, JAK2		
Eosinophilic Neoplasm	PDGFRA, PDGFRB, FGFR1, JAK2, BCR::ABL1		
Myelodysplastic Syndrome (MDS)	D5S23, D5S721/EGR1, CEP7/D7S486, CEP8, D20S108		
Ph-like B-ALL	CRLF2, JAK2, P2RY8, PDGFRB		
T-Cell Receptor	TRB, TRA/D		
Oncology FISH Tests			
FISH Probes Names	Chromosomes/Loci	Most Commonly Associated Diseases	FFPE
11q23q24	11q23q24	High grade B-cell lymphoma, DLBCL	√
ALK	2p23	Anaplastic large cell lymphoma, lung cancer	√
ANGPTL1	1p36.3	Oligodendroglioma	√
ATM	11q22.3	CLL	
BCL2	18q21.3	B-cell neoplasms	√
BCL6	3q27.3	Lymphoma, DLBCL	√
BCR-ABL1	9q34.1/22q11.2	ALL, AML, CML	
BCR-ABL1/ASS1	9q34.11/22q11.23/9q34.12	CML, ALL, AML	
BIRC3(API2)-MALT1	11q22.2/18q21.32	MALT/marginal zone lymphoma	√
CBFB	16q22.1	AML	
CCND1/IGH	11q13.3/14q32.3	Mantle cell lymphoma	√
CCND1/MYEOV-IGH	11q13.3/14q32.3	Multiple myeloma	
CCND3-IGH	t(6;14), 6p21.1/14q32.3	Multiple myeloma, DLBCL	
CDKN2A/CEP9	9p21.3/Centromere 9	ALL, brain tumors	√
CDKN2C/CKS1B	1p32.3/1q21.3	Multiple myeloma, MPN	
Centromere 3	Centromere 3	Lymphoma	√
Centromere 4	Centromere 4	ALL	√
Centromere 10	Centromere 10	ALL	
Centromere 12	Centromere 12	CLL	
Centromere 18	Centromere 18	Lymphoma	√
CEPX/Yq12	Centromere X/Yq12	Sex mismatched transplant/chimerism	
CRLF2	Xp22.3 and Yp11.3	B-ALL	
D17Z1	Centromere 17	ALL	
D20S108	20q12	MDS	
D5S23, D5S721/CEP 9/CEP15	5p15/Centromere 9/Centromere 15	Multiple myeloma	
D5S23, D5S721/EGR1	5p15.2/5q31.2	AML, MDS	
D7Z1/D7S486	Centromere 7/7q31	AML, MDS	
D8Z2	Centromere 8	AML, MDS	
DEK-NUP214	t(6;9), 6p22.3/9q34.12-q34.13	AML	
DUSP22/IRF4	6q25	Lymphoma	√
EGFR	7p12	Brain tumor	√
ERBB2 (HER2)	17q12	Breast cancer, GI cancer	√
ERG	21q22.2	Multiple myeloma, AML	
ETV6 (TEL)	12p13.2	ALL, AML	
ETV6-RUNX1 (TEL/AML1)	t(12;21), 12p13.2/21q22.12	ALL	
FGFR1	8p11.23	Eosinophilia, MPN	
FGFR3-IGH	t(4;14), 4p16.3/14q32.3	Lymphoma, multiple myeloma	
GLTSC1R	19q13	Oligodendroglioma	√
IGH	14q32.3	B-cell disorders	√
IGH-BCL2	t(14;18), 14q32.3/18q21.3	B-cell disorders	√
IGL	22q11.2	B-cell disorder	√
JAK2	9p24.1	B-ALL, myeloid neoplasms	√
KMT2A (MLL)	11q23.3	ALL, AML, MDS	
MAFB-IGH	t(14;20), 14q32.3/20q12	Multiple myeloma	
MAF-IGH	t(14;16), 14q32.3/16q23.2	Multiple myeloma	√
MALT1-IGH	t(14;18), 14q32.3/18q21.3	MALT/marginal zone lymphoma	√
MECOM (EV11)	3q26.2	AML, MDS	

Oncology FISH Tests			
FISH Probes	Chromosomes/Loci	Disease	FFPE
MYB	6q23	CLL	
MYC	8q24.21	Lymphoma	√
MYC-IGH	8q24.21/14q32.3	Burkitt lymphoma, DLBCL	√
N-MYC/CEP2	2p24.3/Centromere 2	CLL	
P2RY8	Xp22.3 and Yp11.3	B-ALL	
PDGFRA	4q12	Eosinophilia, MPN	
PDGFRB	5q32-q33.1	Eosinophilia, MPN, B-ALL	
PL174	Centromere 11/11q23.3/11q24.3	High grade B-cell lymphoma with 11q aberrations	√
PML-RARA	15q24.1/17q21.2	APL	
RB1	13q14.3	CLL, multiple myeloma, retinoblastoma	
ROS1	6q22.1	Lung cancer	√
RUNX1-RUNX1T1	t(8;21), 8q21.3/21q22	AML	
TCF3(E2A)-PBX1	1q23.3/19p13.3	B-ALL	
TCL1A	14q32.2	PLL	
TP53/D17Z1	17p13.1/Centromere 17	AML, CLL, Multiple myeloma	√
TP63	3q28	Lymphoma	√
TP73	1p36	Oligodendroglioma	√
TRA/D (TCRAD)	14q11.2	T-cell disorders	
TRB (TORB)	7q34	T-cell disorders	√
ZN4F43	19p13	Oligodendroglioma	√
Constitutional FISH Tests			
FISH Probes	Chromosomes/Loci	Diseases/conditions	
1p36/1q25 TELVYSION	1p36/1q25	1p deletion/duplication syndrome	
TUPLE1 (HIRA)	22q11.2	DiGeorge/VCF syndrome	
Centromere 1	Centromere 1	molar pregnancy	√ only
Centromere 11	Centromere 11	molar pregnancy	√ only
Aneuploid Testing			
Prenatal aneuploidy testing	X, Y, 13, 18, 21	Aneuploidy	
Postnatal aneuploidy testing	X, Y, 13, 18, 21	Aneuploidy	
Postnatal gender determination	X, Y, SRY/Yp11.2	Aneuploidy	
POC aneuploidy testing	X, Y, 13, 15, 16, 18, 21, 22	Aneuploidy	