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Target	Disease	Locus	Application
Lymphomas			
BCL2	Diffuse Large B-Cell Lymphoma (DLBCL) Follicular Lymphoma	18q21	Aids in diagnosis and classification of follicular lymphoma (FL), diffuse large B-cell lymphoma (DLBCL), and other aggressive B-cell lymphomas
BCL6	Diffuse Large B-Cell Lymphoma (DLBCL) Follicular Lymphoma	3q27	Aids in diagnosis and classification of mature B-cell lymphomas
IGH	Non-Hodgkin Lymphomas Diffuse Large B-Cell Lymphoma (DLBCL) Follicular Lymphoma	14q32	Immunoglobulin heavy chain (IgH) rearrangement can be identified in about 70% of DLBCL cases, 50% of NHLS and 80% of Follicular Lymphomas.
IGH/BCL2	Diffuse Large B-Cell Lymphoma (DLBCL) Follicular Lymphoma	t(14;18)	Found in 15 to 30% of DLBCL and 50% of Follicular Lymphomas.
IGH/CCND1	Mantle Cell Lymphoma (MCL)	t(11;14)	To confirm the diagnosis of mantle cell lymphoma.
IGH/BCL2	Follicular Lymphoma (FL)	t(14;18)	To provide additional support to the diagnosis of follicular lymphoma.
IGH/MYC	Burkitt's Lymphoma	t(8:14)	To confirm Burkitt's lymphoma.
MYC	Burkitt's Lymphoma/Leukemia	8q24	To confirm Burkitt's lymphoma, including t(8;14), t(2;8) and t(8;22).
MALT1	MALT Lymphoma	t(11;18)	To confirm the diagnosis of MALT lymphoma.
Leukemias			
BCR/ABL	Acute Myeloid Leukemia (AML) AML with BCR/ABL	t(9;22)	Unfavorable prognosis, in acute myeloid leukemia, blastic CML and some ALLs. Detected in about 2-3% of AML patients.
Chromosome 8	AML with Multilineage Dysplasia	cen 8	Intermediate prognosis.
FIP1L1 – CHIC2 – PDGFRA	Eosinophilia	4q12	Deletion of CHIC2 generates FIPIL1-PEGFRA fusion gene, observed in patients with HES, CEL and CMPD.
PDGFRB	Eosinophilia	5q31-q33	PDGFRB rearrangements are seen in myeloproliferative neoplasms with eosinophilia where they can be associated with tyrosine kinase inhibitor responsiveness.
PML/RARA	Acute Promyelocytic Leukemia (APL)	t(15;17)	Favorable prognosis, Diagnostic for acute promyelocytic leukemia. Detected in about 5-8% of AML patients. Treatment with all-trans retinoic acid (ATRA) and other chemotherapy agents can induce remission and terminate disseminated intravascular coagulation (DIC).
FGFR1	Eosinophilia	8p11	Poor prognosis.
Myelodysplastic Syndromes (MDS)			
EGR1		5q31	Associated with favorable prognosis. 5q- is found in 10-15% of MDS
D7S48		7q31	Unfavorable prognosis.
Chromosome 8		cen 8	Intermediate prognosis.
20q12		20q-	Favorable prognosis if it is the sole abnormality. A chromosomal 20q deletion is seen in 5% of primary MDS.
TP53		17p13	Unfavorable prognosis. Deletion is usually found in transformation to AML.

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Myeloproliferative Disorders			
BCR/ABL	Chronic Myeloid Leukemia (CML)	t(9;22)	Diagnostic for CML and also seen in a subset of ALL and AML.
Chromosome 8		cen 8	Associated with accelerated/blast phase of CML.
Solid Tumors			
HER2	Breast Cancer (PathVysion ®) (Vysis)	17q11.2-712	Unfavorable prognosis. Prognostic for amplification/overexpression of HER2. Stratifies patients for Herceptin® therapy.
ALK	Lung Cancer	2p23	Predicts response to targeted therapy in patients with NSCLC with ALK gene rearrangement
ROS1	Lung Cancer, Thyroid Cancer	6q22	Predicts response to targeted therapy in patients with NSCLC with ROS1 gene rearrangement
RET	Lung Cancer	10q11.21	RET translocation aids in the differential diagnosis of atypical thyroid lesions.
c-MET	Kindey Cancer, Lung Cancer	7q31	Diagnostic marker
Chromosome 3	Bladder Cancer (UroVysion ®)	cen 3	Detects aneuploidy for chromosomes 3,7,17 and loss of the 9p21 locus as an aid both in initial diagnosis and for monitoring tumor recurrence.
Chromosome 7	Bladder Cancer (UroVysion ®)	cen 7	Detects aneuploidy for chromosomes 3,7,17 and loss of the 9p21 locus as an aid both in initial diagnosis and for monitoring tumor recurrence.
Chromosome 17	Bladder Cancer (UroVysion ®)	cen 17	Detects aneuploidy for chromosomes 3,7,17 and loss of the 9p21 locus as an aid both in initial diagnosis and for monitoring tumor recurrence.
p16	Bladder Cancer (UroVysion ®)	9p21	Detects aneuploidy for chromosomes 3,7,17 and loss of the 9p21 locus as an aid both in initial diagnosis and for monitoring tumor recurrence.
SS18 (SYT)	Synovial Sarcoma	18q11.2	t(x;18) translocation is very specific and sensitive for the diagnosis of SS
EWSR1	Ewing's Sarcoma	22q12	Translocation of EWSR/Fli1 is observed in over 85% of Ewing's sarcomas
FOXO1 (FKHR)	Alveolar Rhabdomyosarcoma	13q14	To confirm the diagnosis of Alveolar Rhabdomysarcoma
1p36 19q13	Oligodendroglioma	1p36/ 1q25 19q13/ 19p13	Deletion of either (or both) 1p36 and 19p13 supports the diagnosis of oligodendroglioma and is associated with a better prognosis
EGFR	Lung Cancer, Breast Cancer	7p12	EGFR gene status is related to response to anti-EGFR therapy
DNA Ploidy	Multiple tumors, and molar pregnancy	CEP 3 CEP 7	Multiple tumors, and molar pregnancy
Topoisomerase II Alpha	Breast Cancer	17q21-22	To predict the response to adriamycin-based chemotherapy
TMPRSS2/ERG	Prostate Cancer	22q22.3 21q22.3	Poor prognostic indicator in prostate cancer patients with the translocation
ERG	Prostate Cancer	21q22.3	Poor prognosis in post-prostatectomy patients with increased copy numbers
PTEN	Endometrial Cancer, Prostate Cancer, Brain Cancer	10q23	Prognostic marker

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MDM2	Soft tissue Cancer, Bone Cancer, and Brain Cancer	12q15	Aids in the differential diagnosis of lipomatous neoplasms.
Plasma Cell Dyscrasias			
IGH/CCND1	Multiple Myeloma (MM)	t(11;14)	Favorable prognosis compared to other IgH rearrangements. t(11;14) is associated with cyclin D1 overexpression.
IGH	Multiple Myeloma (MM)	14q32	Rearrangements of the IgH gene occur in 34.7% of plasma cell myeloma cases and is among the most frequent chromosomal change.
TP53	Multiple Myeloma (MM)	17p13	Alterations of the TP53 gene in hematological malignancies are generally associated with a poor prognosis
RB1	Multiple Myeloma (MM)	13q34	Deletion of 13q is found in a wide variety of hematologic malignancies, and it has been commonly associated with unfavorable prognosis for Multiple Myeloma patients.
FGFR3/IGH	Multiple Myeloma (MM)	t(4;14)	translocation is found in approximately 10% of myeloma patients.