

PARAFFIN TISSUE FISH TESTS

LYMPHOMA

Anaplastic Large Cell Lymphoma

Rearrangement of ALK

Burkitt Lymphoma

Rearrangement of MYC
IGH@/MYC rearrangement [t(8;14)]

Diffuse Large B Cell Lymphoma / Burkitt Lymphoma

Rearrangement of MYC
IGH@/MYC rearrangement [t(8;14)]
Rearrangement of BCL2
Rearrangement of BCL6

Follicular Lymphoma

Rearrangement of BCL2

General Lymphoma Panel

Rearrangement of IGH@
Rearrangement of MYC
Rearrangement of BCL2
Rearrangement of BCL6
IGH@/MYC rearrangement [t(8;14)]
IGH@/CCND1 rearrangement [t(11;14)]

Mantle Cell Lymphoma

IGH@/CCND1 rearrangement [t(11;14)]

Marginal Zone B Cell Lymphoma

Rearrangement of BCL6
Rearrangement of IGH@
Rearrangement of MALT1
Loss of 7q31
Gain of 3q27
Gain of chromosome 12
Loss of TP53
IGH@/CCND1 rearrangement [t(11;14)]

T Cell Leukemia/Lymphoma

Rearrangement of TRA/D@
Rearrangement of TRB@
Rearrangement of TRG@
Loss of 6q

NEUROLOGICAL MALIGNANCY

High Grade Glioma / Glioblastoma Multiforme

Loss of 1p / 19q
Amplification of EGFR
Loss of PTEN
Loss of TP53

Low Grade Glioma / Oligodendroglioma

Loss of 1p / 19q

Medulloblastoma

Amplification of MYC
Amplification of MYCN
Imbalance of 17q or i(17q)
Loss of 6q

Meningioma

Loss of chromosome 14
Loss of chromosome 22

Neuroblastoma

Amplification of MYCN
Loss of 1p
Loss of 11q

Schwannoma

Loss of chromosome 22

SOFT TISSUE MALIGNANCY

Alveolar Soft Part Sarcoma

Rearrangement of TFE3

Angiomatoid fibrous histiocytoma

Rearrangement of EWSR1
Rearrangement of FUS

Chondromyxoid Fibroma

Loss of 6q

Clear Cell Sarcoma / Melanoma of the Soft Parts

Rearrangement of EWSR1

Desmoplastic Small Round Cell

Rearrangement of EWSR1

Endometrial Sarcoma

Rearrangement of JAZF1

Ewing's Sarcoma / Peripheral Primitive Neuroectodermal Tumour / Askins Tumour

Rearrangement of EWSR1
Rearrangement of FUS

Extraskeletal Chondrosarcoma

Rearrangement of EWSR1

Hibernoma

Loss of 11q

Infantile Fibrosarcoma

Rearrangement of ETV6

Inflammatory Myofibroblastic Tumour

Rearrangement of ALK

Lipoma Not Otherwise Specified

Loss of 13q

Lipoma Spindle Cell

Loss of 13q
Loss of 16q

Liposarcoma Dedifferentiated

Amplification of MDM2

Liposarcoma Myxoid / Round Cell

Rearrangement of DDIT3
Rearrangement of EWSR1
Rearrangement of FUS

Liposarcoma Well-Differentiated

Amplification of MDM2

Low Grade Fibromyxoid Sarcoma

Rearrangement of FUS

Rhabdoid Tumour

Loss of 22q11

Rhabdomyosarcoma Alveolar

Rearrangement of FOXO1

Rhabdomyosarcoma Embryonal

Gain of chromosome 2
Gain of chromosome 8
Gain of chromosome 20

Synovial Sarcoma

Rearrangement of SS18

URINARY SYSTEM MALIGNANCY

Clear Cell Carcinoma

Loss of chromosome 3

Congenital Mesoblastic Nephroma

Rearrangement of ETV6

Renal Cell Chromophobe / Oncocytoma

Loss of 3p
Loss of chromosome 13
Loss of chromosome 17
Loss of chromosome 21

Renal Translocation Carcinoma

Rearrangement of TFE3

Renal Papillary Cell Carcinoma

Gain of chromosome 7
Gain of chromosome 17

Renal Transitional Cell Carcinoma

Gain of chromosome 3
Gain of chromosome 7
Gain of chromosome 9
Gain of chromosome 17

OTHER

Breast Cancer

Amplification of ERBB2 (HER2)
Amplification of TOP2A

Breast Cancer Secretory

Rearrangement of ETV6

Cervical - Squamous Cell Carcinoma

Gain of TERC

Chimerism Determination

XX,XY

Choroidal Melanoma

Loss of 1p
Loss of chromosome 3
Imbalance of 6p or i(6q)
Gain of 8q
Loss of 10q

Gastric Cancer

Amplification of ERBB2 (HER2)
Amplification of EGFR

Gastrointestinal Tumour (GIST)

Loss of chromosome 14
Loss of chromosome 22

Germ Cell Tumour

Imbalance of 12p or i(12p)

Lung Cancer

Amplification of EGFR
Amplification of ERBB2 [HER2]
Rearrangement of ALK

Lung Cancer - Pleuropulmonary Blastoma

Gain of chromosome 2
Gain of chromosome 8
Gain of chromosome 10
Gain of chromosome 20

Lung Cancer - Mesothelioma

Loss of 9p
Loss of 6q

Ploidy status - Hydropic Abortus

Aneuploidy X,Y,13,18,21
Aneuploidy 15,16,22

Ploidy status - Products of Conception

Aneuploidy X,Y,13,18,21

Melanoma

Imbalance of 6q or i(6q)
Gain of 11q

HAEMATOLOGICAL FISH TESTS

MYELOID DISORDERS

Acute Myeloid Leukaemia (AML)

RUNX1T1/RUNX1 rearrangement [t(8;21)]
PML/RARA rearrangement [t(15;17)]
Rearrangement of CBFβ [t(16;16)] or inv(16)
Rearrangement of MLL (11q23)
Rearrangement of MECOM (3q26)
Monosomy 5 or loss of 5q31
Monosomy 7 or loss of 7q31

Secondary AML

Rearrangement of MLL (11q23)
Monosomy 5 or loss of 5q31
Monosomy 7 or loss of 7q31

Myeloproliferative Disease

BCR/ABL1 rearrangement [t(9;22)]
Trisomy 8
Trisomy 9
Loss of 13q14
Loss of 20q12

Hypereosinophilic Syndrome

Rearrangement of PDGFRA (4q12)
Rearrangement of PDGFRB (5q33)
Rearrangement of FGFR1 (8p12)

Myelodysplastic Syndrome

Monosomy 5 or loss of 5q31
Monosomy 7 or loss of 7q31
Trisomy 8
Loss of 9
Loss of 20

CML

BCR/ABL1 rearrangement [t(9;22)]
Loss of 17p/i(17q)
Trisomy 8
Trisomy 9

Lymphoid Disorders

Acute Lymphoid Leukaemia (ALL) - Childhood

Rearrangement of MLL (11q23)
RUNX1/ETV6 rearrangement [t(12;21)]
TCF3/PBX1 rearrangement [t(1;19)]
Hyperdiploidy
iamp (21)

ALL Adult

BCR/ABL1 rearrangement [t(9;22)]
Hyperdiploidy/near haploidy
Rearrangement of MLL (11q23)
TCF3/PBX1 rearrangement [t(1;19)]

Chronic Lymphocytic Leukaemia or B-Lymphoproliferative Disease unspecified

Loss of ATM
Loss of TP53
Loss of 13q14
Trisomy 12
IGH@/CCND1 rearrangement [t(11;14)] (exclude mantle cell)

B-Prolymphocytic leukaemia (B-PLL)

Loss of ATM
Loss of TP53
Loss of 13q14
Trisomy 12
Rearrangement of IGH@

Adult T-cell Leukaemia / Lymphoma (ATLL)

Rearrangement of TRA/D@
Rearrangement of TRB@
Rearrangement of TRG@

T-cell Prolymphocytic Leukaemia (T-PLL)

Rearrangement of TRA/D@
Rearrangement of TRB@
Rearrangement of TRG@

Lymphoma

(Confirmed infiltrated bone marrow aspirate)

NHL unspecified

Rearrangement of IGH@

Follicular Lymphoma

Rearrangement of BCL2
IGH@/MYC rearrangement [t(8;14)] (if transforming)

Mantle Cell Lymphoma

IGH@/CCND1 rearrangement [t(11;14)]

Anaplastic Large Cell Lymphoma

Rearrangement of ALK

Burkitt Lymphoma

Rearrangement of MYC
IGH@/MYC rearrangement [t(8;14)]

T Cell Lymphoma

Rearrangement of TRA/D@
Rearrangement of TRB@
Rearrangement of TRG@

Diffuse Large B-cell Lymphoma

IGH@/MYC rearrangement [t(8;14)]
Rearrangement of BCL2
Rearrangement of BCL6

MALT (Mucosa Associated Lymphoid Tissue)

Rearrangement of MALT1

Splenic marginal zone lymphoma

Monosomy 7 or loss of 7q31
Rearrangement of BCL6
Loss of TP53

Lymphoplasmacytic lymphoma

Rearrangement of IGH@

MYELOMA AND RELATED DISORDERS

Myeloma / Monoclonal Gammopathy of Undetermined Significance / Plasma Cell Leukaemia

IGH@/FGFR3 rearrangement [t(4;14)]
IGH@/MAF rearrangement [t(14;16)]
IGH@/CCND1 rearrangement [t(11;14)]
Loss of 13q14
Loss of TP53
Hypodiploidy / Hyperdiploidy

TRANSPLANT

Pre Bone Marrow Transplant screen

Monosomy 5 or loss of 5q31
Monosomy 7 or loss of 7q31
Loss of RB1

Post Bone Marrow Transplant (Chimerism studies)

Donor vs recipient (sex mismatch)
Donor vs recipient - allo or auto
(previous cytogenetic abnormality detectable by FISH)

PRENATAL FISH TESTS

Aneuscreen

Aneuploidy X, Y, 13, 18, 21

Loss of Pregnancy Screen

Aneuploidy X, Y, 13, 18, 21
Aneuploidy 15, 16, 22

POSTNATAL FISH TESTS

22q13.3 Deletion Syndrome

Loss of ARSA (22q13.3)

Cri-du-chat Syndrome

Loss of 5p

Di George, Velo-cardio-facial, CATCH22 Syndrome

Loss of HIRA (22q11)

Prader-Willi / Angelman Syndrome

Loss of SNRPN (15q11)

Smith-Magenis Syndrome

Loss of FLII (17p11)

Miller-Dieker Syndrome

Loss of PAFAH1B1 (17p13)

Retinoblastoma

Loss of RB1

Sex determination

Loss of SRY

Williams-Buren Syndrome / Supra Valvula Aortic Stenosis

Loss of ELN (7q11)

CONVENTIONAL G-BANDED ANALYSIS

Cancer Cytogenetics

Bone Marrow
Peripheral Blood*
Tumour tissue

Constitutional Cytogenetics

Peripheral Blood

Prenatal Cytogenetics

Chorionic Villus
Amniotic Fluid
Products of Conception

* Peripheral blood for cancer cytogenetics is only recommended in specific disease types. Please consult directly with the laboratory.