

Molecular diagnostic tests available for bone and soft tissue tumours at Rizzoli Institute

Tumor	Molecular alteration	Validated tissue	Molecular method	FISH probes
ALK -Rearranged Sarcoma	ALK rearranged	FFPET		ALK
Alveolar Rhabdomyosarcoma	t(1;13)PAX7-FKHR t(2;13)PAX3-FKHR	frozen/FFPET frozen/FFPET	qRT-PCR qRT-PCR	FOXO1
Alveolar Soft Part Sarcoma	t(X;17)ASPL-TFE3	frozen/FFPET	qRT-PCR	TFE3
Aneurysmal bone cyst	t(16;17)CDH11-USP6 t(17;17)COL1A1-USP6	FFPET FFPET		USP6 USP6, COL1A1
Angiomatoid Fibrous Histiocytoma	t(2;22) EWS-CREB1 t(12;16) FUS-ATF1 t(12;22) EWS-ATF1	FFPET FFPET FFPET		EWSR1, FUS
BCOR ITD	BCOR rearranged	frozen/FFPET	RT-PCR + Sanger	
CIC-Rearranged Sarcoma	t(4;19) e t(10;14)CIC-DUX4	FFPET	NGS	CIC
Clear Cell Sarcoma	t(12;22)EWS-ATF t(2;22)EWS-CREB1	frozen/FFPET frozen/FFPET	qRT-PCR qRT-PCR	EWSR1
Chondroblastoma	H3F3B variant	frozen/FFPET	RT-PCR + Sanger	
Chondrosarcoma	IDH1 and IDH2 variant	frozen/FFPET	RT-PCR + Sanger	
Dermatofibrosarcoma Protuberans	t(17;22) COL1A1-PDGFB	FFPET		COL1A1, PDGFB
Desmoplastic Small Round Cell Tumor	t(11;22) EWS-WT1	frozen/FFPET	qRT-PCR	EWSR1, WT1
Desmoid fibromatosis	CTNNB1 variant	frozen/FFPET	RT-PCR + Sanger	n.a.
Epithelioid Haemangiioendothelioma	t(1;3)WWWTR1-CAMTA1 t(X;11) YAP1-TFE3	FFPET FFPET		WWTR1 TFE3
Ewing sarcoma	t(11;22) EWSR1-FLI1 t(21;22) EWSR1-ERG	frozen/FFPET frozen/FFPET	qRT-PCR qRT-PCR	EWSR1, FUS, ERG
Extraskelatal Mixoid Chondrosarcoma	t(9;22)EWS-NR4A3 t(9;17)TAF2N-NR4A3 t(9;15)TCF12-NR4A3	FFPET FFPET FFPET		NR4A3
Fibrous dysplasia	GNAS variant	frozen/FFPET	RT-PCR + Sanger	n.a.
GLI1-Rearranged Sarcoma	GLI1 rearranged	FFPET		GLI1
Giant Cell Tumor of Bone	H3F3A variant	frozen/FFPET	RT-PCR + Sanger	n.a.
Infantile Sarcoma	t(12;15) ETV6-NTRK3	frozen/FFPET	qRT-PCR	ETV6, NTRK3
Intimal Sarcoma	MDM2 amplification	FFPET	n.a.	MDM2
Low-Grade Central Osteosarcoma	MDM2 amplification	FFPET	n.a.	MDM2
Low Grade Fibromyxoid Sarcoma- Sclerosing Epitelioid Fibrosarcoma	t(7;16)FUS-CREB3L2 t(11;16)FUS-CREB3L1 t(7;22)EWS-CREB3L2 t(11;22)EWS-CREB3L1	FFPET FFPET FFPET FFPET		EWSR1, FUS
Malignant Peripheral Nerve Sheath Tumor (MPNST)	MDM2 amplification	n.a.	n.a.	MDM2
Mesenchimal Chondrosarcoma	t(8;8)HEY1-NCOA2	frozen/FFPET	qRT-PCR	NCOA2
Mixoid Round Cell Liposarcoma	t(12;16) FUS-DDIT3 t(12;22)EWS-DDIT3	FFPET FFPET		DDIT3, FUS, EWSR1
Mioepitelioma	t(1;22) EWS-PBX1 t(19;22) EWS-ZNF444 t(6;22) EWS-POU5F1	FFPET FFPET FFPET		EWSR1
NTRK-rearranged sarcoma	NTRKs rearranged	FFPET		NTRK1,2,3
Parosteal Osteosarcoma	MDM2 amplification	n.a.	n.a.	MDM2
Round cell sarcoma with EWSR1-non-ETS fusions / Undifferentiated sarcoma	FUSION PLEX SARCOMA PANEL	frozen/ffpet	NGS	EWSR1, FUS
Sarcoma with BCOR-Genetic Alteration	t(X;X)BCOR-CCNB3	frozen/ffpet	qRT-PCR	BCOR
Sclerosing Rhabdomyosarcoma	MYOD1 variant	frozen/FFPET	RT-PCR + Sanger	
Synovial Sarcoma	t(X;18) SS18-SSX	frozen/ffpet	qRT-PCR	SS18
Solitary Fibrous Tumor	inv(12)(q13q13) NAB2/STAT6	frozen/ffpet	qRT-PCR	n.a.
Well/Dedifferentiated Liposarcoma	MDM2 amplification	n.a.	n.a.	MDM2

Legend: RT-PCR, reverse-transcriptase Polimerase Chain Reaction; qRT-PCR, quantitative RT-PCR; FISH, fluorescence in situ hybridisation; FFPET, formalin-fixed paraffin-embedded tissue; n.a, not available; NGS, Next Generation Sequencing.