

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

Tumor	Molecular alteration	Molecular method	Validated tissue	FISH probes
Alveolar Rhabdomyosarcoma	t(1;13)PAX7-FKHR	qRT-PCR	frozen/FFPET	FOXO1
	t(2;13)PAX3-FKHR	qRT-PCR	frozen/FFPET	FOXO1
Alveolar Soft Part Sarcoma	t(X;17)ASPL-TFE3	qRT-PCR	frozen/FFPET	TFE3
Aneurysmal bone cyst	t(16;17)CDH11-USP6	RT-PCR qRT-PCR	frozen/FFPET	USP6
	t(17;17)COL1A1-USP6	qRT-PCR	frozen	COL1A1
Angiomatoid Fibrous Histiocytoma	t(2;22) EWS-CREB1	RT-PCR	frozen/FFPET	EWSR1
	t(12;16) FUS-ATF1	n.a.	n.a.	FUS
	t(12;22) EWS-ATF1	qRT-PCR	frozen/FFPET	EWSR1
CIC-Rearranged Sarcoma	t(4;19) e t(10;14)CIC-DUX4	qRT-PCR	frozen/FFPET	CIC
Clear Cell Sarcoma	t(12;22)EWS-ATF	RT-PCR qRT-PCR	frozen/FFPET	EWSR1
	t(2;22)EWS-CREB1	RT-PCR	frozen/FFPET	EWSR1
Chondroblastoma	H3F3B variant	RT-PCR + Sanger	frozen/FFPET	n.a.
Chondrosarcoma	IDH1 and IDH2 variant	RT-PCR + Sanger	frozen/FFPET	n.a.
Dermatofibrosarcoma Protuberans	t(17;22) COL1A1-PDGFB	qRT-PCR	frozen	COL1A1 / PDGFB
Desmoplastic Small Round Cell Tumor	t(11;22) EWS-WT1	RT-PCR qRT-PCR	frozen/FFPET	EWSR1 / WT1
Epithelioid Haemangioendothelioma	t(1;3)WWWTR1-CAMTA1	RT-PCR qRT-PCR	frozen/FFPET	WWWTR1
	t(X;11) YAP1-TFE3	RT-PCR	frozen	TFE3
Epithelioid Haemangioma	t(19;19) ZFP36-FOSB	qRT-PCR	frozen/FFPET	n.a.
	t(1;14) FOS-LMNA	n.a.	n.a.	FOS
Ewing sarcoma	t(11;22) EWSR1-FLI1	RT-PCR qRT-PCR	frozen/FFPET	EWSR1
	t(21;22) EWSR1-ERG	RT-PCR qRT-PCR	frozen/FFPET	EWSR1 / ERG
	t(17;22) EWSR1-E1AF	RT-PCR	frozen	EWSR1
	t(7;22) EWSR1-ETV1	RT-PCR	frozen/FFPET	EWSR1
	t(17;22) EWSR1-ETV4	RT-PCR	frozen/FFPET	EWSR1
	t(2;22) EWSR1-FEV	RT-PCR	frozen	EWSR1
	t(2;22) EWSR1-FEV	RT-PCR	frozen/FFPET	EWSR1
	t(16;21) FUS-ERG	RT-PCR qRT-PCR	frozen/FFPET	FUS / ERG
Extraskelatal Mixoid Chondrosarcoma	t(9;22)EWS-NR4A3	RT-PCR qRT-PCR	frozen/FFPET	NR4A3
	t(9;17)TAF2N-NR4A3	qRT-PCR	frozen/FFPET	NR4A3
	t(9;15)TCF12-NR4A3	qRT-PCR	frozen/FFPET	NR4A3
Fibrous dysplasia	GNAS variant	RT-PCR + Sanger	frozen/FFPET	n.a.
Giant Cell Tumor of Bone	H3F3A variant	RT-PCR + Sanger	frozen/FFPET	n.a.
Infantile Sarcoma	t(12;15) ETV6-NTRK3	qRT-PCR	frozen/FFPET	ETV6
Intimal Sarcoma	MDM2 amplification	n.a.	FFPET	MDM2
Low-Grade Central Osteosarcoma	MDM2 amplification	n.a.	FFPET	MDM2
Low Grade Fibromyxoid Sarcoma / Sclerosing Epithelioid Fibrosarcoma	t(7;16)FUS-CREB3L2	RT-PCR	frozen	FUS
	t(11;16)FUS-CREB3L1	RT-PCR	frozen	FUS
	t(7;22)EWS-CREB3L2	RT-PCR	frozen	EWSR1
	t(11;22)EWS-CREB3L1	RT-PCR	frozen	EWSR1
Malignant Peripheral Nerve Sheath Tumor (MPNST)	MDM2 amplification	n.a.	n.a.	MDM2
Mesenchimal Chondrosarcoma	t(8;8)HEY1-NCOA2	RT-PCR	frozen/FFPET	NCOA2
Mixoid Round Cell Liposarcoma	t(12;16) FUS-DDIT3	RT-PCR qRT-PCR	frozen/ffpet	DDIT3 / FUS
	t(12;22)EWS-DDIT3	RT-PCR qRT-PCR	frozen/ffpet	DDIT3 / EWSR1
Mioepitelioma	t(1;22) EWS-PBX1	RT-PCR	frozen	EWSR1
	t(19;22) EWS-ZNF444	RT-PCR	frozen	EWSR1
	t(6;22) EWSR1-POU5F1	RT-PCR	frozen	EWSR1
Parosteal Osteosarcoma	MDM2 amplification	n.a.	n.a.	MDM2
Pseudomiogenico Haemangioendothelioma	t(7;19)Serpine1-FOSB	RT-PCR	frozen	n.a.
Round cell sarcoma with EWSR1-non-ETS fusions / Undifferentiated sarcoma	t(1;22) EWSR1-PATZ1	RT-PCR	frozen/ffpet	EWSR1
	t(4;22) EWSR1-SMARCA5	RT-PCR	frozen	EWSR1
	t(2;22) EWSR1-SP3	RT-PCR	frozen	EWSR1
	EWSR1-NFATC2	RT-PCR	frozen/ffpet	EWSR1
	FUS-NFATC2	n.a.	n.a.	FUS
Sarcoma with BCOR-Genetic Alteration	t(X;X)BCOR-CCNB3	qRT-PCR	frozen/ffpet	BCOR
Synovial Sarcoma	t(X;18) SS18-SSX	qRT-PCR	frozen/ffpet	SS18
Solitary Fibrous Tumor	inv(12)(q13q13) NAB2/STAT6	RT-PCR	frozen/ffpet	n.a.
Well/Dedifferentiated Liposarcoma	MDM2 amplification	n.a.	n.a.	MDM2

**Legend:** RT-PCR, reverse-transcriptase Polymerase Chain Reaction; qRT-PCR, quantitative RT-PCR; FISH, fluorescence in situ hybridisation; FFPET, formalin-fixed paraffin-embedded tissue; n.a., not applicable.