

Sarcoma Diagnosis and Classification

Order GTC Solid Tumor Expression/Fusion Profile

GTC Solid Tumor Expression/Fusion Profiling provides highly informative and actionable data for patients with sarcoma.

Sarcomas are heterogeneous group of cancer that arises in the connective tissue. Diagnosis and subclassification of sarcoma can be very difficult due to overlapping in histological and immunohistological features. Determining the molecular abnormalities, especially fusion genes and chromosomal translocations in a specific sarcoma can be very helpful in diagnosis, subclassification and determining therapy. Sarcoma is a rare cancer type, but it is relatively more common in pediatric patients. In general, the presence of a specific translocation in one sarcoma type does not occur in any other types of sarcoma, thus it is very helpful to test for the driver abnormality in subclassification of sarcomas. Using NGS is the most comprehensive and cost-effective approach to determine the driver molecular abnormality in a cancer expected to be a sarcoma. Our NGS assay analyzes the most common genes involved in translocations reported in sarcoma and covers all partner genes in a fashion similar to break apart probes in FISH testing. Below is a list of the most common translocations in sarcoma. Additional translocations can also be detected with our assay. In addition, mutations in various genes can also be detected and will be reported.

Tumor Type	Translocation	Gene(s)
Aneurysmal bone cyst	t(16;17)(q22;p13)	CDH11-USP6
	t(1;17)(p34.3;p13)	THRAP3-USP6
	t(3;17)(q21;p13)	CNBP-USP6
	t(9;17)(q22;p13)	OMD-USP6
	t(17;17)(q21;p13)	COL1A1-USP6
Angiofibroma	t(5;8)(p15;q13)	AHRR-NCOA2
Angiomatoid fibrous histiocytoma	t(12;22)(q13;q12)	EWSR1-ATF1
	t(2;22)(q33;q12)	EWSR1-CREB1
	t(12;16)(q13;p11)	FUS-ATF1
Alveolar rhabdomyosarcoma	t(2;13)(q35;q14)	PAX3-FKHR
	t(2;2)(q35;p23)	PAX3-NCOA1
	t(X;2)(q35;q13)	PAX3-AFX
	t(1;13)(p36;q14)	PAX7-FKHR
Alveolar soft-part sarcoma	der(17)t(X;17)(p11;q25)	ASPSR1-TFE3
Chondroid lipoma	t(11;16)(q13;p13)	C11orf95-MKL2
Clear-cell sarcoma	t(12;22)(q13;q12)	EWSR1-ATF1
	t(2;22)(q33;q12)	EWSR1-CREB1
Congenital/infantile fibrosarcoma	t(12;15)(p13;q25)	ETV6-NTRK3
Dermatofibrosarcoma protuberans/giant cell fibroblastoma	t(17;22)(q21;q13)	COL1A1-PDGFB

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Desmoplastic small round-cell tumor	t(11;22)(p13;q12)	EWSR1-WT1
Endometrial stromal sarcoma	t(7;17)(p15;q11)	JAZF1-JJAZ1(SUZ12)
	t(6;7)(p21;p15)	JAZF1-PHF1
	t(6;10)(p21;p11)	EPC-PHF1
	t(t(10;17)(q22;p13)	YWHAE-FAM22A/B
	t(1;6)(p32~34;p21)	MEAF6-PHF1
Epithelioid hemangioendothelioma	t(1;3)(p36;q25)	WWTR1-CAMTA1
	t(X;11)(p11.2;q22.1)	TFE3-YAP1
Ewing sarcoma; Primitive neuroectodermal tumor (PNET)	t(11;12)(q24;q12)	EWSR1-FLI1
	t(21;22)(q22;q12)	EWSR1-ERG
	t(7;22)(p22;q12)	EWSR1-ETV1
	t(2;22)(q33;q12)	EWSR1-FEV
	t(17;22)(q12;q12)	EWSR1-E1AF
	inv(22)(q21;12)	EWSR1-ZSG
	t(16;21)(p11;q22)	FUS-ERG
Ewing-like bone sarcoma	inv(X)(p11.4p11.22)	BCOR-CCNB3
	t(20;22)(q13;q12)	EWSR1-NFATC2
Extraskeletal myxoid chondrosarcoma	t(9;22)(q22;q12)	EWSR1-NR4A3
	t(9;17)(q22;q11)	TAF15-NR4A3
	t(9;15)(q22;q21)	TCF12-NR4A3
	t(3;9)(q11;q22)	TFG-NR4A3
Giant cell fibroblastoma	t(17;22)(q21;q13)	COL1A1-PDGFB
Inflammatory myofibroblastic tumor	t(1;2)(q22;p23)	TPM3-ALK
	t(2;19)(p23;p13)	TPM4-ALK
	t(2;17)(p23;q23)	CLTC-ALK
	t(2;2)(p23;q13)	RANBP2-ALK
	t(2;11)(p23;p15)	CARS-ALK
	inv(2)(p23;q35)	ATIC-ALK
	t(2;4)(p23;q21)	SEC31A-ALK
Lipoblastoma	t(7;8)(p22;q13)	COL1A2-PLAG1
Lipoma	t(3;12)(q27-28;q14-q15)	HMGA2-LPP
		LPP-HMGA2
Low-grade fibromyxoid sarcoma	t(7;16)(q33;p11)	FUS-CREB3L2
	t(11;16)(p11;p11)	FUS-CREB3L1
Meningeal hemangiopericytoma	inv(12)(q13q13)	NAB2-STAT6

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Mesenchymal chondrosarcoma	t(8;8)(q13;q21)	HEY1-NCOA2
Myoepithelial tumor of	t(6;22)(p21;q12)	EWSR1-POU5F1
soft tissue and bone	t(19;22)(q13;q12)	EWSR1-ZNF444
	t(1;22)(q23;q12)	EWSR1-PBX1
	t with 16p11	FUS
Myxoid/round-cell liposarcoma	t(12;16)(q13;p11)	FUS-DDIT3
	t(12;22)(q13;q12)	EWSR1-DDIT3
Nodular fasciitis	t(17;22)(p13;q13)	MYH9-USP6
Pericytoma	t(7;12)(p22;q13)	ACTB-GLI1
Pulmonary myxoid sarcoma	t(2;22)(q34;q12)	EWSR1-CREB1
Sclerosing epithelioid	t(7;16)(p22;q24)	FUS-CREB3L2
fibrosarcoma	t(11;16)(p13;p11.2)	FUS-CREB3L1
Solitary fibrous tumor	inv(12)(q13q13)	NAB2-STAT6
Spindle cell rhabdomyosarcoma	t(6;8)(p21;q13)	SRF-NCOA2
	t(8;11)(q13;p15)	TEAD-NCOA2
Synovial sarcoma	t(X;18)(p11;q11)	SS18-SSX1
		SS18-SSX2
		SS18-SSX4
BCOR-rearranged sarcoma	inv(X)(p11.4p11.22)	BCOR-CCNB3
		BCOR-MAML3
		ZC3H7B-BCOR
CIC-rearranged sarcoma (Undifferentiated small round cell/Blue cell tumor)	t(4;19)(q35;q13) or t(10;19)(q26;q13)	CIC-DUX4
	t(X;19)(q13;q13.3)	CIC-FOXO4

Specimen Requirements:

-FFPE: 1 H&E slide and 8-10 unstained slides, 5-7 microns of tissue fixed with 10% NBF fixative.

Shipping:

Ship using cold pack. Ship with overnight delivery.

Turn Around Time:

7-10 days

