

Molecular In My Pocket™ ...

ONCOLOGY: Pediatric Soft Tissue Tumors

Prepared by the Association for Molecular Pathology Training and Education Committee

Tissue Type	Tumor Type	Gene/Biomarker	Alteration	Significance	Primary Assays
Adipocytic	Lipoblastoma	<i>PLAG1, HMGA2</i>	fusion	diagnosis	FISH, NGS (DNA or RNA)
Myofibroblastic	Alveolar Rhabdomyosarcoma	<i>PAX3/7-FOXO1</i>	fusion	diagnosis, prognosis	FISH, NGS (DNA or RNA)
	Embryonal Rhabdomyosarcoma	<i>KRAS, NRAS, HRAS, FGFR4</i>	activating sequence variants	diagnosis	NGS (DNA)
	Spindle Cell Rhabdomyosarcoma	<i>MYOD1</i>	sequence variant	diagnosis, prognosis	NGS (DNA)
	Spindle Cell Rhabdomyosarcoma	<i>VGLL2, NCOA2</i>	fusions	diagnosis, prognosis	NGS (DNA or RNA)
	Solitary Fibrous Tumor	<i>NAB2-STAT6</i>	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Sclerosing Epithelioid Fibrosarcoma	<i>EWSR1-CREB3L1</i>	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Infantile Myofibroma	<i>PDGFRB</i>	sequence variant	diagnosis, treatment	NGS (DNA)
	Inflammatory Myofibroblastic Tumor	<i>ALK, RET, ROS1</i>	fusions	diagnosis, prognosis	IHC, FISH, NGS (DNA or RNA)
	Infantile Fibrosarcoma	<i>ETV6-NTRK3, other NTRK fusions</i>	fusion	diagnosis, treatment	RT-PCR, FISH, NGS
	Nodular Fasciitis, Cranial Fasciitis	<i>USP6</i>	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Desmoid-Type Fibromatosis	<i>CTNNB1 (sporadic), APC (germline)</i>	sequence variant	diagnosis	NGS (DNA)
	Dermatofibrosarcoma Protuberans / Giant Cell Fibroblastoma	<i>COL1A1-PDGFB</i>	fusion	diagnosis	FISH, NGS
	Vascular	Pseudomyogenic Hemangioendothelioma	<i>FOSB</i>	fusion	diagnosis
Epithelioid Hemangioendothelioma		<i>WWTR1-CAMTA1, YAP1-TFE3</i>	fusion	diagnosis	FISH, RT-PCR, NGS (DNA or RNA)
Bone	Aneurysmal Bone Cyst	<i>USP6</i>	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Osteosarcoma	<i>TP53</i>	fusion, sequence variant, deletion	diagnosis	NGS (DNA)
	Giant Cell Tumor Of Bone	<i>H3F3A G34W, G34L</i>	sequence variant	diagnosis	NGS (DNA)
Cartilage	Mesenchymal Chondrosarcoma	<i>HEY1-NCOA2</i>	fusion	diagnosis	NGS (RNA), FISH
	Chondroblastoma	<i>H3F3A K36M</i>	sequence variant	diagnosis	NGS (DNA)
Miscellaneous	Ewing Sarcoma	<i>EWSR1</i>	fusions	diagnosis	FISH, NGS (DNA or RNA)
	Ewing-Like Sarcoma	<i>CIC-DUX4, BCOR-CCNB3</i>	fusions	diagnosis	NGS (DNA or RNA)
	Synovial Sarcoma	<i>SS18, SS18L1</i>	fusions	diagnosis	FISH, NGS (DNA or RNA)
	Desmoplastic Small Round Cell Tumor	<i>EWSR1-WT1</i>	fusion	diagnosis	RT-PCR, FISH, NGS
	Alveolar Soft Part Sarcoma	<i>ASPSR1-TFE3</i>	fusion	diagnosis	RT-PCR, FISH, NGS
	Angiomatoid Fibrous Histiocytoma	<i>ATF1/CREB1-FUS/EWSR1</i>	fusion	diagnosis	RT-PCR, FISH, NGS (RNA)

Miscellaneous (Continued...)	NUT Midline Carcinoma	<i>BRD3-NUTM1, BRD4-NUTM1</i>	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Chordoma (Poorly Differentiated)	<i>SMARCB1 (INI1)</i>	loss (sequence variant, partial deletion)	diagnosis, prognosis	IHC, CMA, NGS, MLPA
	Rhabdoid (Extra-Renal)	<i>SMARCB1 (INI1) and SMARCA4 (BRG1)</i>	loss (sequence variant, partial deletion)	diagnosis	IHC, CMA, NGS, MLPA
	Epithelioid Sarcoma	<i>SMARCB1 (INI1) and SMARCA4 (BRG1)</i>	loss (sequence variant, partial deletion)	diagnosis, treatment	IHC, CMA, NGS, MLPA
Multi-system	DICER1 syndrome	<i>DICER1</i>	loss-of-function sequence variant or deletion + hotspot sequence variant	diagnosis, familial cancer risk	NGS (DNA)
	CMMRD	<i>MLH1, MSH2, PMS2, MSH6</i>	loss-of-function sequence variant or deletion	diagnosis, familial cancer risk, treatment	IHC, NGS (DNA), MSI
	Gorlin	<i>SUFU, PTCH1</i>	loss-of-function sequence variants, deletion/duplication	diagnosis, familial cancer risk	sequencing, CMA, MLPA
	MEN 2A/2B	<i>RET</i>	gain-of-function sequence variants	diagnosis, familial cancer risk	sequencing
	Li Fraumeni Syndrome	<i>TP53</i>	loss-of-function sequence variant or deletion	diagnosis, familial cancer risk	NGS (DNA)

Abbreviations:

CGH Comparative Genomic Hybridization

FISH Fluorescent *in situ* Hybridization

MSI Microsatellite Instability

RT-PCR Reverse Transcription PCR

CMA Chromosomal Microarray Analysis

IHC Immunohistochemistry

NGS Next Generation Sequencing

ddPCR Digital Droplet PCR

MLPA Multiplex Ligation-dependent Probe Amplification

PCR Polymerase Chain Reaction



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