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	*PLAG1, HMGA2*	fusion	diagnosis	FISH, NGS (DNA or RNA)
Myofibroblastic	Alveolar Rhabdomyosarcoma	*PAX3/7-FOXO1*	Fusion	diagnosis, prognosis	FISH, NGS (DNA or RNA)
	Embryonal Rhabdomyosarcoma	*KRAS, NRAS, HRAS, FGFR4*	activating sequence variants	diagnosis	NGS (DNA)
	Spindle Cell Rhabdomyosarcoma	*MYOD1*	sequence variant	diagnosis, prognosis	NGS (DNA)
	Spindle Cell Rhabdomyosarcoma	*VGLL2, NCOA2*	Fusions	diagnosis, prognosis	NGS (DNA or RNA)
	Solitary Fibrous Tumor	*NAB2-STAT6*	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Sclerosing Epithelioid Fibrosarcoma	*EWSR1-CREB3L1*	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Infantile Myofibroma	*PDGFRB*	sequence variant	diagnosis, treatment	NGS (DNA)
	Inflammatory Myofibroblastic Tumor	*ALK, RET, ROS1*	fusions	diagnosis, prognosis	IHC, FISH, NGS (DNA or RNA)
	Infantile Fibrosarcoma	*ETV6-NTRK3, other NTRK fusions*	fusion	diagnosis, treatment	RT-PCR, FISH, NGS
	Nodular Fasciitis, Cranial Fasciitis	*USP6*	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Desmoid-Type Fibromatosis	*CTNNB1 (sporadic), APC (germline)*	sequence variant	diagnosis	NGS (DNA)
	Dermatofibrosarcoma Protuberans / Giant Cell Fibroblastoma	*COL1A1-PDGFB*	fusion	diagnosis	FISH, NGS
Vascular	Pseudomyogenic Hemangioendothelioma	*FOSB*	fusion	diagnosis	NGS (DNA or RNA)
	Epithelioid Hemangioendothelioma	*WWTR1-CAMTA1, YAP1-TFE3*	fusion	diagnosis	FISH, RT-PCR, NGS (DNA or RNA)
Bone	Aneurysmal Bone Cyst	*USP6*	fusion	diagnosis	FISH, NGS (DNA or RNA)
	Osteosarcoma	*TP53*	fusion, sequence variant, deletion	diagnosis	NGS (DNA)
	Giant Cell Tumor Of Bone	*H3F3A G34W, G34L*	sequence variant	diagnosis	NGS (DNA)
Cartilage	Mesenchymal Chondrosarcoma	*HEY1-NCOA2*	fusion	diagnosis	NGS (RNA), FISH
	Chondroblastoma	*H3F3A K36M*	sequence variant	diagnosis	NGS (DNA)
Miscellaneous	Ewing Sarcoma	*EWSR1*	fusions	diagnosis	FISH, NGS (DNA or RNA)
	Ewing-Like Sarcoma	*CIC-DUX4, BCOR-CCNB3*	fusions	diagnosis	NGS (DNA or RNA)
	Synovial Sarcoma	*SS18, SS18L1*	fusions	diagnosis	FISH, NGS (DNA or RNA)
	Desmoplastic Small Round Cell Tumor	*EWSR1-WT1*	fusion	diagnosis	RT-PCR, FISH, NGS
	Alveolar Soft Part Sarcoma	*ASPSCR1-TFE3*	fusion	diagnosis	RT-PCR, FISH, NGS
	Angiomatoid Fibrous Histiocytoma	*ATF1/CREB1-FUS/EWSR1*	fusion	diagnosis	RT-PCR, FISH, NGS (RNA)

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