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* (rare)	fusion	diagnosis	IHC (PLAG1), FISH, NGS (DNA, RNA)
Skeletal Muscle	Alveolar Rhabdomyosarcoma	*PAX3::FOXO1*, *PAX7::FOXO1*	fusion	diagnosis, prognosis	FISH, RT-PCR, NGS (DNA, RNA)
	Embryonal Rhabdomyosarcoma	*HRAS, KRAS, NRAS; FGFR4; NF1*	activating sequence variant	diagnosis	NGS (DNA)
	Spindle Cell / Sclerosing Rhabdomyosarcoma	*MYOD1*	sequence variant (p.L122R)	diagnosis, prognosis	NGS (DNA), ddPCR
		VGLL2, NCOA2	fusion	diagnosis, prognosis	NGS (DNA, RNA)
Myofibroblastic	Dermatofibrosarcoma Protuberans / Giant Cell Fibroblastoma	*COL1A1::PDGFB*	fusion	diagnosis	FISH, NGS (DNA, RNA)
	Desmoid-Type Fibromatosis	*CTNNB1* (sporadic) *APC* (germline)	sequence variant	diagnosis, familial cancer risk	NGS (DNA)
	Infantile Fibrosarcoma	*ETV6::NTRK3*, other *NTRK* fusions	fusion	diagnosis, treatment	FISH, RT-PCR, NGS (DNA, RNA)
	Inflammatory Myofibroblastic Tumor	*ALK, NTRK3, ROS1*	fusion	diagnosis, prognosis, treatment	IHC (ALK), FISH, NGS (DNA, RNA)
	Low-grade Fibromyxoid Sarcoma / Sclerosing Epithelioid Fibrosarcoma	*FUS::CREB3L2* *EWSR1::CREB3L1*	fusion	diagnosis	FISH, NGS (DNA, RNA)
	Nodular Fasciitis, Cranial Fasciitis	*USP6*	fusion	diagnosis	FISH, NGS (DNA, RNA)
Pericytic	Infantile Myofibroma / Myofibromatosis	*PDGFRB*	sequence variant	diagnosis, treatment	NGS (DNA)
Vascular	Pseudomyogenic Hemangioendothelioma	*FOSB*	fusion	diagnosis	NGS (DNA, RNA)
	Epithelioid Hemangioendothelioma	*WWTR1::CAMTA1, YAP1::TFE3*	fusion	diagnosis	FISH, RT-PCR, NGS (DNA, RNA)
Bone	Aneurysmal Bone Cyst	*USP6*	fusion	diagnosis	FISH, NGS (DNA, RNA)
	Giant Cell Tumor of Bone	*H3-3A (H3F3A)* p.G35W (p.G34W), p.G35L (p.G34L)	sequence variant	diagnosis	IHC (H3.3 G34W), NGS (DNA)
	Osteosarcoma	*TP53*	fusion, sequence variant, deletion	diagnosis	NGS (DNA, RNA)
Cartilage	Chondroblastoma	*H3-3B (H3F3B)* p.K37M (p.K36M)	sequence variant	diagnosis	IHC (H3.3 K36M), NGS (DNA)
	Mesenchymal Chondrosarcoma	*HEY1::NCOA2*	fusion	diagnosis	FISH, NGS (DNA, RNA)
Miscellaneous	Ewing Sarcoma	*EWSR1::FLI1*, *EWSR1::ERG*, other FET-ETS fusions	fusion	diagnosis	RT-PCR, FISH, NGS (DNA, RNA)
	CIC-Rearranged Sarcoma	*CIC::DUX4*, other *CIC* fusions	fusion	diagnosis	FISH, NGS (DNA, RNA)
	Sarcoma with *BCOR* Genetic Alterations	*BCOR::CCNB3*, *BCOR* internal tandem duplication (ITD)	fusion, internal tandem duplication	diagnosis	IHC (BCOR), NGS (DNA, RNA)
	Alveolar Soft Part Sarcoma	*ASPSCR1::TFE3*	fusion	diagnosis	IHC (TFE3), RT-PCR, FISH, NGS (DNA, RNA)
	Angiomatoid Fibrous Histiocytoma	*EWSR1::CREB1* (90%), *EWSR1::ATF1*	fusion	diagnosis	FISH, NGS (DNA, RNA)
	Chordoma (Poorly Differentiated)	*SMARCB1* (*INI1*)	loss (usually partial deletion)	diagnosis, prognosis	IHC (INI1), CMA, NGS (DNA), MLPA
	Desmoplastic Small Round Cell Tumor	*EWSR1::WT1*	fusion	diagnosis	RT-PCR, FISH, NGS (DNA, RNA)

Miscellaneous (Continued…)	Epithelioid Sarcoma	*SMARCB1* (*INI1*; >95%), *SMARCA4* (*BRG1*)	loss (usually deletion)	diagnosis	IHC (INI1, BRG1), CMA, NGS (DNA), MLPA
	NTRK-Rearranged Spindle Cell Neoplasm	*NTRK1/2/3*	fusion	diagnosis, treatment	FISH, NGS (DNA, RNA)
	NUT Carcinoma	*BRD4::NUTM1* (80%), *BRD3::NUTM1* (15%)	fusion	diagnosis	IHC (NUT), FISH, NGS (DNA, RNA)
	Rhabdoid Tumor (Extra-Renal)	*SMARCB1* (*INI1*; >95%), *SMARCA4* (*BRG1*)	loss (usually deletion)	diagnosis, familial cancer risk	IHC (INI1, BRG1), CMA, NGS (DNA), MLPA
	Synovial Sarcoma	*SS18::SSX1, SS18::SSX2, SS18::SSX4*	fusion	diagnosis	RT-PCR, FISH, NGS (DNA, RNA)
Multi-system	*DICER1* Syndrome	*DICER1*	loss-of-function sequence variant or deletion (germline)	diagnosis, familial cancer risk	NGS (DNA)
	Constitutional Mismatch Repair Deficiency Syndrome (CMMRD)	*MLH1, MSH2, PMS2, MSH6*	loss-of-function sequence variant or deletion (germline, biallelic)	diagnosis, familial cancer risk, treatment	IHC (MLH1, MSH2, PMS2, MSH6), NGS (DNA), MSI
	Gorlin Syndrome	*SUFU, PTCH1*	loss-of-function sequence variant, deletion/duplication (germline)	diagnosis, familial cancer risk	NGS (DNA), CMA, MLPA
	Multiple Endocrine Neoplasia (MEN) 2A/2B	*RET*	gain-of-function sequence variant (germline)	diagnosis, familial cancer risk	NGS (DNA)
	Li-Fraumeni Syndrome	*TP53*	loss-of-function sequence variant (germline)	diagnosis, familial cancer risk	NGS (DNA)

Abbreviations:

CMA Chromosomal Microarray Analysis
IHC Immunohistochemistry
NGS Next Generation Sequencing
ddPCR Digital Droplet PCR
MLPA Multiplex Ligation-dependent Probe Amplification
PCR Polymerase Chain Reaction
FISH Fluorescent *in situ* Hybridization
MSI Microsatellite Instability
RT-PCR Reverse Transcription PCR

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