

Molecular In My Pocket™ ...

ONCOLOGY: Pediatric Solid Tumors

Prepared by the Association for Molecular Pathology Training and Education Committee

Tissue Type	Tumor Type	Gene/Biomarker	Alteration	Significance	Primary Assays
Thyroid	Papillary Thyroid Carcinoma	<i>BRAF</i>	sequence variants, fusions	diagnosis, treatment	sequencing
		<i>RET</i>	fusions (usually RET-PTC)	diagnosis	NGS (RNA)
		<i>NTRK1, NTRK2, NTRK3</i>	fusions	diagnosis, treatment	IHC (screening), NGS (RNA)
		<i>ALK</i>	fusions	diagnosis, prognosis	NGS (RNA), FISH
		<i>RAS</i>	sequence variants	diagnosis	sequencing
		<i>DICER1</i>	sequence variants	familial cancer risk	sequencing
		<i>AKT1</i>	sequence variants	diagnosis, prognosis	sequencing
		<i>TERT</i>	sequence variants	diagnosis, prognosis	sequencing
		<i>TP53</i>	sequence variants	diagnosis, prognosis	sequencing
	*Cribiform Morular Variant	<i>APC</i>	loss-of-function variants	diagnosis, familial cancer risk	sequencing
		<i>CTNNB1</i>	sequence variants	diagnosis	sequencing
	Follicular Thyroid Carcinoma	<i>PPARG</i>	fusions	diagnosis, treatment	NGS (RNA)
		<i>RAS</i>	sequence variants	diagnosis, treatment	sequencing
		<i>PIK3CA</i>	sequence variants	diagnosis	sequencing
	With Multiple Adenomas/Adenomatous Nodules	<i>PTEN</i>	sequence variants	diagnosis, familial cancer risk	sequencing
	Medullary Thyroid Carcinoma	<i>RET</i>	sequence variants	diagnosis, familial cancer risk	sequencing
		<i>ALK</i>	fusions (CCDC6)	diagnosis, treatment	NGS (RNA)
NIFTP	<i>RAS</i>	sequence variants	diagnosis	sequencing	
Lung	Pleuropulmonary Blastoma	<i>DICER1</i>	loss-of-function sequence variant or deletion + hotspot sequence variant	diagnosis, familial cancer risk	NGS (DNA)
GI	Gastrointestinal Stromal Tumor	<i>SDHB, KIT exon 9/11, PDGFRA</i>	sequence variants	diagnosis, familial cancer risk, treatment	IHC, NGS (DNA)
Liver	Hepatoblastoma	<i>CTNNB1</i>	activating sequence variants, deletion of exon 3	diagnosis (more common in hepatoblastoma than in hepatocellular carcinoma)	IHC, NGS (DNA)
	Fibrolamellar Hepatocellular Carcinoma	<i>DNAJB1-PRKACA</i>	fusion	diagnosis	FISH, NGS (DNA or RNA)
Kidney	Wilms Tumor	<i>1q gain, 1p/16q LOH</i>	copy gain, LOH	prognosis	CMA, NGS (DNA)
	Clear Cell Sarcoma Of Kidney	<i>BCOR, YWHAE</i>	BCOR internal tandem duplication (ITD), YWHAE fusion	diagnosis	NGS (RNA or DNA)
	Congenital Mesoblastic Nephroma	<i>NTRK3</i>	ETV6-NTRK3 fusion, alternate NTRK fusions	diagnosis, treatment	ETV6 FISH or RT-PCR for classic fusion, NGS (RNA or DNA) for classic or alternate fusions
	Translocation Renal Cell Carcinoma	<i>TFE3, TFEB</i>	Fusion	diagnosis	FISH, NGS (RNA or DNA)

Kidney <i>(continued...)</i>	Metanephric Adenoma	<i>BRAF</i>	V600E, other exon 15 mutations	diagnosis	IHC, sequencing
	Rhabdoid Tumor	<i>SMARCB1 (INI1), SMARCA4 (BRG1)</i>	loss (sequence variant, partial deletion)	diagnosis	IHC, CMA, NGS, MLPA
	Pediatric Cystic Nephroma	<i>DICER1</i>	loss-of-function mutations	familial cancer risk	NGS (DNA)
Ovary tumors	Juvenile Granulosa Cell Tumor	<i>AKT1</i>	in-frame duplication, sequence variants	diagnosis	sequencing
Eye	Retinoblastoma	<i>RB1</i>	inactivating mutation, loss, or loss of heterozygosity (LOH)	diagnosis, familial cancer risk if germline	Sanger sequencing, MLPA, CMA, NGS (DNA)
PNS	Paraganglioma	<i>SDHA, SDHB, SDHC, SDHD</i>	sequence variants, loss of protein expression	diagnosis, familial cancer risk	IHC, NGS (DNA)
Adrenal	Neuroblastoma	<i>MYCN</i>	amplification	prognosis	FISH, NGS (DNA)
		<i>ALK</i>	mutation, amplification	prognosis, treatment	NGS
		<i>ATRX</i>	mutations	prognosis	NGS
		<i>1p/11q LOH</i>	LOH	prognosis	CMA, NGS
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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