Molecular In My Pocket™

Solid Tumors (Sporadic)

Prepared by the Association for Molecular Pathology Training and Education Committee

Abbreviations

AMP	Anchored Multiplex PCR	ISH	*In Situ* Hybridization	MMR	Mismatch Repair	TKI	Tyrosine Kinase Inhibitor
CA	Carcinoma	FISH	Fluorescence ISH	MSI	Microsatellite Instability		
CGX	Cytogenetics	GI	Gastrointestinal	NGS	Next-Generation Sequencing		
IHC	Immunohistochemistry	LOF	Loss of Function	SGA	Single Gene Assay		

Tissue Type	Disease	Gene	Common Genomic Variant(s)	Significance	Primary Assays
Tissue-Agnostic (pan-cancer FDA approval)	All Solid Tumors	MSI testing *MLH1, MSH2, MSH6, PMS2*	MSI-High, Low, or Stable MMR deficiency	responsiveness to immunotherapy	MSI testing, NGS IHC
	All Solid Tumors	Tumor mutation burden (TMB)	TMB-High, Low	responsiveness to immunotherapy	NGS (large panel)
	All Solid Tumors	*NTRK1, NTRK2, NTRK3*	rearrangements	responsiveness to NTRK inhibitors	NGS, AMP, FISH
	All Solid Tumors	*BRAF*	V600E	responsiveness to RAF/MEK inhibitors	IHC, SGAs, NGS
	All Solid Tumors	*RET*	rearrangements	responsiveness to RET inhibitors	NGS, AMP, FISH
	All Solid Tumors	*ERBB2* (HER2)	amplification/overexpression	responsiveness to trastuzumab-deruxtecan	IHC, FISH/ISH, NGS
Adrenal	Neuroblastoma	*MYCN*	amplification	prognosis	FISH, CGX
Bile Duct	Cholangiocarcinoma	*IDH1*	*IDH1* R132	responsiveness to IDH1 inhibitors	SGAs, NGS
		FGFR2	rearrangements	responsiveness to FGFR inhibitors	NGS, AMP, FISH
Brain	**See "Neuropathology" and "Molecular Biomarkers of the Central Nervous System" cards**				
Breast	**See "Breast Cancer" card**				
Cervix	Squamous cell CA	High-risk HPV	high-risk types 16, 18, other	diagnosis, prognosis	IHC (p16), ISH, viral typing
Endometrium	Endometrial CA	MSI testing	MSI-High, Low, or Stable	response to immunotherapy, prognosis (molecular subtype)	MSI testing, NGS
		MLH1, MSH2, MSH6, PMS2	LOF	response to immunotherapy, prognosis (molecular subtype)	IHC
		MLH1	promoter hypermethylation	somatic etiology (not Lynch Syndrome)	Promoter methylation
		POLE	exonuclease domain	prognosis (molecular subtype)	SGAs, NGS
		TP53	LOF	diagnosis (serous carcinoma), prognosis (molecular subtype)	IHC, SGAs, NGS
		ERBB2 (HER2)	amplification/overexpression; S310, L755 mutations	responsiveness to trastuzumab	FISH, IHC, SGAs, NGS
Eye	Uveal Melanoma	*GNAQ, GNA11*	Q209 mutations	diagnosis	SGAs, NGS
GI Tract	Gastrointestinal Stromal Tumor (GIST)	*KIT* (CD117)	exon 9 variants	responsiveness to TKIs, associated with 1' resistance	SGAs, NGS ***IHC not a proxy***
			exon 11 in-frame indels; D820; less frequently exons 8, 17	responsiveness to TKIs	
			exon 14	resistance to first-line TKIs	
			exon 13	responsiveness to TKIs as 1' variant; as 2' variant confers resistance to TKIs	
		PDGFRA	exon 12, 14, 18 variants	responsiveness to TKIs	SGAs, NGS
			exon 18 D842V	resistance to TKIs	
		SDHA, SDHB, SDHC, SDHD	LOF	resistance to TKIs, risk of hereditary cancer	SGAs, NGS, IHC for SDHB
GI Tract (Lower)	**See "Molecular Biomarkers of Colorectal Cancer" card**				
Kidney	Translocation-Associated Renal Cell CA	*TFE3* rearrangement	translocations involving Xp11	diagnosis	FISH, CGX
Liver	Hepatocellular Adenoma	*CTNNB1* (Beta-Catenin)	exon 3, exon 7, exon 8 mutations	diagnosis of beta-catenin-activated subtype	IHC, SGAs, NGS
		HNF1A	LOF	diagnosis of *HNF1A*-inactivated subtype	SGAs, NGS
		IL6ST, STAT3, JAK1, GNAS1, ROS1	gain of function variants	diagnosis of inflammatory subtype	SGAs, NGS
		GLI1	*INHBE::GLI1*	diagnosis of sonic hedgehog-activated subtype	NGS, AMP, FISH
Lung	**See "Molecular Biomarkers of Lung Cancer" card**				
Oropharynx	Squamous Cell CA	High-risk HPV	high-risk virus, type 16, 18, other	prognosis	IHC (p16), ISH, viral typing
Ovary	**See "Molecular Biomarkers of Ovarian Cancer" card**				
Prostate	Prostate CA	*BRCA1, BRCA2, ATM*, and other genes involved in the HRR pathway	LOF	responsiveness to PARP inhibitor	SGAs, NGS
		AR	SNVs, amplification	diagnosis; prognosis; resistance to androgen deprivation	IHC, SGAs, NGS, FISH
		TMPRSS2 rearrangement	*TMPRSS2::ERG*; t(21;21)	diagnosis	FISH, NGS
Skin	Merkel Cell CA	Merkel Cell Polyoma Virus	Virus detection	diagnosis	ISH, Viral Test
	See "Molecular Biomarkers in Cutaneous Melanoma" card				
Soft Tissue	**See "Bone and Soft Tissue Tumors Part 1" and "Bone and Soft Tissue Tumors Part 2" cards**				
Stomach & Esophagus	Gastric and Esophageal Adenocarcinoma	*ERBB2* (HER2)	amplification/overexpression; S310, L755	responsiveness to trastuzumab	FISH, IHC; NGS, SGAs
Thymus	Thymic CA	*KIT* (CD117)	exon 11 variants; D820	responsiveness to TKIs	IHC, SGAs, NGS
Thyroid	**See "Molecular Biomarkers of Thyroid Cancer" card**				
Urinary Tract	Urothelial CA	*FGFR3* rearrangement or mutation	*FGFR3::TACC3*; S249	responsiveness to FGFR inhibitors	NGS, AMP

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