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Clinical Solid Tumor Molecular Oncology: Selected Tests by Tumor Type

This table is for quick reference only. Clinical decision making, including diagnosis and therapy, should not be based solely on this information. The information should be considered in conjunction with clinical information, imaging, and laboratory studies. Additional reading and investigation should be undertaken regarding the tabular entries before information is used in the clinical setting.

Tumor Type	Gene/Loci	Somatic Alteration	Clinical Use	References
Colorectal Adenocarcinoma				
	KRAS codons 12, 13, 59, 61, 117, 146	Mutation	Lack of response to EGFR monoclonal antibodies (p.G13D may be an exception)	2–3, 11–12, 14, 24, 31, 49, 52
	NRAS codons 12, 13, 59, 61, 117, 146	Mutation	Lack of response to EGFR monoclonal antibodies	11, 14, 52
	BRAF	p.V600E mutation	MSI stratification, prognostic factor, possible reduced response to EGFR monoclonal antibodies but insufficient evidence	3, 10–11, 31, 44, 46
	MLH1	Promoter methylation	Indicates sporadic MSI tumor	3, 10
	PIK3CA	Mutation	Possible improved survival with postoperative aspirin therapy	13, 29
Lung Adenocarcinoma				
	EGFR exons 18–21	Mutation	Response to EGFR inhibitors	5, 32, 35, 37, 38
	EGFR	p.T790M and some exon 20 insertion mutations	Resistance to EGFR inhibitors	5, 26, 30, 39, 53
	KRAS codons 12, 13, 61	Mutation	Exclusion of *EGFR* mutation	5, 8, 30, 42
	BRAF p.V600E	Mutation	Possible response to BRAF inhibitor	40
	ALK	Rearrangement	Response to TKI	5, 8, 28, 30
	RET	Rearrangement	Response to TKI	15, 17
	ROS1	Rearrangement	Response to TKI	4, 8
	MET	Amplification	Resistance to EGFR inhibitors	5, 8, 16
Breast Carcinoma				
	HER2/ERBB2	Amplification	Response to HER2 monoclonal antibodies	18, 51
Gastric Adenocarcinoma				
	HER2/ERBB2	Amplification	Response to HER2 monoclonal antibodies	45
Thyroid Carcinoma				
Papillary Thyroid Carcinoma / Anaplastic Thyroid Cancer	*BRAF*	p.V600E mutation	Preoperative FNA diagnosis and prognosis, potential therapeutic target	9, 36, 43
	NRAS, HRAS, KRAS	Mutation	Preoperative FNA diagnosis	36
	RET-PTC	Rearrangement	Preoperative FNA diagnosis	36
Follicular Thyroid Carcinoma	*NRAS, HRAS, KRAS*	Mutation	Preoperative FNA diagnosis	36
	PAX8-PPAR$_{\gamma}$	Rearrangement	Preoperative FNA diagnosis	36
Melanoma				
Cutaneous & Mucosal	*BRAF* codon 600	Mutation	Response to BRAF inhibitors	19–20, 33
	KIT	Mutation	Response to TKI	7
Uveal	*GNAQ* or *GNA11*	Mutation	Diagnostic	50
	Chromosome 3	Loss (monosomy)	Unfavorable prognosis	23
GIST				
	KIT	Mutation	Response to TKI	41
	PDGFRA	Mutation	Response to TKI	41
	BRAF p.V600E	Mutation	Possible imatinib resistance	1, 34
CNS Neoplasms				
Glioma	*MGMT*	Promoter methylation	Favorable response to alkylating agents	21
	IDH1 and *IDH2*	Mutation	Distinguishes reactive gliosis from glioma, favorable prognosis	27, 54
Oligodendroglioma	Chromosome 1p and 19q	Co-deletion	Favorable prognosis and response to therapy	6, 22
Pilocytic Astrocytoma	*BRAF*	Duplication/ fusion and p.V600E mutation (extracere-bellar)	Diagnostic	27, 47
Pleomorphic Xanthoastrocytoma and Ganglioglioma	*BRAF*	p.V600E mutation	Diagnostic	47
Cholangiocarcinoma/Pancreatic Carcinoma				
	KRAS codons 12, 13, 61	Mutation	Preoperative bile duct brushing diagnosis	25
Oropharyngeal Squamous Cell Carcinoma				
	HR HPV-related	Positive detection	Favorable response to chemoradiation therapy	48

Source: Allison M. Cushman-Vokoun, MD, PhD

MSI = Microsatellite Instability; TKI = Tyrosine-Kinase Inhibitors; HR HPV= High-Risk Human Papillomavirus
This table is meant to be a list of selected tests and is not a comprehensive resource.

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