

APPENDIX **XIV**

*TUMOR ASSOCIATED
MUTATIONS IN PIK3CA*

Domain	Mutation	Tumor Type
	p.G12D	Large intestine adenocarcinoma
	p.P17S	Large intestine adenocarcinoma
	p.I31M	Breast adenoid cystic carcinoma
ABD	p.R38H	Large intestine adenocarcinoma
ABD	p.R38C	Large intestine adenocarcinoma
ABD	p.Q60K	Breast carcinoma
ABD	p.E81K	Large intestine adenocarcinoma
ABD	p.R88Q	Large intestine adenocarcinoma
		Astrocytoma grade IV
		Breast lobular carcinoma
		Oligodendroglioma grade III
		Leiomyosarcoma
		Cervical carcinoma
		Endometrioid carcinoma
ABD	p.R93W	Large intestine adenocarcinoma
ABD	p.P104R	Large intestine adenocarcinoma
ABD	p.P104 G106>R	Large intestine adenocarcinoma
ABD	p.G106V	Large intestine adenocarcinoma
ABD	p.G106 R108del	Lung SCC cell line (NCI-H69)
ABD	p.R108P	Large intestine adenocarcinoma
ABD	p.R108del	Large intestine adenocarcinoma
ABD	p.R108H	Large intestine adenocarcinoma
	p.E109del	Large intestine adenocarcinoma
	p.E110K	Breast ductal carcinoma

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	p.K111del	Large intestine adenocarcinoma
	p.K111R	Lung SCC cell line (NCI-H1048)
	p.K111N	Large intestine adenocarcinoma Breast ductal carcinoma
	p.K111E	Lung adenocarcinoma
	p.K111 I112>N	Large intestine adenocarcinoma
	p.I112N	Endometrioid carcinoma
	p.L113del	Astrocytoma grade IV
	p.G118D	Large intestine adenocarcinoma Astrocytoma grade IV Lung carcinoma
	p.G122D	Large intestine adenocarcinoma
	p.P124T	Large intestine adenocarcinoma
	p.P124L	Urinary tract carcinoma
	p.D133V	Hepatocellular carcinoma
	p.P134S	Large intestine adenocarcinoma
	p.S158L	Large intestine adenocarcinoma
	p.H160N	Large intestine adenocarcinoma
RBD	p.S173Y	Large intestine adenocarcinoma
RBD	p.K179T	Large intestine adenocarcinoma
RBD	p.K184E	Large intestine adenocarcinoma
	p.P298T	Astrocytoma grade IV
	p.R310C	Astrocytoma grade IV
	p.T324I	Pancreas ductal carcinoma
	p.L339F	Esophagus squamous cell carcinoma
	p.Y343C	Upper aerodigestive tract squamous cell carcinoma
	p.V344G	Astrocytoma grade IV
	p.N345S	Astrocytoma grade IV
	p.N345I	Breast ductal carcinoma
	p.N345K	Large intestine adenocarcinoma Breast ductal carcinoma
	p.D350H	Astrocytoma grade IV
	p.C378F	Astrocytoma grade IV
	p.C378R	Large intestine adenocarcinoma Astrocytoma grade IV
	p.S405F	Breast ductal carcinoma
	p.E418K	Breast carcinoma
	p.C420R	Large intestine adenocarcinoma

		Ductal carcinoma
		Breast lobular carcinoma
	p.P449T	Large intestine carcinoma
	p.G451 L456>V	Breast carcinoma
	p.E453K	Large intestine adenocarcinoma
		Astrocytoma grade IV
		Gastric carcinoma
	p.E453Q	Large intestine adenocarcinoma
Helical	p.H510H	Large intestine adenocarcinoma
Helical	p.R524M	Neuroblastoma
Helical	p.R537R	Large intestine adenocarcinoma
Helical	p.D538N	Melanoma
Helical	p.P539R	Breast carcinoma
Helical	p.E542G	Large intestine adenocarcinoma
Helical	p.E542K	Upper aerodigestive tract SQCC
		Large intestine adenocarcinoma
		Glioma
		Breast carcinoma
		Endometrioid carcinoma
		Cervix SCC
		Stomach adenocarcinoma
		Thyroid follicular carcinoma
		Large intestine adenocarcinoma
		Breast lobular carcinoma
		NSCLC
		Oligodendroglioma
		Thyroid papillary carcinoma
		Penis carcinoma
		Seborrheic keratosis
		Ovary serous carcinoma
		Esophagus SQCC
		Urinary tract transitional cell carcinoma
		Neuroectodermal medulloblastoma
Helical	p.E542V	Large intestine adenocarcinoma
		Endometrioid carcinoma
		Breast lobular carcinoma
		Breast carcinoma
		Large intestine tubulovillous adenoma

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Helical	p.E542Q	Endometrioid adenocarcinoma Lung squamous cell carcinoma Large intestine adenocarcinoma
Helical	p.I543V	DLBCL
Helical	p.E545V	Large intestine adenocarcinoma
Helical	p.E545Q	Large intestine adenocarcinoma Esophageal squamous cell carcinoma
Helical	p.E545D	ATCLL Large intestine adenocarcinoma Endometrioid carcinoma Esophageal squamous cell carcinoma
Helical	p.E545G	DLBCL Large intestine adenocarcinoma Epidermal nevus Breast ductal carcinoma Endometrioid carcinoma Seborrheic keratosis Upper aerodigestive tract SQCC Urinary tract transitional cell carcinoma and cell line
Helical	p.E545A	Large intestine adenocarcinoma Astrocytoma Endometrioid carcinoma Breast carcinoma Ovary carcinoma
Helical	p.E545K	PCM Lung adenocarcinoma Astrocytoma Carcinosarcoma–malignant mesodermal mixed tumor Cervix carcinoma cell line (ME-180) Cervix SQCC cell line (OMC-1) Kidney clear cell carcinoma Breast ductal carcinoma Endometrioid carcinoma Ovary carcinoma Ovary mucinous carcinoma Ovary clear cell carcinoma Ovary serous carcinoma

		Large intestine adenocarcinoma
		Stomach adenocarcinoma
		Breast lobular carcinoma
		Breast carcinoma cell line
		Mixed adenosquamous carcinoma
		Large intestine mucinous carcinoma
		Lung NSCLC
		Oligodendroglioma grade III
		Thyroid papillary carcinoma
		Seborrheic keratosis
		Endometrioid serous carcinoma
		Upper aerodigestive tract SQCC
		Esophagus SQCC
		penis carcinoma
		Urinary tract transitional cell carcinoma
		kidney carcinoma cell line (BFTC-909)
		Melanoma
		Biliary tract carcinoma cell line (HuH-28)
Helical	p.Q546L	Adenocarcinoma
		Squamous cell carcinoma
Helical	p.Q546H	Endometrioid carcinoma
Helical	p.Q546K	Lung SCC cell line (NCI-N417)
		Large intestine adenocarcinoma
		Endometrioid carcinoma
		Lung adenocarcinoma
Helical	p.Q546E	Large intestine adenocarcinoma
		Breast ductal carcinoma
		Endometrioid carcinoma
Helical	p.Q546P	Large intestine adenocarcinoma
		Astrocytoma grade III
		Oligodendroglioma grade III
Helical	p.Q546R	Large intestine adenocarcinoma
		Breast ductal carcinoma
		Endometrioid carcinoma
Helical	p.D549N	Large intestine adenocarcinoma
		Breast ductal carcinoma
		Endometrioid carcinoma
Helical	p.W552G	Pancreatic carcinoma

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Helical	p.S553fs*7	DLBCL
Helical	p.Q661K	Large intestine adenocarcinoma
	p.H701P	Breast carcinoma
		Astrocytoma grade IV
	p.K733R	Large intestine adenocarcinoma
		Glioma
Kinase	p.C901F	Large intestine adenocarcinoma
		Stomach adenocarcinoma
Kinase	p.F909L	Breast carcinoma
		Large intestine adenocarcinoma
Kinase	p.E982D	Autonomic ganglia carcinoma
Kinase	p.A987T	Large intestine adenocarcinoma
		Neuroblastoma
Kinase	p.M1004I	Large intestine adenocarcinoma
		Astrocytoma grade IV
Kinase	p.G1007R	Large intestine adenocarcinoma
		Urinary tract carcinoma
		Endometrioid carcinoma
Kinase	p.G1007D	DLBCL
Kinase	p.S1008P	Large intestine adenocarcinoma
Kinase	p.P1011P	Large intestine adenocarcinoma
Kinase	p.P1011S	Stomach adenocarcinoma
Kinase	p.E1012E	Endometrioid carcinoma
Kinase	p.S1015F	Pancreas ductal carcinoma
	p.D1017H	Endometrioid carcinoma
	p.Y1021H	Endometrioid carcinoma
	p.Y1021C	Large intestine adenocarcinoma
		Stomach adenocarcinoma
		Astrocytoma grade IV
		Endometrioid carcinoma
	p.Y1021N	Astrocytoma grade IV
	p.R1023Q	Large intestine adenocarcinoma
		Glioma
	p.T1025T	Endometrioid carcinoma
		Hepatocellular carcinoma
		Ovarian serous carcinoma
		Upper aerodigestive tract SQCC

	Breast carcinoma cell line (HCC1500)
p.T1025N	Large intestine adenocarcinoma Astrocytoma grade IV
p.T1025A	Glioma
p.T1025S	Endometrioid carcinoma Breast adenocarcinoma Endometrioid giant cell carcinoma
p.L1026P	DLBCL
p.D1029E	Lung squamous cell carcinoma
p.E1034G	Hepatocellular carcinoma
p.A1035T	Endometrioid carcinoma
p.A1035V	Endometrioid carcinoma Large intestine adenocarcinoma
p.L1036S	Endometrioid carcinoma
p.Y1038C	Large intestine adenocarcinoma
p.M1040I	DLBCL
p.M1043T	Stomach adenocarcinoma
p.M1043I	Large intestine adenocarcinoma Astrocytoma grade IV Endometrioid carcinoma
p.M1043V	Endometrioid carcinoma Upper aerodigestive tract squamous cell carcinoma Glioma Breast carcinoma
p.N1044K	Large intestine adenocarcinoma Breast carcinoma
p.N1044S	Astrocytoma grade IV
p.N1044D	Endometrioid carcinoma Glioma
p.D1045N	DLBCL Adenocarcinoma
p.A1046E	Large intestine adenocarcinoma
p.A1046A	Adenocarcinoma
p.A1046V	Breast ductal carcinoma
p.A1046T	Large intestine adenocarcinoma
p.H1047Y	Adenocarcinoma Astrocytoma grade IV

p.H1047R	Ductal carcinoma
	DLBCL
	Endometrial clear cell carcinoma
	Breast ductal carcinoma
	Breast ductal carcinoma in situ
	Breast lobular carcinoma
	Endometrioid carcinoma
	Hepatocellular carcinoma
	Intestinal adenocarcinoma
	Ovarian large cell carcinoma
	Ovary serous carcinoma
	Mixed endometrioid and clear cell carcinoma
	Mixed serous and endometrioid carcinoma
	Lung NSCLC
	Lung SCC cell line (NCI-H1048)
	Upper aerodigestive tract SQCC
	Oligodendroglioma
	Astrocytoma
	Thyroid papillary carcinoma
	Seborrheic keratosis
	Melanoma
	Stomach adenocarcinoma
	Large intestine adenocarcinoma
Hepatocellular carcinoma	
Urinary tract transitional cell carcinoma (MGH-U4)	
p.H1047Q	Pancreas ductal carcinoma
	Endometrioid carcinoma
	Ewing's sarcoma
	Melanoma
	Medulloblastoma
p.H1047L	Meningioma
	DLBCL
	Adenocarcinoma
	Astrocytoma grade IV
	Breast ductal carcinoma
	Breast ductal carcinoma in situ

	Endometrioid carcinoma
	Hepatocellular carcinoma
	Breast lobular carcinoma
	Large intestine carcinoma
	Urinary tract transitional cell carcinoma
	Ovary mucinous carcinoma cell line (OAW-42)
p.H1047T	Lymph. Neo. Breast carcinoma Breast carcinoma in situ Glioma
p.H1048Q	Adenocarcinoma
p.H1048L	Breast ductal carcinoma
p.G1049A	Adenocarcinoma
p.G1049S	Astrocytoma grade IV Large intestine carcinoma Upper aerodigestive tract squamous cell carcinoma
p.G1049R	Large intestine adenocarcinoma Breast ductal carcinoma Thyroid papillary carcinoma Squamous cell carcinoma Glioma Endometrioid carcinoma cell line (HEC-1)
p.G1049G	Ovarian carcinoma
p.G1050D	Endometrioid carcinoma
p.T1052K	Endometrioid carcinoma
p.H1065Y	Medulloblastoma
p.H1065fs*6	Medulloblastoma Upper aerodigestive tract squamous cell carcinoma
p.H1065L	Endometrioid carcinoma
p.A1066V	Urinary tract transitional cell carcinoma
p.N1068fs*4	Adenocarcinoma Hepatocellular carcinoma
p. 542 54 E>K	Breast carcinoma
p.?	Large intestine adenocarcinoma Astrocytoma grade IV

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	Upper aerodigestive tract squamous cell carcinoma
p.*1069	Endometrioid carcinoma
1069insWKDN	Ovarian carcinoma
	Glioma

SOURCES

Forbes SA, Bhamra G, Bamford S, Dawson E, Kok C, Clements J, Menzies A, Teague JW, Futreal PA, Stratton MR. The Catalog of Somatic Mutations in Cancer (COSMIC). *Curr Protocols Hum Genet.* 2008 Apr; Chapter 10: Unit 10.11.