

APPENDIX **IX**

*TUMOR ASSOCIATED
MUTATIONS IN KIT*

Domain	Mutation	Tumor Type
Ig	p.D52N	CML, MDS
Ig	p.E73K	Genital tract seminoma
Ig	p.T96M	Genital tract seminoma
Ig	p.T417 R420>R	AML
Ig	p.T417 R420>SVIVG	AML
Ig	p.T417 D419>I	AML
Ig	p.T417 D419>L	AML
Ig	p.T417 D419>N	AML
Ig	p.T417 D419>V	AML
Ig	p.T417 D419>Y	AML
Ig	p.T417 D419>RA	AML
Ig	p.T417 D419>RG	AML
Ig	p.T417 Y418>H	AML
Ig	p.Y418 D419>S	AML
Ig	p.Y418 D419>G	AML
Ig	p.Y418 D419insFF	AML
Ig	p.D419 L421>F	AML
Ig	p.D419 R420>SSDG	AML
Ig	p.D419del	AML
Ig	p.P456S	GIST
Ig	p.F469L	Ewing's sarcoma
Ig	p.S480fs*47	GIST
Ig	p.N486D	Ewing's sarcoma
Ig	p.V489A	Ewing's sarcoma

2 TARGETING PROTEIN KINASES FOR CANCER THERAPY

Ig	p.E490 F504>DHIVVSLTF	GIST
Ig	p.N495I	Lung SQCC
Ig	p.V497V	GIST
Ig	p.K499K	PMF
Ig	pA502 Y503dup	GIST
	p.Y503 F504insAY	GIST
	p.F506 A507insAYFNF	AML
	p.V530I	AML
	p.I538M	PCV
	p.M541L	Breast phyllode tumor
	p.Y547 K557>Q	GIST
	p.L548 K557>Q	GIST
	p.L548 E554>QK	GIST
	p.Q549 V555>I	GIST
Juxtamembrane	p.K550I	GIST
Juxtamembrane	p.K550N	GIST
Juxtamembrane	p.K550R	GIST
Juxtamembrane	p.K550 P551>I	GIST
Juxtamembrane	p.K550 K558del	GIST
Juxtamembrane	p.K550 V559del	GIST
Juxtamembrane	p.K550 V555>I	GIST
Juxtamembrane	p.K550 W557del	GIST
Juxtamembrane	p.K550 W557>FL	GIST
Juxtamembrane	p.K550 V555>QRI	GIST
Juxtamembrane	p.K550 V555>F	GIST
Juxtamembrane	p.K550 P551del	GIST
Juxtamembrane	p.K550 Q556>L	GIST
Juxtamembrane	p.K550 V555del	GIST
	p.K550 W558>IL	GIST
Juxtamembrane	p.K550 E554del	GIST
Juxtamembrane	p.K550 Q556>II	GIST
Juxtamembrane	p.K550 V560>L	GIST
Juxtamembrane	p.K550 K558>Q	GIST
Juxtamembrane	p.K550 K558>G	GIST
Juxtamembrane	p.K550 Q556del	GIST
Juxtamembrane	p.P551 V559del	GIST
Juxtamembrane	p.P551 Q556del	GIST

Juxtamembrane	p.P551 W557>R	GIST
Juxtamembrane	p.P551 Y553del	GIST
Juxtamembrane	p.P551 E554>H	GIST
	p.P551S	DLBCL
Juxtamembrane	p.P551T	GIST
Juxtamembrane	p.P551 V555>L	GIST
Juxtamembrane	p.P551 Q556>HV	GIST
Juxtamembrane	p.P551 M552>L	GIST
Juxtamembrane	p.P551 W557del	GIST
Juxtamembrane	p.P551 E554del	GIST
Juxtamembrane	p.P551 V555del	GIST
Juxtamembrane	p.P551 E561>Q	GIST
Juxtamembrane	p.P551del	GIST
Juxtamembrane	p.M552 E561>K	GIST
Juxtamembrane	p.M552 Q556del	GIST
Juxtamembrane	p.M552 V560del	GIST
Juxtamembrane	p.M552 K558>NE	GIST
Juxtamembrane	p.M552 K558del	GIST
Juxtamembrane	p.M552 Y553del	GIST
Juxtamembrane	p.M552 Y553del + E554K	GIST
Juxtamembrane	p.M552 Q556>	GIST
Juxtamembrane	p.M552 V559>I	GIST
Juxtamembrane	p.M552 I563del	GIST
Juxtamembrane	p.M552 W557del	GIST
Juxtamembrane	p.M552 V555>I	GIST
Juxtamembrane	p.M552 V555del	GIST
		Seminoma
Juxtamembrane	p.M552 K558>T	GIST
Juxtamembrane	p.M552 V559>IT	GIST
Juxtamembrane	p.M552T	NKTCL
Juxtamembrane	p.M552V	GIST
Juxtamembrane	p.M552I	GIST
Juxtamembrane	p.M552K	GIST
Juxtamembrane	p.M552L	GIST
Juxtamembrane	p.M552 E554del	GIST
Juxtamembrane	p.M552 E554>K	GIST
Juxtamembrane	p.M552 E554>I	GIST
Juxtamembrane	p.M552 Q556>T	GIST
Juxtamembrane	p.M552 Q556>K	GIST

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Juxtamembrane	p.Y553 Q556del	GIST
Juxtamembrane	p.M552 W557del	GIST
Juxtamembrane	p.Y553 K558del	GIST
Juxtamembrane	p.Y553 V559del	GIST
	p.Y553 P573del	GIST
Juxtamembrane	p.Y553V	GIST
Juxtamembrane	p.Y553C	GIST
Juxtamembrane	p.Y553N	GIST
		Melanoma
Juxtamembrane	p.Y553 K558>L	GIST
Juxtamembrane	p.Y553 W557del	GIST
Juxtamembrane	p.Y553 T574>S	GIST
Juxtamembrane	p.Y553 K558>SE	GIST
Juxtamembrane	p.E554 V559del	GIST
		Melanoma
Juxtamembrane	p.E554K	NKTCL
Juxtamembrane	p.E554G	DLBCL
Juxtamembrane	p.E554D	GIST
Juxtamembrane	p.E554 K558del	GIST
Juxtamembrane	p.E554 K557>G	GIST
Juxtamembrane	p.E554 Q556del	GIST
Juxtamembrane	p.E554 V560>K	GIST
Juxtamembrane	p.E554 V560>D	GIST
Juxtamembrane	p.E554 V555del	GIST
Juxtamembrane	p.V555 V560del	GIST
Juxtamembrane	p.V555 P573del	GIST
		Seminoma
Juxtamembrane	p.V555 I571del	GIST
Juxtamembrane	p.V555 V559del	GIST
Juxtamembrane	p.V555 Q556>E	GIST
Juxtamembrane	p.V555 Y570del	GIST
Juxtamembrane	p.V555 Q556del	GIST
Juxtamembrane	p.V555 D572del	GIST
Juxtamembrane	p.V555 I563del	GIST
Juxtamembrane	p.V555 K558del	GIST
Juxtamembrane	p.V555 V560>H	GIST
Juxtamembrane	p.V555 V560>T	GIST
Juxtamembrane	p.V555 E562del	GIST
Juxtamembrane	p.V555V	GIST

Juxtamembrane	p.Q556 D572del	GIST
Juxtamembrane	p.Q556 V559del	GIST
Juxtamembrane	p.Q556 V559>H	GIST
Juxtamembrane	p.Q556H	GIST
Juxtamembrane	p.Q556 V559>HF	GIST
Juxtamembrane	p.Q556 V559>HT	GIST
Juxtamembrane	p.Q556 K558>HT	GIST
Juxtamembrane	p.Q556 V560>H	GIST
Juxtamembrane	p.Q556 V560>F	GIST
Juxtamembrane	p.Q556 W557>E	GIST
Juxtamembrane	p.Q556 D572>H	GIST
Juxtamembrane	p.Q556 V559>PF	GIST
Juxtamembrane	p.Q556 L576del	GIST
Juxtamembrane	p.Q556 T574del	GIST
Juxtamembrane	p.Q556 K558>H	GIST
Juxtamembrane	p.Q556 K558>N	GIST
Juxtamembrane	p.Q556 K558>T	GIST
Juxtamembrane	p.Q556 N564>H	GIST
Juxtamembrane	p.Q556 W557del	GIST
Juxtamembrane	p.Q556 E561del	GIST
Juxtamembrane	p.Q556 E561>HH	GIST
Juxtamembrane	p.Q556 K558del	GIST
Juxtamembrane	p.Q556 V560del	GIST
Juxtamembrane	p.Q556E + W557 K558del	GIST
Juxtamembrane	p.W557 V560>C	GIST
Juxtamembrane	p.W557 V560>F	GIST
Juxtamembrane	p.W557del	GIST
Juxtamembrane	p.W557 K558>E	GIST
Juxtamembrane	p.W557 K558>S	GIST
Juxtamembrane	p.W557 V560del	GIST
Juxtamembrane	p.W557 T574del	GIST
Juxtamembrane	p.W557S	GIST
Juxtamembrane	p.W557R	GIST
		Melanoma
Juxtamembrane	p.W557G	GIST
Juxtamembrane	p.W557C	Seminoma
Juxtamembrane	p.W557 I563>FP	GIST
Juxtamembrane	p.W557 K558>FP	GIST
Juxtamembrane	p.W557 K558>FQ	GIST

6 TARGETING PROTEIN KINASES FOR CANCER THERAPY

Juxtamembrane	p.W557 Y570del	GIST
Juxtamembrane	p.W557 K558>CP	GIST
Juxtamembrane	p.W557 E562del	GIST
Juxtamembrane	p.W557 Q575del	GIST
Juxtamembrane	p.W557 N564del	GIST
Juxtamembrane	p.W557 K558>Y	GIST
Juxtamembrane	p.W557 P573>S	GIST
Juxtamembrane	p.W557*	MDS
Juxtamembrane	p.W557 I563>T	GIST
Juxtamembrane	p.W557 K558del	GIST
Juxtamembrane	p.W557 V559delinsF	GIST
Juxtamembrane	p.W557 V559>I	GIST
Juxtamembrane	p.W557 V559>F	GIST
Juxtamembrane	p.W557 V559del	GIST
Juxtamembrane	p.W557 E561del	GIST
Juxtamembrane	p.W557 T575>CP	GIST
Juxtamembrane	p.W557 E561>PS	GIST
Juxtamembrane	p.W557 D572del	GIST
Juxtamembrane	p.W557 V559>C	GIST
Juxtamembrane	p.W557 I571del	GIST
Juxtamembrane	p.W557 V559>E	GIST
Juxtamembrane	p.K558 K>NQ	GIST
Juxtamembrane	p.K558 V559insS	GIST
Juxtamembrane	p.K558 V559>QP	GIST
Juxtamembrane	p.K558 D572>N	GIST
Juxtamembrane	p.K558 I563del	GIST
Juxtamembrane	p.K558 E562del	GIST
Juxtamembrane	p.K558 V560del	GIST
Juxtamembrane	p.K558 G565>R	GIST
Juxtamembrane	p.K558*	PTCL
Juxtamembrane	p.K558 G565>A	GIST
Juxtamembrane	p.K558K	GIST
Juxtamembrane	p.K558N	GIST
		Melanoma
Juxtamembrane	p.K558N + V559del	GIST
Juxtamembrane	p.K558N + V559P577del	GIST
Juxtamembrane	p.K558 V559del	GIST
Juxtamembrane	p.K558 G565del	GIST
Juxtamembrane	p.K558 E561del	GIST

Juxtamembrane	p.K558 P573>R	GIST
Juxtamembrane	p.K558>NP	GIST
Juxtamembrane	p.K558 N564>R	GIST
Juxtamembrane	p.K558 V560>I	GIST
Juxtamembrane	p.K558 V560>N	GIST
Juxtamembrane	p.K558 V560>M	GIST
Juxtamembrane	p.K558 N564del	GIST
Juxtamembrane	p.K558 V559>I	GIST
Juxtamembrane	p.K558 V559>N	GIST
Juxtamembrane	p.K558 V559>S	GIST
Juxtamembrane	p.K558 K558insNP	GIST
Juxtamembrane	p.K558 D572del	GIST
Juxtamembrane	p.K558delinsNP	GIST
Juxtamembrane	p.K558delinsQP	GIST
Juxtamembrane	p.K558delinsNQ	GIST
Juxtamembrane	p.V559D	GIST
Juxtamembrane	p.V559del	GIST
Juxtamembrane	p.V559A	GIST
		NKTCL
		Melanoma
Juxtamembrane	p.V559I	ATCLL
		DLBCL
		MCN
		NKTCL
		Systemic mastocytosis
Juxtamembrane	p.V559G	GIST
Juxtamembrane	p.V559 N566>D	GIST
Juxtamembrane	p.V559 G565del	GIST
Juxtamembrane	p.V559 E561del	GIST
Juxtamembrane	p.V559 V560del	GIST
Juxtamembrane	p.V559 E562del	GIST
Juxtamembrane	p.V560 I571del	GIST
Juxtamembrane	p.V560del	GIST
		Thymic carcinoma
Juxtamembrane	p.V560 Y578del	GIST
Juxtamembrane	p.V560 L576del	GIST
Juxtamembrane	p.V560V	GIST
Juxtamembrane	p.V560A	GIST

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Juxtamembrane	p.V560G	GIST
		MCN
Juxtamembrane	p.V560D	GIST
		MCN
		Seminoma
Juxtamembrane	p.V560E	GIST
Juxtamembrane	p.E561 Y578del	GIST
Juxtamembrane	p.E561del	GIST
Juxtamembrane	p.E561K	NKTCL
		GIST
Juxtamembrane	p.E561 D579>P	GIST
Juxtamembrane	p.E562 P573del	GIST
Juxtamembrane	p.E562A	GIST
Juxtamembrane	p.E562K	NKCTL
Juxtamembrane	p.E562 V569>D	GIST
Juxtamembrane	p.I563 T574del	GIST
Juxtamembrane	p.I563 L576del	GIST
Juxtamembrane	p.I563 N564>MI	GIST
Juxtamembrane	p.I563 D572del	GIST
Juxtamembrane	p.N564 P577del	GIST
Juxtamembrane	p.N564 Y578del	GIST
Juxtamembrane	p.N564 L576del	GIST
Juxtamembrane	p.N564 P573>TS	GIST
Juxtamembrane	p.N564K	CML
Juxtamembrane	p.N564H	GIST
Juxtamembrane	p.N564 T574del	GIST
Juxtamembrane	p.G565 T574>P	GIST
Juxtamembrane	p.G565R	GIST
Juxtamembrane	p.G565 D579>V	GIST
Juxtamembrane	p.N566D	DLBCL
		Melanoma
Juxtamembrane	p.N567 P577del	GIST
Juxtamembrane	p.N567S	GIST
Juxtamembrane	p.N567K	Lung SQCC
Juxtamembrane	p.N567H	GIST
Juxtamembrane	p.N567 P573del	GIST
Juxtamembrane	p.N567 L576>E	GIST
Juxtamembrane	p.N567 L576>KV	GIST
Juxtamembrane	p.Y568 L576>CV	GIST

Juxtamembrane	p.Y568D	GIST
Juxtamembrane	p.Y568C	GIST
Juxtamembrane	p.Y568 T574del	GIST
Juxtamembrane	p.V569 D572del	GIST
Juxtamembrane	p.V569 Q575>E	GIST
Juxtamembrane	p.V569A	GIST
Juxtamembrane	p.V569G	Melanoma
Juxtamembrane	p.V569 T574del	GIST
Juxtamembrane	p.V569 L576>G	GIST
Juxtamembrane	p.V569 L576del	GIST
Juxtamembrane	p.V569 Q575del	GIST
Juxtamembrane	p.Y570 L576del	GIST
Juxtamembrane	p.Y570 Y578del	GIST
Juxtamembrane	p.571 572>GE	AML
Juxtamembrane	p.I571 D572ins14	AML
Juxtamembrane	p.D572Y	GIST
Juxtamembrane	p.P573 D579del	GIST
Juxtamembrane	p.P573 T574insDP	GIST
Juxtamembrane	p.P573 T574insYIDP	GIST
Juxtamembrane	p.T574 H580del	GIST
Juxtamembrane	p.T574I	PTCL
Juxtamembrane	p.T574 Q575ins12	GIST
Juxtamembrane	p.T574 Q575insPT	GIST
Juxtamembrane	p.Q575 L576ins17	AML
Juxtamembrane	p.L576 P577insPTQL	GIST
Juxtamembrane	p.L576P	GIST
		Seminoma
		Melanoma
		Thymic carcinoma
Juxtamembrane	p.L576 P577ins15	GIST
Juxtamembrane	p.L576 P577insQL	GIST
Juxtamembrane	p.L576del	GIST
Juxtamembrane	p.L576F	GIST
Juxtamembrane	p.P577 Y578del	GIST
Juxtamembrane	p.P577 Y578insTQLP	GIST
Juxtamembrane	p.P577S	ATCLL
Juxtamembrane	p.P577K	GIST
Juxtamembrane	p.P577L	GIST
Juxtamembrane	p.Y578 D579insXXX	GIST

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Juxtamembrane	p.Y578 D579insENY	GIST
Juxtamembrane	p.Y578 D579insPY	GIST
Juxtamembrane	p.Y578 D579insELPY	GIST
Juxtamembrane	p.D579 H580insPTQLPYD	GIST
Juxtamembrane	p.D579 H580insPTQLPY	GIST
Juxtamembrane	p.D579 H580insIDPTQLPYD	GIST
Juxtamembrane	p.D579del	GIST
Juxtamembrane	p.D579E	GIST
Juxtamembrane	p.D579 H580insTQLPYD	GIST
	p.H580del	GIST
Juxtamembrane	p.H580 K581insPYDH	GIST
Juxtamembrane	p.H580 K581insYDH	GIST
Juxtamembrane	p.H580 K581insHPYD	GIST
Juxtamembrane	p.H580 R586del	GIST
Juxtamembrane	p.K581 W582insDPTQLPYDH	GIST
Juxtamembrane	p.W582 E583insDHWEN	GIST
Juxtamembrane	p.583 584>STQLPYDHWKE	GIST
Juxtamembrane	p.F584 P585insLPYDHWKEF	GIST
Juxtamembrane	p.F584 P585ins13	GIST
Juxtamembrane	p.F584 P585insREF	GIST
Juxtamembrane	p.F584S	GIST
Juxtamembrane	p.P585 R586insEIS	GIST
Juxtamembrane	p.P585 R586insLPYDHWKEFP	GIST
Juxtamembrane	p.P585 R586ins12	GIST
Juxtamembrane	p.P585 R586ins11	GIST
Juxtamembrane	p.R586I	CMML
Juxtamembrane	p.R586 N587ins13	GIST
Juxtamembrane	p.R586 N587ins12	GIST
	p.N587 R588ins15	GIST
	p.R588 L589ins17	GIST
	p.R588 L589insDHWKEFPRN	GIST
	p.R588 L589ins14	GIST
Kinase	p.L589 S590insKTLGAGAFGK	GIST
Kinase	p.L589 S590ins13	GIST
Kinase	p.L589 S590ins15	GIST
Kinase	p.L589 S590ins17	GIST
Kinase	p.S590 F591ins13	GIST
Kinase	p.S590 F591ins14	GIST
Kinase	p.S590N	GIST

Kinase	p.591 592>AYDHKWEFPRNRLSF	GIST
Kinase	p.F591 G592ins21	GIST
Kinase	p.F591 G592ins15	GIST
Kinase	p.G592 K593ins13	GIST
Kinase	p.G592 K593ins15	GIST
Kinase	p.K593 T594insFBPRNSLSFG	GIST
Kinase	p.G596D	MDS
Kinase	p.A617S	AMLAMDS
Kinase	p.P627L	GIST
Kinase	p.R634R	PTCL
Kinase	p.R634W	Melanoma
Kinase	p.A636V	Seminoma
Kinase	p.K642E	GIST Melanoma
Kinase	p.V643A	GIST
Kinase	p.L647P	GIST
Kinase	p.M651V	GIST
Kinase	p.I653T	GIST
Kinase	p.V654A	GIST
Kinase	p.T670E	GIST
Kinase	p.T670I	GIST
	p.D696G	PCV
	p.K704 N705del	GIST
	p.S709F	GIST
	p.S715del	GIST
	p.D716N	GIST
	p.K735M	PCV
	p.D737N	Large intestine adenocarcinoma
	p.I748T	AML
	p.T753A	PCV
Kinase	p.I798I	CML Seminoma Melanoma
Kinase	p.L799L	Seminoma
Kinase	p.T801I	Seminoma
Kinase	p.R804W	Large intestine adenocarcinoma
Kinase	p.I805I	Seminoma
Kinase	p.C809R	AMLAMDS
Kinase	p.C809G	GIST

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Kinase	p.A814S	AML
Kinase	p.A814T	MCN
Kinase	p.R815 D816insVI	Mastocytosis
Kinase	p.D816?	AML
Kinase	p.D816G	AML GIST
Kinase	p.D816Y	AML AMLAMDS CML MCN Dysgerminoma Nonseminoma, seminoma
Kinase	p.D816V	AML (M1,M2,M4,M4Eo, M5, MA) AMLTR MCN Dysgerminoma Seminoma Cutaneous mastocytosis
Kinase	p.D816N	NKTCL
Kinase	p.D816H	AML AMLAMDS MCN Dysgerminoma Mixed germinoma Seminoma Melanoma
Kinase	p.D816I	AML
Kinase	p.D816F	MCN
Kinase	p.D816E	GIST Seminoma
Kinase	p.D816A	Seminoma
Kinase	p.I817V	
Kinase	p.K818R	Seminoma
Kinase	p.D820G	MCN PCV Seminoma
Kinase	p.D820E	GIST
Kinase	p.D820H	GIST Seminoma
Kinase	p.D820N	NKTCL

Kinase	p.D820V	Seminoma
Kinase	p.D820Y	GIST
Kinase	p.N822Y	GIST
		Seminoma
Kinase	p.N822T	AML
Kinase	p.N822K	GIST
		Seminoma
		AML
Kinase	p.N822H	GIST
Kinase	p.Y823D	GIST
		Seminoma
		AML
		Melanoma
Kinase	p.Y823C	Seminoma
Kinase	p.Y823N	Seminoma
Kinase	p.V825A	AML
		ATCLL
		DLBCL
		NKTCL
Kinase	p.V825I	AML
		PCV
Kinase	p.A829P	Testis nonseminoma
Kinase	p.E839K	MCN
Kinase	p.D842V	PCV
		GIST
Kinase	p.L859P	PCV
Kinase	p.S868G	PCV
Kinase	p.K918E	PCV
	p.E925G	PCV
	p.V950A	PCV

Note that synonymous mutations have been excluded.

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