

Mutations in SNU-503

SNU503_LARGE_INTESTINE													
Gene	Protein Change	Chr	Start	End	NCBI Build	Variant Classification	Variant Type	Reference Allele	Tumor Allele 1	dbSNP ID	Annotation Transcript	Other Transcripts	Mutation Assessor
ADAM28		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	AM28_uc011laa...	
ALK	p.S171N	2	30143014	30143014	37	missense_Mutati	SNP	C	T		uc002rmy.2		view
ALPK1		4	113347609	113347610	37	Intron	Ins	-	T	rs68137453	uc003iap.3	LPK1_uc003ian....	
ALPK2	p.EDTST1006de	18	56204388	56204402	37	In_Frame_Del	Del	CAGTT...	-	rs44017;rs11762	uc002lhj.3	LPK2_uc002lhk....	
APC	p.K1543*	5	112175918	112175918	37	missense_Mutati	SNP	A	T		uc010jby.2	PC_uc011cvt.1_	view
ATP10A	p.R244C	15	25981213	25981213	37	missense_Mutati	SNP	G	A	rs114003800	uc010ayu.2		view
AURKC		19	57742531	57742532	37	5'UTR	Ins	-	C		uc002qoe.2	JRKC_uc002qoc....	
BRD2		6	32948607	32948608	37	3'UTR	Ins	-	T	49414;rs11419!	uc010juh.2	BD2_uc003ocn.3...	
CABC1	p.G73V	1	227152741	227152741	37	missense_Mutati	SNP	G	T		uc001hqm.1	CBC1_uc010pvp	view
CACNB2		10	18828721	18828722	37	3'UTR	Ins	-	AA	rs3841459	uc001ipr.2	CNB2_uc009xjz...	
CAS5	p.F1899L	15	40921504	40921504	37	missense_Mutati	SNP	T	C		uc010bbs.1	CAS5_uc010bbt	view
CDK11B		1	1654066	1654068	37	Intron	Del	GCG	-		uc001agv.1	K11B_uc001aha...	
CDK14	p.R452L	7	90747439	90747440	37	missense_Mutati	DNP	CG	TT	rs74839397	uc003uky.2	DK14_uc003ukz....	
CHRNA5		15	78885615	78885616	37	3'UTR	Ins	-	TA	rs72534078	uc002bdy.2	RNA3_uc002bea...	
CHRN4	p.P400S	15	78921449	78921449	37	missense_Mutati	SNP	G	A		uc002bed.1	RNB4_uc002be	view
CHUK	p.A200T	10	101978856	101978856	37	missense_Mutati	SNP	C	T		uc001kqp.2		view
CLTCL1	p.V1201_splice	22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977	uc002zpb.2	TCL1_uc011agv...	
CREB3L2	p.T100del	7	137612914	137612916	37	In_Frame_Del	Del	TGG	-		uc003vtw.2	REB3L2_uc003vt...	
CSMD3	p.G1474V	8	113563043	113563043	37	missense_Mutati	SNP	C	A		uc003ynu.2	MD3_uc003yns	view
CSMD3	p.P1047H	8	113662443	113662443	37	missense_Mutati	SNP	G	T		uc003ynu.2	MD3_uc003yns	view
CSMD3		8	114326774	114326774	37	Intron	SNP	C	A		uc003ynu.2	MD3_uc003ynt	view
CTNNA2	p.L17F	2	79878733	79878733	37	missense_Mutati	SNP	G	T		uc010ysh.1	NNA2_uc010ys	view
CUBN	p.P2706T	10	16943405	16943405	37	missense_Mutati	SNP	G	T		uc001ioo.2	JBN_uc009xjq.1	view
CYLD		16	50821016	50821016	37	Intron	SNP	T	G		uc002egp.1	LD_uc010cbs.1	view
DHER		5	79924857	79924858	37	3'UTR	Ins	-	T		uc003kgx.1	HFR_uc011ctl.1...	
DST	p.V3418_splice	6	56401739	56401739	37	Splice_Site_SNF	SNP	C	A		uc003pdf.2	ST_uc003pcz.3_	view
EIF3H		8	117670990	117670990	37	Intron	SNP	C	T		uc003yob.2	F3H_uc003yoa.	view
EIF4B	p.P83T	12	53412677	53412677	37	missense_Mutati	SNP	C	A		uc010snu.1	F4B_uc009zmp	view
EP300		22	41575030	41575031	37	3'UTR	Ins	-	T		uc003azl.3		
EPHA6		3	97367230	97367231	37	Intron	Ins	-	A	rs115253553	uc010how.1	PHA6_uc003drs....	
ERCC3	p.D60Y	2	128051145	128051145	37	missense_Mutati	SNP	C	A		uc002toh.1	CC3_uc002toe	view
ETV1		7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc011jq.1	TV1_uc011jxn.1...	
FGFR10P		6	167453519	167453520	37	3'UTR	Ins	-	T	rs115519488	uc003qvj.2	CR6_uc003qvl.2...	

FGFR2	p.A512T	10	123260370	123260370	37	issense_Mutati	SNP	C	T		uc010qtj.1	gFR2_uc010qtg	view
FLI1		11	128638217	128638217	37	Intron	Del	G	-		uc010sbu.1	LI1_uc010sbt.1...	
FMN2	p.V793M	1	240370477	240370477	37	issense_Mutati	SNP	G	A		uc010pye.1	FN2_uc010pyd.	view
FN1		2	216244182	216244185	37	Intron	Del	GTAC	-		uc002vfa.2	v1_uc002vfb.2...	
FSCB	p.E311D	14	44975258	44975258	37	issense_Mutati	SNP	C	A		uc001wvn.2		view
GLI3	p.R667L	7	42012039	42012039	37	issense_Mutati	SNP	C	A		uc011kbh.1	LI3_uc011kbg.1	view
GPR112	p.D2657del	X	135474445	135474447	37	In_Frame_Del	Del	GAT	-	rs3030295	uc004ezu.1	R112_uc010nsb...	
GPR87	p.R246Q	3	151012297	151012297	37	issense_Mutati	SNP	C	T		uc003eyt.2	ED12L_uc011bn	view
GRAMD1B	p.D51G	11	123448203	123448203	37	issense_Mutati	SNP	A	G		uc001pyw.2	AMD1B_uc001f	view
GRIA3		X	122318386	122318387	37	Splice_Site_Ins	Ins	-	G	rs66632982	uc004etq.3	RIA3_uc004etr...	
GRIN2B	p.W197L	12	13906671	13906671	37	issense_Mutati	SNP	C	A		uc001rbt.2		view
GUCY2C		12	14827464	14827466	37	Intron	Del	ATC	-	rs525754;rs3083	uc001rcd.2	JCY2C_uc009zhz...	
HOXD10	p.N5D	2	176981574	176981574	37	issense_Mutati	SNP	A	G		uc002ukj.2		view
ILK		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	K_uc001mef.2...	
ITPR2	p.R470_splice	12	26834804	26834805	37	Splice_Site_Ins	Ins	-	ACTC	696203;rs59199	uc001rhg.2		
KDM5A	p.T30A	12	498170	498170	37	issense_Mutati	SNP	T	C		uc001qif.1	M5A_uc001qie	view
LATS2		13	21620222	21620223	37	5'UTR	Ins	-	A	rs40127;rs11572	uc009zsz.2	LTS2_uc001unr....	
MAF		16	79628537	79628537	37	Intron	Del	T	-		uc002ffm.2	AF_uc002ffn.2...	
MAML3	p.Q767_splice	4	140651585	140651587	37	Splice_Site_Del	Del	CTG	-		uc003ihz.1	GST2_uc010ioi....	
MAP2K1		15	66679649	66679650	37	5'UTR	Ins	-	C	rs67229745	uc010bhq.2		
MAP3K1	p.T949del	5	56177849	56177851	37	In_Frame_Del	Del	CAA	-		uc003jqw.3		
MECOM	p.S607I	3	168833840	168833840	37	issense_Mutati	SNP	C	A		uc011bpj.1	COM_uc010hw	view
MLL3	p.E3430L	7	151860373	151860374	37	issense_Mutati	DNP	TC	AA		uc003wla.2	LL3_uc003wkz.2...	
MLLT3		9	20346378	20346378	37	3'UTR	Del	A	-		uc003zoe.2	LLT3_uc011lne....	
MRE11A	p.A595P	11	94180385	94180385	37	issense_Mutati	SNP	C	G		uc001peu.2	E11A_uc001pe	view
MSH6	p.T1355fs	2	48033981	48033982	37	Frame_Shift_Ins	Ins	-	TTGA		uc002rwd.3	SH6_uc010fbj.2...	
MYLK	p.E1066del	3	123419117	123419119	37	In_Frame_Del	Del	TTC	-	rs72491150	uc003ego.2	YLK_uc011bjw.1...	
NAV3	p.H2372Q	12	78604255	78604255	37	issense_Mutati	SNP	C	A		uc001syg.2	AV3_uc001syo.2	view
NCAM1		11	112832307	112832308	37	5'UTR	Ins	-	A	rs117108942	uc009yyq.1	AM1_uc001pno....	
NCAM1		11	112832340	112832340	37	5'UTR	Del	C	-		uc009yyq.1	AM1_uc001pno....	
NCAM1		11	113075169	113075169	37	Novo_Start_Ins	SNP	G	A		uc009yyq.1	AM1_uc001pnc	view
NEK3	p.K313_splice	13	52718050	52718051	37	Splice_Site_Ins	Ins	-	T	rs72514762	uc001vgh.2	EK3_uc001vvg.2...	
NR1H2	p.176_177insQ	19	50881820	50881821	37	In_Frame_Ins	Ins	-	AAC	rs783852;rs11235	uc010enw.2	R1H2_uc002prv....	
NUMA1	p.Q1527*	11	71723970	71723970	37	Insense_Mutati	SNP	G	A		uc001orl.1	MA1_uc009ysv	view
NUP98	p.S325P	11	3784245	3784245	37	issense_Mutati	SNP	A	G		uc001lyh.2	UP98_uc001lyi.	view
OR2L13	p.M227T	1	248263357	248263357	37	issense_Mutati	SNP	T	C		uc001ids.2		view
P2RX7	p.A185A	12	121603181	121603181	37	Silent	SNP	C	T		uc001tzm.2	P2RX7_uc001tzn.	view
P2RX7	p.E531*	12	121622408	121622408	37	Insense_Mutati	SNP	G	T		uc001tzm.2	P2RX7_uc001tzn.	view

PAX3		2	223066017	223066018	37	Intron	Ins	-	C	rs115619343	uc002vmt.1	X3_uc002vmy.1...	
PAX3		2	223158653	223158653	37	Intron	Del	T	-	rs112753212	uc002vmt.1	X3_uc002vmy.1...	
PCDH15		10	55581490	55581491	37	3'UTR	Ins	-	A		uc010qhy.1	DH15_uc010qhq...	
PCM1	p.L1288V	8	17830115	17830115	37	issense_Mutati	SNP	C	G		uc003wyi.3	M1_uc011kyh...	view
PDGFRA		4	55131002	55131002	37	Intron	Del	A	-		uc003han.3	GFRA_uc003haa...	
PDGFRB	p.K967E	5	149498315	149498315	37	issense_Mutati	SNP	T	C		uc003lro.2	IGFRB_uc010jht...	view
PDZRN4	p.G818W	12	41967630	41967630	37	issense_Mutati	SNP	G	T		uc010skn.1	ZRN4_uc001rm...	view
PGR		11	100933598	100933599	37	Intron	Ins	-	AATGA	rs35148438	uc001pgh.2	iR_uc001pgg.2...	
PINK1	p.R578S	1	20977172	20977172	37	issense_Mutati	SNP	G	C		uc001bdm.2	NK1_uc001bdn...	view
PRKD2		19	47200599	47200599	37	Intron	SNP	G	A		uc010xye.1	PKD2_uc002pfe...	view
RBPJ		4	26431822	26431822	37	Intron	Del	A	-	rs76214390	uc003grx.1	BPJ_uc003gry.1...	
RECQL4	p.R766fs	8	145738768	145738768	37	frame_Shift_De	Del	G	-		uc003zdz.2		
REL	p.T433I	2	61149108	61149108	37	issense_Mutati	SNP	C	T		uc002sam.1	EL_uc002san.1...	view
ROBO1	p.E1647D	3	78649263	78649263	37	issense_Mutati	SNP	C	A		uc003dqe.2	ROBO1_uc003dqt...	view
ROS1	p.G1415W	6	117674231	117674231	37	issense_Mutati	SNP	C	A		uc003pxp.1	OS1_uc011ebi.1...	view
SACS	p.S832P	13	23915521	23915521	37	issense_Mutati	SNP	A	G		uc001uon.2	ACS_uc001uoo.2...	view
SGK196	p.T149N	8	42977413	42977413	37	issense_Mutati	SNP	C	A		uc003xpw.2		view
STAT1	p.R304H	2	191859820	191859820	37	issense_Mutati	SNP	C	T		uc002usj.2	STAT1_uc010fse...	view
STK31		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sws.3	STK31_uc003swt...	
TAOK3		12	118588761	118588762	37	3'UTR	Ins	-	T	rs116872184	uc001twx.2	OK3_uc001twv...	
TEC		4	48139307	48139310	37	3'UTR	Del	TTAT	-	rs79098351	uc003gxz.2		
TNK2		3	195596501	195596502	37	Intron	Ins	-	AG	rs79861797	uc003ftv.1	NK2_uc003fvq.1...	
TNK2	p.R326L	3	195609021	195609021	37	issense_Mutati	SNP	C	A		uc003ftv.1	NK2_uc003fvs.1...	view
TNN	p.Y381H	1	175052978	175052978	37	issense_Mutati	SNP	T	C		uc001gkl.1	TNN_uc010pmx.1...	view
TP53	p.R273L	17	7577120	7577120	37	issense_Mutati	SNP	C	A	rs28934576	uc002gim.2	TP53_uc002gig.1...	view
TP63		3	189507518	189507519	37	Intron	Ins	-	AGAG	702062;rs79714	uc003fry.2	TP63_uc003frx.2...	
TRIM33		1	114940215	114940216	37	3'UTR	Ins	-	G	rs68133208	uc001eew.2	TRIM33_uc010owr...	
TRIM37		17	57076621	57076621	37	Intron	SNP	G	A		uc002iwy.3	TRIM37_uc002iw...	view
TRPS1		8	116635872	116635873	37	5'UTR	Ins	-	A	rs76893349	uc003yny.2	TRPS1_uc011lhy...	

