

## Mutations in SW480

SW480\_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
<a href="#">ADAM12</a>	p.P168L	10	127806716	127806716	37	issense_Mutati	SNP	G	A		uc001ljk.2	AM12_uc010q	<a href="#">view</a>
<a href="#">ADAM17</a>		2	9630245	9630245	37	3'UTR	Del	T	-		uc002qzu.2	AM17_uc010yiz.1...	
<a href="#">ADAM28</a>		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	AM28_uc011aa...	
<a href="#">ADAM32</a>	p.I662T	8	39112015	39112015	37	issense_Mutati	SNP	T	C		uc003xmt.3	AM32_uc011lc	<a href="#">view</a>
<a href="#">AKAP12</a>	p.L1531_1532ins	6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819	uc011eep.1	AP12_uc003qoe...	
<a href="#">AKAP6</a>	p.G193_splice	14	33014434	33014434	37	Splice_Site_SNP	SNP	A	C		uc001wrq.2	AKAP6_uc010aml	<a href="#">view</a>
<a href="#">AKAP9</a>	p.S3861F	7	91737843	91737843	37	issense_Mutati	SNP	C	T		uc003ulg.2	AKAP9_uc003ulf	<a href="#">view</a>
<a href="#">ALPK2</a>	p.EDTST1006de	18	56204388	56204402	37	In_Frame_Del	Del	CAGTT...	-	rs44017;rs11762	uc002lhj.3	ALPK2_uc002lhk...	
<a href="#">APC</a>	p.Q1338*	5	112175303	112175303	37	insense_Mutati	SNP	C	T		uc010jby.2	APC_uc011cvt.1	<a href="#">view</a>
<a href="#">ARFGEF1</a>	p.A983V	8	68152428	68152428	37	issense_Mutati	SNP	G	A		uc003xxo.1	ARFGEF1_uc003x	<a href="#">view</a>
<a href="#">ATM</a>	p.R2461P	11	108201015	108201015	37	issense_Mutati	SNP	G	C		uc001pkb.1	ATM_uc009yxr.1	<a href="#">view</a>
<a href="#">AURKC</a>		19	57742494	57742507	37	5'UTR	Del	GGACG...	-	rs112039303;rs112039304	uc002qoe.2	AURKC_uc002qoc...	
<a href="#">BCL9</a>	p.P514fs	1	147091501	147091501	37	In_Frame_Shift_De	Del	C	-		uc001epq.2	BCL9_uc010ozr.1...	
<a href="#">BTG1</a>		12	92537706	92537707	37	3'UTR	Ins	-	T	rs33983967	uc001tby.3	BTG1_uc001tbv.1...	
<a href="#">C15ORF2</a>	p.P462L	15	24922399	24922399	37	issense_Mutati	SNP	C	T		uc001ywo.2		<a href="#">view</a>
<a href="#">CARD10</a>	p.KE272del	22	37906309	37906314	37	In_Frame_Del	Del	CTCCT...	-		uc003asx.1	CARD10_uc003ast...	
<a href="#">CDK11B</a>		1	1654066	1654068	37	Intron	Del	GCG	-		uc001agv.1	CDK11B_uc001aha...	
<a href="#">CHD9</a>	p.R1784C	16	53326804	53326804	37	issense_Mutati	SNP	C	T		uc002ehb.2	CHD9_uc002egy.2	<a href="#">view</a>
<a href="#">CNTN5</a>		11	99426889	99426889	37	5'UTR	SNP	G	A		uc001pga.2	CNTN5_uc009yvw	<a href="#">view</a>
<a href="#">COL3A1</a>	p.R1432*	2	189876393	189876393	37	insense_Mutati	SNP	C	T		uc002uqj.1		<a href="#">view</a>
<a href="#">CSNK1A1L</a>		13	37678315	37678315	37	3'UTR	SNP	A	G		uc001uwm.1		<a href="#">view</a>
<a href="#">CSNK1G3</a>	p.V102I	5	122909101	122909101	37	issense_Mutati	SNP	G	A		uc003ktl.2	CSNK1G3_uc003k	<a href="#">view</a>
<a href="#">CTBP2</a>	p.K974fs	10	126678123	126678123	37	In_Frame_Shift_De	Del	T	-		uc001lie.3	CTBP2_uc009yak...	
<a href="#">CXCL12</a>		10	44873992	44873993	37	Intron	Ins	-	CT	rs117617571	uc001jbh.2	CXCL12_uc001jbf...	
<a href="#">DBF4</a>	p.E653*	7	87537410	87537410	37	insense_Mutati	SNP	G	T		uc003ujf.1	DBF4_uc003ujh.1	<a href="#">view</a>
<a href="#">DCLK3</a>	p.S492F	3	36763128	36763128	37	issense_Mutati	SNP	G	A	rs34904965	uc003cqi.2		<a href="#">view</a>
<a href="#">DNAH8</a>	p.A3677V	6	38905867	38905867	37	issense_Mutati	SNP	C	T		uc003ooe.1	DNAH8_uc003ooe	<a href="#">view</a>
<a href="#">DYRK1A</a>		21	38884966	38884967	37	3'UTR	Ins	-	T	rs72430212	uc002ywk.2	DYRK1A_uc002ywi...	
<a href="#">ECM1</a>	p.E172K	1	150483393	150483393	37	issense_Mutati	SNP	G	A		uc001euu.2	ECM1_uc010pce.	<a href="#">view</a>
<a href="#">EP300</a>		22	41574970	41574972	37	3'UTR	Del	GTA	-	rs76489032	uc003azl.3		
<a href="#">EPHA8</a>	p.G936V	1	22927870	22927870	37	issense_Mutati	SNP	G	T		uc001bfx.1		<a href="#">view</a>
<a href="#">EPS15</a>	p.L146R	1	51929409	51929409	37	issense_Mutati	SNP	A	C		uc001csq.1	EPS15_uc009vyz.	<a href="#">view</a>
<a href="#">ERBB2</a>	p.E1229*	17	37884214	37884214	37	insense_Mutati	SNP	G	T		uc002hso.2	ERBB2_uc002hsm	<a href="#">view</a>

<a href="#">ESR2</a>	p.S39R	14	64749589	64749589	37	issense_Mutati	SNP	T	G		uc001xha.1	R2_uc001xgu.2	<a href="#">view</a>
<a href="#">FBN2</a>	p.A750_splice	5	127704874	127704874	37	Splice_Site_SNF	SNP	C	T		uc003kuu.2	N2_uc003kuv.2	<a href="#">view</a>
<a href="#">FGFR1OP</a>		6	167453519	167453520	37	3'UTR	Ins	-	T	rs115519488	uc003qvj.2	R6_uc003qvl.2...	
<a href="#">FGFR3</a>		4	1809128	1809129	37	3'UTR	Del	GT	-		uc003gdu.2	FR3_uc003gdr....	
<a href="#">FLI1</a>		11	128638217	128638217	37	Intron	Del	G	-		uc010sbu.1	L1_uc010sbt.1...	
<a href="#">FMN2</a>	p.I1668N	1	240601441	240601441	37	issense_Mutati	SNP	T	A		uc010pye.1	N2_uc010pyd.	<a href="#">view</a>
<a href="#">FN1</a>		2	216244182	216244185	37	Intron	Del	GTAC	-		uc002vfa.2	N1_uc002vfb.2_...	
<a href="#">GLI3</a>	p.A1139T	7	42005256	42005256	37	issense_Mutati	SNP	C	T		uc011kbh.1	L3_uc011kbg.1	<a href="#">view</a>
<a href="#">GPC3</a>		X	132888208	132888208	37	Intron	Del	A	-		uc010nrm.1	C3_uc004exd.1...	
<a href="#">GRAMD1B</a>	p.T290S	11	123476139	123476139	37	issense_Mutati	SNP	A	T		uc001pyw.2	AMD1B_uc001p...	<a href="#">view</a>
<a href="#">GRIA3</a>		X	122318386	122318387	37	Splice_Site_Ins	Ins	-	G	rs66632982	uc004etq.3	RIA3_uc004etr....	
<a href="#">GUCY2C</a>		12	14827464	14827466	37	Intron	Del	ATC	-	rs525754;rs3083	uc001rcd.2	JCY2C_uc009zhz...	
<a href="#">HDAC2</a>		6	114262093	114262093	37	3'UTR	SNP	G	T		uc003pwd.1	A2_uc003pwc	<a href="#">view</a>
<a href="#">HIF1A</a>		14	62213929	62213930	37	3'UTR	Ins	-	T	rs67890419	uc001xfs.2	F1A_uc001xfq....	
<a href="#">HNF1A</a>		12	121434630	121434631	37	Intron	Ins	-	TCATTCAT	rs75100977	uc010sxn.1	F1A_uc001tze....	
<a href="#">IL1RAP</a>	p.A43S	3	190321979	190321979	37	issense_Mutati	SNP	G	T		uc003fsq.2	1RAP_uc003fsk	<a href="#">view</a>
<a href="#">ILK</a>		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	K_uc001mef.2_...	
<a href="#">ISG15</a>	p.A40T	1	949478	949478	37	issense_Mutati	SNP	G	A		uc001acj.3		<a href="#">view</a>
<a href="#">ITPR2</a>	p.R470_splice	12	26834804	26834805	37	Splice_Site_Ins	Ins	-	ACTC	rs696203;rs59199	uc001rhg.2		
<a href="#">KIAA1804</a>	p.K497N	1	233497978	233497978	37	issense_Mutati	SNP	G	T		uc001hvt.3	AA1804_uc001f...	<a href="#">view</a>
<a href="#">KRAS</a>	p.G12V	12	25398284	25398284	37	issense_Mutati	SNP	C	A		uc001rgp.1	RAS_uc001rgq.1	<a href="#">view</a>
<a href="#">LAMA1</a>	p.L405fs	18	7042192	7042192	37	Frame_Shift_De	Del	G	-		uc002knm.2	MA1_uc010wzj....	
<a href="#">LRGUK</a>	p.R528C	7	133884008	133884008	37	issense_Mutati	SNP	C	T		uc003vrm.1		<a href="#">view</a>
<a href="#">LRP1B</a>	p.C2993F	2	141253190	141253190	37	issense_Mutati	SNP	C	A		uc002tvj.1		<a href="#">view</a>
<a href="#">LRP2</a>	p.L2204H	2	170063619	170063619	37	issense_Mutati	SNP	A	T		uc002ues.2		<a href="#">view</a>
<a href="#">LRRK2</a>	p.I1260_splice	12	40699585	40699585	37	Splice_Site_SNF	SNP	A	C		uc001rmg.3	RK2_uc009zjw.	<a href="#">view</a>
<a href="#">MAP3K1</a>	p.T949del	5	56177849	56177851	37	In_Frame_Del	Del	CAA	-		uc003jqw.3		
<a href="#">MAP3K14</a>	p.R218_splice	17	43364293	43364294	37	Splice_Site_Ins	Ins	-	G	rs66766023	uc002iiv.1	AP3K14_uc010da...	
<a href="#">MAPK11</a>		22	50705157	50705157	37	Intron	SNP	C	T		uc003bkr.2	APK11_uc010ha	<a href="#">view</a>
<a href="#">MLLT6</a>		17	36881873	36881873	37	3'UTR	Del	C	-		uc002hqi.3	LLT6_uc002hqq....	
<a href="#">MSH3</a>	p.PPA66del	5	79950742	79950750	37	In_Frame_Del	Del	CCCCC...	-	rs484572;rs1047	uc003kgz.2	HFR_uc011ctl.1...	
<a href="#">MSH6</a>	p.E221D	2	48025785	48025785	37	issense_Mutati	SNP	A	C	rs41557217	uc002rwd.3	SH6_uc002rwc...	<a href="#">view</a>
<a href="#">MSMB</a>	p.W112*	10	51562390	51562390	37	insense_Mutati	SNP	G	A		uc001jiq.2	ARG_uc001jih.2	<a href="#">view</a>
<a href="#">MTUS2</a>	p.P578T	13	29600537	29600537	37	issense_Mutati	SNP	C	A		uc001usl.3		<a href="#">view</a>
<a href="#">MYB</a>	p.Q560*	6	135520157	135520157	37	insense_Mutati	SNP	C	T		uc003qfh.2	YB_uc003qfc.2_...	<a href="#">view</a>
<a href="#">MYH11</a>	p.A1958A	16	15797914	15797914	37	Silent	SNP	T	C		uc002ddx.2	H11_uc002ddv...	<a href="#">view</a>
<a href="#">MYST4</a>	p.L1104_1105ins	10	76781905	76781906	37	In_Frame_Ins	Ins	-	GAA	rs111976843	uc001jwn.1	ST4_uc001jwm....	
<a href="#">NAV3</a>		12	78604356	78604356	37	3'UTR	SNP	G	A		uc001syv.2	AV3_uc001syo.2	<a href="#">view</a>

<a href="#">NCAM1</a>		11	112832307	112832308	37	5'UTR	Ins	-	A	rs117108942	uc009yyq.1	AM1_uc001pno....	
<a href="#">NCAM1</a>		11	112832340	112832340	37	5'UTR	Del	C	-		uc009yyq.1	AM1_uc001pno....	
<a href="#">NCOA3</a>	p.QQ1272del	20	46279815	46279823	37	In_Frame_Del	Del	GCAGC...	-		uc002xtk.2	COA3_uc010ght...	
<a href="#">NEK3</a>	p.K313_splice	13	52718050	52718051	37	Splice_Site_Ins	Ins	-	T	rs72514762	uc001vgh.2	EK3_uc001vvg.2...	
<a href="#">NEK6</a>		9	127113415	127113415	37	3'UTR	SNP	G	A		uc004boh.2	EK6_uc004bof.2	<a href="#">view</a>
<a href="#">NFIA</a>		1	61921185	61921186	37	3'UTR	Ins	-	T	270124;rs7613:	uc010oos.1	FIA_uc001czy.2...	
<a href="#">NPR1</a>		1	153665919	153665920	37	3'UTR	Ins	-	C	rs66682713	uc001fcs.3	PR1_uc010pdz.1...	
<a href="#">NR1H2</a>	p.176_177insQ	19	50881822	50881823	37	In_Frame_Ins	Ins	-	CAG	1751954;rs1052	uc010enw.2	NR1H2_uc002prv....	
<a href="#">NTRK2</a>		9	87636649	87636649	37	3'UTR	SNP	C	T		uc004anz.1	NRK2_uc004aoa	<a href="#">view</a>
<a href="#">NYNRIN</a>	p.P114L	14	24877217	24877217	37	issense_Mutati	SNP	C	T		uc001wpf.3		<a href="#">view</a>
<a href="#">PAX3</a>		2	223158653	223158653	37	Intron	Del	T	-	rs112753212	uc002vmt.1	PAX3_uc002vmy.1...	
<a href="#">PGGT1B</a>		5	114548071	114548072	37	3'UTR	Ins	-	C		uc003kqw.3	PGT1B_uc003kqx...	
<a href="#">PIK3C2G</a>		12	18800990	18800990	37	3'UTR	Del	A	-	rs16914368	uc010sib.1	PIK3C2G_uc001rd...	
<a href="#">PIKFYVE</a>	p.K1454*	2	209200620	209200620	37	insense_Mutati	SNP	A	T		uc002vcz.2	KFYVE_uc002vc	<a href="#">view</a>
<a href="#">PKHD1</a>		6	51949755	51949755	37	5'UTR	SNP	T	G		uc003pah.1	PKHD1_uc003pai	<a href="#">view</a>
<a href="#">PPARD</a>	p.R258Q	6	35392251	35392251	37	issense_Mutati	SNP	G	A		uc003okm.2	PPARD_uc003okl	<a href="#">view</a>
<a href="#">PRKDC</a>	p.L1244fs	8	48805816	48805817	37	In_Frame_Shift_In	Ins	-	G	rs67588121	uc003xqi.2	PRKDC_uc003xqi....	
<a href="#">PRRX1</a>		1	170705428	170705431	37	3'UTR	Del	ACAA	-		uc001ghf.2	PRRX1_uc001ghe....	
<a href="#">RAPGEF2</a>		4	160189200	160189201	37	5'UTR	Ins	-	T	rs72011511	uc003iqg.3		
<a href="#">RASGRF1</a>	p.R220Q	15	79339307	79339307	37	issense_Mutati	SNP	C	T		uc002beq.2	RASGRF1_uc002b	<a href="#">view</a>
<a href="#">RECQL4</a>	p.R766fs	8	145738768	145738768	37	In_Frame_Shift_De	Del	G	-		uc003zjd.2		
<a href="#">RHPN2</a>	p.Q30fs	19	33535248	33535251	37	In_Frame_Shift_De	Del	GTTT	-		uc002nuf.2	RHPN2_uc010xro....	
<a href="#">RIMS2</a>	p.E310K	8	104898421	104898421	37	issense_Mutati	SNP	G	A		uc003yls.2	RIMS2_uc003ylp.	<a href="#">view</a>
<a href="#">RIMS2</a>		8	105264087	105264088	37	3'UTR	Ins	-	A	rs78110727	uc003yls.2	RIMS2_uc003ylp....	
<a href="#">RIOK1</a>	p.R534S	6	7417569	7417569	37	issense_Mutati	SNP	A	C		uc003mxn.2	RIOK1_uc003mxo	<a href="#">view</a>
<a href="#">RIPK2</a>	p.P228A	8	90784009	90784009	37	issense_Mutati	SNP	C	G		uc003yee.2	RIPK2_uc003yef.	<a href="#">view</a>
<a href="#">ROS1</a>	p.P1136L	6	117681543	117681543	37	issense_Mutati	SNP	G	A		uc003pxp.1	ROS1_uc011ebi.1	<a href="#">view</a>
<a href="#">ROS1</a>	p.L936Q	6	117687244	117687244	37	issense_Mutati	SNP	A	T		uc003pxp.1	ROS1_uc011ebi.1	<a href="#">view</a>
<a href="#">RPS6KA5</a>		14	91360733	91360734	37	Intron	Ins	-	T	rs77438853	uc001xys.2	RPS6KA5_uc010tw...	

