

### Mutations in SJSA-1

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="#">AAK1</a>	p.541_542QQ>Q	2	69741754	69741756	37	In_Frame_Del	Del	TGT	-	rs55712143	uc002sfp.2	AAK1_uc010fdk.2...	
<a href="#">ADAM17</a>		2	9630245	9630245	37	3'UTR	Del	T	-		uc002qzu.2	IAH1_uc010yiz.1...	
<a href="#">ADAM28</a>		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	ADAM28_uc011laa...	
<a href="#">AFF3</a>	p.R1099Q	2	100175401	100175401	37	Missense_Mutation	SNP	C	T		uc002taf.2	AFF3_uc002tag.2...	<a href="#">view</a>
<a href="#">AKAP12</a>	p.1531_1532insE	6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819	uc011eep.1	AKAP12_uc003qoe...	
<a href="#">AKAP9</a>	p.1335_1336insQ	7	91652178	91652179	37	In_Frame_Ins	Ins	-	AAC	rs34756483	uc003ulg.2	AKAP9_uc003ule...	
<a href="#">ASTN1</a>	p.V1082M	1	176852113	176852113	37	Missense_Mutation	SNP	C	T		uc001glc.2	ASTN1_uc001glb...	<a href="#">view</a>
<a href="#">AURKC</a>		19	57742494	57742507	37	5'UTR	Del	GGACG...	-	:67926438;rs112039303;rs5909495	uc002qoe.2	AURKC_uc002qoc...	
<a href="#">BMP4</a>		14	54416601	54416602	37	3'UTR	Ins	-	TT	rs74495140;rs76335800	uc001xal.3	BMP4_uc010aoh.2...	
<a href="#">BMPR1A</a>	p.T33S	10	88649849	88649849	37	Missense_Mutation	SNP	C	G		uc001kdy.2		<a href="#">view</a>
<a href="#">BRCA2</a>		13	32973072	32973073	37	3'UTR	Ins	-	G		uc001uub.1		
<a href="#">CACNA1F</a>	p.815_815E>EE	X	49076224	49076225	37	In_Frame_Ins	Ins	-	TCC		uc004dmb.2	CACNA1F_uc010ni...	
<a href="#">CACNB2</a>		10	18828721	18828722	37	3'UTR	Ins	-	AA	rs3841459	uc001ipr.2	CACNB2_uc009xjz...	
<a href="#">CCR6</a>		6	167417112	167417112	37	5'UTR	Del	C	-		uc003qvl.2	FGFR1OP_uc011eg...	
<a href="#">CDC73</a>		1	193091168	193091168	37	5'UTR	SNP	C	T		uc001gtb.2		<a href="#">view</a>
<a href="#">CHD1</a>	p.P1684del	5	98192165	98192167	37	In_Frame_Del	Del	AGG	-	rs79267787;rs61749618	uc003knf.2	CHD1_uc010jbn.2...	
<a href="#">CLTCL1</a>	p.V1201_splice	22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977	uc002zpb.2	CLTCL1_uc011agv...	
<a href="#">CNTN6</a>		3	1339681	1339686	37	Intron	Del	GTTTT...	-		uc003boz.2	CNTN6_uc010hbo...	
<a href="#">CREB3L2</a>	p.T100del	7	137612914	137612916	37	In_Frame_Del	Del	TGG	-		uc003vtw.2	CREB3L2_uc003vt...	
<a href="#">CYFIP1</a>	p.Q222_splice	15	22933659	22933660	37	Splice_Site_Ins	Ins	-	AAGG		uc001yus.2	CYFIP1_uc001yut...	
<a href="#">DDX10</a>	p.788_789insDD	11	108788634	108788635	37	In_Frame_Ins	Ins	-	TGATGA		uc001pkm.2	DDX10_uc001pkl...	
<a href="#">EDN1</a>		6	12290729	12290729	37	5'UTR	Del	A	-		uc003nae.3	EDN1_uc010jpb.2...	
<a href="#">EIF4G1</a>	p.EE471del	3	184039744	184039749	37	In_Frame_Del	Del	GAAGA...	-		uc010hxx.2	EIF4G1_uc003fno...	
<a href="#">EP300</a>	p.T891P	22	41546056	41546056	37	Missense_Mutation	SNP	A	C		uc003azl.3		<a href="#">view</a>
<a href="#">EPHA6</a>		3	97367230	97367231	37	Intron	Ins	-	A	rs115253553	uc010how.1	EPHA6_uc003drs...	
<a href="#">EPHB6</a>		7	142568771	142568771	37	3'UTR	Del	C	-		uc011kst.1	EPHB6_uc011ksu...	
<a href="#">ERBB3</a>	p.E173Q	12	56480410	56480410	37	Missense_Mutation	SNP	G	C		uc001sjh.2	ERBB3_uc009zpj...	<a href="#">view</a>
<a href="#">ESR2</a>		14	64694154	64694154	37	3'UTR	SNP	A	C		uc001xgu.2	ESR2_uc001xgv.2...	<a href="#">view</a>
<a href="#">ESR2</a>	p.Q393K	14	64716312	64716312	37	Missense_Mutation	SNP	G	T		uc001xha.1	ESR2_uc001xgu.2...	<a href="#">view</a>
<a href="#">ETV1</a>		7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc011jq.1	ETV1_uc011jxn.1...	
<a href="#">FGFR1OP</a>		6	167453519	167453520	37	3'UTR	Ins	-	T	rs115519488	uc003qvj.2	CCR6_uc003qvl.2...	
<a href="#">FGFR3</a>		4	1809128	1809129	37	3'UTR	Del	GT	-		uc003gdu.2	FGFR3_uc003gdr...	
<a href="#">FLI1</a>		11	128638217	128638217	37	Intron	Del	G	-		uc010sbu.1	FLI1_uc010sbt.1...	
<a href="#">FLT1</a>	p.N425H	13	29001892	29001892	37	Missense_Mutation	SNP	T	G		uc001usb.3	FLT1_uc010aar.1...	<a href="#">view</a>
<a href="#">FMN2</a>	p.917_918insPP	1	240370839	240370840	37	In_Frame_Ins	Ins	-	CCCCCT		uc010pye.1	FMN2_uc010pyd.1...	
<a href="#">FOXP1</a>		3	71008341	71008342	37	3'UTR	Ins	-	T	rs11433724;rs114233617	uc003dol.2	FOXP1_uc003dom...	
<a href="#">FURIN</a>	p.S758F	15	91424996	91424996	37	Missense_Mutation	SNP	C	T		uc002bpu.1	FES_uc010uqj.1...	<a href="#">view</a>
<a href="#">GPC3</a>	p.L157V	X	132888072	132888072	37	Missense_Mutation	SNP	A	C		uc010nm.1	GPC3_uc004exd.1...	<a href="#">view</a>
<a href="#">GPR112</a>	p.D2657del	X	135474445	135474447	37	In_Frame_Del	Del	GAT	-	rs3030295	uc004ezu.1	GPR112_uc010nsb...	
<a href="#">GRIA3</a>		X	122318386	122318387	37	Splice_Site_Ins	Ins	-	G	rs66632982	uc004etq.3	GRIA3_uc004etr...	
<a href="#">GUCY2C</a>		12	14827464	14827466	37	Intron	Del	ATC	-	rs72525754;rs3083879	uc001rcd.2	GUCY2C_uc009zhz...	
<a href="#">HDAC4</a>		2	240048387	240048387	37	Intron	SNP	C	G		uc002vyk.3	HDAC4_uc010fyz...	<a href="#">view</a>
<a href="#">HIF1A</a>		14	62213929	62213930	37	3'UTR	Ins	-	T	rs67890419	uc001xfs.2	HIF1A_uc001xfq...	
<a href="#">HSP90AA1</a>		14	102549281	102549281	37	Intron	SNP	A	G		uc001ykv.3	HSP90AA1_uc001y...	<a href="#">view</a>
<a href="#">ILK</a>		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	ILK_uc001mef.2...	
<a href="#">KALRN</a>		3	124393482	124393482	37	Intron	SNP	C	T		uc003ehg.2	KALRN_uc003ehi...	<a href="#">view</a>
<a href="#">KLK7</a>		19	51480672	51480675	37	3'UTR	Del	GGTT	-		uc002puo.2	KLK7_uc002pup.2...	

<a href="#">LRRC7</a>	p.S90N	1	70257805	70257805	37	Missense_Mutation	SNP	G	A		uc001dep.2	LRRC7_uc001deo....	<a href="#">view</a>
<a href="#">MAGEB16</a>	p.P8T	X	35820335	35820335	37	Missense_Mutation	SNP	C	A		uc010ngt.1		<a href="#">view</a>
<a href="#">MAMDC4</a>	p.P99fs	9	139747878	139747881	37	Frame_Shift_Del	Del	CTCA	-		uc004cjs.2	MAMDC4_uc011mej...	
<a href="#">MAML2</a>	p.604_607QQQ>Q	11	95825375	95825383	37	In_Frame_Del	Del	TGCTG...	-	rs113349418	uc001pfi.1		
<a href="#">MAML3</a>	p.Q767_splice	4	140651585	140651587	37	Splice_Site_Del	Del	CTG	-		uc003ihz.1	MGST2_uc010ioi....	
<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	uc002iiw.1	MAP3K14_uc010da...	
<a href="#">MAP3K4</a>	p.A1199del	6	161519351	161519353	37	In_Frame_Del	Del	CTG	-		uc003qtn.2	MAP3K4_uc010kkc...	
<a href="#">MAPK8IP2</a>		22	51049312	51049312	37	3'UTR	SNP	A	G		uc003bmx.2	MAPK8IP2_uc003b...	<a href="#">view</a>
<a href="#">MCL1</a>		1	150549800	150549801	37	3'UTR	Ins	-	TGGTGG		uc001euz.2	MCL1_uc010pch.1...	
<a href="#">MDM2</a>	p.E333D	12	69233134	69233134	37	Missense_Mutation	SNP	G	T		uc001sui.2	MDM2_uc009zri.2...	<a href="#">view</a>
<a href="#">MKL1</a>	p.D522H	22	40814878	40814878	37	Missense_Mutation	SNP	C	G		uc003ayv.1	MKL1_uc003ayw.1...	<a href="#">view</a>
<a href="#">MLLT6</a>		17	36881873	36881873	37	3'UTR	Del	C	-		uc002hqi.3	MLLT6_uc002hqq....	
<a href="#">MN1</a>	p.G1019R	22	28193477	28193477	37	Missense_Mutation	SNP	C	T		uc003adj.2		<a href="#">view</a>
<a href="#">MSI2</a>		17	55709379	55709380	37	Intron	Ins	-	A	rs60931600;rs28700576	uc002iuz.1	MSI2_uc010wnm.1...	
<a href="#">MTUS2</a>		13	30002788	30002790	37	Intron	Del	CTC	-		uc001usl.3	MTUS2_uc001usm....	
<a href="#">MYH10</a>		17	8379034	8379034	37	3'UTR	SNP	G	A		uc002glm.2	MYH10_uc002gll....	<a href="#">view</a>
<a href="#">MYH11</a>	p.T1553M	16	15814850	15814850	37	Missense_Mutation	SNP	G	A		uc002ddx.2	MYH11_uc002ddv....	<a href="#">view</a>
<a href="#">MYO18B</a>	p.Q1485fs	22	26287291	26287292	37	Frame_Shift_Ins	Ins	-	A		uc003abz.1	MYO18B_uc003aca...	
<a href="#">MYST4</a>	p.E1104del	10	76781906	76781908	37	In_Frame_Del	Del	GAA	-	rs79644703	uc001jwn.1	MYST4_uc001jwm....	
<a href="#">MYST4</a>		10	76790936	76790936	37	3'UTR	Del	A	-		uc001jwn.1	MYST4_uc001jwo....	
<a href="#">NCAM1</a>		11	112832307	112832308	37	5'UTR	Ins	-	A	rs117108942	uc009yyq.1	NCAM1_uc001pno....	
<a href="#">NCAM1</a>		11	112832340	112832340	37	5'UTR	Del	C	-		uc009yyq.1	NCAM1_uc001pno....	
<a href="#">NLRP3</a>		1	247582066	247582066	37	5'UTR	SNP	C	T		uc001icr.2	NLRP3_uc001ics....	<a href="#">view</a>
<a href="#">NLRP3</a>		1	247611982	247611982	37	3'UTR	Del	C	-		uc001icr.2	NLRP3_uc001ics....	
<a href="#">NOTCH2</a>		1	120457918	120457918	37	3'UTR	SNP	G	A		uc001eik.2		<a href="#">view</a>
<a href="#">NR1H2</a>	p.176_177insQ	19	50881820	50881821	37	In_Frame_Ins	Ins	-	AAC	rs66783852;rs112359270	uc010enw.2	NR1H2_uc002prv....	
<a href="#">NTRK3</a>		15	88420115	88420115	37	3'UTR	SNP	G	C		uc002bme.1	NTRK3_uc002bmh....	<a href="#">view</a>
<a href="#">NUP153</a>		6	17616285	17616286	37	3'UTR	Ins	-	CAC		uc011dje.1	NUP153_uc003ncd...	
<a href="#">PAX3</a>		2	223066017	223066018	37	Intron	Ins	-	C	rs115619343	uc002vmt.1	PAX3_uc002vmy.1...	
<a href="#">PAX3</a>		2	223158653	223158653	37	Intron	Del	T	-	rs112753212	uc002vmt.1	PAX3_uc002vmy.1...	
<a href="#">PCM1</a>	p.T1279R	8	17830089	17830089	37	Missense_Mutation	SNP	C	G		uc003wyi.3	PCM1_uc011kyh.1...	<a href="#">view</a>
<a href="#">PCNA</a>		20	5099019	5099019	37	Intron	SNP	A	G		uc002wlp.2	PCNA_uc002wlp.2...	<a href="#">view</a>
<a href="#">PIK3C2G</a>		12	18800990	18800990	37	3'UTR	Del	A	-	rs16914368	uc010sib.1	PIK3C2G_uc001rd...	
<a href="#">PIK3R1</a>		5	67587949	67587949	37	Intron	SNP	T	C		uc003jva.2	PIK3R1_uc003jvb...	<a href="#">view</a>
<a href="#">PINK1</a>	p.A443V	1	20975564	20975564	37	Missense_Mutation	SNP	C	T		uc001bdm.2	PINK1_uc001bdn....	<a href="#">view</a>
<a href="#">PRKDC</a>	p.L1244fs	8	48805816	48805817	37	Frame_Shift_Ins	Ins	-	G	rs67588121	uc003xqi.2	PRKDC_uc003xqi....	
<a href="#">RAPGEF2</a>		4	160189200	160189201	37	5'UTR	Ins	-	T	rs72011511	uc003iqg.3		
<a href="#">RECQL4</a>	p.R766fs	8	145738768	145738768	37	Frame_Shift_Del	Del	G	-		uc003zdz.2		
<a href="#">RFC4</a>	p.G284R	3	186508147	186508147	37	Missense_Mutation	SNP	C	G		uc003fqz.2	RFC4_uc011bsc.1...	<a href="#">view</a>
<a href="#">RHOA</a>	p.V43A	3	49412895	49412895	37	Missense_Mutation	SNP	A	G		uc003cwu.2	RHOA_uc010hku.2...	<a href="#">view</a>
<a href="#">RIF1</a>	p.1837_1839EIC>G	2	152321544	152321549	37	In_Frame_Del	Del	AAATT...	-		uc002txm.2	RIF1_uc002txl.2...	
<a href="#">RIPK3</a>		14	24807831	24807831	37	Intron	Del	A	-		uc001wpb.2	RIPK3_uc001wpa....	
<a href="#">ROBO2</a>		3	77666937	77666937	37	Intron	Del	A	-	rs78745603	uc003dpz.2	ROBO2_uc003dpy....	
<a href="#">ROBO2</a>	p.W1024R	3	77651564	77651564	37	Missense_Mutation	SNP	T	C		uc003dpz.2	ROBO2_uc003dpy....	<a href="#">view</a>
<a href="#">ROCK2</a>	p.V1048I	2	11337789	11337789	37	Missense_Mutation	SNP	C	T		uc002rbd.1		<a href="#">view</a>
<a href="#">SCD5</a>		4	83552397	83552397	37	3'UTR	SNP	G	A		uc003hna.2		<a href="#">view</a>
<a href="#">SGK269</a>	p.N118D	15	77473917	77473917	37	Missense_Mutation	SNP	T	C		uc002bcm.2	SGK269_uc002bcn...	<a href="#">view</a>
<a href="#">SOS1</a>		2	39212848	39212848	37	3'UTR	SNP	A	T		uc002rrk.3	SOS1_uc002rrj.3...	<a href="#">view</a>
<a href="#">SOS1</a>	p.R497Q	2	39250079	39250079	37	Missense_Mutation	SNP	C	T		uc002rrk.3	SOS1_uc010ynr.1...	<a href="#">view</a>

<a href="#">SP1</a>	p.G572C	12	53800407	53800407	37	Missense_Mutation	SNP	G	T		uc001scw.2	SP1_uc010sog.1_...	<a href="#">view</a>
<a href="#">SRPK1</a>		6	35802992	35802992	37	3'UTR	Del	G	-		uc003olj.2	SRPK1_uc011dtg...	
<a href="#">STK31</a>		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sws.3	STK31_uc003swt...	