

**Mutations in SF-295**

SF295\_CENTRAL\_NERVOUS\_SYSTEM

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="#">ADAM28</a>		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	ADAM28_uc011laa...	
<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="#">ALPK2</a>	p.EDTST1006del	18	56204388	56204402	37	In_Frame_Del	Del	CAGTT...	-	rs44017;rs11762	uc002lhj.3	ALPK2_uc002lhc....	
<a href="#">ANAPC5</a>		12	121790193	121790195	37	5'UTR	Del	CAC	-		uc001uag.2	ANAPC5_uc001uah...	
<a href="#">ARID1A</a>		1	27107263	27107264	37	3'UTR	Ins	-	C	rs3841356	uc001bmv.1	ARID1A_uc001bmu...	
<a href="#">ASPH</a>	p.A264_splice	8	62550924	62550925	37	Splice_Site_Ins	Ins	-	A	rs5891816	uc003xuj.2	ASPH_uc011leg.1...	
<a href="#">AXL</a>	p.Y703*	19	41762429	41762429	37	Nonsense_Mutation	SNP	C	A		uc010ehj.2	CYP2F1_uc010xvw...	<a href="#">view</a>
<a href="#">BRDT</a>	p.V267I	1	92442768	92442768	37	Missense_Mutation	SNP	G	A		uc010osz.1	BRDT_uc001dok.3...	<a href="#">view</a>
<a href="#">CACNB2</a>		10	18828721	18828722	37	3'UTR	Ins	-	AA	rs3841459	uc001ipr.2	CACNB2_uc009xjz...	
<a href="#">CDK11B</a>		1	1654066	1654068	37	Intron	Del	GCG	-		uc001agv.1	CDK11B_uc001aha...	
<a href="#">CDK14</a>	p.G276_splice	7	90547041	90547041	37	Splice_Site_SNP	SNP	T	C		uc003uky.2	CDK14_uc003ukz....	<a href="#">view</a>
<a href="#">CHD8</a>		14	21853630	21853630	37	3'UTR	Del	T	-	rs78520772	uc001was.1	CHD8_uc001war.1...	
<a href="#">CLP1</a>	p.I416V	11	57428876	57428876	37	Missense_Mutation	SNP	A	G		uc001nkw.2	CLP1_uc010rjw.1...	<a href="#">view</a>
<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="#">CTBP2</a>	p.K974fs	10	126678123	126678123	37	Frame_Shift_Del	Del	T	-		uc001lie.3	CTBP2_uc009yak....	
<a href="#">CXCL12</a>		10	44873992	44873993	37	Intron	Ins	-	CT	rs117617571	uc001jbh.2	CXCL12_uc001jbf...	
<a href="#">EIF4B</a>	p.P472H	12	53431286	53431286	37	Missense_Mutation	SNP	C	A		uc010snu.1	EIF4B_uc001sbh....	<a href="#">view</a>
<a href="#">EP300</a>		22	41574970	41574972	37	3'UTR	Del	GTA	-	rs76489032	uc003azl.3		
<a href="#">ETV1</a>		7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc011jqx.1	ETV1_uc011jxn.1...	
<a href="#">FANCA</a>	p.E1451Q	16	89805026	89805026	37	Missense_Mutation	SNP	C	G		uc002fou.1	ZNF276_uc010ciq...	<a href="#">view</a>
<a href="#">FBXO43</a>	p.G50W	8	101154334	101154334	37	Missense_Mutation	SNP	C	A		uc003yjd.2	FBXO43_uc003yje...	<a href="#">view</a>
<a href="#">FER</a>	p.V713M	5	108516536	108516536	37	Missense_Mutation	SNP	G	A		uc003kop.1	FER_uc011cvg.1_...	<a href="#">view</a>
<a href="#">FGFR1</a>	p.P25P	8	38314890	38314890	37	Silent	SNP	C	T	rs17175757	uc003xlp.2	FGFR1_uc011lbo....	<a href="#">view</a>
<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="#">FMN2</a>	p.PPLPGAGIPPP939del	1	240370914	240370946	37	In_Frame_Del	Del	GCCCC...	-	997328;rs4997:	uc010pye.1	FMN2_uc010pyd.1...	
<a href="#">FN1</a>		2	216244182	216244185	37	Intron	Del	GTAC	-		uc002vfa.2	FN1_uc002vfb.2_...	
<a href="#">FOXP1</a>		3	71008341	71008342	37	3'UTR	Ins	-	T	rs133724;rs11423	uc003dol.2	FOXP1_uc003dom....	
<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	-	rs1525754;rs3083	uc001rcd.2	GUCY2C_uc009zhz...	
<a href="#">HEPH</a>	p.G380V	X	65412038	65412038	37	Missense_Mutation	SNP	G	T		uc011moz.1	HEPH_uc004dwn.2...	<a href="#">view</a>
<a href="#">HIF1A</a>		14	62213929	62213930	37	3'UTR	Ins	-	T	rs67890419	uc001xfs.2	HIF1A_uc001xfq....	
<a href="#">HNF1A</a>		12	121434630	121434631	37	Intron	Ins	-	TCATTCAT	rs75100977	uc010szn.1	HNF1A_uc001tze....	
<a href="#">ICK</a>	p.W418C	6	52876924	52876924	37	Missense_Mutation	SNP	C	A		uc003pbh.2	ICK_uc003pbi.2_...	<a href="#">view</a>
<a href="#">ILK</a>		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	ILK_uc001mef.2_...	
<a href="#">ITPR1</a>		3	4888118	4888118	37	3'UTR	SNP	G	C		uc003bqa.2	ITPR1_uc010hca....	<a href="#">view</a>
<a href="#">ITPR2</a>	p.R505H	12	26818880	26818880	37	Missense_Mutation	SNP	C	T		uc001rhg.2		<a href="#">view</a>
<a href="#">KLK7</a>		19	51480672	51480675	37	3'UTR	Del	GGTT	-		uc002puo.2	KLK7_uc002pup.2...	
<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="#">MAPKAPK3</a>		3	50685575	50685575	37	3'UTR	Del	A	-	rs76026164	uc003day.1	MAPKAPK3_uc003d...	
<a href="#">MYST4</a>	p.EEEE1083del	10	76781852	76781863	37	In_Frame_Del	Del	GAGGA...	-	rs11001248	uc001jwn.1	MYST4_uc001jwm...	
<a href="#">NACA</a>	p.P1306fs	12	57111395	57111396	37	Frame_Shift_Ins	Ins	-	G		uc009zoy.1	NACA_uc001slz.2...	
<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="#">NCOA3</a>	p.QQQ1272del	20	46279815	46279823	37	In_Frame_Del	Del	GCAGC...	-		uc002xtk.2	NCOA3_uc010ght...	
<a href="#">NFIA</a>		1	61921185	61921186	37	3'UTR	Ins	-	T	270124;rs7613	uc010oos.1	NFIA_uc001czy.2...	
<a href="#">NIPBL</a>	p.V1890L	5	37024780	37024780	37	Missense_Mutation	SNP	G	T		uc003jkl.3	NIPBL_uc003jkl....	<a href="#">view</a>
<a href="#">NOTCH1</a>		9	139390479	139390479	37	3'UTR	SNP	C	T		uc004chz.2		<a href="#">view</a>
<a href="#">NPM1</a>		5	170837734	170837734	37	3'UTR	Del	T	-		uc011dex.1	NPM1_uc003mbi.2...	
<a href="#">NR1H2</a>	p.176_177insQ	19	50881820	50881821	37	In_Frame_Ins	Ins	-	AAC	783852;rs11235	uc010enw.2	NR1H2_uc002prv....	
<a href="#">PAX3</a>		2	223158653	223158653	37	Intron	Del	T	-	rs112753212	uc002vmt.1	PAX3_uc002vmy.1...	
<a href="#">PCDH15</a>	p.I116K	10	56129022	56129022	37	Missense_Mutation	SNP	A	T		uc010qhy.1	PCDH15_uc010qhq...	<a href="#">view</a>
<a href="#">PGR</a>		11	100933598	100933599	37	Intron	Ins	-	AATGA	rs35148438	uc001pgh.2	PGR_uc001pgg.2_...	
<a href="#">PIK3C2G</a>		12	18801075	18801075	37	3'UTR	SNP	A	G		uc010sib.1	PIK3C2G_uc001rd...	<a href="#">view</a>
<a href="#">PLD1</a>	p.K647*	3	171395413	171395413	37	Nonsense_Mutation	SNP	T	A		uc003fhs.2	PLD1_uc003fht.2...	<a href="#">view</a>
<a href="#">PRKCA</a>		17	64298959	64298960	37	5'UTR	Ins	-	G	rs71160558	uc002jfp.1	PRKCA_uc002jfo...	
<a href="#">PRKDC</a>	p.L1244fs	8	48805816	48805817	37	Frame_Shift_Ins	Ins	-	G	rs67588121	uc003xqi.2	PRKDC_uc003xqi....	
<a href="#">PTEN</a>	p.R233*	10	89717672	89717672	37	Nonsense_Mutation	SNP	C	T		uc001kfb.2		<a href="#">view</a>
<a href="#">RECQL4</a>	p.R766fs	8	145738768	145738768	37	Frame_Shift_Del	Del	G	-		uc003zdz.2		
<a href="#">RPS6KA1</a>	p.T449N	1	26887320	26887320	37	Missense_Mutation	SNP	C	A		uc001bms.1	RPS6KA1_uc001bm...	<a href="#">view</a>
<a href="#">SLC6A8</a>		X	152957656	152957657	37	Intron	Ins	-	A		uc004fib.3	SLC6A8_uc004fic...	
<a href="#">STK31</a>		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sws.3	STK31_uc003swt....	
<a href="#">TAF15</a>	p.R408C	17	34171525	34171525	37	Missense_Mutation	SNP	C	T		uc002hkd.2	TAF15_uc002hkc....	<a href="#">view</a>
<a href="#">TNK2</a>		3	195595583	195595583	37	Intron	SNP	G	A		uc003vft.1	TNK2_uc003fvq.1...	<a href="#">view</a>
<a href="#">TP53</a>	p.R248Q	17	7577538	7577538	37	Missense_Mutation	SNP	C	T	rs11540652	uc002gim.2	TP53_uc002gig.1...	<a href="#">view</a>
<a href="#">UBE2O</a>	p.K531del	17	74395563	74395565	37	In_Frame_Del	Del	TTC	-		uc002jrm.3	UBE2O_uc002jrn....	
<a href="#">VEGFC</a>	p.S419_splice	4	177605082	177605084	37	Splice_Site_Del	Del	TCA	-		uc003ius.1		
<a href="#">WHSC1</a>	p.V805A	4	1956963	1956963	37	Missense_Mutation	SNP	T	C		uc003gdz.3	WHSC1_uc003geb....	<a href="#">view</a>
<a href="#">ZMYND10</a>		3	50378764	50378764	37	3'UTR	SNP	C	A		uc003dag.1	RASSF1_uc003dad...	<a href="#">view</a>

