

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
ADAM28		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	ADAM28_uc011laa...	
AKAP12	p.L531_1532insE	6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819	uc011eep.1	AKAP12_uc003qoe...	
AKAP9	p.L335_1336insQ	7	91652178	91652179	37	In_Frame_Ins	Ins	-	AAC	rs34756483	uc003ulg.2	AKAP9_uc003ule...	
ALPK2	p.EDT5T1006del	18	56204388	56204402	37	In_Frame_Del	Del	CAGT...	-	rs4017rs11762	uc002lhj.3	ALPK2_uc002lkh...	
ANAPC5		12	121790193	121790195	37	5'UTR	Del	CAC	-		uc001uag.2	ANAPC5_uc001uah...	
ARID1A		1	27107263	27107264	37	3'UTR	Ins	-	C	rs3841356	uc001bmv.1	ARID1A_uc001bmu...	
ASPH	p.A264_splice	8	62550924	62550925	37	Splice_Site_Ins	Ins	-	A	rs5891816	uc003xuj.2	ASPH_uc011leg.1...	
AXL	p.Y703*	19	41762429	41762429	37	Nonsense_Mutation	SNP	C	A		uc010ehj.2	CYP2F1_uc010xvw...	view
BRD1	p.V267I	1	92442768	92442768	37	Missense_Mutation	SNP	G	A		uc010osz.1	BRD1_uc001dsk.3...	view
CACNB2		10	18828721	18828722	37	3'UTR	Ins	-	AA	rs3841459	uc001ipr.2	CACNB2_uc009jz...	
CDK11B		1	1654066	1654068	37	Intron	Del	GCG	-		uc001agv.1	CDK11B_uc001aha...	
CDK14	p.G276_splice	7	90547041	90547041	37	Splice_Site_SNP	SNP	T	C		uc003uky.2	CDK14_uc003ukz...	view
CHD8		14	21853630	21853630	37	3'UTR	Del	T	-	rs78520772	uc001was.1	CHD8_uc001war.1...	
CLP1	p.L416V	11	57428876	57428876	37	Missense_Mutation	SNP	A	G		uc001nkw.2	CLP1_uc010rjw.1...	view
CLIC1	p.V1201_splice	22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977	uc002zpb.2	CLIC1_uc011agv...	
CNTN6		3	1339681	1339686	37	Intron	Del	GTTT...	-		uc003bez.2	CNTN6_uc010hbo...	
CTBP2	p.K974fs	10	126678123	126678123	37	Frame_Shift_Del	Del	T	-		uc001lle.3	CTBP2_uc009yak...	
CXCL12		10	44873992	44873993	37	Intron	Ins	-	CT	rs117617571	uc001jph.2	CXCL12_uc001jbf...	
EIF4B	p.P472H	12	53431286	53431286	37	Missense_Mutation	SNP	C	A		uc010snu.1	EIF4B_uc001sbb...	view
EP300		22	41574970	41574972	37	3'UTR	Del	GTA	-	rs76489032	uc003azl.3		
ETV1		7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc001jxq.1	ETV1_uc011jxn.1...	
FANCA	p.E1451Q	16	89805026	89805026	37	Missense_Mutation	SNP	C	G		uc002fou.1	ZNF276_uc010ciq...	view
FBXO43	p.G50W	8	101154334	101154334	37	Missense_Mutation	SNP	C	A		uc003yjd.2	FBXO43_uc003yje...	view
FER	p.V713M	5	108516536	108516536	37	Missense_Mutation	SNP	G	A		uc003kop.1	FER_uc011cvg.1...	view
FGFR1	p.P25P	8	38314890	38314890	37	Silent	SNP	C	T	rs17175757	uc003xhp.2	FGFR1_uc011lbo...	view
FGFR1OP		6	167453519	167453520	37	3'UTR	Ins	-	T	rs115519488	uc003qvj.2	CCR6_uc003qvj.2...	
FGR3		4	1809128	1809129	37	3'UTR	Del	GT	-		uc003gedu.2	FGR3_uc003gedr...	
FMN2	p.PPLPGAGPPP939del	1	240370914	240370946	37	In_Frame_Del	Del	GCCCC...	-	997328rs4997	uc010pye.1	FMN2_uc010pyd.1...	
FN1		2	216244182	216244185	37	Intron	Del	GTAC	-		uc002vfa.2	FN1_uc002vfb.2...	
FOXP1		3	71008341	71008342	37	3'UTR	Ins	-	T	133724rs11423	uc003dol.2	FOXP1_uc003dom...	
GRIA3		X	122318386	122318387	37	Splice_Site_Ins	Ins	-	G	rs66632982	uc004etq.3	GRIA3_uc004etr...	
GUCY2C		12	14827464	14827466	37	Intron	Del	ATC	-	rs25754rs3083	uc001rcd.2	GUCY2C_uc009zfb...	
HEPH	p.G380V	X	65412038	65412038	37	Missense_Mutation	SNP	G	T		uc011mox.1	HEPH_uc004dwn.2...	view
HIF1A		14	62213929	62213930	37	3'UTR	Ins	-	T	rs67890419	uc001kfs.2	HIF1A_uc001kfq...	
HNF1A		12	121434630	121434631	37	Intron	Ins	-	TCATTGAT	rs75100977	uc010sxn.1	HNF1A_uc001tze...	
ICK	p.W418C	6	52876924	52876924	37	Missense_Mutation	SNP	C	A		uc003pbh.2	ICK_uc003pbj.2...	view
ILK		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	ILK_uc001mef.2...	
ITPR1		3	4888118	4888118	37	3'UTR	SNP	G	C		uc003bqa.2	ITPR1_uc010hca...	view
ITPR2	p.R505H	12	26818880	26818880	37	Missense_Mutation	SNP	C	T		uc001rhg.2		view
KLK7		19	51480672	51480675	37	3'UTR	Del	GGTT	-		uc002puo.2	KLK7_uc002pup.2...	
MAML3	p.O767_splice	4	140651585	140651587	37	Splice_Site_Del	Del	CTG	-		uc003ihz.1	MGST2_uc010loi...	
MAP3K1	p.T949del	5	56177849	56177851	37	In_Frame_Del	Del	CAA	-		uc003qaw.3		

MAP3K14	p.R218_splice	17	43364293	43364294	37	Splice_Site_Ins	Ins	-	G	rs66766023	uc002iiv.1	MAP3K14_uc010da...
MAPKAPK3		3	50685575	50685575	37	3'UTR	Del	A	-	rs76026164	uc003day.1	MAPKAPK3_uc003d...
MYST4	p.EEEE1083del	10	76781852	76781863	37	In_Frame_Del	Del	GAGGA...	-	rs11001248	uc001jwm.1	MYST4_uc001jwm...
NACA	p.P1306fs	12	57111395	57111396	37	Frame_Shift_Ins	Ins	-	G		uc009zoy.1	NACA_uc001siz.2...
NCAM1		11	112832307	112832308	37	5'UTR	Ins	-	A	rs117108942	uc009yyq.1	NCAM1_uc001pno...
NCAM1	p.OQQ1272del	11	112832340	112832340	37	5'UTR	Del	C	-		uc009yyq.1	NCAM1_uc001pno...
NCOA3		20	46279815	46279823	37	In_Frame_Del	Del	GGAGC...	-		uc002xtk.2	NCOA3_uc010ght...
NFIA		1	61921185	61921186	37	3'UTR	Ins	-	T	270124;rs7613	uc010oes.1	NFIA_uc001ocy.2...
NIPBL	p.V489L	5	37024780	37024780	37	Missense_Mutation	SNP	G	T		uc003jkl.3	NIPBL_uc003jkk...
NOTCH1		9	139390479	139390479	37	3'UTR	SNP	C	T		uc004czh.2	
NPM1		5	170837734	170837734	37	3'UTR	Del	T	-		uc011dex.1	NPM1_uc003mbi.2...
NR1H2	p.L76_177msQ	19	50881820	50881821	37	In_Frame_Ins	Ins	-	AAC	rs3852;rs11235	uc010enw.2	NR1H2_uc002prv...
PAX3		2	223158653	223158653	37	Intron	Del	T	-	rs112753212	uc002vmt.1	PAX3_uc002vmy.1...
PCDH15	p.L116K	11	56129022	56129022	37	Missense_Mutation	SNP	A	T		uc010qhy.1	PCDH15_uc010qhq...
PGR		12	100933598	100933599	37	Intron	Ins	-	AATGA	rs35148438	uc001pgh.2	PGR_uc001pge.2...
PIK3C2G		12	18801075	18801075	37	3'UTR	SNP	A	G		uc010sib.1	PIK3C2G_uc001rd...
PLD1	p.K647*	3	171395413	171395413	37	Nonsense_Mutation	SNP	T	A		uc003fhs.2	PLD1_uc003ft.2...
PRKA		17	64298959	64298960	37	5'UTR	Ins	-	G	rs71160558	uc002jfp.1	PRKCA_uc002jfo...
PRKDC	p.L1244fs	8	48805816	48805817	37	Frame_Shift_Ins	Ins	-	G	rs67588121	uc003xqi.2	PRKDC_uc003xqi...
PTEN	p.R233*	10	89717672	89717672	37	Nonsense_Mutation	SNP	C	T		uc001kfb.2	
RECQL4	p.R766fs	8	145738768	145738768	37	Frame_Shift_Del	Del	G	-		uc003zdl.2	
RPS6KA1	p.T449N	1	26887320	26887320	37	Missense_Mutation	SNP	C	A		uc001bms.1	RPS6KA1_uc001bm...
SLC6A8		X	152957656	152957657	37	Intron	Ins	-	A		uc004fib.3	SLC6A8_uc004fib...
STK31		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sww.3	STK31_uc003swf...
TAF15	p.R408C	17	34171525	34171525	37	Missense_Mutation	SNP	C	T		uc002hkd.2	TAF15_uc002hkc...
TNK2		3	195595583	195595583	37	Intron	SNP	G	A		uc003fvf.1	TNK2_uc003fvq.1...
TP53	p.R248Q	17	7577538	7577538	37	Missense_Mutation	SNP	C	T	rs11540652	uc002gim.2	TP53_uc002gig.1...
UBE2O	p.K531del	17	74395563	74395565	37	In_Frame_Del	Del	TTC	-		uc002jrm.3	UBE2O_uc002jrm...
VEGFC	p.S419_splice	4	177605082	177605084	37	Splice_Site_Del	Del	TCA	-		uc003ius.1	
WHSC1	p.V805A	4	1956963	1956963	37	Missense_Mutation	SNP	T	C		uc003gdt.3	WHSC1_uc003geb...
ZMYND10		3	50378764	50378764	37	3'UTR	SNP	C	A		uc003dag.1	RASSF1_uc003dad...

