

Mutations in SH-SY5Y

SHSY5Y_AUTONOMIC_GANGLIA

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
ADAM17		2	9630245	9630245	37	3'UTR	Del	T	-		uc002qzu.2	H1_uc010yiz.1...	
ADAM28		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	AM28_uc011laa...	
AKAP12	p.1531_1532ins	6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819	uc011eep.1	AP12_uc003qoe...	
AKAP9	p.1335_1336ins	7	91652178	91652179	37	In_Frame_Ins	Ins	-	AAC	rs34756483	uc003ulg.2	AKAP9_uc003ule...	
ALPK1		4	113347609	113347610	37	Intron	Ins	-	T	rs68137453	uc003iap.3	ALPK1_uc003ian...	
AMZ2		17	66246229	66246229	37	Intron	SNP	T	A		uc002jgs.1	AMZ2_uc002jgr.1	view
ANAPC5	p.A504T	12	121758193	121758193	37	missense_Mutati	SNP	C	T		uc001uag.2	ANAPC5_uc010sz...	view
ARID1A	p.G1254G	1	27099883	27099883	37	Silent	SNP	C	T		uc001bmv.1	ARID1A_uc001bm...	view
AXIN1	p.V827A	16	338231	338231	37	missense_Mutati	SNP	A	G		uc002cgp.1	AXIN1_uc002cgq...	view
AXL		19	41749740	41749740	37	Intron	SNP	C	T		uc010ehj.2	P2F1_uc010xv...	view
CASP10	p.S296F	2	202072871	202072871	37	missense_Mutati	SNP	C	T		uc002ujx.1	CASP10_uc010zhi...	view
CDK17		12	96673965	96673965	37	3'UTR	SNP	A	G		uc001tep.1		view
CHD1	p.P1684del	5	98192165	98192167	37	In_Frame_Del	Del	AGG	-	267787;rs6174...	uc003knf.2	CHD1_uc010jbn.2...	
CHEK2	p.T410fs	22	29091857	29091857	37	In_Frame_Shift_De	Del	G	-		uc003adt.1	CHEK2_uc003ads...	
CLTCL1	p.V1201_splice	22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977	uc002zpb.2	CLTCL1_uc011agv...	
CXCL12		10	44873992	44873993	37	Intron	Ins	-	CT	rs117617571	uc001jhb.2	CXCL12_uc001jbf...	
CYP3A4		7	99381749	99381749	37	5'UTR	SNP	G	C		uc003urv.1	CYP3A4_uc003urv	view
DDR2		1	162750211	162750212	37	3'UTR	Del	TT	-		uc001gcf.2	DDR2_uc001gcf.2...	
DNAH8	p.Q578*	6	38750903	38750903	37	missense_Mutati	SNP	C	T		uc003ooe.1		view
DNAH8		6	38952247	38952250	37	Intron	Del	CACA	-		uc003ooe.1	DNAH8_uc003oog...	
ETV1		7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc011jqx.1	ETV1_uc011jxn.1...	
FGFR1OP		6	167453519	167453520	37	3'UTR	Ins	-	T	rs115519488	uc003qvj.2	FGFR1OP_uc003qvl.2...	
FGFR3		4	1809128	1809129	37	3'UTR	Del	GT	-		uc003gdu.2	FGFR3_uc003gdr...	
FLI1		11	128638217	128638217	37	Intron	Del	G	-		uc010sbu.1	FLI1_uc010sbt.1...	
GUCY2C		12	14827464	14827466	37	Intron	Del	ATC	-	rs1525754;rs3083	uc001rcd.2	GUCY2C_uc009zhz...	
HDAC1		1	32793474	32793474	37	Intron	SNP	G	A		uc001bvb.1	HDAC1_uc010ohc	view
HEPH	p.H1071Q	X	65483490	65483490	37	missense_Mutati	SNP	C	A		uc011moz.1	HEPH_uc004dwn...	view
ILK		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	ILK_uc001mef.2...	
ITPR2	p.R470_splice	12	26834804	26834805	37	Splice_Site_Ins	Ins	-	ACTC	696203;rs5919...	uc001rhg.2		
JUND	p.I269M	19	18391488	18391488	37	missense_Mutati	SNP	G	C		uc002nip.2		view
KLK7		19	51480672	51480675	37	3'UTR	Del	GGTT	-		uc002puo.2	KLK7_uc002pup.2...	
LETM2		8	38265796	38265798	37	3'UTR	Del	ACT	-		uc003xlm.1	LETM2_uc003xll...	

LRRK1	p.A95T	15	101523754	101523754	37	issense_Mutati	SNP	G	A		uc002bwr.2	RK1_uc010usb	view
MAML3	p.Q767_splice	4	140651585	140651587	37	Splice_Site_Del	Del	CTG	-		uc003ihz.1	GST2_uc010ioi...	
MAP3K14	p.R218_splice	17	43364293	43364294	37	Splice_Site_Ins	Ins	-	G	rs66766023	uc002iiw.1	MAP3K14_uc010da...	
MAPKAPK3		3	50685575	50685575	37	3'UTR	Del	A	-	rs76026164	uc003day.1	MAPKAPK3_uc003d...	
MLL3	p.P2467R	7	151876961	151876961	37	issense_Mutati	SNP	G	C		uc003wla.2	MLL3_uc003wkz.2	view
MLLT3		9	20346377	20346378	37	3'UTR	Ins	-	A	rs66943046	uc003zoe.2	MLLT3_uc011lne...	
MPL	p.R199S	1	43805147	43805147	37	issense_Mutati	SNP	G	C		uc001ciw.2	PL_uc001civ.2	view
MSH3	p.PPA66del	5	79950742	79950750	37	In_Frame_Del	Del	CCCCC...	-	rs1484572;rs1047	uc003kgz.2	HFR_uc011ctl.1...	
MYO18B	p.V618V	22	26168462	26168462	37	Silent	SNP	G	A		uc003abz.1	MYO18B_uc003ac	view
MYST3	p.K198R	8	41905903	41905903	37	issense_Mutati	SNP	T	C		uc010lxb.2	MYST3_uc010lxc	view
NCAM1		11	112832307	112832308	37	5'UTR	Ins	-	A	rs117108942	uc009yyq.1	NCAM1_uc001pno...	
NCAM1		11	112832340	112832340	37	5'UTR	Del	C	-		uc009yyq.1	NCAM1_uc001pno...	
NCOA3		20	46282339	46282340	37	3'UTR	Ins	-	AT		uc002xtk.2	NCOA3_uc002xtl...	
NEK3	p.K313_splice	13	52718050	52718051	37	Splice_Site_Ins	Ins	-	T	rs72514762	uc001vgh.2	NEK3_uc001vvg.2...	
NFKB1	p.D917_splice	4	103534740	103534741	37	Splice_Site_Ins	Ins	-	A	rs35795162	uc011cep.1	NFKB1_uc011ceq...	
NLRP3		1	247611982	247611982	37	3'UTR	Del	C	-		uc001icr.2	NLRP3_uc001ics...	
NPR1		1	153665919	153665920	37	3'UTR	Ins	-	C	rs66682713	uc001fcs.3	NPR1_uc010pdz.1...	
NR1H2	p.L176_177insQ	19	50881822	50881823	37	In_Frame_Ins	Ins	-	CAG	rs751954;rs1052	uc010enw.2	NR1H2_uc002prv...	
NUDT11		X	51239296	51239309	37	Start_Codon_Del	Del	TCCTC...	-		uc010njt.2		
PAX3		2	223158653	223158653	37	Intron	Del	T	-	rs112753212	uc002vmt.1	PAX3_uc002vmy.1...	
PCM1		8	17813249	17813249	37	Intron	SNP	C	T		uc003wyi.3	PCM1_uc011kyh.	view
PDE4DIP		1	144994926	144994926	37	5'UTR	SNP	G	T		uc001elw.3	PDE4DIP_uc009wi	view
PDGFRA		4	55130162	55130162	37	Intron	Del	T	-		uc003han.3	PDGFRA_uc003haa...	
PGR		11	100933598	100933599	37	Intron	Ins	-	AATGA	rs35148438	uc001pgh.2	PGR_uc001pgg.2...	
PIK3C2G	p.P129del	12	18435399	18435401	37	In_Frame_Del	Del	CCC	-		uc010sib.1	PIK3C2G_uc001rd...	
PIK3C2G		12	18800990	18800990	37	3'UTR	Del	A	-	rs16914368	uc010sib.1	PIK3C2G_uc001rd...	
PLCG1	p.F1172I	20	39802411	39802411	37	issense_Mutati	SNP	T	A		uc002xjo.1	PLCG1_uc002xjp.	view
PRKDC	p.L1244fs	8	48805816	48805817	37	In_Frame_Shift_Ins	Ins	-	G	rs67588121	uc003xqi.2	PRKDC_uc003xqj...	
RECQL4	p.R766fs	8	145738768	145738768	37	In_Frame_Shift_Del	Del	G	-		uc003zdz.2		
RPS6KL1		14	75373626	75373627	37	3'UTR	Ins	-	TGAT		uc001xqx.1	RPS6KL1_uc001xq...	
SACS	p.H1260Y	13	23914237	23914237	37	issense_Mutati	SNP	G	A		uc001uon.2	SACS_uc001uoo.2	view
SMARCA4	p.R973W	19	11134251	11134251	37	issense_Mutati	SNP	C	T		uc010dxo.2	SMARCA4_uc002m	view
SORL1		11	121500401	121500401	37	3'UTR	Del	A	-		uc001pxx.2	SORL1_uc010rzp...	
STK31		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sws.3	STK31_uc003swt...	
TLK1		2	171850178	171850179	37	3'UTR	Ins	-	AA	rs71008739	uc002ugo.2	TLK1_uc002ugn.2...	
TRPS1		8	116635872	116635873	37	5'UTR	Ins	-	A	rs76893349	uc003yny.2	TRPS1_uc011lhy...	
VEGFC	p.S419_splice	4	177605082	177605084	37	Splice_Site_Del	Del	TCA	-		uc003ius.1		

