

Mutations in JIMT-1

JIMT1_BREAST													
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	JH1_uc010yiz.1...	
ADAM28		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	AM28_uc011aa...	
AKAP12	p.1531_1532insE	6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819	uc011eep.1	AP12_uc003qoe...	
AKAP9	p.1335_1336insQ	7	91652178	91652179	37	In_Frame_Ins	Ins	-	AAC	rs34756483	uc003ulg.2	KAP9_uc003ule....	
ALS2	p.L723F	2	202608982	202608982	37	Missense_Mutation	SNP	T	G		uc002uy0.2	S2_uc002uy0.2	view
ARID1A	p.Q586del	1	27058045	27058047	37	In_Frame_Del	Del	CAG	-		uc001bmv.1	ID1A_uc001bmt...	
ASPH	p.A264_splice	8	62550924	62550925	37	Splice_Site_Ins	Ins	-	A	rs5891816	uc003xuj.2	SPH_uc011leg.1...	
BCL3		19	45262908	45262908	37	3'UTR	SNP	G	A		uc010xxe.1		view
BCR		22	23631813	23631814	37	Intron	Ins	-	C	rs66503844	uc002zww.2	JR_uc002zwx.2...	
CABC1		1	227171585	227171585	37	Intron	Del	T	-		uc001hqm.1	JBC1_uc001hqn...	
CLTCL1	p.V1201_splice	22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977	uc002zpb.2	JTCL1_uc011agv...	
CXCL12		10	44873992	44873993	37	Intron	Ins	-	CT	rs117617571	uc001jhb.2	JCL12_uc001jbf...	
DIP2C		10	415689	415689	37	Intron	SNP	C	G		uc001ifp.2	IP2C_uc009xhi.	view
DNAH8	p.E1899A	6	38831685	38831685	37	Missense_Mutation	SNP	A	C		uc003ooe.1		view
DST		6	56473206	56473206	37	Intron	SNP	A	G		uc003pdf.2	JT_uc003pcz.3...	view
EPHA3		3	89448689	89448689	37	Intron	SNP	G	C		uc003dqy.2	HA3_uc003dqx	view
EPHB3	p.P859S	3	184298796	184298796	37	Missense_Mutation	SNP	C	T		uc003foz.2		view
ETV1		7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc011jxq.1	JV1_uc011jxn.1...	
FASN		17	80036961	80036961	37	3'UTR	SNP	G	C		uc002kdu.2	JSN_uc002kdv...	view
FGFR1OP		6	167453519	167453520	37	3'UTR	Ins	-	T	rs115519488	uc003qvj.2	JR6_uc003qvl.2...	
FLI1		11	128638217	128638217	37	Intron	Del	G	-		uc010sbu.1	JL1_uc010sbt.1...	
GATS		7	99799294	99799294	37	3'UTR	SNP	A	C		uc003uuua.3	JAG3_uc010lgs.	view
GRIA3		X	122318386	122318387	37	Splice_Site_Ins	Ins	-	G	rs66632982	uc004etq.3	JRIA3_uc004etr...	
GRIN2A	p.D785N	16	9892137	9892137	37	Missense_Mutation	SNP	C	T		uc002czo.3	JIN2A_uc010uyr	view
GUCY2C		12	14827464	14827466	37	Intron	Del	ATC	-	?525754;rs3083	uc001rcd.2	JCY2C_uc009zh...	
HNF1A		12	121434630	121434631	37	Intron	Ins	-	TCATTCA	rs75100977	uc010szn.1	JNF1A_uc001tze...	
HSP90B1	p.E791del	12	104341188	104341190	37	In_Frame_Del	Del	GAA	-	rs117491163	uc001tkb.1	P90B1_uc010sw...	
ILK		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	KJ_uc001mef.2...	
LAMA1	p.I1547F	18	6999468	6999468	37	Missense_Mutation	SNP	T	A		uc002knm.2	MA1_uc010wz...	view
MAF	p.V271L	16	79632989	79632989	37	Missense_Mutation	SNP	C	A		uc002ffm.2	AFJ_UC002ffn.2...	view
MAML3	p.Q767_splice	4	140651585	140651587	37	Splice_Site_Del	Del	CTG	-		uc003ihz.1	JGST2_uc010iol...	
MAP3K1	p.T949del	5	56177849	56177851	37	In_Frame_Del	Del	CAA	-		uc003jqw.3	JMAP3K1...	
MAP3K14	p.R218_splice	17	43364293	43364294	37	Splice_Site_Ins	Ins	-	G	rs66766023	uc002iiw.1	JMAP3K14 uc010da...	
MAP4K5		14	50886558	50886558	37	3'UTR	SNP	G	A		uc001wya.2	JP4K5 uc001wy...	view
MAPKAPK3		3	50685575	50685575	37	3'UTR	Del	A	-	rs76026164	uc003day.1	JPKAPK3 uc003d...	

MCF2		X	138724730	138724730	37	Intron	SNP	T	A		uc011mwo.1	CF2_uc004fau.1	view
MELK	p.L320H	9	36651780	36651780	37	Missense_Mutation	SNP	T	A		uc003zzn.2	ELK_uc011pm.1	view
NCAM1		11	112832307	112832308	37	5'UTR	Ins	-	A	rs117108942	uc009yyq.1	AM1_uc001pno....	
NCAM1		11	112832340	112832340	37	5'UTR	Del	C	-		uc009yyq.1	AM1_uc001pno....	
NEK3	p.K313_splice	13	52718050	52718051	37	Splice_Site_Ins	Ins	-	T	rs72514762	uc001vgh.2	EK3_uc001vgg.2...	
NFIA		1	61921185	61921186	37	3'UTR	Ins	-	T	270124;rs7613	uc010oos.1	FIA_uc001czy.2...	
NFKB1	p.D917_splice	4	103534740	103534741	37	Splice_Site_Ins	Ins	-	A	rs35795162	uc011cep.1	KB1_uc011ceq....	
NIN	p.G2027_splice	14	51192786	51192786	37	Splice_Site_SNP	SNP	T	C		uc001wyi.2	N_uc001wyj.2	view
NR1H2	p.176_177insQ	19	50881820	50881821	37	In_Frame_Ins	Ins	-	AAC	783852;rs11235	uc010enw.2	R1H2_uc002prv....	
ODZ1		X	124097626	124097626	37	5'UTR	SNP	C	A		uc010nqy.2	OZ1_uc011muj.	view
ODZ1		X	124097633	124097633	37	5'UTR	SNP	A	T		uc010nqy.2	OZ1_uc011muj.	view
PDE4DIP	p.L2318_splice	1	144854212	144854212	37	Splice_Site_SNP	SNP	C	T		uc001elw.3	PF10_uc009wi	view
PDE4DIP	p.Q1665fs	1	144873963	144873963	37	Frame_Shift_Del	Del	T	-		uc001elw.3	PF10_uc009wir...	
PGGT1B		5	114548071	114548072	37	3'UTR	Ins	-	C		uc003kqw.3	GT1B_uc003kqx...	
PIK3C2G	p.P129del	12	18435399	18435401	37	In_Frame_Del	Del	CCC	-		uc010sib.1	K3C2G_uc001rd...	
PIK3CA	p.C420R	3	178927980	178927980	37	Missense_Mutation	SNP	T	C		uc003fjk.2		view
PINK1	p.V350M	1	20972141	20972141	37	Missense_Mutation	SNP	G	A		uc001bdm.2	NK1_uc001bdn	view
PKHD1	p.S1833L	6	51882310	51882310	37	Missense_Mutation	SNP	G	A		uc003pah.1	KHD1_uc003pai	view
PRKDC	p.L1244fs	8	48805816	48805817	37	Frame_Shift_Ins	Ins	-	G	rs67588121	uc003xqi.2	RKDC_uc003xqj....	
PTPRT	p.G903R	20	40790024	40790024	37	Missense_Mutation	SNP	C	G		uc010ggi.2	PRT uc002xkg	view
RECQL4	p.R766fs	8	145738768	145738768	37	Frame_Shift_Del	Del	G	-		uc003zdj.2		
SIK3	p.N586N	11	116734411	116734411	37	Silent	SNP	G	A		uc001ppy.2	K3_uc001ppz.2	view
SPEG	p.G1383S	2	220337818	220337818	37	Missense_Mutation	SNP	G	A		uc010fwg.2		view
STK31		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sws.3	K31_uc003swt....	
TP53	p.R248W	17	7577539	7577539	37	Missense_Mutation	SNP	G	A		uc002gim.2	P53_uc002gig.1	view
TP63		3	189507518	189507519	37	Intron	Ins	-	AGAG	702062;rs7971	uc003frj.2	P63_uc003frx.2...	
TRIO	p.Y2117C	5	14481356	14481356	37	Missense_Mutation	SNP	A	G		uc003jff.2	RIO_uc003jfg.2	view
TRPM7	p.M1043V	15	50891355	50891355	37	Missense_Mutation	SNP	T	C		uc001zyt.3	PM7_uc010bev	view
VEGFC	p.S419_splice	4	177605082	177605084	37	Splice_Site_Del	Del	TCA	-		uc003ius.1		

