

Mutations in SK-MEL-28

SKMEL28_SKIN															
Gene	Protein Change		Chr	Start	End	NCBI Build	Variant		Reference Variant Type	Tumor Allele	Allele 1	dbSNP ID	Annotation Transcript	Other Transcripts	Mutation Assessor
	Change	Gene					Classification	Variant Type							
ABCA3	p.P138L	16	2374439	2374439	37	issense_Mutati	SNP	G	A			uc002cpv.1	BCA3_uc010bsk	view	
ACACA		17	35766564	35766564	37	5'UTR	Del	A	-			uc002hno.2	ACA_uc002hnn....		
ADAM28		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134		uc003xdy.2	AM28_uc011aa...		
ADAMTS19	p.D1084V	5	129040041	129040041	37	issense_Mutati	SNP	A	T			uc003kvb.1	AMTS19_uc010...	view	
AKAP12	.1531_1532ins	6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819		uc011eep.1	AP12_uc003qoe...		
ALDH7A1		5	125930919	125930919	37	5'UTR	SNP	A	G			uc003ktx.2	DH7A1_uc003k	view	
ALPK1		4	113347609	113347610	37	Intron	Ins	-	T	rs68137453		uc003iap.3	LPK1_uc003ian...		
APC	p.S130G	5	112103053	112103053	37	issense_Mutati	SNP	A	G			uc010jby.2	PC_uc011cvt.1...	view	
ARFGEF1	p.H256Y	8	68204232	68204232	37	issense_Mutati	SNP	G	A			uc003xxo.1		view	
ARID1A	p.P1456L	1	27101085	27101085	37	issense_Mutati	SNP	C	T			uc001bmv.1	ID1A_uc001bm	view	
ASPH	p.A264_splice	8	62550924	62550925	37	Splice_Site_Ins	Ins	-	A	rs5891816		uc003xuj.2	SPH_uc011leg.1...		
ATP11A		13	113536404	113536405	37	3'UTR	Del	TG	-			uc001vsj.3	TP11A_uc001vsi...		
AURKC		19	57742494	57742507	37	5'UTR	Del	GGACG...	-	};rs112039303;I		uc002qoe.2	JRKC_uc002qoc....		
BCR		22	23632777	23632778	37	Intron	Ins	-	T			uc002zww.2	CR_uc002zwx.2...		
BCR		22	23631813	23631814	37	Intron	Ins	-	C	rs66503844		uc002zww.2	CR_uc002zwx.2...		
BLM	p.T221I	15	91293160	91293160	37	issense_Mutati	SNP	C	T			uc002bpr.2	M_uc010uqh.1...	view	
BRAF	p.V600E	7	140453136	140453136	37	issense_Mutati	SNP	A	T	rs113488022		uc003vvw.3		view	
CARS	p.R96R	11	3063473	3063473	37	Silent	SNP	A	C			uc001lxf.2	ARS_uc001lxe.2	view	
CDK4	p.R24C	12	58145431	58145431	37	issense_Mutati	SNP	G	A	rs11547328		uc001spv.2	DK4_uc010ssb.1	view	
CDKN2C		1	51435977	51435977	37	Intron	SNP	C	T			uc001csf.2	KN2C_uc001cs...	view	
CEP110		9	123939569	123939569	37	3'UTR	SNP	T	C			uc004bkx.1	P110_uc004blk	view	
CHEK2	p.E437K	22	29091777	29091777	37	issense_Mutati	SNP	C	T			uc003adat.1	KE2_uc003ads	view	
CHEK2		22	29120751	29120751	37	Intron	SNP	T	A			uc003adat.1	KE2_uc003ads	view	
CLTC1	p.P253S	22	19226836	19226836	37	issense_Mutati	SNP	G	A			uc002zpb.2	TCL1_uc011agv	view	
CLTC1	p.V1201_splice	22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977		uc002zpb.2	TCL1_uc011agv...		
CNTN6		3	1339681	1339686	37	Intron	Del	GTTT...	-			uc003boz.2	ITN6_uc010hbo....		
CSMD3	p.S2119C	8	113358413	113358413	37	issense_Mutati	SNP	T	A			uc003ynu.2	MD3_uc003yns	view	
DST	p.E4564G	6	56362700	56362700	37	issense_Mutati	SNP	T	C			uc003pdf.2	ST_uc003pcz.3...	view	
DYRK1A		21	38884966	38884967	37	3'UTR	Ins	-	T	rs72430212		uc002ywtk.2	RK1A_uc002yw...		
EDN1		6	12290729	12290729	37	5'UTR	Del	A	-			uc003nae.3	N1_uc010jpb.2...		
EGFR	p.P753S	7	55242487	55242487	37	issense_Mutati	SNP	C	T			uc003tqk.2	GFR_uc010kzg.1	view	
EP300		22	41574970	41574972	37	3'UTR	Del	GTA	-	rs76489032		uc003azl.3			
EPHA6		3	97367230	97367231	37	Intron	Ins	-	A	rs115253553		uc010how.1	HA6_uc003drs....		

EPHA8		1	22928305	22928305	37	3'UTR	SNP	G	T		uc001bfx.1	view
FLI1		11	128638217	128638217	37	Intron	Del	G	-		uc010sbu.1	LI1_uc010sbt.1...
FN1		2	216244182	216244185	37	Intron	Del	GTAC	-		uc002vfa.2	J1_uc002vfb.2...
GPR112	p.S53N	X	135405024	135405024	37	issense_Mutati	SNP	G	A		uc004ezu.1	R112_uc010nsl
GPR112	p.D2657del	X	135474445	135474447	37	In_Frame_Del	Del	GAT	-	rs3030295	uc004ezu.1	R112_uc010nsb...
GRIA3		X	122318386	122318387	37	Splice_Site_Ins	Ins	-	G	rs66632982	uc004etq.3	RIA3_uc004etr....
GUCY2C		12	14827464	14827466	37	Intron	Del	ATC	-	?525754;rs3083	uc001rcd.2	JCY2C_uc009zhz...
HDAC1	p.K476Q	1	32798622	32798622	37	issense_Mutati	SNP	A	C		uc001bvb.1	DAC1_uc010ohf
HEPH	p.P159S	X	65393484	65393484	37	issense_Mutati	SNP	C	T		uc011moz.1	PH_uc004dwn.
HIF1A		14	62213929	62213930	37	3'UTR	Ins	-	T	rs67890419	uc001xfs.2	IF1A_uc001xfq....
HNF1A		12	121434630	121434631	37	Intron	Ins	-	TCATTCA	rs75100977	uc010szn.1	VF1A_uc001tze....
INRNPAB1		7	26240410	26240410	37	5'UTR	SNP	C	T		uc003srx.3	RNPA2B1_uc00
ILK		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	K_uc001mef.2...
ITPR2	p.R470_splice	12	26834804	26834805	37	Splice_Site_Ins	Ins	-	ACTC	696203;rs59199	uc001rhg.2	
JAK2	p.E592K	9	5072624	5072624	37	issense_Mutati	SNP	G	A		uc010mhm.2	K2_uc003ziw.2
JAK3	p.P693L	19	17945782	17945782	37	issense_Mutati	SNP	G	A		uc002nhn.3	K3_uc010ebh.2
KIAA1409	p.P435L	14	94038319	94038319	37	issense_Mutati	SNP	C	T		uc001yb.v1	AA1409_uc001
KLK7		19	51480672	51480675	37	3'UTR	Del	GGTT	-		uc002puo.2	.K7_uc002pup.2...
LRP1B	p.R2161*	2	141458137	141458137	37	issense_Mutati	SNP	G	A		uc002tvj.1	
LRP2	p.T2086S	2	170068502	170068502	37	issense_Mutati	SNP	T	A		uc002ues.2	
MAF		16	79628537	79628537	37	Intron	Del	T	-		uc002ffm.2	AF_uc002ffn.2...
MAML3	p.Q767_splice	4	140651585	140651587	37	Splice_Site_Del	Del	CTG	-		uc003ihz.1	GST2_uc010ioi....
MAP3K1	p.T949del	5	56177849	56177851	37	In_Frame_Del	Del	CAA	-		uc003jqw.3	
MAPK14		6	36076345	36076354	37	3'UTR	Del	GTGTG...	-	rs80147786	uc003olp.2	APK14_uc003olq...
MAPKAPK3		3	50685575	50685575	37	3'UTR	Del	A	-	rs76026164	uc003day.1	APKAPK3_uc003d...
MAPKBP1		15	42111746	42111746	37	Intron	Del	G	-		uc001zok.3	APKBP1_uc001zo...
MOS	p.S194L	8	57025961	57025961	37	issense_Mutati	SNP	G	A		uc011leb.1	
MPPED2	p.R229S	11	30435854	30435854	37	issense_Mutati	SNP	T	A		uc001msr.2	PED2_uc001ms
MYH11	p.E1203K	16	15826486	15826486	37	issense_Mutati	SNP	C	T		uc002ddx.2	H11_uc002ddv
NAV3	p.A239fs	12	78388627	78388630	37	Frame_Shift_De	Del	CAAC	-		uc001syp.2	AV3_uc001syo.2...
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	uc001vgg.2	EK3_uc001vgg.2...
NEK9	p.R142H	14	75587850	75587850	37	issense_Mutati	SNP	C	T		uc001xrl.2	
NLRP3	p.D305N	1	247587658	247587658	37	issense_Mutati	SNP	G	A	rs121908153	uc001icr.2	LRP3_uc001ics...
NR1H2	p.176_177insQ	19	50881820	50881821	37	In_Frame_Ins	Ins	-	AAC	?83852;rs11235	uc010enw.2	R1H2_uc002prv....
NR1H2	p.176_177insQ	19	50881822	50881823	37	In_Frame_Ins	Ins	-	CAG	L751954;rs1052	uc010enw.2	R1H2_uc002prv....
NRK	p.G1029R	X	105168796	105168796	37	issense_Mutati	SNP	G	A		uc004emd.2	RK_uc010npc.1

NUP210	p.L621P	3	13407516	13407516	37	issense_Mutati	SNP	A	G		uc003bxv.1	JP210_uc003bx	view
OBSCN	p.E4661A	1	228505725	228505725	37	issense_Mutati	SNP	A	C		uc009xez.1	3SCN_uc001hsn	view
P2RX7	p.T205_splice	12	121603241	121603241	37	Splice_Site_SN	SNP	G	A		uc001tzm.2	2RX7_uc001tzn.	view
PAK7	p.D88N	20	9561520	9561520	37	issense_Mutati	SNP	C	T		uc002wnl.2	K7_uc002wnk.	view
PAX3		2	223066017	223066018	37	Intron	Ins	-	C	rs115619343	uc002vmt.1	X3_uc002vmy.1...	
PDE4DIP	p.Q1665fs	1	144873963	144873963	37	Frame_Shift_De	Del	T	-		uc001elw.3	3PF10_uc009wir...	
PDE4DIP	p.L601fs	1	144915624	144915624	37	Frame_Shift_De	Del	G	-	rs1778112	uc001elw.3	3PF10_uc009wir...	
PDE4DIP	p.E243fs	1	144923729	144923729	37	Frame_Shift_De	Del	T	-		uc001elw.3	3PF10_uc009wir...	
PDZRN4	p.S188I	12	41903672	41903672	37	issense_Mutati	SNP	G	T		uc010skn.1	ZRN4_uc001rm	view
PIK3C2B		1	204423610	204423610	37	Intron	Del	A	-		uc001haw.2	K3C2B_uc010pq...	
PIK3C2G		12	18800990	18800990	37	3'UTR	Del	A	-	rs16914368	uc010sib.1	K3C2G_uc001rd...	
PKN2		1	89271763	89271763	37	Intron	SNP	T	G		uc001dmn.2	V2_uc001dmm.	view
PRKCA		17	64298959	64298960	37	5'UTR	Ins	-	G	rs71160558	uc002jfp.1	RKCA_uc002jfo....	
PRKDC	p.L1244fs	8	48805816	48805817	37	Frame_Shift_In	Ins	-	G	rs67588121	uc003xqi.2	RKDC_uc003xaj....	
PTEN	p.T167A	10	89711881	89711881	37	issense_Mutati	SNP	A	G		uc001kfb.2		view
PTPRU		1	29652275	29652275	37	3'UTR	SNP	C	T		uc001bru.2	'PRU_uc001brv	view
RECQL4	p.R766fs	8	145738768	145738768	37	Frame_Shift_De	Del	G	-		uc003zdj.2		
RPS6KA5		14	91360733	91360734	37	Intron	Ins	-	T	rs77438853	uc001xys.2	S6KA5_uc010tw...	
SACS	p.F2678L	13	23909981	23909981	37	issense_Mutati	SNP	G	C		uc001uon.2	CS_uc001uo...	view
9-Sep		17	75494787	75494788	37	3'UTR	Ins	-	C	rs5822173	uc002jts.3	:PT9_uc010wtk....	
SNRK	p.G271E	3	43381859	43381859	37	issense_Mutati	SNP	G	A		uc003cms.3	JRK_uc003cmt.	view
STK25		2	242435659	242435662	37	Intron	Del	AAGG	-	?15138;rs75497	uc002wbm.2	K25_uc002wbk....	
STK31		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sws.3	'K31_uc003swt....	
TLK1		2	171850178	171850179	37	3'UTR	Ins	-	AA	rs71008739	uc002ugo.2	.K1_uc002ugn.2...	
TNFRSF1B		1	12267184	12267184	37	3'UTR	SNP	G	C		uc001att.2	IFRSF1B_uc001...	view
TP53	p.L145R	17	7578495	7578496	37	issense_Mutati	DNP	CA	AC		uc002gim.2	P53_uc002gig.1...	
TRAF2		9	139793173	139793173	37	5'UTR	SNP	T	G		uc010nbu.2	RAF2_uc010nbv	view
TSC1	.1043_1044ins	9	135771987	135771988	37	In_Frame_Ins	Ins	-	GCT		uc004cca.2	;C1_uc004ccb.3...	
TTN	p.E32971K	2	179393863	179393863	37	issense_Mutati	SNP	C	T		uc010zfg.1	TN_uc010zfh.1	view

