

Mutations in K-562

K562_HAEMATOPOIETIC_AND_LYMPHOID_TISSUE															
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
	Change	Change					Classification	Variant Type							
AAK1	c.541_542QQ>C		2	69741754	69741756	37	In_Frame_Del	Del	TGT	-	rs55712143	uc002sfz.2	AK1_uc010fdk.2...		
ADAM22	p.F412_splice		7	87763642	87763642	37	Splice_Site_SNF	SNP	A	T		uc003ujp.1	AM22_uc003uj	view	
ADAM22	p.D561E		7	87778333	87778333	37	missense_Mutation	SNP	T	G		uc003ujp.1	AM22_uc003uj	view	
ADAM28			8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	AM28_uc011aa...		
AKAP12	c.1531_1532ins		6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819	uc011eep.1	AP12_uc003qoe...		
AKT3	p.G37*		1	243858956	243858956	37	missense_Mutation	SNP	C	A		uc001iab.1	KT3_uc001hzz.1	view	
ALPK2	p.P682fs		18	56205373	56205374	37	Frame_Shift_In	Ins	-	G		uc002lhj.3	LPK2_uc002lh...		
ASPH	p.A264_splice		8	62550924	62550925	37	Splice_Site_Ins	Ins	-	A	rs5891816	uc003xuj.2	SPH_uc011leg.1...		
BRDT	p.R536*		1	92446579	92446579	37	missense_Mutation	SNP	C	T		uc010osz.1	DTD_uc001dok.3	view	
BRSK2	p.G461S		11	1471022	1471022	37	missense_Mutation	SNP	G	A		uc001lrm.2	RSK2_uc009ycv.	view	
C15ORF2			15	24924588	24924588	37	3'UTR	SNP	C	T		uc001yw0.2		view	
CBL	p.D792H		11	119169190	119169190	37	missense_Mutation	SNP	G	C		uc001pwe.2		view	
CDC42BPG			11	64591867	64591867	37	3'UTR	SNP	G	A		uc001obs.3		view	
CDH2	p.S691L		18	25565101	25565101	37	missense_Mutation	SNP	G	A		uc002kwg.2	H2_uc010xbn.3	view	
CDH2	p.Y345C		18	25573588	25573588	37	missense_Mutation	SNP	T	C		uc002kwg.2	H2_uc010xbn.3	view	
CHD1	p.P1684del		5	98192165	98192167	37	In_Frame_Del	Del	AGG	-	267787;rs61749	uc003knf.2	ID1_uc010jbn.2...		
CHD5	c.1626_1627EK>G		1	6181198	6181199	37	missense_Mutation	DNP	TC	GA		uc001amb.1	ID5_uc001alz.1...		
CHD5	p.F892S		1	6196598	6196598	37	missense_Mutation	SNP	A	G		uc001amb.1	D5_uc001ama.	view	
CIT	p.E469Q		12	120221852	120221852	37	missense_Mutation	SNP	C	G		uc001txj.1	IT_uc001txh.1	view	
CLTCL1	p.V1201_splice		22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977	uc002zpb.2	.TCL1_uc011agv...		
CREB3L2	p.T100del		7	137612914	137612916	37	In_Frame_Del	Del	TGG	-		uc003vtw.2	REB3L2_uc003vt...		
CSMD3	p.F1516L		8	113562916	113562916	37	missense_Mutation	SNP	A	C		uc003ynu.2	MD3_uc003yns	view	
CTBP2	p.K974fs		10	126678123	126678123	37	Frame_Shift_De	Del	T	-		uc001lie.3	TBP2_uc009yak...		
CXCL12			10	44873992	44873993	37	Intron	Ins	-	CT	rs117617571	uc001jbh.2	KCL12_uc001bjf...		
DNAH8			6	38952247	38952250	37	Intron	Del	CACA	-		uc003ooe.1	IAH8_uc003oog...		
DST	p.M4809I		6	56354381	56354381	37	missense_Mutation	SNP	C	G		uc003pdf.2	ST_uc003pcz.3	view	
DYRK1A			21	38884966	38884967	37	3'UTR	Ins	-	T	rs72430212	uc002yw.k.2	RK1A_uc002yw...		
EIF3A	p.K765I		10	120810736	120810736	37	missense_Mutation	SNP	T	A		uc001ldu.2	F3A_uc010qsu.	view	
EPHA6			3	97367230	97367231	37	Intron	Ins	-	A	rs115253553	uc010how.1	H6 uc003drs...		
ETV1			7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc011jxq.1	TV1_uc011jxn.1...		
FAM118A			22	45736388	45736388	37	3'UTR	SNP	C	T		uc003bfz.3	M118A_uc003b	view	
FGFR3			4	1809128	1809129	37	3'UTR	Del	GT	-		uc003gdu.2	GFR3_uc003gdr...		
FLI1			11	128638217	128638217	37	Intron	Del	G	-		uc010sbu.1	LI1_uc010sbt.1...		

FZD1	p.93_94insP	7	90894459	90894460	37	In_Frame_Ins	Ins	-	CCG		uc003ula.2	
GCK		7	44184604	44184605	37	3'UTR	Ins	-	G		uc003tkk.1	CK_uc003tkh.1...
GCK		7	44198722	44198722	37	5'UTR	SNP	C	T		uc003tkk.1	CK_uc003tkj.1...
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....
HDAC8	p.L160L	X	71715076	71715076	37	Silent	SNP	C	A		uc004eau.2	AC8_uc011mqe...
HDLBP	p.R226S	2	242195793	242195794	37	issense_Mutati	DNP	GC	AA		uc002waz.2	DLBP_uc002wba....
HNF1A		12	121434630	121434631	37	Intron	Ins	-	TCATTCA	rs75100977	uc010szn.1	NF1A_uc001tze....
HOXA9	p.D192N	7	27204503	27204503	37	issense_Mutati	SNP	C	T		uc003syt.2	
HOXD11	p.E294K	2	176973733	176973733	37	issense_Mutati	SNP	G	A		uc002uki.2)XD11_uc010fq...
IL21R		16	27460779	27460780	37	3'UTR	Ins	-	TG	rs66690088	uc002doq.1	21R_uc002dor....
ILK		11	6630029	6630029	37	Intron	Del	C	-		uc001mee.2	K_uc001mef.2...
ITGB4	p.A1033V	17	73739929	73739929	37	issense_Mutati	SNP	C	T		uc002jgp.2	GB4_uc002jph...
ITGB6	p.N463S	2	160994217	160994217	37	issense_Mutati	SNP	T	C		uc002ubh.2	GB6_uc010fou...
ITPR2	p.G2257_splice	12	26589152	26589152	37	Splice_Site_SN	SNP	G	T		uc001rhg.2	TPR2_uc009zjg...
JUND		19	18391142	18391143	37	3'UTR	Ins	-	G	rs71816628	uc002nip.2	
KLK7		19	51480672	51480675	37	3'UTR	Del	GGTT	-		uc002puo.2	.K7_uc002pup.2...
LRRC7	p.P728T	1	70503803	70503803	37	issense_Mutati	SNP	C	A		uc001dep.2	RC7_uc009wbg...
LSP1	p.G180S	11	1901417	1901417	37	issense_Mutati	SNP	G	A		uc001luj.2	SP1_uc001luj.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	
MAP3K14	p.R218_splice	17	43364293	43364294	37	Splice_Site_Ins	Ins	-	G	rs66766023	uc002iiw.1	AP3K14_uc010da...
MAP3K2	p.L89_splice	2	128088083	128088083	37	Splice_Site_SN	SNP	T	A		uc002toj.1	AP3K2_uc010fl...
MAP4K5	p.A257P	14	50930820	50930820	37	issense_Mutati	SNP	C	G		uc001wya.2	AP4K5_uc001wy...
MAPKBP1		15	42111753	42111754	37	Intron	Ins	-	C	rs3034891	uc001zok.3	\PKBP1_uc001zo...
MN1	o.533_533Q>Q0	22	28194933	28194934	37	In_Frame_Ins	Ins	-	TGC	rs45480998	uc003adj.2	
MSH3	p.PPA66del	5	79950742	79950750	37	In_Frame_Del	Del	CCCCC...	-	rs484572;rs1047	uc003kgz.2	HFR_uc011ctl.1...
MTMR11		1	149900874	149900874	37	3'UTR	SNP	T	A		uc001eti.3	F3B4_uc001eti...
MYO18B	p.E1932A	22	26346379	26346379	37	issense_Mutati	SNP	A	C		uc003abz.1	'O18B_uc003ac...
MYST3	p.R1877C	8	41790109	41790109	37	issense_Mutati	SNP	G	A		uc010lxz.2	YST3_uc010lxc...
MYT1		20	62871860	62871861	37	3'UTR	Ins	-	CCGCA		uc002yii.2	IYT1_uc002yii.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....
NCOA3	p.Q1276del	20	46279864	46279866	37	In_Frame_Del	Del	CAA	-	rs32443;rs28529	uc002xtk.2	COA3_uc010ght....
NCOA4		10	51589252	51589252	37	3'UTR	SNP	A	G		uc009xon.2	ARG_uc001jih.2...
NEK11		3	130893613	130893613	37	Intron	SNP	A	T		uc003eny.2	EK11_uc003enx...
NEK3	p.K313_splice	13	52718050	52718051	37	Splice_Site_Ins	Ins	-	T	rs72514762	uc001vgh.2	EK3_uc001vgg.2...

NOTCH1	p.G326V	9	139413165	139413165	37	issense_Mutati	SNP	C	A		uc004chz.2		view
NR1H2	p.176_177insQ	19	50881822	50881823	37	In_Frame_Ins	Ins	-	CAG	L751954;rs1052	uc010enw.2	R1H2_uc002prv....	view
OBSCN	p.S5208F	1	228511278	228511278	37	issense_Mutati	SNP	C	T		uc009xez.1	BSCN_uc001hsn	view
P4HB	p.R345L	17	79804327	79804327	37	issense_Mutati	SNP	C	A		uc002kbn.1	IHB_uc002kbl.1	view
PDGFRA	p.Q551_splice	4	55140793	55140793	37	Splice_Site_SNPs	SNP	G	A		uc003han.3	GFRA_uc003ha	view
PML		15	74326772	74326772	37	Intron	SNP	C	T		uc002awv.2	IL_uc002awm.2	view
PML		15	74327683	74327683	37	Intron	SNP	C	T		uc002awv.2	IL_uc002awm.2	view
PNCK		X	152938410	152938411	37	Intron	Del	AC	-		uc011myu.1	ICK_uc011myt.1...	
PRKCB	p.G294E	16	24124353	24124353	37	issense_Mutati	SNP	G	A		uc002dme.2	KCB_uc002dmc	view
PRKDC	p.L1244fs	8	48805816	48805817	37	Frame_Shift_In:	Ins	-	G	rs67588121	uc003xqi.2	RKDC_uc003xqj....	
PTK6		20	62164170	62164170	37	Intron	Del	C	-		uc002yfg.2	TK6_uc011aay.1...	
RARB	p.R144L	3	25542776	25542776	37	issense_Mutati	SNP	G	T		uc011awl.1	ARB_uc003cdi.1	view
RARB	p.G110G	3	25542675	25542675	37	Silent	SNP	C	A		uc011awl.1	ARB_uc003cdi.1	view
RECQL4	p.R766fs	8	145738768	145738768	37	Frame_Shift_De	Del	G	-		uc003zdj.2		
RIMS2	p.G155*	8	104778530	104778530	37	issense_Mutati	SNP	G	T		uc003ylp.2		view
ROR1	p.N765H	1	64644017	64644017	37	issense_Mutati	SNP	A	C		uc001dbj.2		view
ROS1	p.R360I	6	117715410	117715410	37	issense_Mutati	SNP	C	A		uc003pxp.1	OS1_uc011ebi.1	view
RPS6KA4	p.L326V	11	64132842	64132842	37	issense_Mutati	SNP	C	G		uc001oae.2	S6KA4_uc001o...	view
RPS6KL1		14	75373626	75373627	37	3'UTR	Ins	-	TGAT		uc001xqx.1	S6KL1_uc001xq...	
SIK1	p.P188R	21	44841184	44841184	37	issense_Mutati	SNP	G	C		uc002zdf.2		view
SLCO4A1	p.R171C	20	61288317	61288317	37	issense_Mutati	SNP	C	T		uc002ydb.1	CO4A1_uc002yb...	view
SOX10	p.L144M	22	38374141	38374141	37	issense_Mutati	SNP	G	T		uc003aun.1	LR2F_uc003aur	view
STK10	p.E68K	5	171583747	171583747	37	issense_Mutati	SNP	C	T		uc003mbo.1		view
STK31		7	23872024	23872025	37	3'UTR	Ins	-	TTTG	rs66787534	uc003sws.3	TK31_uc003swt....	
TACC3		4	1739519	1739527	37	Intron	Del	GCCAT...	-	rs114226356	uc003gdo.2	ACC3_uc010ibz....	
TEC		4	48139307	48139310	37	3'UTR	Del	TTAT	-	rs79098351	uc003gxz.2		
TIAM1	p.L906M	21	32554909	32554909	37	issense_Mutati	SNP	G	T		uc002yow.1	AM1_uc011adk	view
TNFRSF9	p.M1T	1	8000053	8000053	37	issense_Mutati	SNP	A	G		uc001aot.2		view
TNRC6B	p.Q1328del	22	40697177	40697179	37	In_Frame_Del	Del	GCA	-	rs114291931	uc011aor.1	RC6B_uc003aym...	

