

Mutations in LS 180

LS180_LARGE_INTESTINE													
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
AAK1	p.P884fs	2	69706110	69706111	37	Frame_Shift_Ins	Ins	-	G		uc002sfp.2		
AAK1	p.S41_542QQ>Q	2	69741754	69741756	37	In_Frame_Del	Del	TGT	-	rs55712143	uc002sfp.2	AAK1_uc010fdk.2...	
ABCC3	p.P1056S	17	48753737	48753737	37	Missense_Mutation	SNP	C	T		uc002isl.2	ABCC3_uc002isn...	view
ACACB	p.A1284fs	12	109661678	109661681	37	Frame_Shift_Del	Del	CAAA	-		uc001tob.2	ACACB_uc001toc...	
ACVR1C		2	158390344	158390344	37	3'UTR	Del	A	-		uc002tzk.3	ACVR1C_uc002tzl...	
ACVR2A	p.K435fs	2	148683686	148683686	37	Frame_Shift_Del	Del	A	-		uc002twg.2	ACVR2A_uc010zbn...	
ADAM17		2	9630245	9630245	37	3'UTR	Del	T	-		uc002qzu.2	IAH1_uc010yiz.1...	
ADAM28		8	24211994	24211997	37	3'UTR	Del	CTAT	-	rs10610134	uc003xdy.2	ADAM28_uc011laa...	
ADAMTS2	p.M501I	5	178580504	178580504	37	Missense_Mutation	SNP	C	T		uc003mjw.2	DAMTS2_uc011dg...	view
ADHFE1	p.P266H	8	67364250	67364250	37	Missense_Mutation	SNP	C	A		uc003xwb.3	DHFE1_uc003xwd...	view
ADK	p.I310del	10	76429988	76429990	37	In_Frame_Del	Del	ATT	-		uc001jwi.2	ADK_uc010qlb.1...	
AKAP12	p.1531_1532insE	6	151674116	151674117	37	In_Frame_Ins	Ins	-	GAG	rs34737819	uc011eep.1	AKAP12_uc003qoe...	
AKAP9		7	91670248	91670249	37	Intron	Ins	-	T		uc003ulg.2	AKAP9_uc003ule...	
ALDH7A1		5	125930919	125930919	37	5'UTR	SNP	A	G		uc003ktx.2	ALDH7A1_uc003kt...	view
ALPK2	p.EDTST1006del	18	56204388	56204402	37	In_Frame_Del	Del	CAGTT...	-	rs44017;rs11762	uc002lhj.3	ALPK2_uc002lhk...	
APC	p.R1788C	5	112176653	112176653	37	Missense_Mutation	SNP	C	T		uc010jby.2	APC_uc011cvt.1...	view
ARHGAP29		1	94643144	94643145	37	Intron	Del	TT	-		uc001dqj.3	ARHGAP29_uc009w...	
ARID1A	p.K1093fs	1	27097688	27097688	37	Frame_Shift_Del	Del	A	-		uc001bmw.1	ARID1A_uc001bmt...	
ATF2	p.V183A	2	175979496	175979496	37	Missense_Mutation	SNP	A	G		uc002ujl.2	ATF2_uc010fqv.2...	view
ATP10A	p.K1016R	15	25940007	25940007	37	Missense_Mutation	SNP	T	C		uc010ayu.2		view
ATP10A	p.K747E	15	25958926	25958926	37	Missense_Mutation	SNP	T	C		uc010ayu.2		view
AXIN2	p.R834W	17	63526126	63526126	37	Missense_Mutation	SNP	G	A		uc002jfi.2	AXIN2_uc002jfh...	view
BAI1	p.R437fs	8	143558834	143558835	37	Frame_Shift_Ins	Ins	-	C		uc003ywm.2		
BAI3	p.A891V	6	69944988	69944988	37	Missense_Mutation	SNP	C	T		uc003pev.3	BAI3_uc010kak.2...	view
BAX	p.M38fs	19	49458971	49458971	37	Frame_Shift_Del	Del	G	-		uc002plf.1	BAX_uc002plg.1...	
BCKDK	p.A338D	16	31123267	31123267	37	Missense_Mutation	SNP	C	A		uc002eaw.3	BCKDK_uc002eav...	view
BCL11A	p.P159L	2	60695878	60695878	37	Missense_Mutation	SNP	G	A		uc002sae.1	BCL11A_uc002sab...	view
BCL9	p.G513fs	1	147091500	147091501	37	Frame_Shift_Ins	Ins	-	C		uc001epq.2	BCL9_uc010ozr.1...	
BCL9	p.S907fs	1	147092681	147092681	37	Frame_Shift_Del	Del	C	-		uc001epq.2	BCL9_uc010ozr.1...	
BLM		15	91303326	91303328	37	Intron	Del	TTT	-		uc002bpr.2	BLM_uc010uqh.1...	
BMPR2	p.E581fs	2	203420130	203420130	37	Frame_Shift_Del	Del	A	-		uc002uzf.3	BMPR2_uc010ftr...	
BRAF		7	140434295	140434295	37	3'UTR	Del	A	-		uc003vvc.3		
BRAF	p.D211G	7	140507839	140507839	37	Missense_Mutation	SNP	T	C		uc003vvc.3		view
BRCA2	p.T3030fs	13	32954023	32954023	37	Frame_Shift_Del	Del	A	-		uc001uub.1		
BRD2		6	32948599	32948599	37	3'UTR	Del	C	-		uc010juh.2	BRD2_uc003ocn.3...	
BRD3	p.P24fs	9	136918528	136918529	37	Frame_Shift_Ins	Ins	-	G		uc004cew.2	BRD3_uc004cex.2...	

BRIP1	p.D184Y	17	59924539	59924539	37	Missense_Mutation	SNP	C	A		uc002izk.1	view	
BUB1		2	111395467	111395468	37	3'UTR	Ins	-	A		uc002tgc.2	3UB1_uc010yxh.1...	
CABC1	p.V448A	1	227171881	227171881	37	Missense_Mutation	SNP	T	C		uc001hqm.1	CABC1_uc001hqm...	view
CAMK1G	p.G206S	1	209781259	209781259	37	Missense_Mutation	SNP	G	A		uc001hhd.2	AMK1G_uc001hhf...	view
CARD10	p.K918N	22	37887872	37887872	37	Missense_Mutation	SNP	C	A		uc003asx.1	CARD10_uc003ast...	view
CARS	p.I445V	11	3040431	3040431	37	Missense_Mutation	SNP	T	C		uc001lxf.2	CARS_uc001lxe.2...	view
CCND2		12	4409271	4409271	37	3'UTR	Del	A	-		uc001qmo.2		
CD160	p.G66V	1	145704220	145704220	37	Missense_Mutation	SNP	C	A		uc001eol.1	IBPF10_uc001emp...	view
CD40		20	44751957	44751957	37	Intron	SNP	A	C		uc002xrg.1	CD40_uc002xrf.1...	view
CD40LG		X	135741644	135741644	37	3'UTR	SNP	C	T		uc004faa.2	CD40LG_uc010nsd...	view
CDC7	p.F142fs	1	91977242	91977242	37	Frame_Shift_Del	Del	T	-		uc001doe.2	CDC7_uc001dof.2...	
CDH11	p.N605I	16	64984750	64984750	37	Missense_Mutation	SNP	T	A		uc002eoi.2	CDH11_uc010cdn...	view
CDK6	p.R144H	7	92355046	92355046	37	Missense_Mutation	SNP	C	T		uc011khw.1	CDK6_uc010lez.2...	view
CEBPA	p.T337M	19	33792311	33792311	37	Missense_Mutation	SNP	G	A		uc002nun.2	OC80054_uc002n...	view
CEP110	p.Y393S	9	123875835	123875835	37	Missense_Mutation	SNP	A	C		uc004bkk.1	EP110_uc004bkw...	view
CEP110	p.R584I	9	123886309	123886309	37	Missense_Mutation	SNP	G	T		uc004bkk.1	EP110_uc004bky...	view
CEP110	p.E1800fs	9	123924525	123924525	37	Frame_Shift_Del	Del	A	-		uc004bkk.1	CEP110_uc004blb...	
CHD1	p.P1684del	5	98192165	98192167	37	In_Frame_Del	Del	AGG	-	267787;rs6174	uc003knf.2	CHD1_uc010jbn.2...	
CHD1	p.K210fs	5	98236745	98236745	37	Frame_Shift_Del	Del	T	-		uc003knf.2		
CHD8		14	21853630	21853633	37	3'UTR	Del	TTTT	-	520772;rs7830	uc001was.1	CHD8_uc001war.1...	
CHD9	p.R961C	16	53276755	53276755	37	Missense_Mutation	SNP	C	T		uc002ehb.2	CHD9_uc002egy.2...	view
CHD9		16	53284016	53284016	37	Intron	SNP	T	A		uc002ehb.2	CHD9_uc002egy.2...	view
CIC	p.Y249H	19	42791859	42791859	37	Missense_Mutation	SNP	T	C		uc002tof.1		view
CLPTM1L	p.W374fs	5	1325892	1325892	37	Frame_Shift_Del	Del	A	-		uc003jch.2	CLPTM1L_uc003jc...	
CLTC	p.V488I	17	57741284	57741284	37	Missense_Mutation	SNP	G	A		uc002ixr.1	CLTC_uc002ixp.2...	view
CLTCL1	p.V1201_splice	22	19189003	19189004	37	Splice_Site_Ins	Ins	-	C	rs11386977	uc002zpb.2	CLTCL1_uc011agv...	
CNTN6		3	1339681	1339686	37	Intron	Del	GTTTT...	-		uc003boz.2	CNTN6_uc010hbo...	
COX6C	p.K38_splice	8	100904134	100904134	37	Splice_Site_SNP	SNP	A	G		uc003yiy.1		view
CSNK1A1	p.A213V	5	148892675	148892675	37	Missense_Mutation	SNP	G	A		uc003lqw.1	CSNK1A1_uc011dc...	view
CSNK1E	p.A384V	22	38690182	38690182	37	Missense_Mutation	SNP	G	A		uc003avj.2	CSNK1E_uc003avk...	view
CSPP1	p.R1130C	8	68102962	68102962	37	Missense_Mutation	SNP	C	T		uc003xxi.2	ARFGEF1_uc003xx...	view
CSPP1	p.P1214P	8	68107699	68107699	37	Silent	SNP	C	A		uc003xxi.2	ARFGEF1_uc003xx...	view
CTNNB1	p.S45F	3	41266137	41266137	37	Missense_Mutation	SNP	C	T		uc010hia.1	CTNNB1_uc003ckp...	view
DAPK1	p.S706T	9	90296433	90296433	37	Missense_Mutation	SNP	T	A		uc004apc.2	DAPK1_uc004apd...	view
DBN1		5	176899343	176899343	37	5'UTR	SNP	C	A		uc003mgx.2	DBN1_uc003mgy.2...	view
DCLK1	p.V632A	13	36379885	36379885	37	Missense_Mutation	SNP	A	G		uc001uvf.2	DCLK1_uc001uve...	view
DDR2	p.P519H	1	162741865	162741865	37	Missense_Mutation	SNP	C	A		uc001gcf.2	DDR2_uc001gcg.2...	view
DDX10	p.R365fs	11	108562720	108562720	37	Frame_Shift_Del	Del	A	-		uc001pkm.2	DDX10_uc001pkl...	
DDX10		11	108788854	108788854	37	Intron	Del	T	-		uc001pkm.2	DDX10_uc001pkl...	
DDX10		11	108811343	108811343	37	3'UTR	SNP	G	T		uc001pkm.2		view

DDX6	p.N289_splice	11	118629613	118629614	37	Splice_Site_Ins	Ins	-	G		uc001pub.2	DDX6_uc001pua.2...	
DFFA	p.R246G	1	10523162	10523162	37	Missense_Mutation	SNP	T	C		uc001arj.2	DFFA_uc001ark.2...	view
DOCK3	p.T1850fs	3	51417604	51417604	37	Frame_Shift_Del	Del	C	-		uc011bds.1		
DST		6	56472854	56472854	37	Intron	SNP	G	T		uc003pdf.2	DST_uc003pcz.3...	view
DST		6	56480319	56480319	37	Intron	SNP	G	A		uc003pdf.2	DST_uc003pcz.3...	view
DST		6	56480530	56480530	37	Intron	SNP	C	T		uc003pdf.2	DST_uc003pcz.3...	view
DST	p.M923T	6	56504154	56504154	37	Missense_Mutation	SNP	A	G		uc003pdf.2	DST_uc003pcz.3...	view
DST	p.K764N	6	56505040	56505040	37	Missense_Mutation	SNP	T	G		uc003pdf.2	DST_uc003pcz.3...	view
DSTYK		1	205116598	205116598	37	3'UTR	SNP	T	C		uc001hbw.2	DSTYK_uc001hbx...	view
DYRK1A	p.R559C	21	38884217	38884217	37	Missense_Mutation	SNP	C	T		uc002ywk.2	DYRK1A_uc002ywi...	view
DYRK1A		21	38884966	38884967	37	3'UTR	Ins	-	T	rs72430212	uc002ywk.2	DYRK1A_uc002ywi...	
DYRK3		1	206822391	206822391	37	3'UTR	SNP	A	G		uc001hej.2	DYRK3_uc001hek...	view
EEF1D	p.P349fs	8	144671355	144671355	37	Frame_Shift_Del	Del	G	-		uc003yyp.1	EEF1D_uc003yyp...	
EIF2AK4	p.T462P	15	40259911	40259911	37	Missense_Mutation	SNP	A	C		uc001zkm.1	EIF2AK4_uc001zk...	view
EP300	p.G30V	22	41489097	41489097	37	Missense_Mutation	SNP	G	T		uc003azl.3		view
EP300		22	41574970	41574972	37	3'UTR	Del	GTA	-	rs76489032	uc003azl.3		
EPHA6		3	97367230	97367231	37	Intron	Ins	-	A	rs115253553	uc010how.1	EPHA6_uc003drs...	
EPS15		1	51822220	51822221	37	3'UTR	Del	AA	-		uc001csq.1	EPS15_uc009vyz...	
ERBB3		12	56492196	56492197	37	Intron	Del	AA	-	rs77351535	uc001sjh.2	ERBB3_uc009zoi...	
ETV1		7	14028708	14028708	37	5'UTR	Del	A	-	rs77190713	uc011jxq.1	ETV1_uc011jxn.1...	
EWSR1		22	29687622	29687622	37	Intron	Del	A	-		uc003aev.2	EWSR1_uc003aet...	
FANCM	p.M1566T	14	45657008	45657008	37	Missense_Mutation	SNP	T	C		uc001wwd.3	FANCM_uc010anf...	view
FASN		17	80036847	80036847	37	3'UTR	SNP	G	A		uc002kdu.2	FASN_uc002kdv.1...	view
FBN2	p.Q2777K	5	127597463	127597463	37	Missense_Mutation	SNP	G	T		uc003kuu.2		view

