



2.382	0.981
5.04	1
1.163	1
2.789	1
1.957	0.959
-2.568	0.008
2.011	0.081
0.648	0.078
-2.788	0
2.124	0.972
5.713	0.981
-0.18	0.015
0.033	0.008
1.538	0.021
-1.487	0.001
3.48	0.944
0.417	0.498
0.021	0.346
1.925	0.34
-0.456	0.09
0.068	0.11
5.367	0.431
0.67	0.215
-0.733	0.056
1.454	0.089
0.663	0.088
0.436	0.088
0.272	0.092
1.714	0.115
0.136	0.023
1.428	0.027
0.34	0.019
-0.027	0.019
1.47	0.582
2.661	0.981
-0.553	0.789
1.502	0.916
3.082	0.928
-0.794	0.742
2.991	0.99
1.566	0.99
0.634	0.979
0.377	0.969
0.713	0.967
0.138	0.962
2.32	0.968
0.444	0.937
0.153	0.916
1.679	0.916
1.779	0.748
-0.84	0.016
0.565	0.018
1.626	0.021
-4.166	0
2.13	0.969
2.183	0.975
0.357	0.946
-0.977	0.023
1.912	0.074
-0.471	0.07
3.839	0.949
2.187	0.946
-0.147	0.754
0.336	0.778
3.949	0.936
-0.635	0.313
5.731	0.952
1.117	0.881
-0.088	0.002
-0.454	0
0.504	0
-1.855	0
5.657	0.92
1.618	0.886
0.338	0.471
1.015	0.361
-1.194	0.021
0.469	0.076
0.245	0.069
0.245	0.062
-0.42	0
-0.075	0
-1.331	0
-1.88	0
3.069	0.962
2.359	0.966
0.232	0.897
2.208	0.985
1.4	0.978
-0.468	0.926
3.87	1
3.414	1
1.136	0.994
1.999	0.967
-0.072	0.29
-0.003	0.271
0.643	0.301
1.893	0.324
0.353	0.24
2.043	0.292
1.607	0.276
0.068	0.051
0.417	0.021
0.343	0.007
-2.808	0
-0.174	0.001
0.162	0.015
0.041	0.081
2.1	0.925
2.404	0.953
0.779	0.939
0.917	0.918
1.072	0.859
-0.246	0.62
2.304	0.641

1.153	0.453
-0.208	0.009
-0.115	0.001
-0.063	0.001
-0.037	0
0.335	0
-0.194	0
0.016	0.003
(flanking)	1.455 0.948
<a href="#">exon sizes</a> and/or inspect your position(s) in <a href="#">igLUCSC Genome Browser</a>	
splice sites	effect gDNA position score wt detection sequence exon-intron border
	Acc gained 472 0.47 mu: GAGTCTTTAACTTGGTGGATATGGGGTGGTTATGGTC
distance from splice site	179
Kozak consensus sequence altered?	N/A
conservation protein level for non-synonymous changes	species match gene aa alignment
	Human mutated partly conserved 71GGFGSRSLVNLGGSKISISIVA RGGGRG SGFGGG YG GGGFGGGGFGGGGFGGGG IGGGGFGGFGSGGGGFGGGGFGGGGFGGGYGPVCP
	Ptrogodytes no homologue 38 --GSAGIINYQ-RRRTTSSSTR RSGG-----GGGRFSGCG GGGGFGA----GGGFSRSLVNLGGYGGYGPV
	Mmulatta partly conserved <a href="#">ENSMUG00000010962</a> 72GGFGSRSLVNLGGSKISISIVA RGGGR- SFGGGG YG GGGFGGGGFGGGGFGGGGFGGGI GGGGFGGFGSGGGGFGGGGFGGGGFGGGYGPVCP
	Fcatus partly conserved <a href="#">ENSEFCAG00000009291</a> 72GGFGSRSLVNLGGSKISISIVA RGGGRGSGFGGG GGGFGGGGFGGGGFGGGG FGGGFGGGGFGGGGFGGGGFGGGGFGGGYGPV
	Mmusculus partly conserved <a href="#">ENSMUSG00000048334</a> 72SGFGSKSLMNLGGGRSISKVA GGGGFCGGFGGGSYG GGGFGGGYGGGGFGGGG FGGGFGGSGFGGGLGGGGGFGSGGGFGGGRFSG
	Ggallus no homologue
	Trubripes no homologue
	Dreftio no homologue
	Dmelanogaster no homologue
	Celegans no homologue
	Xtropicalis no homologue
protein features	start (aa) end (aa) feature details
	2 151 COMPBias Gly/Phe/Ser-rich. lost
	2 179 REGION Head. lost
	82 82 MOD_RES Omega-N-methylarginine. lost
	180 215 REGION Coil 1A. might get lost (downstream of altered splice site)
	180 489 REGION Rod. might get lost (downstream of altered splice site)
	201 201 CONFLICT L->M (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	206 206 CONFLICT Q->K (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	216 234 REGION Linker 1. might get lost (downstream of altered splice site)
	236 326 REGION Coil 1b. might get lost (downstream of altered splice site)
	238 238 CONFLICT L->S (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	276 276 MOD_RES N6,N6-dimethyllysine. might get lost (downstream of altered splice site)
	295 295 MOD_RES Phosphotyrosine. might get lost (downstream of altered splice site)
	297 297 MOD_RES Phosphothreonine. might get lost (downstream of altered splice site)
	327 350 REGION Linker 2. might get lost (downstream of altered splice site)
	344 344 MOD_RES Phosphoserine (By similarity). might get lost (downstream of altered splice site)
	344 345 CONFLICT SL->QF (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	351 489 REGION Coil 2. might get lost (downstream of altered splice site)
	403 403 CONFLICT R->H (in Ref. 4; BAG38898). might get lost (downstream of altered splice site)
	404 404 CONFLICT V->M (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	433 433 SITE Sluiter. might get lost (downstream of altered splice site)
	447 447 CONFLICT L->M (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	463 463 CONFLICT R->C (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	466 466 CONFLICT Q->H (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	460 644 REGION Tail. might get lost (downstream of altered splice site)
	502 641 COMPBias Gly/Ser-rich. might get lost (downstream of altered splice site)
	504 504 CONFLICT S->T (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	511 512 CONFLICT TI->SM (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	564 564 CONFLICT G->S (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	613 613 CONFLICT I->S (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
	638 638 CONFLICT T->S (in Ref. 9; AAA36153). might get lost (downstream of altered splice site)
length of protein	deletion of more than 2 AA
AA sequence altered	yes
position of stopcodon in wt / mu CDS	1935 / 1737
position (AA) of stopcodon in wt / mu AA sequence	645 / 579
position of stopcodon in wt / mu cDNA	1994 / 1796
poly(A) signal	N/A
conservation nucleotide level for at changes - no scoring up to now	N/A
position of start ATG in wt / mu cDNA	60 / 60
chromosome	12
strand	-1
last intron/exon boundary	1570
theoretical NMD boundary in CDS	1460
length of CDS	1935
coding sequence (CDS) position	215 / 414
cDNA position (for insdel: last normal base / first normal base)	274 / 473
gDNA position (for insdel: last normal base / first normal base)	274 / 473
chromosomal position (for insdel: last normal base / first normal base)	53073719 / 53073918
original gDNA sequence snippet	AGTCTGTTAACTTGGTGGATATGGGGTGGTTATGGTC
altered gDNA sequence snippet	AGTCTGTTAACTTGGTGGATATGGGGTGGTTATGGTC
original cDNA sequence snippet	AGTCTGTTAACTTGGTGGATATGGGGTGGTTATGGTC
altered cDNA sequence snippet	AGTCTGTTAACTTGGTGGATATGGGGTGGTTATGGTC
wildtype AA sequence	MSRQFSRSRG YRSGGGFSG SAGIINYQR TTSSTRRSG GGGGRFSSG GGGGSFGAGG GFGSRSLVNL GSKSISISV ARGGGRSGGF GGTGGGGFG GGGFGGGFG GGIIGGGGFG GFGSGGGFG GGGGGGGG GGGGFCGPG CIGVITKMS LLDRLNVID PELDKWRE REQIKSLNQ FASFDKVRF LEQNKVLOT KWELLOVDI STRHNLEPY FESFINLR RVDLKSDDQ RLDELNMQ DMVEDYRKY EDEINRRTA ENFVITKQ VDGAYTKYD LQALNLNQ EDIFLTKLY AELSQQVQI SEVWLSND NRRLLDSE IAEVWAYED LAQSKAEAE SLYQSKYEL QITAGRHDQ VNSKIEISE LNRVQRLS EIDNKKQIS NLQSTSDAE ORGENALKDA NKNLQLEDA LQAKEDLAR LLRDVDELN TKLALDLEIA TYRLLLEGE SRSQGEAPN VSVSVSTHT TSSGGGSRG GGGYSGSG SYSSGGGSG SGGGGGGRR SYSGGSSYG SGGGSYSGG GGGGSGYOS GSSGGYRG SGGGGGSSG GSGGGSSG GSIIGROSSS GGVKSSGSS SVKVFSTYS QVTR*
mutated AA sequence	MSRQFSRSRG YRSGGGFSG SAGIINYQR TTSSTRRSG GGGGRFSSG GGGGSFGAGG GFGSRSLVNL GGGGSGGPG CPGGIDVIT INSLDPLN VEIDREIVK KSRBERIKS LNNQFASFD KVRFLQDQ VLQKWLLO QVDITSTRHN LEPYFESFIN LNRNVDLQ SDQSLDSEL KNDQEDYD RNYEDEIN RINAENFYI LKQVDGAYM TKVDLQKLD NLQEDFLT ALYQAELESQ QIQISQETWV LSKNDRSLD LQSLAEKKA QYEDAKSK AEASLYOSK YEELQITAGR HDQSVNSKI EISELNRVQ RLRSIEDNK KQSNLQDSI SDAERQENA LKDNKLNLD LEDLQAKE DLARLLRDY ELNMTKALD LEIATYRLL EGEERMSQE CAPNHSVHS TSHTSISGG SRGGGGGNG GGGSYSGG GSYSGGGGG GRRGYSYSG SYSGGGGSG GGGGGGGHG SYSGSSSGG YRGGSGGGG GSSGGRRGG GSSGSGTGR GSSSGVKSS GSSSVKFSV TYSQVTR*
speed	0.13 s

All positions are in basepairs (bp) if not explicitly stated differently.  
 AA:aa: amino acid; CDS: coding sequence; mu: mutated; NMD: nonsense-mediated mRNA decay; nt: nucleotide; wt: wildtype; TGP: 1000 Genomes Project