

Mutations *Mus musculus*

Name	Mutation	Nucleotide change ¹	Amino Acid change/Predicted Consequence
DelV265	Deletion	c.966DelTCG	Deletion of V265
T267G	Missense	c.970GAC>TGG	Thr267Gly
W268G	Missense	c.974T>G	Trp268Gly
K286E	Missense	c.998A>G	Lys268Glu
D294N	Missense	c.1052G>A	Asp294Asn
T328P	Missense	c.1154A>C	Thr328Pro
T328I	Missense	c.1155C>T	Thr328Ile
R330G	Missense	c.1160A>G	Arg330Gly
E341D	Missense	c.1195G>T	Glu341Asp
c.1253Del31bp	Deletion	c.1253Del31bp	Truncation after G360
c.1261-1287 duplication	Duplication	c.1261-1287 duplication	Frameshift after G372
H371D	Missense	c.1283C>G	His371Asp
G372S	Missense	c.1286G>A	Gly372Ser
H371fs	Frameshift	c.1287DelG	Frameshift after H371
P372fs	Frameshift	c.1292DelG	Frameshift after P372
L386fs	Frameshift	c.1330DelC	Frameshift after L386
A392fs	Frameshift	c.1351DelC	Frameshift after A392
D393fs	Frameshift	c.1351InsT	Frameshift after D393
M396fs	Frameshift	c.1363InsCG	Frameshift after M369
F398fs	Frameshift	c.1367DelAT	Frameshift after F398
S403X	Nonsense	c.1377DelCTTC	Stop at S403
DelL429-S437	Deletion	c.1458Del27bp	Deletion L429-S437

1) Nucleotide 1 is the first base of the published *M. musculus* cDNA sequence: U58988.

(AKUdatabase; <http://www.alkaptonuria.cib.csic.es/>)

(from: Manning et al. 1999, PNAS,96:11928-11933)