# Phenotypic Mouse Allele Sequence Variant **Annotation at Mouse Genome Informatics**

MGI

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## ABSTRACT

The power of the mouse as a model for human disease can only be fully exploited if researchers are able to find suitable mouse models for their human disease of interest. Many human diseases are ultimately caused by simple genomic mutations (single or multiple nucleotide variations (SNVs, MNVs) small insertions or deletions (indels)). However, the large number of genetic variants uncovered from individual patients presents challenges in identifying the causal gene or genomic regions. Although the Mouse Genome Informatics (MGI) database provides gene and genotype connections to phenotype annotations, the sequence context of the genome variants for phenotypic alleles was not yet available.

To provide researchers with a searchable, structured dataset of mouse mutations for comparative analysis, we have started annotating mouse variants, concentrating on SNVs, MNVs and small indels. These variants, characterized by their genomic position and sequence changes, are associated with engineered and spontaneous phenotypic alleles in the MGI database (www.informatics.jax.org). Variant attributes include variant type (insertion, point mutation, etc.) and molecular consequence (frameshift, stop gain, etc.). Data will be available in Human Genome Variation Society (HGVS) notation to provide transcript and protein contexts. Additional variants from large sequencing and mouse mutagenesis projects will be added to complement the manually curated data. By tying variant data and associated phenotype data to the genome, researchers will, in the near future, be able to search using human variants and find models with variants/mutations that result in the same amino acid change, have the same variant effect (missense, etc.), have the same functional impact (pathogenic, etc.), occur in the same protein domain(s), or have the same m of inheritance (recessive, dominant, etc.), resulting in phenotypes similar to a patient.

Variant data and associated phenotypic data is accessible from the Alliance of Genome Resources (www.alliancegenome.org) gene pages.

## **VARIANT ANNOTATION**

- Sequence variant data is mined from publications describing phenotypic alleles, and also provided by large-scale mutagenesis projects.
- Sources most often provide amino-acid changes, and often also transcript-level nucleotide changes.
- Sequence variants are defined by genomic coordinates and reference and variant sequences.
- Source information is entered into the web form, even if information is incorrect.
- Using various tools, source information is validated and
- genomic coordinates and sequence changes are determined. Validation of source data serves as quality control for published data:
  - a common error is using incorrect single-letter amino-acid codes (e.g. L is leucine, not lysine; lysine is K)
  - another common error is using c.466A>G type notation but using absolute transcript coordinates in stead of the proper CDS coordinates

## ACKNOWLEDGMENTS

## www.informatics.jax.org

## **ANNOTATION TOOLS**

VARIANT ANNOTATION WEB FORM to create sequence variant records for phenotypic alleles

lata from source ts20:NM\_001164785.1:c.2281C>T:p. Phe) Adamts20:NM 177431 4:c 22810 Types Types Popup Effects Effects Popu SO:1000008 (point\_mutation SO:0001583 (missense\_variant nultiple variants attached to allel All variants for allele: Abca4<tm2.1 plice acceptor varia lice\_donor\_variar lice\_region\_varia GRCm38 122102742 122102742 541 541 rameshift\_variant Cm38 122126183 122126183 C ameshift\_elongation prime UTR premature start codon gain variant

### **TRANSVAR** web interface to translate between genomic, cDNA/CDS and peptide coordinates and sequence

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M. musculus (MGI)       • File       View         10,000,000       20,000,000         94,341,000       94,3         Variants       94,3         Variants       94,3         Alf Genes       2 (ENSEMBL:ENSMUST00000035342)         NMUST00000155907 (ENSEMBL:ENSMUST000000155907)       94,3         M_001164785.1 (NCBL Gene:NM_001164785.1)       94,3         M_1777431.5 (NCBL Gene:NM_001164785.1)       94,3         M_0017316581.2 (NCBL Gene:XM_001764785.1)       94,3         M_017316581.2 (NCBL Gene:XM_001764785.1)       94,3         M_017316581.2 (NCBL Gene:XM_001761518.1)       94,3         M_017316381.2 (NCBL Gene:XM_001761518.1) <td>W         Help           30,000,000           30,000,000           341,025           94,341,           341,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025<!--</td--><td>40,000,000 Q Q Q ,030 ,030 type 1 motif T P R L Y T T V A C A C C G C T T T G T G G T G C C A A V G R N</td><td>50,000,000</td><td>943409959434111 hts20<bt-2h> G -&gt; A U V V I S C H A G I T G I C A I C T C A A C A G T A G</bt-2h></td><td>7 (123 b) Go 94,341,075 S G L P F W S S L L V F T C T G G T C T T C C A A G A C C A G A A G G</td><td></td></td>	W         Help           30,000,000           30,000,000           341,025           94,341,           341,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025 </td <td>40,000,000 Q Q Q ,030 ,030 type 1 motif T P R L Y T T V A C A C C G C T T T G T G G T G C C A A V G R N</td> <td>50,000,000</td> <td>943409959434111 hts20<bt-2h> G -&gt; A U V V I S C H A G I T G I C A I C T C A A C A G T A G</bt-2h></td> <td>7 (123 b) Go 94,341,075 S G L P F W S S L L V F T C T G G T C T T C C A A G A C C A G A A G G</td> <td></td>	40,000,000 Q Q Q ,030 ,030 type 1 motif T P R L Y T T V A C A C C G C T T T G T G G T G C C A A V G R N	50,000,000	943409959434111 hts20 <bt-2h> G -&gt; A U V V I S C H A G I T G I C A I C T C A A C A G T A G</bt-2h>	7 (123 b) Go 94,341,075 S G L P F W S S L L V F T C T G G T C T T C C A A G A C C A G A A G G	
M. musculus (MGI)       File       View         10,000,000       20,000,000         94,341,000       94,3         Variants       94,3         Variants       94,3         Att Genes       2 (ENSEMBL:ENSMUST00000035342)         NSMUST00000155907 (ENSEMBL:ENSMUST000000155907)       94,3         M_001164785.1 (NCBL_Gene:NM_001164785.1)       94,3         M_177431.5 (NCBL_Gene:NM_001164785.1)       94,3         M_001245594.3 (NCBL_Gene:XM_001781581.2)       94,3         100178157.1 (NCBL_Gene:XM_001781581.2)       94,3         M_017316581.2 (NCBL_Gene:XM_001781581.2)       94,3         M_017316581.2 (NCBL_Gene:XH_001781518.1)       94,3         Gisintegrin-like and metallopeptidase (reprolysin type)       94,3         Reference sequence       2       2       F       A       P         Q       Q       M       S       S       L       I       I         Gamta20 (MGI:2660628)	W         Help           30,000,000           30,000,000           341,025           94,341,           341,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025 </td <td>40,000,000 Q Q Q ,030 ,030 type 1 motif T P R L Y T T V A C A C C G C T T T G T G G T G C C A A V G R N</td> <td>50,000,000 ① 15 ▼ 15 94,341,050 Adam SNV Adam SNV C L P R L T E 3 A Y R D C T T A C C G A G A G A A T G G C T C T A</td> <td>943409959434111 hts20<bt-2h> G -&gt; A U V V I S C H A G I T G I C A I C T C A A C A G T A G</bt-2h></td> <td>7 (123 b) Go 94,341,075 S G L P F W S S L L V F T C T G G T C T T C C A A G A C C A G A A G G</td> <td></td>	40,000,000 Q Q Q ,030 ,030 type 1 motif T P R L Y T T V A C A C C G C T T T G T G G T G C C A A V G R N	50,000,000 ① 15 ▼ 15 94,341,050 Adam SNV Adam SNV C L P R L T E 3 A Y R D C T T A C C G A G A G A A T G G C T C T A	943409959434111 hts20 <bt-2h> G -&gt; A U V V I S C H A G I T G I C A I C T C A A C A G T A G</bt-2h>	7 (123 b) Go 94,341,075 S G L P F W S S L L V F T C T G G T C T T C C A A G A C C A G A A G G	
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M. musculus (MGI)       File       View         10,000,000       20,000,000         94,341,000       94,3         Variants       94,3         Variants       94,3         All Genes       2 (ENSEMBL:ENSMUST00000035342)         NSMUST00000155907 (ENSEMBL:ENSMUST000000155907)       M.001164785.1         M_177431.5 (NCBL Gene:NM_077451.5)       M_00652063.3 (NCBL Gene:XM_0011245594.3)         M_017245594.3 (NCBL Gene:XM_017316581.2)       M_00178151.2 (NCBL Gene:XM_017316581.2)         M_017716581.2 (NCBL Gene:XM_017316581.2)       M_001781581.2 (NCBL Gene:XM_001781581.2)         M_01781581.2 (NCBL Gene:XM_001781581.2)       M_001781581.2 (NCBL Gene:XM_001781581.2)         M_01781582.1 (NCBL Gene:XM_001781581.2)       M_01781581.2 (NCBL Gene:XM_001781581.2)         M_01781582.1 (NCBL Gene:XM_001781581.2)       M_01781582.2 (MGI 2660628)         Gisintegrin-like and metallopeptidase (reprolysin	W         Help           30,000,000           30,000,000           341,025           94,341,           341,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           94,341,           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           40,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025           41,025 </td <td>40,000,000 Q Q Q ,030 ,030 type 1 motif T P R L Y T T V A C A C C G C T T T G T G G T G C C A A V G R N</td> <td>50,000,000</td> <td>943409959434111 hts20<bt-2h> G -&gt; A U V V I S C H A G I T G I C A I C T C A A C A G T A G</bt-2h></td> <td>7 (123 b) Go 94,341,075 S G L P F W S S L L V F T C T G G T C T T C C A A G A C C A G A A G G</td> <td></td>	40,000,000 Q Q Q ,030 ,030 type 1 motif T P R L Y T T V A C A C C G C T T T G T G G T G C C A A V G R N	50,000,000	943409959434111 hts20 <bt-2h> G -&gt; A U V V I S C H A G I T G I C A I C T C A A C A G T A G</bt-2h>	7 (123 b) Go 94,341,075 S G L P F W S S L L V F T C T G G T C T T C C A A G A C C A G A A G G	
M. musculus (MGI)	W         Help           30,000,000           30,000,000           341,025           94,341, <td>40,000,000 Q Q Q Q ,030 type 1 motif T P R L Y T T V A C A C C A C G G T T V G R N C W P Q C W P Q C V V T 2</td> <td>50,000,000</td> <td>943409959434111 hts20 <bt-2h> G -&gt; A L S S V V I S C H S C H S C A C A C A G T A G T T M C N D D L Q * R</bt-2h></td> <td>7 (123 b) 94,341,075 94,040</td> <td></td>	40,000,000 Q Q Q Q ,030 type 1 motif T P R L Y T T V A C A C C A C G G T T V G R N C W P Q C W P Q C V V T 2	50,000,000	943409959434111 hts20 <bt-2h> G -&gt; A L S S V V I S C H S C H S C A C A C A G T A G T T M C N D D L Q * R</bt-2h>	7 (123 b) 94,341,075 94,040	
M. musculus (MGI)  Pile View 10,000,000 20,000,000  94,341,000 94,3 Variants All Genes All Genes 2 (ENSEMBL:ENSMUST0000033342) All Genes NSMUST00000155907 (ENSEMBL:ENSMUST00000155907) M_001164785.1 (NCBLGene:NM_001164785.1) M_177431.5 (NCBLGene:NM_001164785.1) M_0015052063.3 (NCBLGene:XM_001164785.3) M_001781581.2 (NCBLGene:XM_001316581.2) I_001781581.2 (NCBLGene:XM_001316581.2) I_001781581.2 (NCBLGene:XM_001316581.2) I_001781581.2 (NCBLGene:XM_001316581.2) I_001781581.2 (NCBLGene:XM_001781581.2) I_001781581.2 (NCBLGene:	W         Help           30,000,000           30,000,000           341,025           94,341, <td>40,000,000 Q Q Q Q ,030 type 1 motif T P R L Y T T V A C A C C A C G G T T V G R N C W P Q C W P Q C V V T 2</td> <td>50,000,000</td> <td>943409959434111 hts20 <bt-2h> G -&gt; A L S S V V I S C H S C H S C A C A C A G T A G T T M C N D D L Q * R</bt-2h></td> <td>7 (123 b) 94,341,075 94,040</td> <td></td>	40,000,000 Q Q Q Q ,030 type 1 motif T P R L Y T T V A C A C C A C G G T T V G R N C W P Q C W P Q C V V T 2	50,000,000	943409959434111 hts20 <bt-2h> G -&gt; A L S S V V I S C H S C H S C A C A C A G T A G T T M C N D D L Q * R</bt-2h>	7 (123 b) 94,341,075 94,040	
M. musculus (MGI)       • File       View         10,000,000       20,000,000         94,341,000       94,3         Variants       94,3         Variants       94,3         Alf Genes       2 (EN3EMBL:EN3MU3T00000035342)         MSMUST00000155907 (ENSEMBL:ENSMU3T000000155907)       M001164785.1         M_001164785.1 (NCBL Gene:NM_001164785.1)       M_000520863.3)         M_000520863.3 (NCBL Gene:XM_0011245594.3)       M_00178151.2 (NCBL Gene:XM_001781581.2)         M_00178151.2 (NCBL Gene:XM_001781581.2)       M_001781581.2 (NCBL Gene:XM_001781581.2)         M_00178151.1 (NCBL Gene:XM_001781581.2)       M_001781581.2 (NCBL Gene:XM_001781581.2)         M_00178151.2 (NCBL Gene:XM_001781581.2)       M_001781581.2 (NCBL Gene:XM_001781581.2)         M_00178151.1 (NCBL Gene:XM_001781581.2)       M_001781581.2 (NCBL Gene:XM_001781581.2)         M_00178151.2 (NCBL Gene:XM_001781581.2)       M_00178151.2 (NCBL Gene:XM_001781581.2)         M_00178151.1 (NCBL Gene:XM_001781581.2)       M_00178151.2 (NCBL Gene:XM_001781518.1)         M_00178151.2 (NCBL Gene:XM_001781518.1)       M_001781518.1 (NCBL Gene:XM_001781518.1)         M_00178151.2 (NCBL Gene:XM_001781518.1)       M_01781518.1 (NCBL Gene:XM_001781518.1)         M_00178151.2 (NCBL Gene:XM_001781518.1)       M_01781518.1 (NCBL Gene:XM_001781518.1)         M_0178151.2 (NCBL Gene:XM_001781518.1)       M_0	w Help 30,000,000 341,025 94,341, with thrombospondin E A D I A G R H C G C G T C T G T A C C G T C T G T A C C G T C T G T A C C C T C C T G T A C C C T C C T G T A C C V Y	40,000,000 QQQQ ,030 type 1 motif T P R L Y T V V A C A C C A C G G T T G G G G G C C A A G V G R N C W P Q C V V T 2	50,000,00	$\frac{1}{3} + \frac{1}{3} + \frac{1}$	7 (123 b) 94,341,075 9	

-show-all -c 'chr15:94341055G>A' MISSENSE\_VARIANT, MISSENSE\_VARIANT, MISSENSE\_VARIANT, MISSENSE\_VARIANT

,MISSENSE\_VARIANT,NON\_CODING\_TRANSCRIPT\_EXON\_VARIANT,NON\_CODING\_T RANSCRIPT EXON VARIANT

Adamts20:NM\_001164785.1:c.2281C>T:p.(Leu761Phe),Adamts20:NM\_177431.4:c.2281C> T:p.(Leu761Phe),Adamts20:XM\_006520863.3:c.2308C>T:p.(Leu770Phe),Adamts20:XM\_01 1245594.2:c.2281C>T:p.(Leu761Phe),Adamts20:XM\_017316581.1:c.547C>T:p.(Leu183Ph e),Adamts20:XR 001781517.1:n.1037C>T:,Adamts20:XR 001781518.1:n.2350C>T:

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