

# ***Mtm1*** Cas9-KO Strategy

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# Project Overview

**Project Name**

***Mtm1***

**Project type**

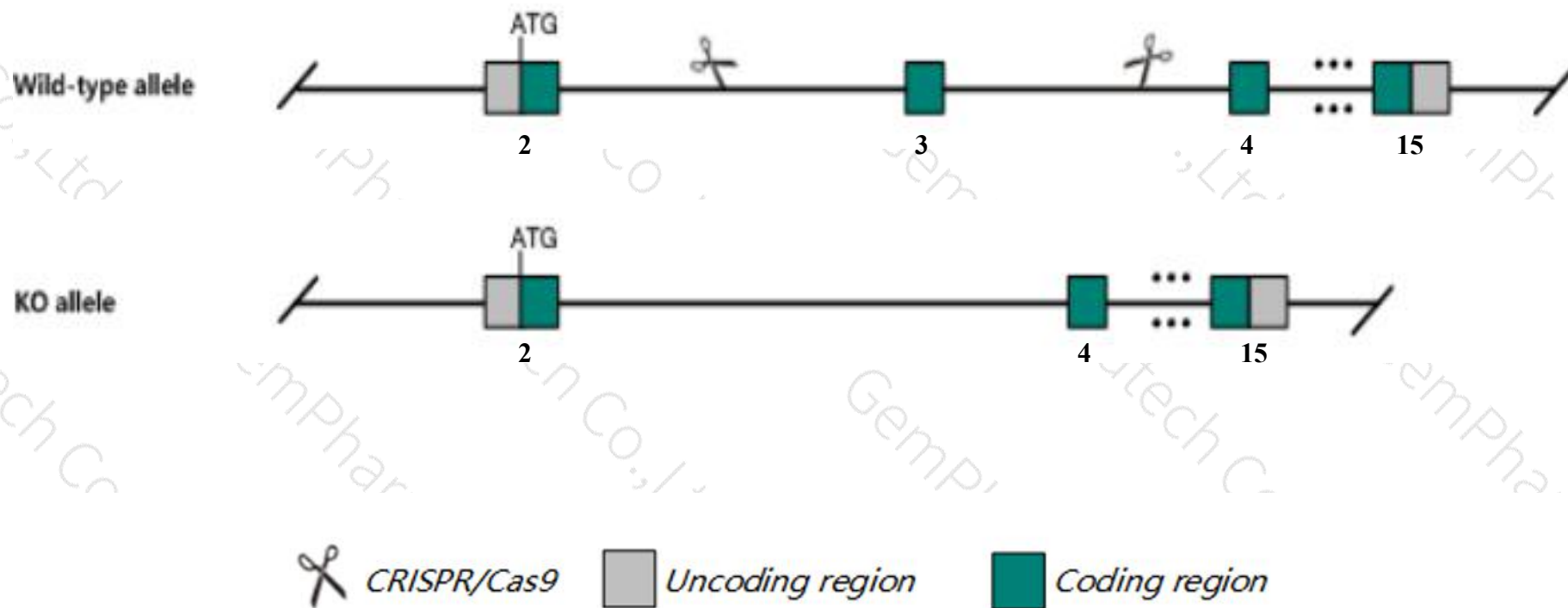
**Cas9-KO**

**Strain background**

**C57BL/6J**

# Knockout strategy

This model will use CRISPR/Cas9 technology to edit the *Mtm1* gene. The schematic diagram is as follows:



- The *Mtm1* gene has 13 transcripts. According to the structure of *Mtm1* gene, exon3 of *Mtm1-213* (ENSMUST00000171933.7) transcript is recommended as the knockout region. The region contains 73bp coding sequence. Knock out the region will result in disruption of protein function.
- In this project we use CRISPR/Cas9 technology to modify *Mtm1* gene. The brief process is as follows: CRISPR/Cas9 system

- According to the existing MGI data, hemizygotes for targeted null mutations develop a generalized, progressive myopathy beginning around 1 month and leading to death at 6-14 weeks of age. mutant mice show amyotrophy with accumulation of central nuclei in skeletal muscle fibers.
- Transcript *Mtm1-208* may not be affected.
- The *Mtm1* gene is located on the ChrX. If the knockout mice are crossed with other mice strains to obtain double gene positive homozygous mouse offspring, please avoid the two genes on the same chromosome.
- This strategy is designed based on genetic information in existing databases. Due to the complexity of biological processes, all risk of the gene knockout on gene transcription, RNA splicing and protein translation cannot be predicted at the existing technology level.



# Gene information (NCBI)

## Mtm1 X-linked myotubular myopathy gene 1 [Mus musculus (house mouse)]

Gene ID: 17772, updated on 13-Mar-2020

### Summary

**Official Symbol** Mtm1 provided by [MGI](#)

**Official Full Name** X-linked myotubular myopathy gene 1 provided by [MGI](#)

**Primary source** [MGI:MGI:1099452](#)

**See related** [Ensembl:ENSMUSG00000031337](#)

**Gene type** protein coding

**RefSeq status** VALIDATED

**Organism** [Mus musculus](#)

**Lineage** Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus

**Also known as** AF073996, Mtm, mKIAA4176

**Expression** Ubiquitous expression in large intestine adult (RPKM 2.8), placenta adult (RPKM 2.7) and 28 other tissues [See more](#)

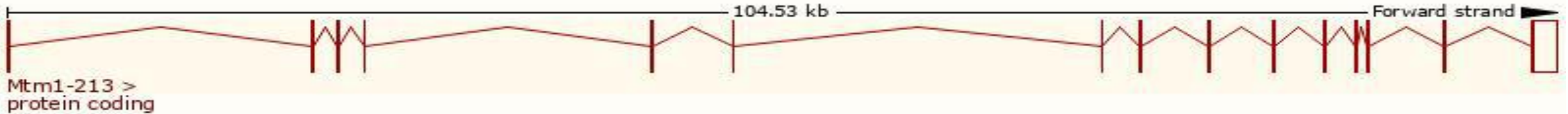
**Orthologs** [human](#) [all](#)

# Transcript information (Ensembl)

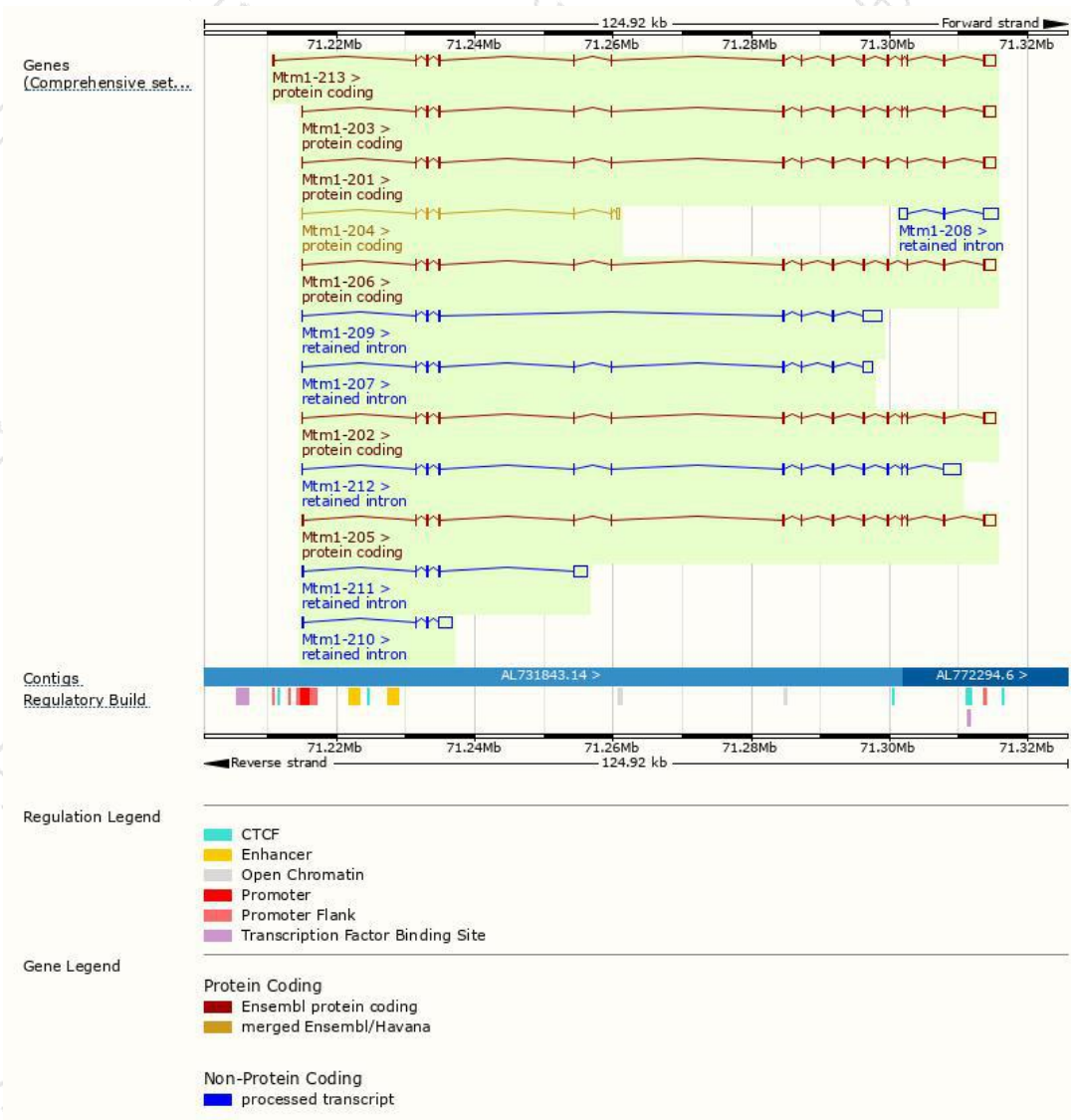
The gene has 13 transcripts,all transcripts are shown below:

Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	Flags
Mtm1-213	<a href="#">ENSMUST00000171933.7</a>	3488	<a href="#">603aa</a>	Protein coding	<a href="#">CCDS30177</a>	<a href="#">Q9Z2C5</a>	TSL:1 GENCODE basic APPRIS is a system to annotate alternatively spliced transcripts based on a range of computational methods to identify the most functionally important transcript(s) of a gene. APPRIS P1
Mtm1-203	<a href="#">ENSMUST00000061970.11</a>	3379	<a href="#">603aa</a>	Protein coding	<a href="#">CCDS30177</a>	<a href="#">Q9Z2C5</a>	TSL:1 GENCODE basic APPRIS is a system to annotate alternatively spliced transcripts based on a range of computational methods to identify the most functionally important transcript(s) of a gene. APPRIS P1
Mtm1-201	<a href="#">ENSMUST00000025391.10</a>	3286	<a href="#">572aa</a>	Protein coding	<a href="#">CCDS53090</a>	<a href="#">B1AW21</a>	TSL:1 GENCODE basic
Mtm1-204	<a href="#">ENSMUST00000101501.9</a>	770	<a href="#">162aa</a>	Protein coding	<a href="#">CCDS53089</a>	<a href="#">Q3UDN6</a>	TSL:1 GENCODE basic
Mtm1-205	<a href="#">ENSMUST00000114617.1</a>	3338	<a href="#">603aa</a>	Protein coding	-	<a href="#">Q9Z2C5</a>	TSL:5 GENCODE basic APPRIS is a system to annotate alternatively spliced transcripts based on a range of computational methods to identify the most functionally important transcript(s) of a gene. APPRIS P1
Mtm1-202	<a href="#">ENSMUST00000033700.11</a>	3316	<a href="#">603aa</a>	Protein coding	-	<a href="#">Q9Z2C5</a>	TSL:5 GENCODE basic APPRIS is a system to annotate alternatively spliced transcripts based on a range of computational methods to identify the most functionally important transcript(s) of a gene. APPRIS P1
Mtm1-206	<a href="#">ENSMUST00000114621.7</a>	3257	<a href="#">572aa</a>	Protein coding	-	<a href="#">B1AW21</a>	TSL:5 GENCODE basic
Mtm1-212	<a href="#">ENSMUST00000156452.7</a>	4111	No protein	Retained intron	-	-	TSL:5
Mtm1-208	<a href="#">ENSMUST00000129722.1</a>	3469	No protein	Retained intron	-	-	TSL:1
Mtm1-209	<a href="#">ENSMUST00000134859.7</a>	3412	No protein	Retained intron	-	-	TSL:1
Mtm1-211	<a href="#">ENSMUST00000152810.1</a>	2353	No protein	Retained intron	-	-	TSL:5
Mtm1-207	<a href="#">ENSMUST00000126208.7</a>	2339	No protein	Retained intron	-	-	TSL:1
Mtm1-210	<a href="#">ENSMUST00000151415.7</a>	2042	No protein	Retained intron	-	-	TSL:1

The strategy is based on the design of *Mtm1-213* transcript,the transcription is shown below:

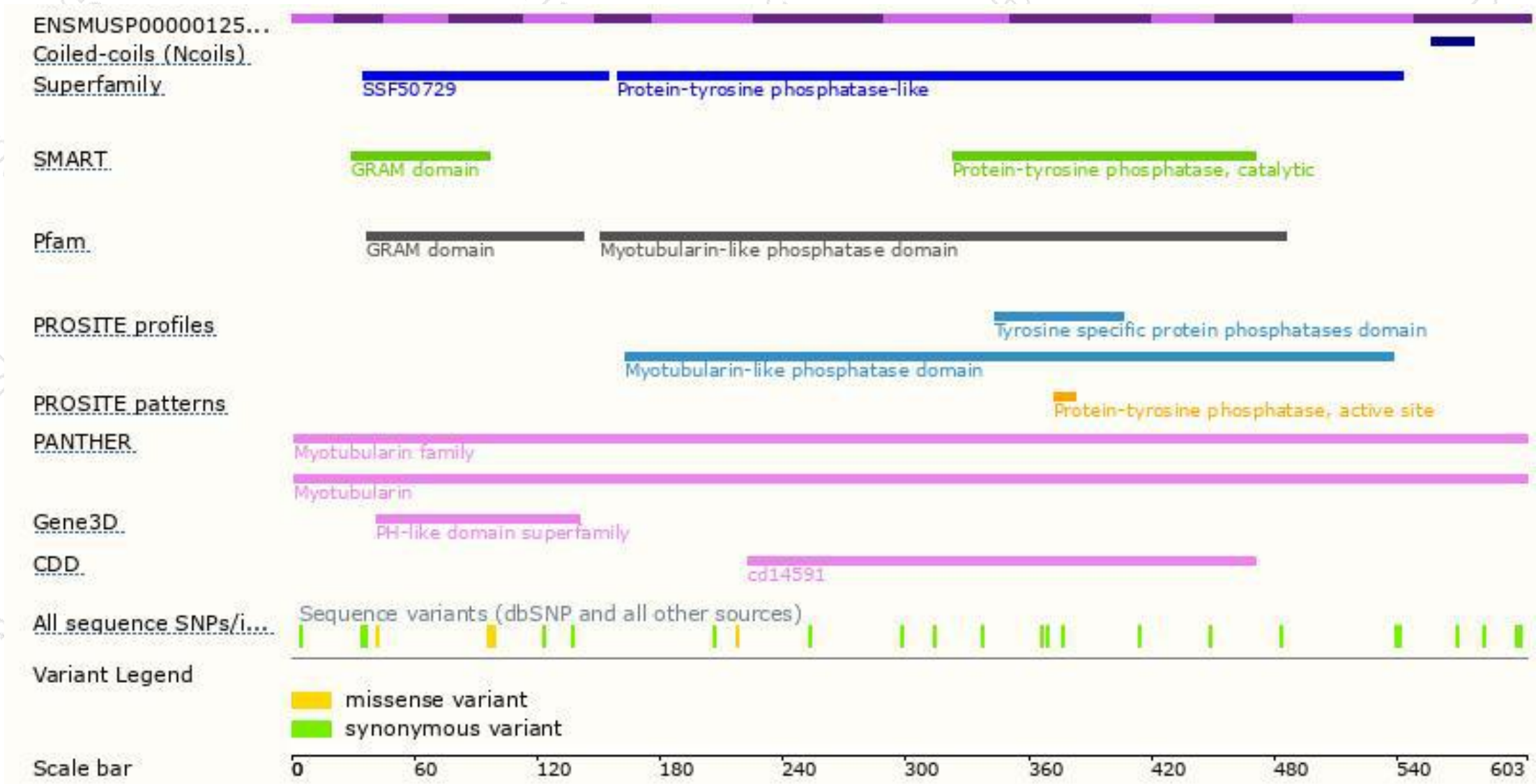


# Genomic location distribution

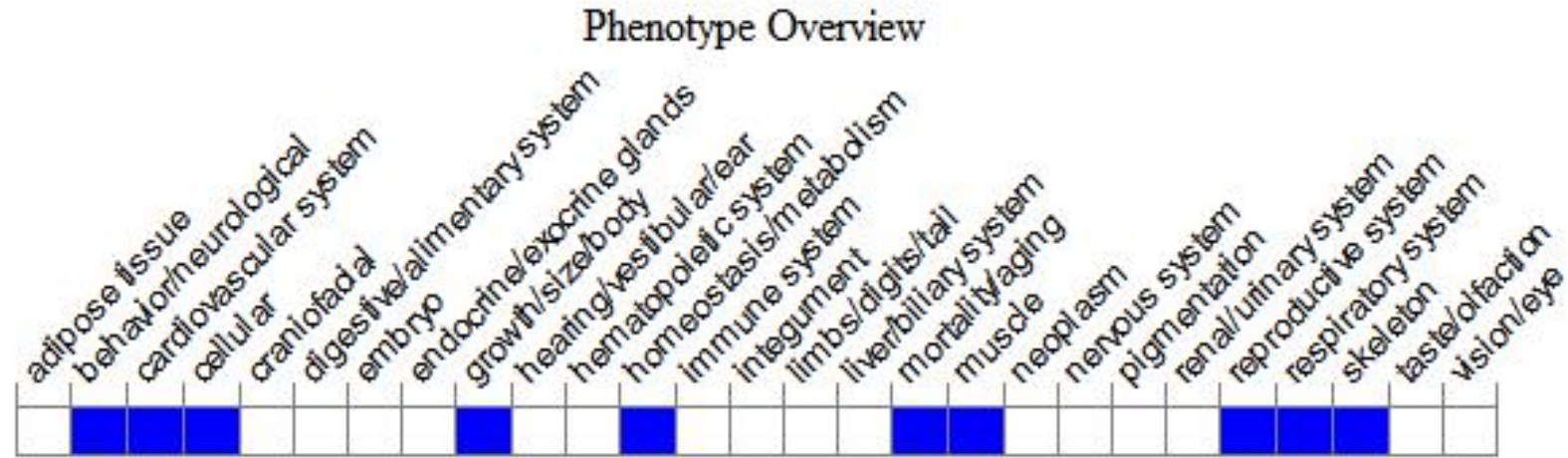




# Protein domain



# Mouse phenotype description(MGI )



*Phenotypes affected by the gene are marked in blue. Data quoted from MGI database(<http://www.informatics.jax.org/>).*

According to the existing MGI data, hemizygotes for targeted null mutations develop a generalized, progressive myopathy beginning around 1 month and leading to death at 6-14 weeks of age. Mutant mice show amyotrophy with accumulation of central nuclei in skeletal muscle fibers.

If you have any questions, you are welcome to inquire.

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