GemPharmatech™

Col6a1 Cas9-CKO Strategy

Designer: Xiangli Bian

Reviewer: Jiaojiao Yan

Design Date: 2023-7-19

Overview

Target Gene Name

- *Col6a1*

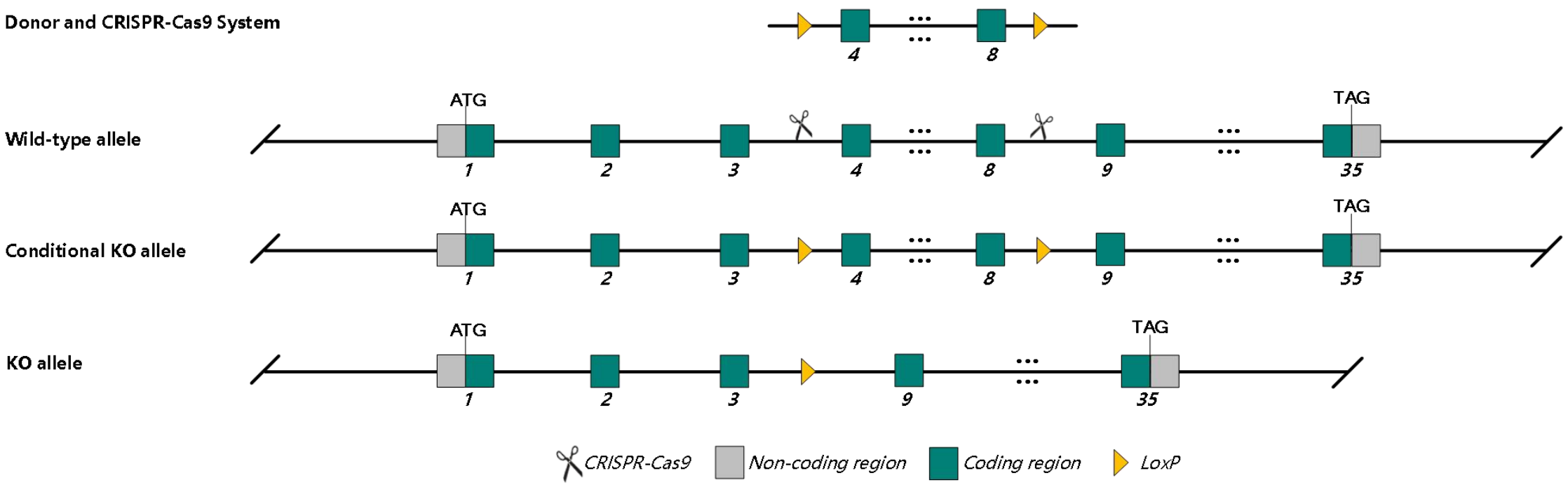
Project Type

- Cas9-CKO

Genetic Background

- C57BL/6JGpt

Strain Strategy



Schematic representation of CRISPR-Cas9 engineering used to edit the *Col6a1* gene.

Technical Information

- The *Col6a1* gene has 2 transcripts. According to the structure of *Col6a1* gene, exon 4-8 of *Col6a1*-201 (ENSMUST00000001147.5) is recommended as the knockout region. The region contains 376 bp of coding sequence. Knocking out the region will result in disruption of gene function.
- In this project we use CRISPR-Cas9 technology to modify *Col6a1* gene. The brief process is as follows: CRISPR-Cas9 system and Donor were microinjected into the fertilized eggs of C57BL/6JGpt mice. Fertilized eggs were transplanted to obtain positive F0 mice which were confirmed by PCR and on-target amplicon sequencing. A stable F1-generation mouse strain was obtained by mating positive F0-generation mice with C57BL/6JGpt mice and confirmation of the desired mutant allele was carried out by PCR and on-target amplicon sequencing.
- The flox mice will be knocked out after mating with mice expressing Cre recombinase, resulting in the loss of function of the target gene in specific tissues and cell types.

Gene Information

Col6a1 collagen, type VI, alpha 1 [*Mus musculus* (house mouse)]

Download Datasets

Gene ID: 12833, updated on 18-May-2023

Summary

Official Symbol	Col6a1 provided by MGI
Official Full Name	collagen, type VI, alpha 1 provided by MGI
Primary source	MGI:MGI:88459
See related	Ensembl:ENSMUSG00000001119 AllianceGenome:MGI:88459
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	Col6a-1
Summary	Predicted to enable collagen binding activity and platelet-derived growth factor binding activity. Acts upstream of or within cellular response to amino acid stimulus. Located in extracellular matrix and sarcolemma. Is expressed in several structures, including alimentary system; brain; genitourinary system; heart and pericardium; and sensory organ. Used to study Bethlem myopathy and Ullrich congenital muscular dystrophy. Human ortholog(s) of this gene implicated in Ullrich congenital muscular dystrophy. Orthologous to human COL6A1 (collagen type VI alpha 1 chain). [provided by Alliance of Genome Resources, Apr 2022]
Expression	Broad expression in subcutaneous fat pad adult (RPKM 393.3), ovary adult (RPKM 227.8) and 16 other tissues See more
Orthologs	human all

NEW **Try the new Gene table**
Try the new Transcript table

Genomic context

Location: 10 C1; 10 39.71 cM — See Col6a1 in Genome Data Viewer

Exon count: 35

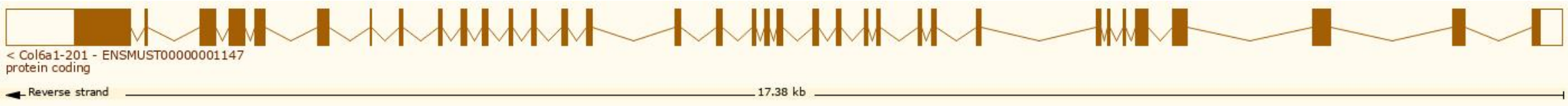
https://www.ncbi.nlm.nih.gov/gene/12833

Transcript Information

The gene has 2 transcripts, all transcripts are shown below:

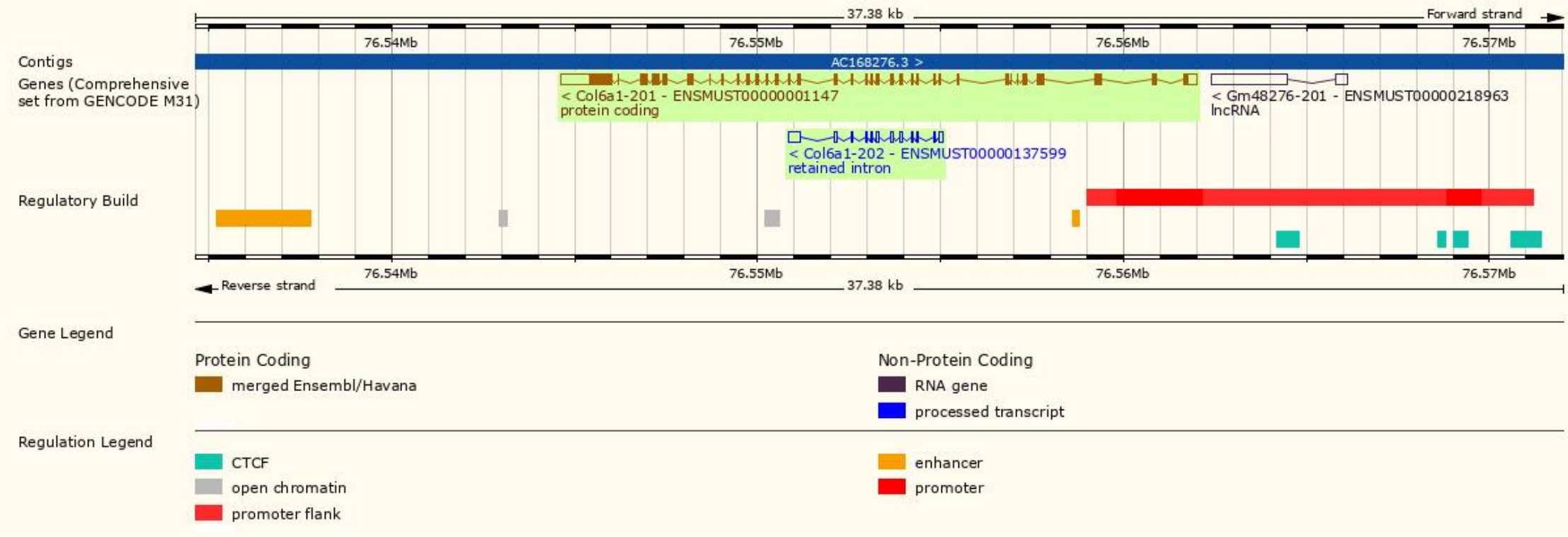
Transcript ID	Name ▲	bp	Protein	Biotype	CCDS	UniProt Match	Flags
ENSMUST00000001147.5	Col6a1-201	4100	1025aa	Protein coding	CCDS23952	Q04857	Ensembl Canonical GENCODE basic APPRIS P1 TSL:1
ENSMUST00000137599.2	Col6a1-202	925	No protein	Retained intron		-	TSL:5

The strategy is based on the design of *Col6a1*-201 transcript, the transcription is shown below:



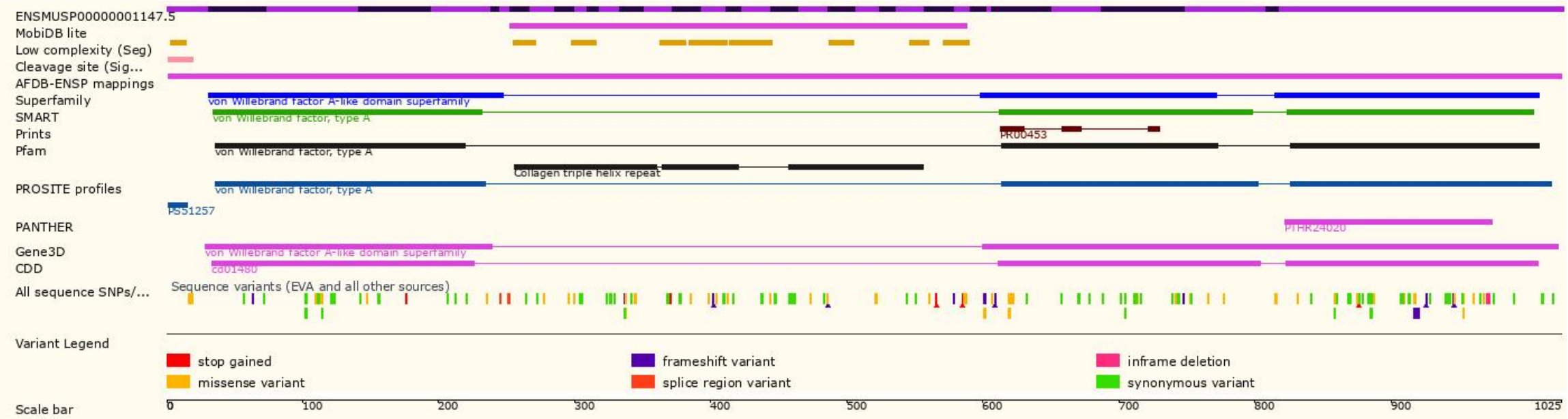
Source: http://asia.ensembl.org/

Genomic Information



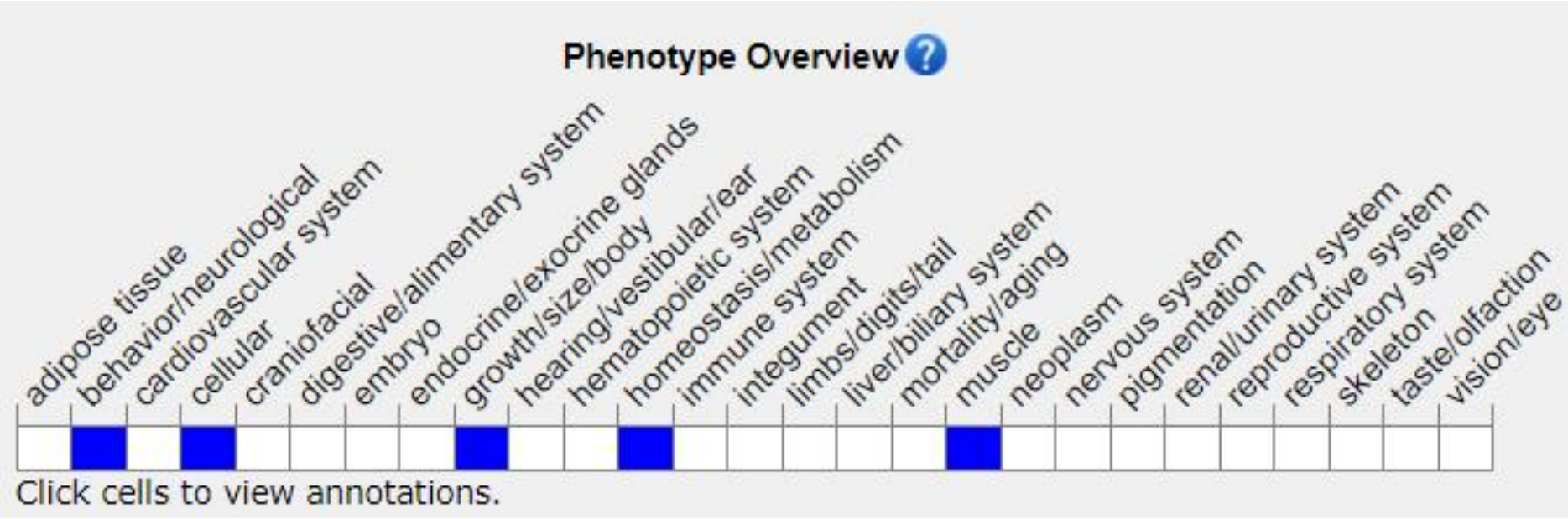
Source: http://asia.ensembl.org/

Protein Information



Source: https://www.ensembl.org

Mouse Phenotype Information (MGI)



Mice homozygous for this targeted mutation display a myopathic disorder that resembles human Bethlem myopathy. Loss of contractile strength in affected muscles is associated with an unexpected latent mitochondrial dysfunction in myofibers, as well as spontaneous apoptosis.

Source: https://www.informatics.jax.org

Important Information

- According to the existing MGI data, mice homozygous for this targeted mutation display a myopathic disorder that resembles human Bethlem myopathy. Loss of contractile strength in affected muscles is associated with an unexpected latent mitochondrial dysfunction in myofibers, as well as spontaneous apoptosis.
- A part of amino acid sequence will still remain at the N-terminal of *Col6a1*-201 transcript.
- This stratergy may not affect *Col6a1*-202 transcript.
- *Col6a1* is located on Chr 10. If the knockout mice are crossed with other mouse strains to obtain double homozygous mutant offspring, please avoid the situation that the second gene is 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 loxp insertion on gene transcription, RNA splicing and protein translation cannot be predicted at the existing technology level.