

Pkhd1 Cas9-KO Strategy

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Project Overview



Project Name

Pkhd1

Project type

Cas9-KO

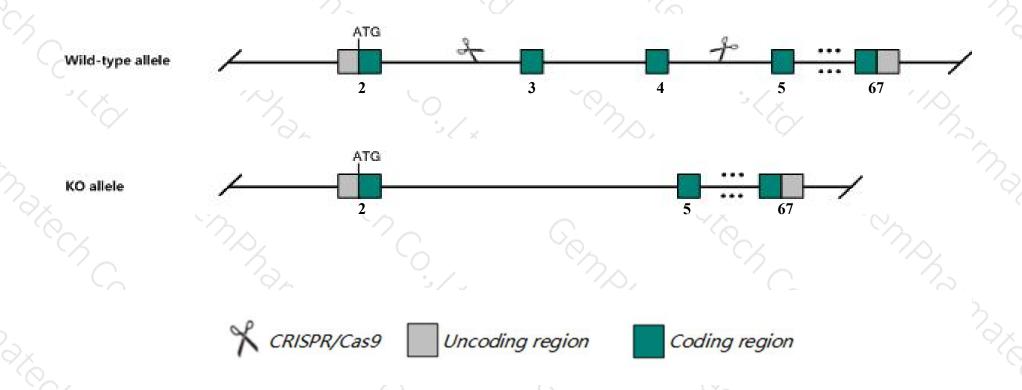
Strain background

C57BL/6JGpt

Knockout strategy



This model will use CRISPR/Cas9 technology to edit the *Pkhd1* gene. The schematic diagram is as follows:



Technical routes



The *Pkhd1* gene has 4 transcripts. According to the structure of *Pkhd1* gene, exon3-exon4 of *Pkhd1-201* (ENSMUST00000088448.11) transcript is recommended as the knockout region. The region contains 220bp coding sequence Knock out the region will result in disruption of protein function.

In this project we use CRISPR/Cas9 technology to modify *Pkhd1* gene. The brief process is as follows: CRISPR/Cas9 system

Notice



According to the existing MGI data, Mice homozygous for a mutation in this gene display variable progressive liver cysts and fibrosis, but do not display kidney cysts and are fertile. Mice homozygous for a hypomorphic and null allele display renal, pancreatic, billiary and liver cysts.

The *Pkhd1* gene is located on the Chr1. 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



Pkhd1 polycystic kidney and hepatic disease 1 [Mus musculus (house mouse)]

Gene ID: 241035, updated on 19-Mar-2019

Summary

☆ ?

Official Symbol Pkhd1 provided by MGI

Official Full Name polycystic kidney and hepatic disease 1 provided by MGI

Primary source MGI:MGI:2155808

See related Ensembl:ENSMUSG00000043760

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 Al118496, Al182499, FPC, Tigm1

Expression Biased expression in kidney adult (RPKM 6.2), genital fat pad adult (RPKM 1.3) and 3 other tissuesSee more

Orthologs <u>human</u> all

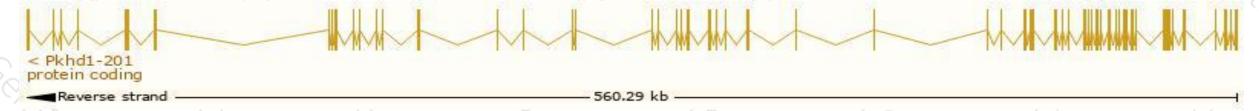
Transcript information Ensembl



The gene has 4 transcripts, all transcripts are shown below:

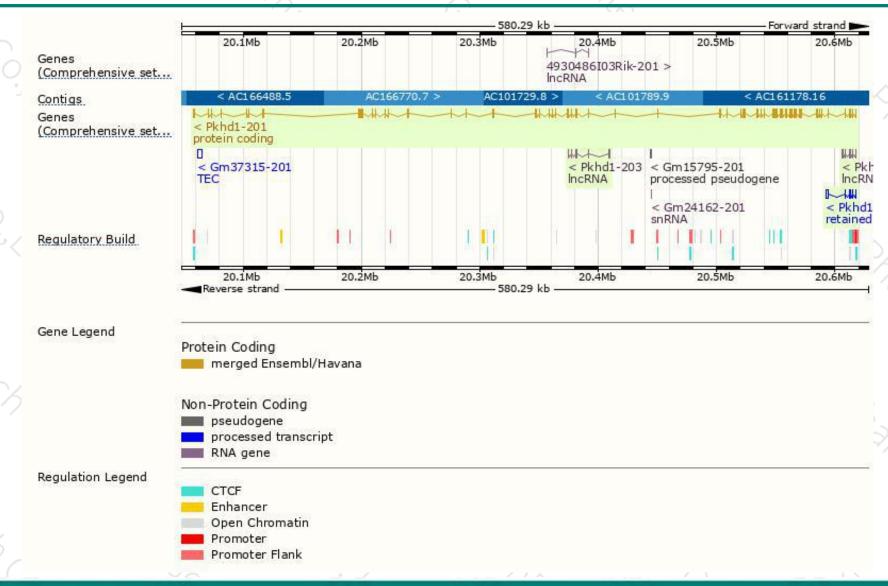
Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	Flags
Pkhd1-201	ENSMUST00000088448.11	12935	4059aa	Protein coding	CCDS14841	E9PZ36	TSL:1 GENCODE basic APPRIS P1
Pkhd1-204	ENSMUST00000147480.7	1636	No protein	Retained intron	5	670	TSL:1
Pkhd1-202	ENSMUST00000128051.1	921	No protein	IncRNA	-	0 <u>4</u> 0	TSL:1
Pkhd1-203	ENSMUST00000132647.1	615	No protein	IncRNA	2	100	TSL:3

The strategy is based on the design of *Pkhd1-201* transcript, The transcription is shown below



Genomic location distribution





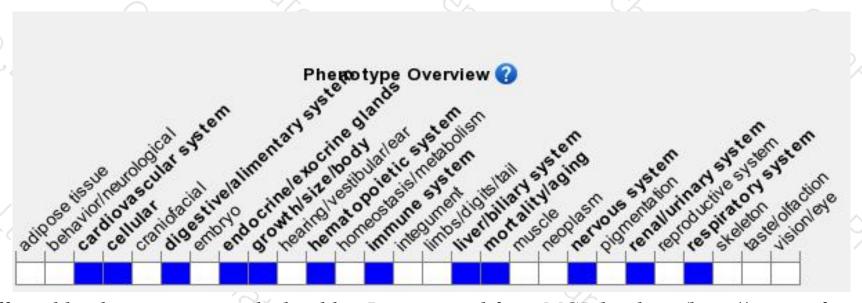
Protein domain





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, Mice homozygous for a mutation in this gene display variable progressive liver cysts and fibrosis, but do not display kidney cysts and are fertile. Mice homozygous for a hypomorphic and null allele display renal, pancreatic, billiary and liver cysts.



If you have any questions, you are welcome to inquire. Tel: 400-9660890





