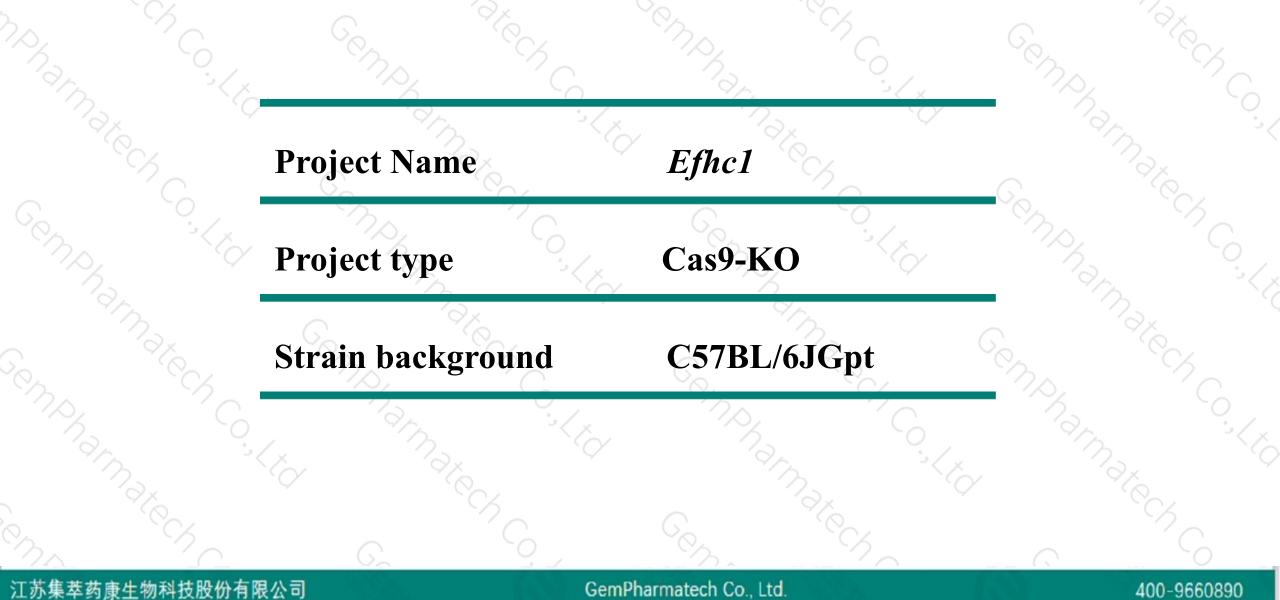


Efhc1 Cas9-KO Strategy

Designer: Reviewer: Design Date: JiaYu Xiaojing Li 2020-2-11

Project Overview

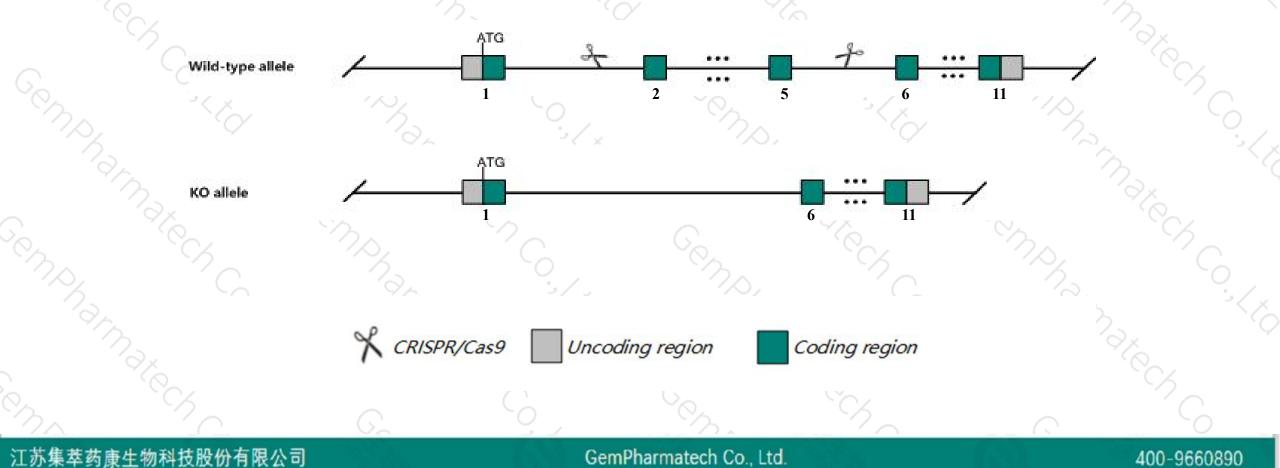




Knockout strategy



This model will use CRISPR/Cas9 technology to edit the *Efhc1* gene. The schematic diagram is as follows:





- The *Efhc1* gene has 2 transcripts. According to the structure of *Efhc1* gene, exon2-exon5 of *Efhc1-201* (ENSMUST00000038447.5) transcript is recommended as the knockout region. The region contains 853bp coding sequence. Knock out the region will result in disruption of protein function.
- > In this project we use CRISPR/Cas9 technology to modify *Efhc1* gene. The brief process is as follows: CRISPR/Cas9 system



- According to the existing MGI data, Mice homozygous or heterozygous for a null mutation display myoclonus and increased susceptibility to pharmacologically induced seizures. Homozygous mice also display enlarged brain ventricles and reduced hippocampal size.
- The *Efhc1* 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)



\$?

Efhc1 EF-hand domain (C-terminal) containing 1 [Mus musculus (house mouse)]

Gene ID: 71877, updated on 31-Jan-2019

Summary

Official Symbol	Efhc1 provided by MGI
Official Full Name	EF-hand domain (C-terminal) containing 1 provided by MGI
Primary source	MGI:MGI:1919127
See related	Ensembl:ENSMUSG00000041809
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	1700029F22Rik, mRib72-1, myoclonin1
Expression	Biased expression in testis adult (RPKM 84.4) and ovary adult (RPKM 3.4)See more
Orthologs	human all

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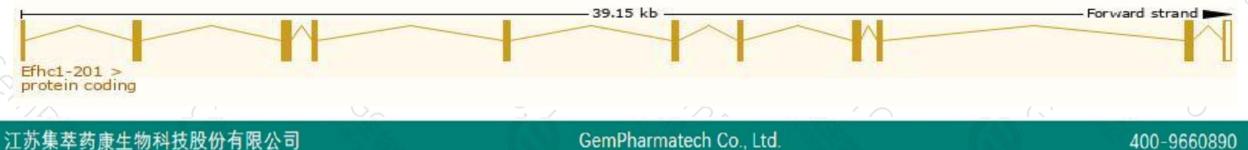
Transcript information (Ensembl)



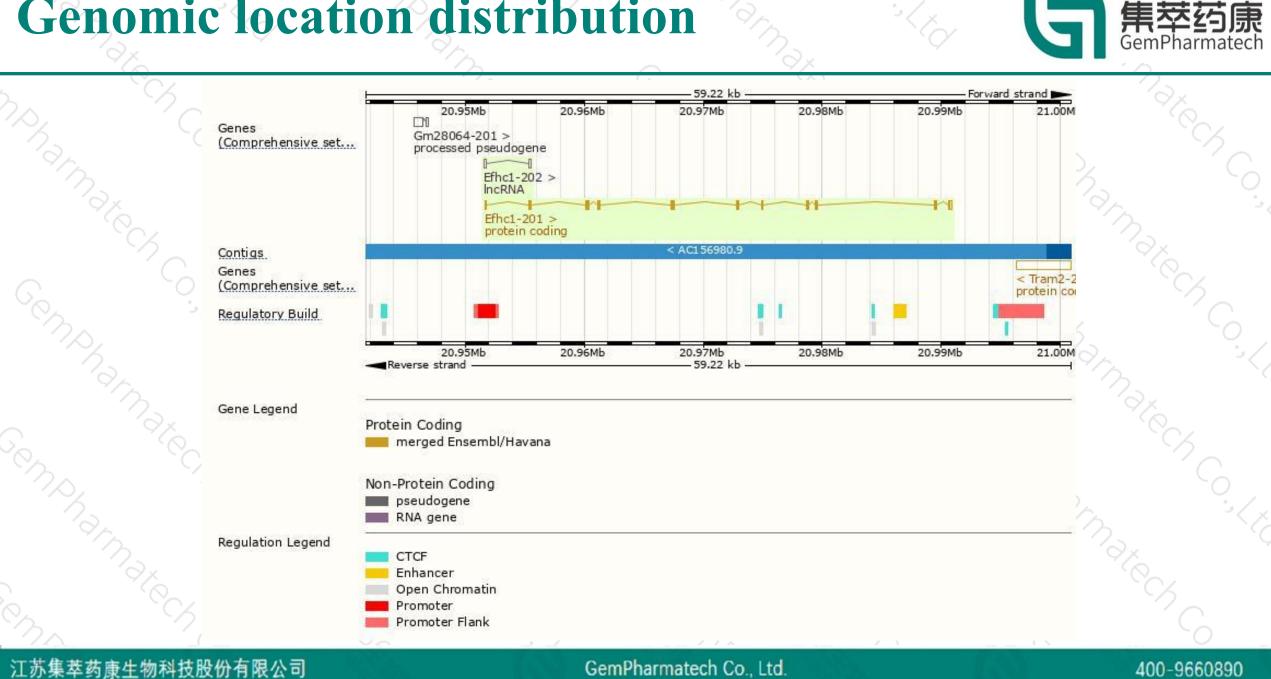
The gene has 2 transcripts, all transcripts are shown below:

Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	Flags	
Efhc1-201	ENSMUST0000038447.5	2146	<u>648aa</u>	Protein coding	CCDS48227	B2CKC6	TSL:1 GENCODE basic APPRIS P1	
Efhc1-202	ENSMUST00000160782.1	357	No protein	IncRNA	-	2 .	TSL:3	

The strategy is based on the design of *Efhc1-201* transcript, The transcription is shown below



Genomic location distribution



Protein domain



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ENSMUSP00000042 Superfamily		a).	-16		12			81. -	EF-ha	nd domain pa
SMART		Uncharacte	rised domain	DM10					• 1	EF-hand doma
Pfam.		Domain of u	nknown funct	tion DUF1126		-	8		-	
PROSITE profiles	11	Uncharacte	rised domain	DM10				-	E	F-hand doma
PANTHER	PTHR12086:S	F9								
Cape2D	EF-hand doma	ain-containing pr	otein EFHC1/	EFHC2/EFHB			- 14			
Gene3D		2,30,29,170							1.1	0.238.10
CDD			30 82 92							EF-hand domi
All sequence SNPs/i	Sequence v	ariants (dbSNP	and all othe	ar sources)	10.1	1 1	1	0.00	1 11 110	013 0
Variant Legend	3									C
		ise variant mous variant								
Scale bar	0 6	0 120	180	240	300	360	420	480	540	648
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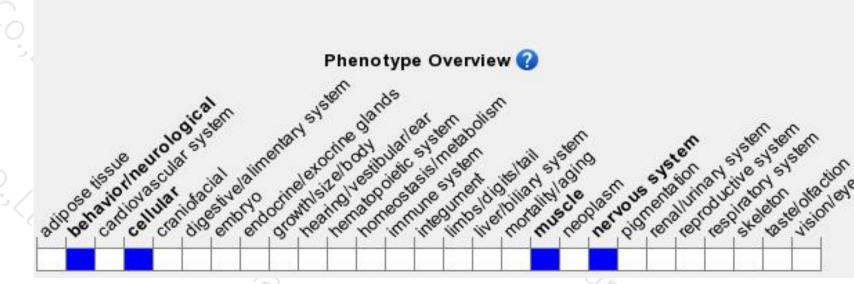
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Mouse phenotype description(MGI)





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 or heterozygous for a null mutation display myoclonus and increased susceptibility to pharmacologically induced seizures. Homozygous mice also display enlarged brain ventricles and reduced hippocampal size.

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



