

GEPHE SUMMARY

RORB (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="RORB">#gephebase-summary-title)	Gephebase Gene	GP00002343	GephelD
	Entry Status	Courtier	Main curator
Published			

PHENOTYPIC CHANGE

Trait Category		Trait	
Behavior (https://www.gephebase.org/search-criteria?/and+Trait Category="Behavior">#gephebase-summary-title)			
gait (saltatorial locomotion) (https://www.gephebase.org/search-criteria?/and+Trait=^gait (saltatorial locomotion)#gephebase-summary-title)		Trait State in Taxon A	
can jump		Trait State in Taxon B	
cannot jump, bipedal gait using their front legs		Ancestral State	
Taxon A		Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Domesticated">#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Oryctolagus cuniculus (#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryctolagus+cuniculus">#gephebase-summary-title)		Oryctolagus cuniculus (#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryctolagus+cuniculus">#gephebase-summary-title)	
rabbit	Common Name	rabbit	Common Name
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	Synonyms	Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagidae	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagidae	Lineage
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)	Parent	Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)	Parent
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)	NCBI Taxonomy ID	9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
	sauteur d'Alfort		Taxon B Description

GENOTYPIC CHANGE

RORB	Generic Gene Name	UniProtKB Canis lupus familiaris
-	Synonyms	GenebankID or UniProtKB
9612.ENSCAFP00000031876 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9612.ENSCAFP00000031876)	String	Canis lupus familiaris
Belongs to the nuclear hormone receptor family.	Sequence Similarities	
	GO - Molecular Function	
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)		
GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)		
GO:0004879 : nuclear receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004879)		

GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)

GO:0008502 : melatonin receptor activity

(<https://www.ebi.ac.uk/QuickGO/term/GO:0008502>)

GO - Biological Process

GO:0006357 : regulation of transcription by RNA polymerase II

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)

GO:0042752 : regulation of circadian rhythm

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042752>)

GO:0046548 : retinal rod cell development

(<https://www.ebi.ac.uk/QuickGO/term/GO:0046548>)

GO:0045668 : negative regulation of osteoblast differentiation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0045668>)

GO:0046549 : retinal cone cell development

(<https://www.ebi.ac.uk/QuickGO/term/GO:0046549>)

GO - Cellular Component

GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

Presumptive Null

Unknown ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=%27Unknown%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27Unknown%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27SNP%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27SNP%27#gephebase-summary-title))

SNP Coding Change

-

Molecular Details of the Mutation

a change from GT to AT in the 5' donor site of intron 9 (chr1: 61,103,503bp). The mutation disrupts the normal splicing of RORB.

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Association Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Association%20Mapping%27#gephebase-summary-title))

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

A loss-of-function mutation in RORB disrupts saltatorial locomotion in rabbits. (2021) (<https://pubmed.ncbi.nlm.nih.gov/33764968>)

Authors

Carneiro M; Vieillard J; Andrade P; Boucher S; Afonso S; Blanco-Aguiar JA; Santos N; Branco J; Esteves PJ; Ferrand N; Kullander K; Andersson L

Abstract

Saltatorial locomotion is a type of hopping gait that in mammals can be found in rabbits, hares, kangaroos, and some species of rodents. The molecular mechanisms that control and fine-tune the formation of this type of gait are unknown. Here, we take advantage of one strain of domesticated rabbits, the sauteur d'Alfort, that exhibits an abnormal locomotion behavior defined by the loss of the typical jumping that characterizes wild-type rabbits. Strikingly, individuals from this strain frequently adopt a bipedal gait using their front legs. Using a combination of experimental crosses and whole genome sequencing, we show that a single locus containing the RAR related orphan receptor B gene (RORB) explains the atypical gait of these rabbits. We found that a splice-site mutation in an evolutionary conserved site of RORB results in several aberrant transcript isoforms incorporating intronic sequence. This mutation leads to a drastic reduction of RORB-positive neurons in the spinal cord, as well as defects in differentiation of populations of spinal cord interneurons. Our results show that RORB function is required for the performance of saltatorial locomotion in rabbits.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Splicing

