

## GEPHE SUMMARY

<p>RORB (<a +rorb"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+RORB"#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002343</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Behavior (<a +behavior"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Behavior"#gephebase-summary-title</a>)</p> <p>gait (saltatorial locomotion) (<a +gait+(saltatorial+locomotion)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+gait+(saltatorial+locomotion)+"#gephebase-summary-title</a>)</p> <p>can jump</p> <p>cannot jump, bipedal gait using their front legs</p> <p>Taxon A</p> <p>Domesticated (<a +domesticated"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated"#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Oryctolagus cuniculus (<a +oryctolagus+cuniculus"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Oryctolagus+cuniculus"#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>rabbit</p> <p>Synonyms</p> <p>Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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; Oryctolagus</p> <p>Parent</p> <p>Oryctolagus () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984</a>)</p> <p>NCBI Taxonomy ID</p> <p>9986 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Oryctolagus cuniculus (<a +oryctolagus+cuniculus"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Oryctolagus+cuniculus"#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>rabbit</p> <p>Synonyms</p> <p>Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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; Oryctolagus</p> <p>Parent</p> <p>Oryctolagus () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984</a>)</p> <p>NCBI Taxonomy ID</p> <p>9986 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Taxon B Description</p> <p>sauteur d'Alfort</p>
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## GENOTYPIC CHANGE

<p>RORB</p> <p>-</p> <p>9612.ENSCAFP00000031876 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9612.ENSCAFP00000031876">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9612.ENSCAFP00000031876</a>)</p> <p>Belongs to the nuclear hormone receptor family.</p> <p>GO - Molecular Function</p> <p>GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001228">https://www.ebi.ac.uk/QuickGO/term/GO:0001228</a>)</p> <p>GO:0008270 : zinc ion binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008270">https://www.ebi.ac.uk/QuickGO/term/GO:0008270</a>)</p> <p>GO:0004879 : nuclear receptor activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004879">https://www.ebi.ac.uk/QuickGO/term/GO:0004879</a>)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p>	<p>F1PET8 (<a href="http://www.uniprot.org/uniprot/F1PET8">http://www.uniprot.org/uniprot/F1PET8</a>)</p> <p>F1PET8 (<a href="https://www.ncbi.nlm.nih.gov/nucore/F1PET8">https://www.ncbi.nlm.nih.gov/nucore/F1PET8</a>)</p>	<p>UniProtKB Canis lupus familiaris</p> <p>GenebankID or UniProtKB Canis lupus familiaris</p>
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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=~Unknown^#gephebase-summary-title>)

Molecular Type

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title>)

SNP Coding Change

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Molecular Details of the Mutation

a change from GT to AT in the 5â€™ 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=~Association Mapping^#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-Aguar 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

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Splicing

