GEPHE SUMMARY

Gephebase Gene
LIPH (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^LIPH^#gephebase-summary-title)

GepheID
GP00000550

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Hair length (https://www.gephebase.org/search-criteria?/and+Trait=^Hair length^#gephebase-summary-title)

Trait State in Taxon A
Oryctolagus cuniculus

Trait State in Taxon B
Oryctolagus cuniculus - rex type with short hair and curled vibrissae

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryctolagus cuniculus^#gephebase-summary-title)

Common Name
rabbit

Synonyms
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits

Rank
species

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; Oryctolagus

Parent
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

NCBI Taxonomy ID
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryctolagus cuniculus^#gephebase-summary-title)

Common Name
rabbit

Synonyms
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits

Rank
species

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; Oryctolagus

Parent
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

NCBI Taxonomy ID
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Liph

Synonyms
P3; H06; lpd1; lpd2; Lpdlr; PLA1B; mPA-PLA1; D16Wsu119e; C130037N08Rik

String
10090.ENSMUSP00000062310 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000062310)

Sequence Similarities
Belongs to the AB hydrolase superfamily. Lipase family.

GO - Molecular Function
GO:0008201 : heparin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008201)
GO:0052689 : carboxylic ester hydrolase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0052689)
GO:0004620 : phospholipase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004620)

GO - Biological Process
GO:0016042 : lipid catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0016042)

UniProtKB Mus musculus
Q8CIV3 (http://www.uniprot.org/uniprot/Q8CIV3)

GenebankID or UniProtKB
NP_001075575 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001075575)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

1bp deletion resulting in frameshift

Experimental Evidence

Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)

Main Reference

A deletion in exon 9 of the LIPH gene is responsible for the rex hair coat phenotype in rabbits (Oryctolagus cuniculus). (2011) (https://pubmed.ncbi.nlm.nih.gov/21552526)

Authors

Diribarne M; Mata X; Chantry-Darmon C; Vaiman A; Auvinet G; Bouet S; Deretz S; Cribiu EP; de Rochambeau H; Allain D; GuÃ©rin G

Abstract

The fur of common rabbits is constituted of 3 types of hair differing in length and diameter while that of rex animals is essentially made up of amazingly soft down-hair. Rex short hair coat phenotypes in rabbits were shown to be controlled by three distinct loci. We focused on the "r1" mutation which segregates at a simple autosomal-recessive locus in our rabbit strains. A positional candidate gene approach was used to identify the rex gene and the corresponding mutation. The gene was primo-localized within a 40 cM region on rabbit chromosome 14 by genome scanning families of 187 rabbits in an experimental mating scheme. Then, fine mapping refined the region to 0.5 cM (Z = 78) by genotyping an additional 359 offspring for 94 microsatellites present or newly generated within the first defined interval. Comparative mapping pointed out a candidate gene in this 700 kb region, namely LIPH (Lipase Member H). In humans, several mutations in this major gene cause alopecia, hair loss phenotypes. The rabbit gene structure was established and a deletion of a single nucleotide was found in LIPH exon 9 of rex rabbits (1362delA). This mutation results in a frameshift and introduces a premature stop codon potentially shortening the protein by 19 amino acids. The association between this deletion and the rex phenotype was complete, as determined by its presence in our rabbit families and among a panel of 60 rex and its absence in all 60 non-rex rabbits. This strongly suggests that this deletion, in a homozygous state, is responsible for the rex phenotype in rabbits.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA001566/9986/