

GEPHE SUMMARY

LIPH (#gephebase-summary-title)	Gephebase Gene	GP00000550	GephelD
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Trait Category			
Morphology (#gephebase-summary-title)	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 (#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Oryctolagus cuniculus (#gephebase-summary-title))	Oryctolagus cuniculus (#gephebase-summary-title))	Oryctolagus cuniculus (#gephebase-summary-title))	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; Oryctolagus	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	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?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

Generic Gene Name			
Liph	Synonyms	Q8C1V3 (http://www.uniprot.org/uniprot/Q8C1V3)	UniProtKB Mus musculus
P3; H06; lpd1; lpd2; Lpdlr; PLA1B; mPA-PLA1; D16Wsu119e; C130037N08Rik	String	NP_001075575 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001075575)	GenebankID or UniProtKB
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:0016042 : lipid catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0016042)	GO - Biological Process		

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; Guérin 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/>