

GEPHE SUMMARY

	Gephebase Gene		GepheID
LIPH (https://www.gephebase.org/search-criteria?/and+GeneGephebase=LIPH^#gephebase-summary-title)		GP00000550	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=Morphology^#gephebase-summary-title)		Trait	
Hair length (https://www.gephebase.org/search-criteria?/and+Trait=Hairlength^#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+TaxonomicStatus=Domesticated^#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Oryctolagus+cuniculus^#gephebase-summary-title)		Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Oryctolagus+cuniculus^#gephebase-summary-title)	
	Common Name		Common Name
rabbit		rabbit	
	Synonyms		Synonyms
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits		Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	
	Rank		Rank
species		species	
	Lineage		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		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		Parent
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)		Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)		9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Liph		Q8CIV3 (http://www.uniprot.org/uniprot/Q8CIV3)	
	Synonyms		GenebankID or UniProtKB
P3; H06; lpd1; lpd2; Lpdlr; PLA1B; mPA-PLA1; D16Wsu119e; C130037N08Rik		NP_001075575 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001075575)	
	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)			

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>)

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes~#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	Molecular Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title)	Aberration Type
1-9 bp	Deletion Size
1bp deletion resulting in frameshift	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	Experimental Evidence
A deletion in exon 9 of the LIPH gene is responsible for the rex hair coat phenotype in rabbits (<i>Oryctolagus cuniculus</i>). (2011) (https://pubmed.ncbi.nlm.nih.gov/21552526)	Main Reference
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	Authors
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.	Abstract
	Additional References

RELATED GEPHE

No matches found.	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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