GEPHE SUMMARY

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

GepheID
GP00000648

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

Trait State in Taxon A
Oryctolagus cuniculus

Trait State in Taxon B
Oryctolagus cuniculus - dilute breeds

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

Common Name
-

Synonyms
-

Rank
genus

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

Parent
Leporidae (rabbits and hares) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9979)

NCBI Taxonomy ID
9984 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
-

Synonyms
-

Rank
genus

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

Parent
Leporidae (rabbits and hares) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9979)

NCBI Taxonomy ID
9984 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Oryctolagus cuniculus - dilute breeds

GENOTYPIC CHANGE

Generic Gene Name
Mlph

Synonyms
In; I1Rk3; Slac-2a; AW228792; D1Wsu84e; l(1)-3Rk; 2210418F23Rik; 5031433I09Rik; Ln; Slac2a

String
10090.ENSMUSP00000027528 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000027528)

Sequence Similarities
-

GO - Molecular Function
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0017137 : Rab GTPase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017137)
GO:0003779 : actin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003779)
GO:0030674 : protein binding, bridging

UniProtKB Mus musculus
Q91V27 (http://www.uniprot.org/uniprot/Q91V27)

GenebankID or UniProtKB
()

(https://www.ebi.ac.uk/QuickGO/term/GO:0030674)
GO:0051010 : microtubule plus-end binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0051010)
GO:0017022 : myosin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017022)
GO:0031489 : myosin V binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031489)

GO - Biological Process

GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0032400 : melanosome localization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032400)
GO:0006605 : protein targeting (https://www.ebi.ac.uk/QuickGO/term/GO:0006605)

GO - Cellular Component

GO:0015629 : actin cytoskeleton (https://www.ebi.ac.uk/QuickGO/term/GO:0015629)
GO:0030425 : dendrite (https://www.ebi.ac.uk/QuickGO/term/GO:0030425)
GO:0048471 : perinuclear region of cytoplasm
(https://www.ebi.ac.uk/QuickGO/term/GO:0048471)
GO:0005815 : microtubule organizing center
(https://www.ebi.ac.uk/QuickGO/term/GO:0005815)
GO:0030864 : cortical actin cytoskeleton
(https://www.ebi.ac.uk/QuickGO/term/GO:0030864)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0001725 : stress fiber (https://www.ebi.ac.uk/QuickGO/term/GO:0001725)
GO:0016461 : unconventional myosin complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0016461)

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

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

Main Reference

A frameshift mutation in the melanophilin gene causes the dilute coat colour in rabbit (Oryctolagus cuniculus) breeds. (2014) (https://pubmed.ncbi.nlm.nih.gov/24320228)

Authors

Fontanesi L; Scotti E; Allain D; Dall'olio S

Abstract

In rabbit, the dilute locus is determined by a recessive mutated allele (d) that causes the dilution of both eumelanic and pheomelanic pigmentations. In mice, similar phenotypes are determined by mutations in the myosin VA, Rab27a and melanophilin (MLPH) genes. In this study, we investigated the rabbit MLPH gene and showed that a mutation in this gene appears responsible for the dilute coat colour in this species. Checkered Giant F1 families segregating for black and grey (diluted or blue) coat colour were first genotyped for a complex indel in intron 1 of the MLPH gene that was completely associated with the coat colour phenotype (Î¸ = 0.00; LOD = 4.82). Then, we sequenced 6357 bp of the MLPH gene in 18 rabbits of different coat colours, including blue animals. A total of 165 polymorphisms were identified: 137 were in non-coding regions and 28 were in coding exons. One of them was a frameshift deletion in exon 5. Genotyping the half-sib families confirmed the complete cosegregation of this mutation with the blue coat colour. The mutation was analysed in 198 rabbits of 23 breeds. All Blue Vienna and all other blue/grey/ash rabbits in other breeds (Californian, Castor Rex, Checkered Giant, English Spot, Fairy Marburg and Fairy Pearly) were homozygous for this deletion. The identification of MLPH as the responsible gene for the dilute locus in rabbit provides a natural animal model for human Griscelli syndrome type 3 and a new mutant to study the role of this gene on pigmentation.

Additional References

RELATED GEPHE

Related Genes

4 (Agouti (ASIP), MC1R, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9984^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Melanophilin (MLPH)^/and+Taxon ID=^9984^/or+Gene Gephebase=^Melanophilin (MLPH)^/and+Taxon ID=^9984^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS