GEPHE SUMMARY

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

GepheID
GP00002332

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
WT

Trait State in Taxon B
dilute coat color

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

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

GenebankID or UniProtKB
()

(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

c.585delG p.L195LfsX123*

Experimental Evidence

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

Main Reference

Two-exon skipping within MLPH is associated with coat color dilution in rabbits. (2013) (https://pubmed.ncbi.nlm.nih.gov/24376820)

Authors

Lehner S; GÃ¤hle M; Dierks C; Stelter R; Gerber J; Brehm R; Distl O

Abstract

Coat color dilution turns black coat color to blue and red color to cream and is a characteristic in many mammalian species. Matings among Netherland Dwarf, Loh, and Lionhead Dwarf rabbits over two generations gave evidence for a monogenic autosomal recessive inheritance of coat colour dilution. Histological analyses showed non-uniformly distributed, large, agglomerating melanin granules in the hair bulbs of coat color diluted rabbits. We sequenced the cDNA of MLPH in two dilute and one black rabbit for polymorphism detection. In both color diluted rabbits, skipping of exons 3 and 4 was present resulting in altered amino acids at p.QGL[37-39]QWA and a premature stop codon at p.K40*. Sequencing of genomic DNA revealed a c.111-5C>A splice acceptor mutation within the polypyrimidine tract of intron 2 within MLPH. This mutation presumably causes skipping of exons 3 and 4. In 14/15 dilute rabbits, the c.111-5C>A mutation was homozygous and in a further dilute rabbit, heterozygous and in combination with a homozygous frame shift mutation within exon 6 (c.585delG). In conclusion, our results demonstrated a colour dilution associated MLPH splice variant causing a strongly truncated protein (p.Q37QfsX4). An involvement of further MLPH-associated mutations needs further investigations.

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=^9986^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism https://omia.org/OMIA000031/9913/