GEPHE SUMMARY

Gephebase Gene
Melanophilin (MLPH)

GepheID
GP00000646

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
Neovison vison

Trait State in Taxon B
Neovison vison - Silverblue

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Neovison vison

Common Name
American mink

Synonyms
Mustela vison; American mink; mink; Mustela vision

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; Laurasiatheria; Carnivora; Caniformia; Mustelidae; Mustelinae; Neovison

Parent
Neovison () - (Rank: genus)

NCBI Taxonomy ID
452646

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Neovison vison

Common Name
American mink

Synonyms
Mustela vison; American mink; mink; Mustela vision

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; Laurasiatheria; Carnivora; Caniformia; Mustelidae; Mustelinae; Neovison

Parent
Neovison () - (Rank: genus)

NCBI Taxonomy ID
452646

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Mlph

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

String
10090.ENSMUSP00000027528

Sequence Similarities
-

GO - Molecular Function
GO:0046872 : metal ion binding
GO:0017137 : Rab GTPase binding
GO:0003779 : actin binding
GO:0030674 : protein binding, bridging
GO:0051010 : microtubule plus-end binding
GO:0017022 : myosin binding
GO:0031489 : myosin V binding

GO - Biological Process
GO:0043473 : pigmentation
GO:0030318 : melanocyte differentiation

UniProtKB Mus musculus
Q91V27

GenebankID or UniProtKB

GO:0032400 : melanosome localization
GO:0006605 : protein targeting

GO - Cellular Component
GO:0015629 : actin cytoskeleton
GO:0030425 : dendrite
GO:0048471 : perinuclear region of cytoplasm
GO:0005815 : microtubule organizing center
GO:0030864 : cortical actin cytoskeleton
GO:0042470 : melanosome
GO:0001725 : stress fiber
GO:0016461 : unconventional myosin complex

Presumptive Null
Yes

Molecular Type
Coding

Aberration Type
Deletion

Deletion Size
100-999 bp

Molecular Details of the Mutation
deletion including intron 7 and exon 8

Experimental Evidence
Candidate Gene

Main Reference
New insights into the melanophilin (MLPH) gene controlling coat color phenotypes in American mink. (2013)

Authors
Cirera S; Markakis MN; Christensen K; Anistoroaei R

Abstract
The mutation causing the Silverblue color type (pp) is one of the most used recessive mutations within American mink (Neovison vison) fur farming, since it is involved in some of the popular color types such as Violet and Saphire which originate from a combination of recessive mutations. In the present study, the genomic and mRNA sequences of the melanophilin (MLPH) gene were studied in Violet, Silverblue and wild-type (wt) mink animals. Although breeding schemes and previous literature indicates that the Violet (aammpp) phenotype is a triple recessive color type involving the same locus as the Silverblue (pp) color type, our findings indicate different genotypes at the MLPH locus. Upon comparison at genomic level, we identified two deletions of the entire intron 7 and of the 5′ end of intron 8 in the sequence of the Silverblue MLPH gene. When investigating the mRNA, the Silverblue animals completely lack exon 8, which encodes 65 residues, of which 47 define the Myosin Va (MYO5A) binding domain. This may cause the incorrect anchoring of the MLPH protein to MYO5A in Silverblue animals, resulting in an improper pigmentation as seen in diluted phenotypes. Additionally, in the MLPH mRNA of wt, Violet and Silverblue phenotypes, part of intron 8 is retained resulting in a truncated MLPH protein, which is 359 residues long in wt and Violet and 284 residues long in Silverblue. Subsequently, our findings point out that the missing actin-binding domain, in neither of the 3 analyzed phenotypes affects the transport of melanosomes or the consequent final pigmentation. Moreover, the loss of the major part of the MYO5A domain in the Silverblue MLPH protein seems to be the responsible for the dilute phenotype. Based on our genomic DNA data, genetic tests for selecting Silverblue and Violet carrier animals can be performed in American mink.

Additional References

RELATED GEPHE

Related Genes
No matches found.
Related Haplotypes
1

COMMENTS