GEPHE SUMMARY

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

GepheID
GP00000647

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
Neovison vison

Trait State in Taxon B
Neovison vison - Violet

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

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

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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 452645)

NCBI Taxonomy ID
452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 452646)

is Taxon A an Infraspecies?
No

Taxon B

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

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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 452645)

NCBI Taxonomy ID
452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 452646)

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

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

-

Molecular Details of the Mutation

Splice donor site variation in exon 8 resulting in a premature stop codon c.901+1G>A

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

New insights into the melanophilin (MLPH) gene controlling coat color phenotypes in American mink. (2013) (https://pubmed.ncbi.nlm.nih.gov/23747352)

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.

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Additional References

Genome analysis identifies the mutant genes for common industrial Silverblue and Hedlund white coat colours in American mink. (2019) (https://pubmed.ncbi.nlm.nih.gov/30872653)

RELATED GEPHE

Related Genes

2 (tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^452646^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Melanophilin (MLPH)^/and+Taxon ID=^452646^/or+Gene Gephebase=^Melanophilin (MLPH)^/and+Taxon ID=^452646^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Splicing @AllelicSeries https://omia.org/OMIA000031/452646/