

GEPHE SUMMARY

	Gephebase Gene		GepheID
Melanophilin (MLPH) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Melanophilin (MLPH)"#gephebase-summary-title)		GP00000646	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category	
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category="Morphology"#gephebase-summary-title)		
	Trait	
Coloration (coat) (<a "="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait=" coat)"#gephebase-summary-title)		
	Trait State in Taxon A	
Neovison vison		
	Trait State in Taxon B	
Neovison vison - Silverblue		
	Ancestral State	
Taxon A		
	Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Domesticated"#gephebase-summary-title)		

Taxon A	Latin Name	Taxon B	Latin Name
Neovison vison (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Neovison vison"#gephebase-summary-title)		Neovison vison (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Neovison vison"#gephebase-summary-title)	
	Common Name		Common Name
American mink		American mink	
	Synonyms		Synonyms
Mustela vison; American mink; mink; Mustela vison species		Mustela vison; American mink; mink; Mustela vison species	
	Rank		Rank
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		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	
	Lineage		Lineage
	Parent		Parent
Neovison () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452645)		Neovison () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452645)	
452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452646)	NCBI Taxonomy ID	452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452646)	NCBI Taxonomy ID
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Mlph		Q91V27 (http://www.uniprot.org/uniprot/Q91V27)	
	Synonyms		GenebankID or UniProtKB
In; l1Rk3; Slac-2a; AW228792; D1Wsu84e; l(1)-3Rk; 2210418F23Rik; 5031433l09Rik; 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			

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

Deletion Size

100-999 bp

Molecular Details of the Mutation

deletion including intron 7 and exon 8

Experimental Evidence

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

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 Sapphire 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 (aammp) 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

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](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](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

<https://omia.org/OMIA000031/452646/@AllelicSeries>

