

## GEPHE SUMMARY

	Gephebase Gene		GepheID
Melanophilin (MLPH) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase="Melanophilin (MLPH)"#gephebase-summary-title)		GP00000649	
	Entry Status	Martin	Main curator
Published			

## PHENOTYPIC CHANGE

	Trait Category		
Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> 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="</a> 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 ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status="Domesticated"#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Oryctolagus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> Synonyms="Oryctolagus"#gephebase-summary-title)		Oryctolagus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> Synonyms="Oryctolagus"#gephebase-summary-title)	
	Common Name		Common Name
-		-	
	Synonyms		Synonyms
-		-	
	Rank		Rank
genus		genus	
	Lineage		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		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		Parent
Leporidae (rabbits and hares) - (Rank: family) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9979">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9979</a> )		Leporidae (rabbits and hares) - (Rank: family) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9979">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9979</a> )	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9984 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984</a> )		9984 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984</a> )	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Oryctolagus cuniculus - dilute breeds	

## GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Mlph		Q91V27 ( <a href="http://www.uniprot.org/uniprot/Q91V27">http://www.uniprot.org/uniprot/Q91V27</a> )	
	Synonyms		GenebankID or UniProtKB
In; I1Rk3; Slac-2a; AW228792; D1Wsu84e; I(1)-3Rk; 2210418F23Rik; 5031433I09Rik; Ln; Slac2a		()	
	String		
10090.ENSMUSP00000027528 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000027528">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000027528</a> )			
	Sequence Similarities		
-			
	GO - Molecular Function		
GO:0046872 : metal ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a> )			
GO:0017137 : Rab GTPase binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0017137">https://www.ebi.ac.uk/QuickGO/term/GO:0017137</a> )			
GO:0003779 : actin binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003779">https://www.ebi.ac.uk/QuickGO/term/GO:0003779</a> )			
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

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

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

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

-

c.111-5C>A (exon skipping resulting in frameshift and truncated protein)

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

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

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; GÄ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=~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](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

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