

GEPHE SUMMARY

Agouti (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] Agouti [^] #gephebase-summary-title)	Gephebase Gene	GP00001975	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category= [^] Morphology [^] #gephebase-summary-title)	Trait Category
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait = [^] Coloration (coat) [^] #gephebase-summary-title)	Trait
Peromyscus maniculatus - New Hampshire - WT	Trait State in Taxon A
Peromyscus maniculatus - New Hampshire - melanic	Trait State in Taxon B
Data not curated	Ancestral State
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status= [^] Intraspecific [^] #gephebase-summary-title)	Taxonomic Status

Taxon A	Latin Name	Taxon B	Latin Name
Peromyscus maniculatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Peromyscus maniculatus [^] #gephebase-summary-title)	Peromyscus maniculatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Peromyscus maniculatus [^] #gephebase-summary-title)	Peromyscus maniculatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Peromyscus maniculatus [^] #gephebase-summary-title)	Peromyscus maniculatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Peromyscus maniculatus [^] #gephebase-summary-title)
North American deer mouse	Common Name	North American deer mouse	Common Name
North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB Mamm 74965; MSB:collector:Mamm:74965; Peromyscus maniculatus	Synonyms	North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB Mamm 74965; MSB:collector:Mamm:74965; Peromyscus maniculatus	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus	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; Rodentia; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus	Lineage
Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10040)	Parent	Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10040)	Parent
10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10042)	NCBI Taxonomy ID	10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10042)	NCBI Taxonomy ID
Yes	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
Peromyscus maniculatus - New Hampshire	Taxon A Description	Peromyscus maniculatus - New Hampshire	Taxon B Description

GENOTYPIC CHANGE

Asip	Generic Gene Name	Q03288 (http://www.uniprot.org/uniprot/Q03288)	UniProtKB Mus musculus
As; ASP; A<y>; ASIP; a	Synonyms	ACV72059 (https://www.ncbi.nlm.nih.gov/nuccore/ACV72059)	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)	String		
-	Sequence Similarities		
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)	GO - Molecular Function		
GO:0031781 : type 3 melanocortin receptor binding			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031781>)
GO:0031782 : type 4 melanocortin receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031782>)

GO - Biological Process

GO:0008343 : adult feeding behavior
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008343>)
GO:0006091 : generation of precursor metabolites and energy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006091>)
GO:0071514 : genetic imprinting (<https://www.ebi.ac.uk/QuickGO/term/GO:0071514>)
GO:0009755 : hormone-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
GO:0032438 : melanosome organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
GO:0032402 : melanosome transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032402>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0048023 : positive regulation of melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
GO:0040030 : regulation of molecular function, epigenetic
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040030>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005623 : cell (<https://www.ebi.ac.uk/QuickGO/term/GO:0005623>)

Presumptive Null
Yes (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title>)
Molecular Type
Gene Loss (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Gene Loss^#gephebase-summary-title>)
Aberration Type
Deletion (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title>)
Deletion Size
10-100 kb
Molecular Details of the Mutation
125kb deletion ; recessive mutation
Experimental Evidence
Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene^#gephebase-summary-title>)
Main Reference
Melanism in peromyscus is caused by independent mutations in agouti. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19649329>)
Authors
Kingsley EP; Manceau M; Wiley CD; Hoekstra HE
Abstract
Identifying the molecular basis of phenotypes that have evolved independently can provide insight into the ways genetic and developmental constraints influence the maintenance of phenotypic diversity. Melanic (darkly pigmented) phenotypes in mammals provide a potent system in which to study the genetic basis of naturally occurring mutant phenotypes because melanism occurs in many mammals, and the mammalian pigmentation pathway is well understood. Spontaneous alleles of a few key pigmentation loci are known to cause melanism in domestic or laboratory populations of mammals, but in natural populations, mutations at one gene, the melanocortin-1 receptor (Mc1r), have been implicated in the vast majority of cases, possibly due to its minimal pleiotropic effects. To investigate whether mutations in this or other genes cause melanism in the wild, we investigated the genetic basis of melanism in the rodent genus Peromyscus, in which melanic mice have been reported in several populations. We focused on two genes known to cause melanism in other taxa, Mc1r and its antagonist, the agouti signaling protein (Agouti). While variation in the Mc1r coding region does not correlate with melanism in any population, in a New Hampshire population, we find that a 125-kb deletion, which includes the upstream regulatory region and exons 1 and 2 of Agouti, results in a loss of Agouti expression and is perfectly associated with melanic color. In a second population from Alaska, we find that a premature stop codon in exon 3 of Agouti is associated with a similar melanic phenotype. These results show that melanism has evolved independently in these populations through mutations in the same gene, and suggest that melanism produced by mutations in genes other than Mc1r may be more common than previously thought.
Additional References

RELATED GEPHE

Related Genes
No matches found.
Related Haplotypes
2 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Agouti^/and+Taxon ID=~10042^/or+Gene Gephebase=~Agouti^/and+Taxon ID=~10042^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000201/10042/>

