GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00001976

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

Peromyscus maniculatus - Alaska - WT

Trait State in Taxon B

Peromyscus maniculatus - Alaska - melanic

Ancestral State

Data not curated

Taxonomic Status

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

Taxon A

Latin Name

Peromyscus maniculatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Peromyscus maniculatus^#gephebase-summary-title)

Common Name

North American deer mouse

Synonyms

North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB Mamm 74965; MSB:collector:Mamm:74965; Peromyscus maniculatis

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; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus

Parent

Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10040)

NCBI Taxonomy ID

10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10042)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Peromyscus maniculatus - Alaska

Taxon B

Latin Name

Peromyscus maniculatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Peromyscus maniculatus^#gephebase-summary-title)

Common Name

North American deer mouse

Synonyms

North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB Mamm 74965; MSB:collector:Mamm:74965; Peromyscus maniculatis

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; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus

Parent

Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10040)

NCBI Taxonomy ID

10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10042)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Peromyscus maniculatus - Alaska

GENOTYPIC CHANGE

Generic Gene Name

Asip

Synonyms

As; ASP; A<y>; ASIP; a

String

10090.ENSMUSP00000029123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000029123)

Sequence Similarities

-

GO - Molecular Function

GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)
GO:0031781 : type 3 melanocortin receptor binding

UniProtKB Mus musculus

Q03288 (http://www.uniprot.org/uniprot/Q03288)

GenebankID or UniProtKB

ACV72059 (https://www.ncbi.nlm.nih.gov/nuccore/ACV72059)

(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

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

Nonsense

Molecular Details of the Mutation

recessive mutation at nucleotide position 193 (in exon 3) resulting in a change from glutamine to a stop codon at amino acid position 65 (Q65term)

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gln	STP	65

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/