GEPHE SUMMARY

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

GepheID
GP00000055

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; dorso-ventral) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat; dorso-ventral)^#gephebase-summary-title)

Trait State in Taxon A
Peromyscus polionotus

Trait State in Taxon B
Peromyscus polionotus - New Hampshire; 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 polionotus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Peromyscus polionotus^#gephebase-summary-title)

Common Name
oldfield mouse

Synonyms
oldfield mouse

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
42413 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 42413)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
oldfield mouse

Synonyms
oldfield mouse

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
42413 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 42413)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Peromyscus polionotus - New Hampshire; melanic

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 (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)

UniProtKB Mus musculus
Q03288 (http://www.uniprot.org/uniprot/Q03288)

GenebankID or UniProtKB
ABV02195 (https://www.ncbi.nlm.nih.gov/nuccore/ABV02195)

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

100-1000 kb

Molecular Details of the Mutation

125kb deletion including 5′ CDS of Agouti

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

1 (MC1R) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^42413^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Agouti^/and+Taxon ID=^42413^/or+Gene Gephebase=^Agouti^/and+Taxon ID=^42413^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS