

GEPHE SUMMARY

Gephebase Gene
MC1R

Entry Status
Published

GepheID
GP00000582

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
Mus musculus

Trait State in Taxon B
Mus musculus

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Mus musculus

Common Name
house mouse

Synonyms
house mouse; mouse; *Mus musculus* Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

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; Muridae; Murinae; Mus; Mus

Parent
Mus () - (Rank: subgenus)

NCBI Taxonomy ID
10090

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Mus musculus

Common Name
house mouse

Synonyms
house mouse; mouse; *Mus musculus* Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

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; Muridae; Murinae; Mus; Mus

Parent
Mus () - (Rank: subgenus)

NCBI Taxonomy ID
10090

is Taxon B an Intraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
MC1R

Synonyms
CMM5; MSH-R; SHEP2; MSHR

String
9606.ENSP00000451605

Sequence Similarities
Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function
GO:0008528 : G protein-coupled peptide receptor activity
GO:0004977 : melanocortin receptor activity
GO:0004980 : melanocyte-stimulating hormone receptor activity
GO:0031625 : ubiquitin protein ligase binding

GO - Biological Process
GO:0007275 : multicellular organism development
GO:0045944 : positive regulation of transcription by RNA polymerase II
GO:0042438 : melanin biosynthetic process
GO:0043473 : pigmentation
GO:0007186 : G protein-coupled receptor signaling pathway

UniProtKB Homo sapiens
Q01726

GenebankID or UniProtKB
CH466525

GO:0051897 : positive regulation of protein kinase B signaling
GO:0019233 : sensory perception of pain
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
GO:0035556 : intracellular signal transduction
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide second messenger
GO:0032720 : negative regulation of tumor necrosis factor production
GO:0010739 : positive regulation of protein kinase A signaling
GO:0090037 : positive regulation of protein kinase C signaling
GO:0009650 : UV protection
GO:0070914 : UV-damage excision repair

GO - Cellular Component

GO:0005886 : plasma membrane
GO:0005887 : integral component of plasma membrane

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

Deletion

Deletion Size

1-9 bp

Molecular Details of the Mutation

1bp deletion at 183; frameshift & premature stop

Experimental Evidence

Candidate Gene

Main Reference

Pigmentation phenotypes of variant extension locus alleles result from point mutations that alter MSH receptor function. (1993)

Authors

Robbins LS; Nadeau JH; Johnson KR; Kelly MA; Roselli-Reh fuss L; Baack E; Mountjoy KG; Cone RD

Abstract

Coat colors in the chestnut horse, the yellow Labrador retriever, the red fox, and one type of yellow mouse are due to recessive alleles at the extension locus. Similarly, dominant alleles at this locus are often responsible for dark coat colors in mammals, such as the melanic form of the leopard, *Panthera pardus*. We show here that the murine extension locus encodes the melanocyte-stimulating hormone (MSH) receptor. In mice, the recessive yellow allele (e) results from a frameshift that produces a prematurely terminated, nonfunctioning receptor. The sombre (Eso and Eso-3J) and tobacco darkening (EtoB) alleles, which both have dominant melanizing effects, results from point mutations that produce hyperactive MSH receptors. The Eso-3J receptor is constitutively activated, while the EtoB receptor remains hormone responsive and produces a greater activation of its effector, adenylyl cyclase, than does the wild-type allele.

Additional References

RELATED GEPHE

Related Genes

1 (SLC45A2=MATP)

Related Haplotypes

4

COMMENTS