GEPHE SUMMARY

Gephebase Gene
MC1R (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MC1R^#gephebase-summary-title)

GepheID
GP00000596

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
Sciurus carolinensis

Trait State in Taxon B
Sciurus carolinensis

Ancestral State
Taxon A

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

Taxon A #1

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

Common Name
gray squirrel

Synonyms
gray squirrel; eastern gray squirrel; Sciurus carolinensis Gmelin, 1788; Sciurus carolinensus

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; Sciuromorpha; Sciuridae; Sciurinae; Sciurini; Sciurus

Parent
Sciurus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10001)

NCBI Taxonomy ID
30640 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30640)

is Taxon A an Infraspecies?
No

Taxon B #1

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

Common Name
gray squirrel

Synonyms
gray squirrel; eastern gray squirrel; Sciurus carolinensis Gmelin, 1788; Sciurus carolinensus

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; Sciuromorpha; Sciuridae; Sciurinae; Sciurini; Sciurus

Parent
Sciurus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10001)

NCBI Taxonomy ID
30640 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30640)

is Taxon B an Infraspecies?
No

Taxon A #2

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

Common Name
fox squirrel

Synonyms
fox squirrel; eastern fox squirrel

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; Sciuromorpha; Sciuridae; Sciurinae; Sciurini; Sciurus

Parent
Sciurus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10001)

Taxon B #2

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

Common Name
fox squirrel

Synonyms
fox squirrel; eastern fox squirrel

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; Sciuromorpha; Sciuridae; Sciurinae; Sciurini; Sciurus

Parent
Sciurus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10001)

NCBI Taxonomy ID

34861

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 34861)

is Taxon A an Infraspecies?

No

NCBI Taxonomy ID

34861

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 34861)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

MC1R

Synonyms

CMM5; MSH-R; SHEP2; MSHR

String

9606.ENSP00000451605
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=
9606.ENSP00000451605)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function

GO:0008528 : G protein-coupled peptide receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0008528)
GO:0004977 : melanocortin receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004977)
GO:0004980 : melanocyte-stimulating hormone receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004980)
GO:0031625 : ubiquitin protein ligase binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031625)

GO - Biological Process

GO:0007275 : multicellular organism development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0051897 : positive regulation of protein kinase B signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0051897)
GO:0019233 : sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0019233)
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007189)
GO:0035556 : intracellular signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0035556)
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide
second messenger (https://www.ebi.ac.uk/QuickGO/term/GO:0007187)
GO:0032720 : negative regulation of tumor necrosis factor production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032720)
GO:0010739 : positive regulation of protein kinase A signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0010739)
GO:0090037 : positive regulation of protein kinase C signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0090037)
GO:0009650 : UV protection (https://www.ebi.ac.uk/QuickGO/term/GO:0009650)
GO:0070914 : UV-damage excision repair
(https://www.ebi.ac.uk/QuickGO/term/GO:0070914)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)

UniProtKB Homo sapiens

Q01726 (http://www.uniprot.org/uniprot/Q01726)

GenebankID or UniProtKB

ACC77631 (https://www.ncbi.nlm.nih.gov/nuccore/ACC77631)

Presumptive Null

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

Molecular Type

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

Aberration Type

Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)

Deletion Size

10-99 bp

Molecular Details of the Mutation

Deletion of 8 a.a.

Experimental Evidence

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

Main Reference

The genetic basis of melanism in the gray squirrel (Sciurus carolinensis). (2009 Nov-Dec) (https://pubmed.ncbi.nlm.nih.gov/19643815)

Authors

McRobie H; Thomas A; Kelly J

Abstract

The black squirrel is a melanic variant of the gray squirrel (Sciurus carolinensis). We found 3 coat color variants in the gray squirrel: the wild-type gray, a jet-black, and a brown-black phenotype. These 3 morphs are due to varying distributions of eumelanin and phaeomelanin pigment in hairs. The melanocortin 1 receptor (MC1R) plays a central role in regulating eumelanin and phaeomelanin production. We sequenced the MC1R gene for all 3 coat color phenotypes and found a 24 base-pair deletion. The gray phenotype was homozygous for the wild-type allele E(+), the jet-black phenotype was homozygous for the MC1R-Delta24 allele E(B), and the brown-black phenotype was heterozygous for the E(+) and E(B) alleles. We conclude that melanism in gray squirrels is associated with the MC1R-Delta24 E(B) allele at amino acid positions 87-94 and that this allele is incompletely dominant to the wild-type allele. We predict that the MC1R-Delta24 E(B) allele encodes a constitutively active or hyperactive receptor.

Additional References

Multiple origins of melanism in two species of North American tree squirrel (Sciurus). (2019) (https://pubmed.ncbi.nlm.nih.gov/31296164)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Introgression @Parallelism likely introgression between fox and gray squirrels