GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00002299

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

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

Trait State in Taxon A

WT

Trait State in Taxon B

dominant black

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

-

Synonyms

-

Rank

subspecies

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Meleagridinae; Meleagris; Meleagris gallopavo

Parent

Lacerta () - (Rank:) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9103)

NCBI Taxonomy ID

165260 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 165260)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Domestic turkey

Taxon B

Latin Name

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

Common Name

-

Synonyms

-

Rank

subspecies

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Meleagridinae; Meleagris; Meleagris gallopavo

Parent

Lacerta () - (Rank:) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9103)

NCBI Taxonomy ID

165260 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 165260)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Domestic turkey

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)

UniProtKB Homo sapiens

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

GenebankID or UniProtKB

()

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)

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.364A>T p.I122F

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ile	Phe	122

Main Reference

Variability of the melanocortin 1 receptor (MC1R) gene explains the segregation of the bronze locus in turkey (Meleagris gallopavo). (2010) (https://pubmed.ncbi.nlm.nih.gov/20634512)

Authors

Vidal O; ViÃ±as J; Pla C

Abstract

By sequencing the full coding region of the turkey melanocortin 1 receptor (MC1R) gene, we have found 4 mutations (c.96G > A, c.364A > T, c.450C > T, and c.887C > T) that are organized in 5 different haplotypes (MC1R*1 to MC1R*5). These haplotypes correlate perfectly with the 3 alleles of the bronze locus (i.e., B, b(+), and b(1)). We suggest that the dominant black phenotype, associated with the B allele, results from the constitutive activation of the receptor, an effect that might be mediated by the missense mutation c.364A > T (p.Ile122Phe). Moreover, we propose that the recessive black-winged bronze phenotype (linked to b(1)) might be produced by 2 deleterious mutations of MC1R (c.96G > A and c.887C > T). This is an unexpected finding because in mammals, MC1R deleterious polymorphisms are usually related with either red or lighter fur colors.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^MC1R^/and+Taxon ID=^165260^/or+Gene Gephebase=^MC1R^/and+Taxon ID=^165260^#gephebase-summary-

title)

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA001494/9103/ @Parallelism @Dominance Unusual MC1R phenotype due to possible constitutive activation / gain-of-function