GEPHE SUMMARY

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

GepheID
GP00002300

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 (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
recessive bronze

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)

Mutation #1

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.887C>T p.A296V

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	Ala	Val	296

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

Mutation #2

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

c.96G>A p.Trp32* stop-gain

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	Trp	STP	32

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)

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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