

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00002300	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)	Trait Category		
Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+feathers^#gephebase-summary-title)	Trait		
WT	Trait State in Taxon A		
recessive bronze	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Meleagris gallopavo mexicana (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Meleagris+gallopavo+mexicana^#gephebase-summary-title)	Latin Name	Meleagris gallopavo mexicana (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Meleagris+gallopavo+mexicana^#gephebase-summary-title)	Latin Name
-	Common Name	-	Common Name
-	Synonyms	-	Synonyms
subspecies	Rank	subspecies	Rank
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	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	Lineage
Lacerta () - (Rank:) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9103)	Parent	Lacerta () - (Rank:) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9103)	Parent
165260 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=165260)	NCBI Taxonomy ID	165260 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=165260)	NCBI Taxonomy ID
Yes	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
Domestic turkey	Taxon A Description	Domestic turkey	Taxon B Description

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	0	GenebankID or UniProtKB
9606.ENSP00000451605 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000451605)	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)	GO - Molecular Function		
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)

Mutation #1

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

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

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

Nonsynonymous

c.887C>T p.A296V

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

	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

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes~#gephebase-summary-title)

Presumptive Null

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>)

Authors

Vidal O; ViÑas 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