GEPHE SUMMARY

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

GepheID
GP00000601

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
Columba livia

Trait State in Taxon B
Columba livia - increased production of pheomelanin (red and yellow pigments)

Ancestral State
Data not curated

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

Taxon A

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

Common Name
rock pigeon

Synonyms
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789

Rank
species

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; Columbiformes; Columbidae; Columba

Parent
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931)

NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
rock pigeon

Synonyms
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789

Rank
species

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; Columbiformes; Columbidae; Columba

Parent
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931)

NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932)

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

UniProtKB Homo sapiens
Q01726 (http://www.uniprot.org/uniprot/Q01726)

GenebankID or UniProtKB
KC819647 (https://www.ncbi.nlm.nih.gov/nuccore/KC819647)

(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

Val85Met (G253A)

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	-	-	-

Main Reference

A Val85Met mutation in melanocortin-1 receptor is associated with reductions in eumelanic pigmentation and cell surface expression in domestic rock pigeons (Columba livia). (2013)
(https://pubmed.ncbi.nlm.nih.gov/23977400)

Authors

Guernsey MW; Ritscher L; Miller MA; Smith DA; SchÃ¶neberg T; Shapiro MD

Abstract

Variation in the melanocortin-1 receptor (Mc1r) is associated with pigmentation diversity in wild and domesticated populations of vertebrates, including several species of birds. Among domestic bird species, pigmentation variation in the rock pigeon (Columbalivia) is particularly diverse. To determine the potential contribution of Mc1r variants to pigment diversity in pigeons, we sequenced Mc1r in a wide range of pigeon breeds and identified several single nucleotide polymorphisms, including a variant that codes for an amino acid substitution (Val85Met). In contrast to the association between Val85Met and eumelanism in other avian species, this change was associated with pheomelanism in pigeons. In vitro cAMP accumulation and protein expression assays revealed that Val85Met leads to decreased receptor function and reduced cell surface expression of the mutant protein. The reduced in vitro function is consistent with the observed association with reduced eumelanic pigmentation. Comparative genetic and cellular studies provide important insights about the range of mechanisms underlying diversity among vertebrates, including different phenotypic associations with similar mutations in different species.

Additional References

RELATED GEPHE

Related Genes

6 (Mlana, ndp (norrin), slc2a11b, SLC45A2=MATP, SOX10, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon

ID=^8932^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS