GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000602

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

Coturnix japonica

Trait State in Taxon B

Coturnix japonica

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

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

Common Name

Japanese quail

Synonyms

Coturnix coturnix Japonicus; Coturnix coturnix japanica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849

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; Galloanserae; Galliformes; Phasianidae; Perdicinae; Coturnix

Parent

Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)

NCBI Taxonomy ID

93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

Japanese quail

Synonyms

Coturnix coturnix Japonicus; Coturnix coturnix japanica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849

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; Galloanserae; Galliformes; Phasianidae; Perdicinae; Coturnix

Parent

Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)

NCBI Taxonomy ID

93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)

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

ABD49210 (https://www.ncbi.nlm.nih.gov/nuccore/ABD49210)

(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

E92K

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Association of a Glu92Lys substitution in MC1R with extended brown in Japanese quail (Coturnix japonica). (2006) (https://pubmed.ncbi.nlm.nih.gov/16734695)

Authors

Nadeau NJ; Minvielle F; Mundy NI

Abstract

We investigated melanocortin 1 receptor (MC1R) as a candidate locus for the Extended brown phenotype in quail, in which there is a general darkening throughout the plumage. An initial screen of variation in MC1R in Extended brown and in wild-type quails revealed two polymorphic non-synonymous sites. One of these sites, a G-to-A substitution leading to a Glu92Lys mutation, was perfectly associated with plumage phenotype; all Extended brown birds were homozygous for Lys92. Co-segregation of the Glu92Lys mutation with the Extended brown phenotype was confirmed in 24 progeny of an E/e(+) x E/e(+) cross. Glu92Lys is likely to be the causative mutation for the increased melanism in Extended brown, given that the same mutation is associated with melanic plumage in many breeds of domestic chicken, as well as in a wild passerine bird (the bananaquit, Coereba flaveola) and laboratory mice. Interestingly, the increase in melanization with the Glu92Lys mutation is less marked in quails than in most other birds and mammals. Phylogenetic results indicate that the Glu92Lys mutation has independently occurred in quail and chicken lineages.

Additional References

RELATED GEPHE

Related Genes

6 (Agouti, Endothelin receptor B, Melanophilin (MLPH), Microphtalmia-associated transcription factor, SLC45A2=MATP, tyrosinase-related protein 1 (TYRP1))
(https://www.gephebase.org/search-criteria?/or+Taxon ID=^93934^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

No matches found.

EXTERNAL LINKS

COMMENTS