GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000270

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

Trait State in Taxon A

Coturnix japonica

Trait State in Taxon B

Coturnix japonica - panda (white-spotting ; recessive)

Ancestral State

Taxon A

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?

Yes

Taxon B Description

Coturnix japonica - panda (white-spotting)

GENOTYPIC CHANGE

Generic Gene Name

Ednrb

Synonyms

ETb; ET-B; ET-BR; ETR-b; Sox10m1

String

10090.ENSMUSP00000022718 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000022718)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family. Endothelin receptor subfamily. EDNRB sub-subfamily.

GO - Molecular Function

GO:0017046 : peptide hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017046)

UniProtKB Mus musculus

P48302 (http://www.uniprot.org/uniprot/P48302)

GenebankID or UniProtKB

XP_015707082 (https://www.ncbi.nlm.nih.gov/nuccore/XP_015707082)

GO:0004962 : endothelin receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004962)
GO:0031702 : type 1 angiotensin receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031702)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0007422 : peripheral nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007422)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0001755 : neural crest cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0001755)
GO:0019934 : cGMP-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0019934)
GO:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0032269 : negative regulation of cellular protein metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0032269)
GO:0042311 : vasodilation (https://www.ebi.ac.uk/QuickGO/term/GO:0042311)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0019722 : calcium-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0019722)
GO:0007204 : positive regulation of cytosolic calcium ion concentration
(https://www.ebi.ac.uk/QuickGO/term/GO:0007204)
GO:0035815 : positive regulation of renal sodium excretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0035815)
GO:0051930 : regulation of sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0051930)
GO:0032496 : response to lipopolysaccharide
(https://www.ebi.ac.uk/QuickGO/term/GO:0032496)
GO:0014070 : response to organic cyclic compound
(https://www.ebi.ac.uk/QuickGO/term/GO:0014070)
GO:0048265 : response to pain (https://www.ebi.ac.uk/QuickGO/term/GO:0048265)
GO:0008217 : regulation of blood pressure
(https://www.ebi.ac.uk/QuickGO/term/GO:0008217)
GO:0048066 : developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048066)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)
GO:0014826 : vein smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:0014826)
GO:0007568 : aging (https://www.ebi.ac.uk/QuickGO/term/GO:0007568)
GO:0071222 : cellular response to lipopolysaccharide
(https://www.ebi.ac.uk/QuickGO/term/GO:0071222)
GO:0086100 : endothelin receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0086100)
GO:0048484 : enteric nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048484)
GO:0035645 : enteric smooth muscle cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0035645)
GO:0042045 : epithelial fluid transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0042045)
GO:0048246 : macrophage chemotaxis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048246)
GO:0014043 : negative regulation of neuron maturation
(https://www.ebi.ac.uk/QuickGO/term/GO:0014043)
GO:0007200 : phospholipase C-activating G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007200)
GO:0060406 : positive regulation of penile erection
(https://www.ebi.ac.uk/QuickGO/term/GO:0060406)
GO:0001934 : positive regulation of protein phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001934)
GO:0035810 : positive regulation of urine volume
(https://www.ebi.ac.uk/QuickGO/term/GO:0035810)
GO:0007497 : posterior midgut development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007497)
GO:0050678 : regulation of epithelial cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0050678)
GO:0031620 : regulation of fever generation
(https://www.ebi.ac.uk/QuickGO/term/GO:0031620)
GO:0006885 : regulation of pH (https://www.ebi.ac.uk/QuickGO/term/GO:0006885)
GO:1990839 : response to endothelin (https://www.ebi.ac.uk/QuickGO/term/GO:1990839)
GO:0019233 : sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0019233)

GO - Cellular Component

GO:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0045121 : membrane raft (https://www.ebi.ac.uk/QuickGO/term/GO:0045121)
GO:0031965 : nuclear membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0031965)

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.995G>A p.R332H

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	Arg	His	332

Main Reference

Endothelin receptor B2 (EDNRB2) is associated with the panda plumage colour mutation in Japanese quail. (2007) (https://pubmed.ncbi.nlm.nih.gov/17313575)

Authors

Miwa M; Inoue-Murayama M; Aoki H; Kunisada T; Hiragaki T; Mizutani M; Ito S

Abstract

The panda mutant in Japanese quail (Coturnix japonica) displays spots of wild-type plumage on a white background and is controlled by an autosomal recessive allele (s). The dotted white is controlled by a third allele (s(dw)) of the s locus with s(dw)/s(dw) quail having less pigmentation than s/s quail. We mapped the s locus to the Japanese quail chromosome 4 (CJA04) in a previous study. The orthologous region of the chicken (Gallus gallus) genome includes endothelin receptor B2 (EDNRB2), an avian-specific paralog of endothelin receptor B (EDNRB). EDNRB mutations in mammals retard the migration of neural crest cells (NCCs), which results in a spotted coat colour and an enteric nervous defect. In the present study, we investigated the association between the s locus and EDNRB2 in Japanese quail. Sequence comparison among transcripts from livers of wild-type, panda and dotted white quail revealed a nucleotide substitution (c.995G>A) leading to a p.R332H amino acid change that was specific to panda, whereas no amino acid substitution was found in dotted white birds. The amino acid position 332 is located in the sixth transmembrane domain and is highly conserved in both avian and mammalian endothelin receptors. The A allele at nucleotide position 995 was specific to panda (s/s) birds among 10 strains, and was mapped to the same chromosomal region as the s locus. Quantitative RT-PCR revealed that EDNRB2 transcripts were reduced in both panda and dotted white mutants compared with wild-type. However, there was no difference between the early embryos of wild-type and panda with respect to the migration of NCCs. The genetic association of EDNRB2 with plumage colour in birds was found for the first time in this study.

Additional References

RELATED GEPHE

Related Genes

6 (Agouti, MC1R, 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)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA000375/93934/