GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00000660

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 - silver

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 - silver

GENOTYPIC CHANGE

Generic Gene Name

Mitf

Synonyms

Wh; bw; mi; vit; BCC2; Bhlhe32; Gsfbcc2; Vitiligo; Bw; Mi; Vit

String

10090.ENSMUSP00000044938 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938)

Sequence Similarities

Belongs to the MiT/TFE family.

GO - Molecular Function

GO:0046983 : protein dimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046983)

UniProtKB Mus musculus

Q08874 (http://www.uniprot.org/uniprot/Q08874)

GenebankID or UniProtKB

ACV31196 (https://www.ncbi.nlm.nih.gov/nuccore/ACV31196)

GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0003682 : chromatin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003682)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000978)
GO:0070888 : E-box binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070888)
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-specific binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003705)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0006357 : regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0006357)
GO:0006355 : regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006355)
GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154)
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:0045893 : positive regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0010628 : positive regulation of gene expression (https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0006351 : transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006351)
GO:0045165 : cell fate commitment (https://www.ebi.ac.uk/QuickGO/term/GO:0045165)
GO:0010468 : regulation of gene expression (https://www.ebi.ac.uk/QuickGO/term/GO:0010468)
GO:0030318 : melanocyte differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0043010 : camera-type eye development (https://www.ebi.ac.uk/QuickGO/term/GO:0043010)
GO:0030316 : osteoclast differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030316)
GO:0042127 : regulation of cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0042127)
GO:0046849 : bone remodeling (https://www.ebi.ac.uk/QuickGO/term/GO:0046849)
GO:0044336 : canonical Wnt signaling pathway involved in negative regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0044336)
GO:0030336 : negative regulation of cell migration (https://www.ebi.ac.uk/QuickGO/term/GO:0030336)
GO:2000144 : positive regulation of DNA-templated transcription, initiation (https://www.ebi.ac.uk/QuickGO/term/GO:2000144)
GO:0065003 : protein-containing complex assembly (https://www.ebi.ac.uk/QuickGO/term/GO:0065003)
GO:0045670 : regulation of osteoclast differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0045670)
GO:2001141 : regulation of RNA biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:2001141)
GO:0016055 : Wnt signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0016055)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0032991 : protein-containing complex (https://www.ebi.ac.uk/QuickGO/term/GO:0032991)

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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

2bp deletion

Experimental Evidence

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

Main Reference

The "silver" Japanese quail and the MITF gene: causal mutation, associated traits and homology with the "blue" chicken plumage. (2010) (https://pubmed.ncbi.nlm.nih.gov/20184729)

Authors

Minvielle F; Bed'hom B; Coville JL; Ito S; Inoue-Murayama M; Gourichon D

Abstract

The MITF (microphthalmia-associated transcription factor) gene has been investigated in mice and various vertebrates but its variations and associated effects have not yet been explored much in birds. The present study describes the causal mutation B at the MITF gene responsible for the "silver" plumage colour in the Japanese quail (Coturnix japonica), and its associated effects on growth and body composition, and tests its allelism with the "blue" plumage colour mutation Bl in Gallus gallus.

The semi dominant B mutation results from a premature stop codon caused by a 2 bp deletion in exon 11 of MITF. Homozygous "white" (B/B) quail which have a white plumage also show a slightly lower growth, lower body temperature, smaller heart, and lighter pectoralis muscles but more abdominal adipose tissue than the recessive homozygous "wild-type" (+/+) and heterozygous "silver" (B/+) quail. Similar observations on cardiac and body growth were made on mice (Mus musculus) homozygous for mutations at MITF. The production of chicken-quail hybrids with a white plumage obtained by crossing Bl/+ chicken heterozygous for the blue mutation with B/B white quail indicated that the mutations were allelic.

The "silver" Japanese quail is an interesting model for the comparative study of the effects of MITF in birds and mammals. Further investigation using a chicken family segregating for the "blue" plumage and molecular data will be needed to confirm if the "blue" plumage in chicken results from a mutation in MITF.

Additional References

Spontaneous transdifferentiation of quail pigmented epithelial cell is accompanied by a mutation in the Mitf gene. (1998) (https://pubmed.ncbi.nlm.nih.gov/9576828)

RELATED GEPHE

Related Genes

6 (Agouti, Endothelin receptor B, MC1R, Melanophilin (MLPH), SLC45A2=MATP, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+TaxonID=^93934^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism https://omia.org/OMIA000915/93934/ @HeterozygoteAdvantage with homozygotes showing slow growth; small heart; osteopetrosis