

GEPHE SUMMARY

|                                                                                                                                                                                                                                                                                                                                                     |  |              |  |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|--------------|--|
| Gephebase Gene                                                                                                                                                                                                                                                                                                                                      |  | GepheID      |  |
| Microphthalmia-associated transcription factor ( <a +microphthalmia-associated+transcription+factor`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene Gephebase=`Microphthalmia-associated transcription factor`#gephebase-summary-title</a> ) |  | GP00000660   |  |
| Entry Status                                                                                                                                                                                                                                                                                                                                        |  | Main curator |  |
| Published                                                                                                                                                                                                                                                                                                                                           |  | Martin       |  |

PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                                                                                                               |  |                                                                                                                                                                                                                                                                                                                                                                                                               |  |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|
| Trait Category                                                                                                                                                                                                                                                                                                                                                                                                |  |                                                                                                                                                                                                                                                                                                                                                                                                               |  |
| Morphology ( <a +morphology`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait Category=`Morphology`#gephebase-summary-title</a> )                                                                                                                                                                       |  | Trait                                                                                                                                                                                                                                                                                                                                                                                                         |  |
| Coloration (feathers) ( <a +coloration+(feathers)`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait=`Coloration (feathers)`#gephebase-summary-title</a> )                                                                                                                                                        |  | Trait State in Taxon A                                                                                                                                                                                                                                                                                                                                                                                        |  |
| Coturnix japonica                                                                                                                                                                                                                                                                                                                                                                                             |  | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                                                                                        |  |
| Coturnix japonica - silver                                                                                                                                                                                                                                                                                                                                                                                    |  | Ancestral State                                                                                                                                                                                                                                                                                                                                                                                               |  |
| Taxon A                                                                                                                                                                                                                                                                                                                                                                                                       |  | Taxonomic Status                                                                                                                                                                                                                                                                                                                                                                                              |  |
| Domesticated ( <a +domesticated`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic Status=`Domesticated`#gephebase-summary-title</a> )                                                                                                                                                             |  |                                                                                                                                                                                                                                                                                                                                                                                                               |  |
| Taxon A                                                                                                                                                                                                                                                                                                                                                                                                       |  | Taxon B                                                                                                                                                                                                                                                                                                                                                                                                       |  |
| Coturnix japonica                                                                                                                                                                                                                                                                                                                                                                                             |  | Coturnix japonica                                                                                                                                                                                                                                                                                                                                                                                             |  |
| ( <a +coturnix+japonica`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=`Coturnix japonica`#gephebase-summary-title</a> )                                                                                                                                                            |  | ( <a +coturnix+japonica`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=`Coturnix japonica`#gephebase-summary-title</a> )                                                                                                                                                            |  |
| Common Name                                                                                                                                                                                                                                                                                                                                                                                                   |  | Common Name                                                                                                                                                                                                                                                                                                                                                                                                   |  |
| Japanese quail                                                                                                                                                                                                                                                                                                                                                                                                |  | Japanese quail                                                                                                                                                                                                                                                                                                                                                                                                |  |
| Synonyms                                                                                                                                                                                                                                                                                                                                                                                                      |  | Synonyms                                                                                                                                                                                                                                                                                                                                                                                                      |  |
| Coturnix coturnix Japonicus; Coturnix coturnix japonica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849                                                                                                                                                                                                                                  |  | Coturnix coturnix Japonicus; Coturnix coturnix japonica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849                                                                                                                                                                                                                                  |  |
| Rank                                                                                                                                                                                                                                                                                                                                                                                                          |  | Rank                                                                                                                                                                                                                                                                                                                                                                                                          |  |
| species                                                                                                                                                                                                                                                                                                                                                                                                       |  | species                                                                                                                                                                                                                                                                                                                                                                                                       |  |
| Lineage                                                                                                                                                                                                                                                                                                                                                                                                       |  | 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; Percidinae; Coturnix |  | 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; Percidinae; Coturnix |  |
| Parent                                                                                                                                                                                                                                                                                                                                                                                                        |  | Parent                                                                                                                                                                                                                                                                                                                                                                                                        |  |
| Coturnix () - (Rank: genus)                                                                                                                                                                                                                                                                                                                                                                                   |  | Coturnix () - (Rank: genus)                                                                                                                                                                                                                                                                                                                                                                                   |  |
| ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9090">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9090</a> )                                                                                                                                                                                                                                                           |  | ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9090">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9090</a> )                                                                                                                                                                                                                                                           |  |
| NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                                                                                                              |  | NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                                                                                                              |  |
| 93934                                                                                                                                                                                                                                                                                                                                                                                                         |  | 93934                                                                                                                                                                                                                                                                                                                                                                                                         |  |
| ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=93934">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=93934</a> )                                                                                                                                                                                                                                                         |  | ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=93934">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=93934</a> )                                                                                                                                                                                                                                                         |  |
| is Taxon A an Intraspecies?                                                                                                                                                                                                                                                                                                                                                                                   |  | is Taxon B an Intraspecies?                                                                                                                                                                                                                                                                                                                                                                                   |  |
| No                                                                                                                                                                                                                                                                                                                                                                                                            |  | Yes                                                                                                                                                                                                                                                                                                                                                                                                           |  |
|                                                                                                                                                                                                                                                                                                                                                                                                               |  | Taxon B Description                                                                                                                                                                                                                                                                                                                                                                                           |  |
|                                                                                                                                                                                                                                                                                                                                                                                                               |  | Coturnix japonica - silver                                                                                                                                                                                                                                                                                                                                                                                    |  |

GENOTYPIC CHANGE

|                                                                                                                                                                                                                 |  |                                                                                                                        |  |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|------------------------------------------------------------------------------------------------------------------------|--|
| Generic Gene Name                                                                                                                                                                                               |  | UniProtKB Mus musculus                                                                                                 |  |
| Mitf                                                                                                                                                                                                            |  | Q08874 ( <a href="http://www.uniprot.org/uniprot/Q08874">http://www.uniprot.org/uniprot/Q08874</a> )                   |  |
| Synonyms                                                                                                                                                                                                        |  | GenebankID or UniProtKB                                                                                                |  |
| Wh; bw; mi; vit; BCC2; Bhlhe32; Gsfbcc2; Vitiligo; Bw; Mi; Vit                                                                                                                                                  |  | ACV31196 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/ACV31196">https://www.ncbi.nlm.nih.gov/nuccore/ACV31196</a> ) |  |
| String                                                                                                                                                                                                          |  |                                                                                                                        |  |
| 10090.ENSMUSP00000044938                                                                                                                                                                                        |  |                                                                                                                        |  |
| ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938</a> ) |  |                                                                                                                        |  |
| Sequence Similarities                                                                                                                                                                                           |  |                                                                                                                        |  |
| Belongs to the MiT/TFE family.                                                                                                                                                                                  |  |                                                                                                                        |  |
| GO - Molecular Function                                                                                                                                                                                         |  |                                                                                                                        |  |
| GO:0046983 : protein dimerization activity                                                                                                                                                                      |  |                                                                                                                        |  |
| ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046983">https://www.ebi.ac.uk/QuickGO/term/GO:0046983</a> )                                                                                                   |  |                                                                                                                        |  |

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>)

|                                                                                                                                                                                                                                                              |                                   |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------|
| Yes ( <a href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title</a> )                                            | Presumptive Null                  |
| Coding ( <a href="https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title</a> )                                       | Molecular Type                    |
| Deletion ( <a href="https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title</a> )                               | Aberration Type                   |
| 1-9 bp                                                                                                                                                                                                                                                       | Deletion Size                     |
| 2bp deletion                                                                                                                                                                                                                                                 | Molecular Details of the Mutation |
| Candidate Gene ( <a href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene^#gephebase-summary-title</a> ) | Experimental Evidence             |
| The “silver” Japanese quail and the MITF gene: causal mutation, associated traits and homology with the “blue” chicken plumage. (2010) ( <a href="https://pubmed.ncbi.nlm.nih.gov/20184729">https://pubmed.ncbi.nlm.nih.gov/20184729</a> )                   | Main Reference                    |

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