GEPHE SUMMARY

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

GepheID
GP00001973

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 - fawn2 and beige phenotypes

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?
No

GENOTYPIC CHANGE

Generic Gene Name
Asip

Synonyms
As; ASP; A<y>; ASIP; a

String
10090.ENSMUSP00000029123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)

Sequence Similarities
-

GO - Molecular Function
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)
GO:0031781 : type 3 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)
GO:0031782 : type 4 melanocortin receptor binding

UniProtKB Mus musculus
Q03288 (http://www.uniprot.org/uniprot/Q03288)

GenebankID or UniProtKB
ACA24932 (https://www.ncbi.nlm.nih.gov/nuccore/ACA24932)

(https://www.ebi.ac.uk/QuickGO/term/GO:0031782)

GO - Biological Process

GO:0008343 : adult feeding behavior
(https://www.ebi.ac.uk/QuickGO/term/GO:0008343)
GO:0006091 : generation of precursor metabolites and energy
(https://www.ebi.ac.uk/QuickGO/term/GO:0006091)
GO:0071514 : genetic imprinting (https://www.ebi.ac.uk/QuickGO/term/GO:0071514)
GO:0009755 : hormone-mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0009755)
GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0032438 : melanosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032438)
GO:0032402 : melanosome transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0032402)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0048023 : positive regulation of melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0048023)
GO:0040030 : regulation of molecular function, epigenetic
(https://www.ebi.ac.uk/QuickGO/term/GO:0040030)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005623 : cell (https://www.ebi.ac.uk/QuickGO/term/GO:0005623)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Insertion Size

10-100 kb

Molecular Details of the Mutation

71-kb tandem duplication that comprises one unchanged copy of ASIP and one copy present in the ITCH-ASIP fusion gene which leads to a transcript coding for a normal ASIP protein

Experimental Evidence

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

Main Reference

Two new structural mutations in the 5′ region of the ASIP gene cause diluted feather color phenotypes in Japanese quail. (2019) (https://pubmed.ncbi.nlm.nih.gov/30987584)

Authors

Robic A; Morisson M; Leroux S; Gourichon D; Vignal A; Thebault N; Fillon V; Minvielle F; Bed'Hom B; Zerjal T; Pitel F

Abstract

In quail, two feather colour phenotypes i.e. fawn-2/beige and yellow are associated with the ASIP locus. The aim of our study was to characterize the structural modifications within this locus that explain the yellow mutation (large deletion) and the fawn-2/beige mutation (assumed to be caused by a different structural modification).

For the yellow phenotype, we identified a complex mutation that involves a 141,162-bp long deletion. For the fawn-2/beige phenotype, we identified a 71-kb tandem duplication that comprises one unchanged copy of ASIP and one copy present in the ITCH-ASIP fusion gene, which leads to a transcript coding for a normal ASIP protein. Although this agrees with previous reports that reported an increased level of ASIP transcripts in the skin of mutant animals, we show that in the skin from fawn-2/beige embryos, this level is higher than expected with a simple duplication of the ASIP gene. Thus, we hypothesize that the 5′ region of the ITCH-ASIP fusion gene leads to a higher transcription level than the 5′ region of the ASIP gene.

We were able to conclude that the fawn-2 and beige phenotypes are caused by the same allele at the ASIP locus. Both of the associated mutations fawn-2/beige and yellow lead to the formation of a fusion gene, which encodes a transcript for the ASIP protein. In both cases, transcription of ASIP depends on the promoter of a different gene, which includes alternative up-regulating sequences. However, we cannot exclude the possibility that the loss of the 5′ region of the ASIP gene itself has additional impacts, especially for the fawn-2/beige mutation. In addition, in several other species including mammals, the existence of other dominant gain-of-function structural modifications that are localized upstream of the ASIP coding sequences has been reported, which supports our hypothesis that repressors in the 5′ region of ASIP are absent in the fawn-2/beige mutant.

Additional References

RELATED GEPHE

Related Genes

6 (Endothelin receptor B, 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

2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Agouti^/and+Taxon ID=^93934^/or+Gene Gephebase=^Agouti^/and+Taxon ID=^93934^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA000201/9615/