GEPHE SUMMARY

Gephebase Gene
Agouti

GephelD
GP00001973

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (feathers)

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

Taxon A

Latin Name
Coturnix japonica

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)

NCBI Taxonomy ID
93934

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Coturnix japonica

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)

NCBI Taxonomy ID
93934

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Asip

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

String
10090.ENSMUSP00000029123

Sequence Similarities
-

GO - Molecular Function
GO:0031779 : melanocortin receptor binding
GO:0031781 : type 3 melanocortin receptor binding
GO:0031782 : type 4 melanocortin receptor binding

GO - Biological Process
GO:0008343 : adult feeding behavior
GO:0006091 : generation of precursor metabolites and energy
GO:0071514 : genetic imprinting
GO:0009755 : hormone-mediated signaling pathway

UniProtKB Mus musculus
Q03288

GenebankID or UniProtKB
ACA24932

GO:0042438 : melanin biosynthetic process
GO:0032438 : melanosome organization
GO:0032402 : melanosome transport
GO:0043473 : pigmentation
GO:0048023 : positive regulation of melanin biosynthetic process
GO:0040030 : regulation of molecular function, epigenetic

GO - Cellular Component
GO:0005576 : extracellular region
GO:0005623 : cell

Presumptive Null
No

Molecular Type
Gene Amplification

Aberration Type
Insertion

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

Main Reference
Two new structural mutations in the 5′ region of the ASIP gene cause diluted feather color phenotypes in Japanese quail. (2019)

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))
Related Haplotypes
2

COMMENTS

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