

GEPHE SUMMARY

Agouti (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^Agouti^#gephebase-summary-title)	Gephebase Gene	GP00000069	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(feathers)^#gephebase-summary-title)		Trait	
Coturnix japonica		Trait State in Taxon A	
Coturnix japonica - yellow phenotype		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Coturnix japonica (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Coturnix+japonica^#gephebase-summary-title)		Coturnix japonica (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Coturnix+japonica^#gephebase-summary-title)	
Japanese quail		Japanese quail	
Coturnix coturnix Japonicus; Coturnix coturnix japonica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849 species		Coturnix coturnix Japonicus; Coturnix coturnix japonica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849 species	
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		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	
Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)		Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)	
93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)		93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)	
No		No	

GENOTYPIC CHANGE

Asip		Q03288 (http://www.uniprot.org/uniprot/Q03288)	
As; ASP; A<y>; ASIP; a		ACA24932 (https://www.ncbi.nlm.nih.gov/nuccore/ACA24932)	
10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)			
-			
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			

(<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

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title>)

Aberration Type

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

Deletion Size

10-100 kb

Molecular Details of the Mutation

141162-bp deletion resulting in novel promoter sharing and expression of a RALY-ASIP fusion transcript

Experimental Evidence

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

Main Reference

Characterization of Japanese quail yellow as a genomic deletion upstream of the avian homolog of the mammalian ASIP (agouti) gene. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18287407>)

Authors

Nadeau NJ; Minvielle F; Ito S; Inoue-Murayama M; Gourichon D; Follett SA; Burke T; Mundy NI

Abstract

ASIP is an important pigmentation gene responsible for dorsoventral and hair-cycle-specific melanin-based color patterning in mammals. We report some of the first evidence that the avian ASIP gene has a role in pigmentation. We have characterized the genetic basis of the homozygous lethal Japanese quail yellow mutation as a >90-kb deletion upstream of ASIP. This deletion encompasses almost the entire coding sequence of two upstream loci, RALY and EIF2B, and places ASIP expression under control of the RALY promoter, leading to the presence of a novel transcript. ASIP mRNA expression was upregulated in many tissues in yellow compared to wild type but was not universal, and consistent differences were not observed among skins of yellow and wild-type quail. In a microarray analysis on developing feather buds, the locus with the largest downregulation in yellow quail was SLC24A5, implying that it is regulated by ASIP. Finally, we document the presence of ventral skin-specific isoforms of ASIP mRNA in both wild-type quails and chickens. Overall, there are remarkable similarities between yellow in quail and lethal yellow in mouse, which involve a deletion in a similar genomic position. The presence of ventral-specific ASIP expression in birds shows that this feature is conserved across vertebrates.

Additional References

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>)

RELATED GEPHE

Related Genes

6 (Endothelin receptor B, MC1R, Melanophilin (MLPH), Microphthalmia-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/>

