

GEPHE SUMMARY

Agouti (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^Agouti^#gephebase-summary-title)	Gephebase Gene	GP00000068	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 - recessive black		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; 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	
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

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

8bp frameshift deletion

Experimental Evidence

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

Main Reference

Recessive black is allelic to the yellow plumage locus in Japanese quail and associated with a frameshift deletion in the ASIP gene. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18287406>)

Authors

Hiragaki T; Inoue-Murayama M; Miwa M; Fujiwara A; Mizutani M; Minvielle F; Ito S

Abstract

The recessive black plumage mutation in the Japanese quail (*Coturnix japonica*) is controlled by an autosomal recessive gene (rb) and displays a blackish-brown phenotype in the recessive homozygous state (rb/rb). A similar black coat color phenotype in nonagouti mice is caused by an autosomal recessive mutation at the agouti locus. An allelism test showed that wild type and mutations for yellow, fawn-2, and recessive black in Japanese quail were multiple alleles (*N, *Y, *F2, and *RB) at the same locus Y and that the dominance relationship was Y*F2 > Y*Y > Y*N > Y*RB. A deletion of 8 bases was found in the ASIP gene in the Y*RB allele, causing a frameshift that changed the last six amino acids, including a cysteine residue, and removed the normal stop codon. Since the cysteine residues at the C terminus are important for disulphide bond formation and tertiary structure of the agouti signaling protein, the deletion is expected to cause a dysfunction of ASIP as an antagonist of alpha-MSH in the Y*RB allele. This is the first evidence that the ASIP gene, known to be involved in coat color variation in mammals, is functional and has a similar effect on plumage color in birds.

Additional References

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/>

