

GEPHE SUMMARY

	Gephebase Gene		GepheID
tyrosinase-related protein 1 (TYRP1) (<a +tyrosinase-related+protein+1+(tyrp1)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title)		GP00001152	
	Entry Status	Martin	Main curator
Published			

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)		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)			
	Trait State in Taxon A		
Coturnix japonica		Trait State in Taxon B	
Coturnix japonica - roux			
	Ancestral State		
Data not curated		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)			
	Taxon A		Taxon B
	Latin Name		Latin Name
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)		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)	
	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) (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)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
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)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Coturnix japonica - roux	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Tyrp1		P07147 (http://www.uniprot.org/uniprot/P07147)	
	Synonyms		GenebankID or UniProtKB
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1		ABW05106 (https://www.ncbi.nlm.nih.gov/nuccore/ABW05106)	
	String		
10090.ENSMUSP00000006151 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)			
	Sequence Similarities		
Belongs to the tyrosinase family.			
	GO - Molecular Function		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)			

GO:0046982 : protein heterodimerization activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046982>)
GO:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)
GO:0004497 : monooxygenase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004497>)

GO - Biological Process

GO:0032438 : melanosome organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
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:0006583 : melanin biosynthetic process from tyrosine
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006583>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0043438 : acetoacetic acid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043438>)
GO:0006582 : melanin metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006582>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0030669 : clathrin-coated endocytic vesicle membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)
GO:0010008 : endosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)
GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)
GO:0033162 : melanosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Phe282Ser

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Phe	Ser	282

Main Reference

Association of a single-nucleotide substitution in TYRP1 with roux in Japanese quail (*Coturnix japonica*). (2007) (<https://pubmed.ncbi.nlm.nih.gov/18028514>)

Authors

Nadeau NJ; Mundy NI; Gourichon D; Minvielle F

Abstract

We investigated TYRP1 as a candidate locus for the recessive, sex-linked roux (br(r)) phenotype in Japanese quail. A screen of the entire coding sequence of TYRP1 in roux and wild-type quail revealed a non-synonymous T-to-C substitution in exon 3, leading to a Phe282Ser mutation. This was perfectly associated with plumage phenotype: all roux birds were homozygous for Ser282. Co-segregation of the Phe282Ser mutation with the roux phenotype was confirmed in three br(r)/BR+ x br(r)/- backcrosses. We found no significant difference in TYRP1 expression between roux and wild-type birds, suggesting that this association is not due to linkage disequilibrium with an unknown regulatory mutation. In addition, the Phe282 amino acid appears to be of functional significance, as it is highly conserved across the vertebrates. This is the first demonstration that TYRP1 has a role in pigmentation in birds.

Additional References

RELATED GEPHE

Related Genes

6 (Agouti, Endothelin receptor B, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, SLC45A2=MATP) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~93934~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001322/93934/>