

GEPHE SUMMARY

	Gephebase Gene	GephelD
Endothelin receptor B2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Endothelin receptor B2^#gephebase-summary-title)	GP00002378	Main curator
Published	Entry Status	Santos

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait		
Coloration (feathers ; white-spotting) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(feathers+;+white-spotting)^#gephebase-summary-title)	Trait State in Taxon A		
Plain colour	Trait State in Taxon B		
White-spotted	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Anas platyrhynchos (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Anas+platyrhynchos^#gephebase-summary-title)		Anas platyrhynchos (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Anas+platyrhynchos^#gephebase-summary-title)	
mallard	Common Name	mallard	Common Name
Anas boschas; Anas domesticus; Anas platyrhynchos f. domestica; mallard; duck; mallard duck; mallard ducks; Anas platyrhynchos Linnaeus 1758; Anas platyrhynchos	Synonyms	Anas boschas; Anas domesticus; Anas platyrhynchos f. domestica; mallard; duck; mallard duck; mallard ducks; Anas platyrhynchos Linnaeus 1758; Anas platyrhynchos	Synonyms
species	Rank	species	Rank
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; Anseriformes; Anatidae; Anatinae; Anas	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; Anseriformes; Anatidae; Anatinae; Anas	Lineage
Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)	Parent	Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)	Parent
8839 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8839)	NCBI Taxonomy ID	8839 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8839)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

EDNRB2	Generic Gene Name	UniProtKB Gallus gallus
-	Synonyms	GenebankID or UniProtKB
-	String	
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities	
GO:0004962 : endothelin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004962)	GO - Molecular Function	
GO:0008217 : regulation of blood pressure (https://www.ebi.ac.uk/QuickGO/term/GO:0008217)	GO - Biological Process	
GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)		
GO:0048484 : enteric nervous system development		

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

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

- (<https://www.gephebase.org/search-criteria/?and+Experimental+Evidence=%Candidate+Gene%#gephebase-summary-title>)

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	His	332

Main Reference

Endothelin Receptor B2 (EDNRB2) Gene Is Associated with Spot Plumage Pattern in Domestic Ducks (*Anas platyrhynchos*). (2015) (<https://pubmed.ncbi.nlm.nih.gov/25955279>)

Authors

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Abstract

Endothelin receptor B subtype 2 (EDNRB2) is a seven-transmembrane G-protein coupled receptor. In this study, we investigated EDNRB2 gene as a candidate gene for duck spot plumage pattern according to studies of chicken and Japanese quail. The entire coding region was cloned by the reverse transcription polymerase chain reaction (RT-PCR). Sequence analysis showed that duck EDNRB2 cDNA contained a 1311 bp open reading frame and encoded a putative protein of 436 amino acids residues. The transcript shared 89%-90% identity with the counterparts in other avian species. A phylogenetic tree based on amino acid sequences showed that duck EDNRB2 was evolutionary conserved in avian clade. The entire coding region of EDNRB2 were sequenced in 20 spot and 20 non-spot ducks, and 13 SNPs were identified. Two of them (c.940G>A and c.995G>A) were non-synonymous substitutions, and were genotyped in 647 ducks representing non-spot and spot phenotypes. The c.995G>A mutation, which results in the amino acid substitution of Arg332His, was completely associated with the spot phenotype: all 152 spot ducks were carriers of the AA genotype and the other 495 individuals with non-spot phenotype were carriers of GA or GG genotype, respectively. Segregation in 17 GA-GG and 22 GA-GA testing combinations confirmed this association since the segregation ratios and genotypes of the offspring were in agreement with the hypothesis. In order to investigate the underlying mechanism of the spot phenotype, MITF gene was used as cell type marker of melanocyte progenitor cells while TYR and TYRP1 gene were used as cell type markers of mature melanocytes. Transcripts of MITF, TYR and TYRP1 gene with expected size were identified in all pigmented skin tissues while PCR products were not obtained from non-pigmented skin tissues. It was inferred that melanocytes are absent in non-pigmented skin tissues of spot ducks.

Additional References

RELATED GEPHE

Related Genes

2 (MC1R, Microphthalmia-associated transcription factor) (<https://www.gephebase.org/search-criteria/?or+TaxonID=%8839%and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

1 (<https://www.gephebase.org/search-criteria/?or+Gene+Gephebase=%Endothelin+receptor+B2%/and+Taxon+ID=%8839%/or+Gene+Gephebase=%Endothelin+receptor+B2%/and+Taxon+ID=%8839%#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

@Parallelism