

GEPHE SUMMARY

	Gephebase Gene	GephelD
Endothelin receptor B2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Endothelin receptor B2^#gephebase-summary-title)	GP00002383	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=%22No%22#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria/?and+Molecular+Type=%22Cis-regulatory%22#gephebase-summary-title>)

Aberration Type

Unknown (<https://www.gephebase.org/search-criteria/?and+Aberration+Type=%22Unknown%22#gephebase-summary-title>)

Molecular Details of the Mutation

"The GWAS results identified a 198 kb (Chr4: 10,149,651 bp to 10,348,068 bp) genetic region that was significantly associated with the black spot phenotype. The conditional GWAS and linkage disequilibrium (LD) analysis further narrowed the ultimate candidate region to 167 kb (Chr4: 10,180,939 bp to 10,348,068 bp). A key gene regulating melanoblast migration and differentiation, EDNRB2 (Endothelin B receptor-like), was found in the candidate region and having significant mRNA expression level changes in embryonic duck skin tissue with different spot sizes. The significant SNPs (single nucleotide polymorphisms) associated with the EDNRB2 gene were annotated, and two mutations (Chr4: 10,180,939 T>C and Chr4: 10,190,671 A>T) were found to result in the loss of binding sites for two trans-factors, XBP1 and cMYB. The phenotypic effect of these two mutations suggested that they can regulate the size of black spots in a dose-dependent manner, and Chr4: 10,180,939 T>C was the major allele locus."

Experimental Evidence

Association Mapping (<https://www.gephebase.org/search-criteria/?and+Experimental+Evidence=%22Association+Mapping%22#gephebase-summary-title>)

Main Reference

Genome-wide association analysis reveals that EDNRB2 causes a dose-dependent loss of pigmentation in ducks. (2021) (<https://pubmed.ncbi.nlm.nih.gov/34034661>)

Authors

Xi Y; Xu Q; Huang Q; Ma S; Wang Y; Han C; Zhang R; Wang J; Liu H; Li L

Abstract

Birds have various plumage color patterns, and spot is a common phenotype. Herein, we conducted genome-wide association studies (GWAS) in a population of 225 ducks with different sized black spots to reveal the genetic basis of this phenomenon.

First, we quantified the black spot phenotype within the duck population. The results showed that the uncolored area of the body surface first appeared on the ventral side. With increasing duck age, the area of the black spots was highly conserved across the whole body surface. The GWAS results identified a 198 kb (Chr4: 10,149,651 bp to 10,348,068 bp) genetic region that was significantly associated with the black spot phenotype. The conditional GWAS and linkage disequilibrium (LD) analysis further narrowed the ultimate candidate region to 167 kb (Chr4: 10,180,939 bp to 10,348,068 bp). A key gene regulating melanoblast migration and differentiation, EDNRB2 (Endothelin B receptor-like), was found in the candidate region and having significant mRNA expression level changes in embryonic duck skin tissue with different spot sizes. The significant SNPs (single nucleotide polymorphisms) associated with the EDNRB2 gene were annotated, and two mutations (Chr4: 10,180,939 T>C and Chr4: 10,190,671 A>T) were found to result in the loss of binding sites for two trans-factors, XBP1 and cMYB. The phenotypic effect of these two mutations suggested that they can regulate the size of black spots in a dose-dependent manner, and Chr4: 10,180,939 T>C was the major allele locus.

Our results revealed that EDNRB2 was the gene responsible for the variation in duck body surface spot size. Chr4: 10,180,939 T>C was the major allele that explained 49.5% (dorsal side) and 32.9% (ventral side) of the variation in duck body surface spot size, while 32.1% (dorsal side) and 19.1% (ventral side) of the variation could be explained by Chr4: 10,190,671 A>T. The trans-factor prediction also suggested that XBP1 and cMYB have the potential to interact with EDNRB2, providing new insights into the mechanism of action of these genes.

Additional References

RELATED GEPHE

Related Genes

2 (MC1R, Microphthalmia-associated transcription factor) (<https://www.gephebase.org/search-criteria/?or+TaxonID=%228839%22+and+Trait=Coloration+and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

1 (<https://www.gephebase.org/search-criteria/?or+Gene+Gephebase=%22Endothelin+receptor+B2%22+and+Taxon+ID=%228839%22+or+Gene+Gephebase=%22Endothelin+receptor+B2%22+and+Taxon+ID=%228839%22#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

@Parallelism