GEPHE SUMMARY

Gephebase Gene
Endothelin receptor B2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Endothelin receptor B2^#gephebase-summary-title)

GepheID
GP00002383

Entry Status
Published

Main curator
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
Anas platyrhynchos (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas platyrhynchos^#gephebase-summary-title)

Common Name
mallard

Synonyms
Anas boschas; Anas domesticus; Anas platyrhynchos f. domestica; mallard; duck; mallard duck; mallard ducks; Anas platyrhynchos Linnaeus 1758; Anas platyrhynchus

Rank
species

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

Parent
Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)

NCBI Taxonomy ID
8839 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8839)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Anas platyrhynchos (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas platyrhynchos^#gephebase-summary-title)

Common Name
mallard

Synonyms
Anas boschas; Anas domesticus; Anas platyrhynchos f. domestica; mallard; duck; mallard duck; mallard ducks; Anas platyrhynchos Linnaeus 1758; Anas platyrhynchus

Rank
species

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

Parent
Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)

NCBI Taxonomy ID
8839 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8839)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
EDNRB2

Synonyms
-

String
-

Sequence Similarities
Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function
GO:0004962 : endothelin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004962)

GO - Biological Process
GO:0008217 : regulation of blood pressure (https://www.ebi.ac.uk/QuickGO/term/GO:0008217)
GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)
GO:0048484 : enteric nervous system development

UniProtKB Gallus gallus
W8VUK4 (http://www.uniprot.org/uniprot/W8VUK4)

GenebankID or UniProtKB
()

(https://www.ebi.ac.uk/QuickGO/term/GO:0048484)

GO - Cellular Component

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

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

Aberration Type

Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#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 Tâ€‰>â€‰C and Chr4: 10,190,671 Aâ€‰>â€‰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 Tâ€‰>â€‰C was the major allele locus."

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#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 198Â kb (Chr4: 10,149,651Â bp to 10,348,068Â 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 167Â kb (Chr4: 10,180,939Â bp to 10,348,068Â 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 Tâ€‰>â€‰C and Chr4: 10,190,671Â Aâ€‰>â€‰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 Tâ€‰>â€‰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 Tâ€‰>â€‰C was the major allele that explained 49.5â€‰% (dorsal side) and 32.9â€‰% (ventral side) of the variation in duck body surface spot size, while 32.1â€‰% (dorsal side) and 19.1â€‰% (ventral side) of the variation could be explained by Chr4: 10,190,671Â Aâ€‰>â€‰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, Microphtalmia-associated transcription factor) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^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