

GEPHE SUMMARY

<p>Endothelin receptor B2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Endothelin+receptor+B2^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002383</p> <p>Santos</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

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

<p>EDNRB2</p> <p>-</p> <p>-</p> <p>Belongs to the G-protein coupled receptor 1 family.</p> <p>GO:0004962 : endothelin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004962)</p> <p>GO:0008217 : regulation of blood pressure (https://www.ebi.ac.uk/QuickGO/term/GO:0008217)</p> <p>GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)</p> <p>GO:0048484 : enteric nervous system development</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p>	<p>UniProtKB Gallus gallus</p> <p>GenebankID or UniProtKB</p> <p>W8VUK4 (http://www.uniprot.org/uniprot/W8VUK4)</p> <p>0</p>
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(<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 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=^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 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, 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