GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00001361

Entry Status

Published

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (feathers)^#gephebase-summary-title)

Trait State in Taxon A

Chicken-EJ and Rhode Island RedY8/NU and Sebright bantam and Large Cochin and others-non mottled

Trait State in Taxon B

Chicken-EJ and CB and Chabo and Uzurao-with mottled plumage

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title)

Common Name

chicken

Synonyms

Gallus gallus domesticus; chicken; bantam; chickens

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; Galliformes; Phasianidae; Phasianinae; Gallus

Parent

Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)

NCBI Taxonomy ID

9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Chicken-EJ and Rhode Island RedY8/NU and Sebright bantam and Large Cochin and others-non mottled

Taxon B

Latin Name

Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title)

Common Name

chicken

Synonyms

Gallus gallus domesticus; chicken; bantam; chickens

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; Galliformes; Phasianidae; Phasianinae; Gallus

Parent

Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)

NCBI Taxonomy ID

9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Chicken-EJ and CB and Chabo and Uzurao-with mottled plumage

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

UniProtKB Gallus gallus

W8VUK4 (http://www.uniprot.org/uniprot/W8VUK4)

GenebankID or UniProtKB

()

(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
(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

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

c.G1272A p.Arg332His

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	Arg	His	332

Main Reference

Endothelin receptor B2 (EDNRB2) is responsible for the tyrosinase-independent recessive white (mo(w)) and mottled (mo) plumage phenotypes in the chicken. (2014)
(https://pubmed.ncbi.nlm.nih.gov/24466053)

Authors

Kinoshita K; Akiyama T; Mizutani M; Shinomiya A; Ishikawa A; Younis HH; Tsudzuki M; Namikawa T; Matsuda Y

Abstract

A mutation that confers white plumage with black eyes was identified in the Minohiki breed of Japanese native chicken (Gallus gallus domesticus). The white plumage, with a few partially pigmented feathers, was not associated with the tyrosinase gene, and displayed an autosomal recessive mode of inheritance against the pigmented phenotype. All F1 offspring derived from crosses with mottled chickens (mo/mo), which show characteristic pigmented feathers with white tips, had plumage with a mottled-like pattern. This result indicates that the white plumage mutation is a novel allele at the mo locus; we propose the gene symbol mo(w) for this mutant allele. Furthermore, the F1 hybrid between the mo(w) /mo(w) chicken and the panda (s/s) mutant of Japanese quail (Coturnix japonica), whose causative gene is the endothelin receptor B2 (EDNRB2) gene, showed a mo(w)/mo(w) chicken-like plumage, suggesting the possibility that the mutations in parental species are alleles of the same gene, EDNRB2. Nucleotide sequencing of the entire coding region of EDNRB2 revealed a non-synonymous G1008T substitution, which causes Cys244Phe amino acid substitution in exon 5 (which is part of the extracellular loop between the putative fourth and fifth transmembrane domains of EDNRB2) in the mutant chicken. This Cys244Phe mutation was also present in individuals of four Japanese breeds with white plumage. We also identified a non-synonymous substitution leading to Arg332His substitution that was responsible for the mottled (mo/mo) plumage phenotype. These results suggest that the EDN3 (endothelin 3)-EDNRB2 signaling is essential for normal pigmentation in birds, and that the mutations of EDNRB2 may cause defective binding of the protein with endothelins, which interferes with melanocyte differentiation, proliferation, and migration.

Additional References

RELATED GEPHE

Related Genes

14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, GRAMD3, MC1R, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, SLCO1B3, SOX10, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9031^/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=^9031^/or+Gene Gephebase=^Endothelin receptor B2^/and+Taxon ID=^9031^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

this is the mottled (mo) locus ; the mutation is recessive ; @AllelicSeries ; https://omia.org/OMIA001904/9031/