GEPHE SUMMARY

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

GepheID
GP00001360

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-Minohiki and others-wild type

Trait State in Taxon B
Chicken-Minohiki and Onagadori and Ohiki and Shokoku and Uzurao-white with a few pigmented feathers

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-Minohiki and others-wild type

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-Minohiki and Onagadori and Ohiki and Shokoku and Uzurao-white with a few pigmented feathers

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.1008G>T p.Cys244Phe

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	Cys	Phe	244

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 ; https://omia.org/OMIA001904/9031/