

GEPHE SUMMARY

	Gephebase Gene	GephelD
Endothelin receptor B2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Endothelin receptor B2^#gephebase-summary-title)	GP00001360	Main curator
Published	Entry Status	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

Taxon A	Latin Name	Taxon B	Latin Name
Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus gallus^#gephebase-summary-title)	Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus gallus^#gephebase-summary-title)	Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus gallus^#gephebase-summary-title)	Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus gallus^#gephebase-summary-title)
chicken	Common Name	chicken	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms	Gallus gallus domesticus; chicken; bantam; chickens	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; Galliformes; Phasianidae; Phasianinae; Gallus	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	Lineage
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)	Parent	Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)	Parent
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)	NCBI Taxonomy ID	9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)	NCBI Taxonomy ID
Yes	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
Chicken-Minohiki and others-wild type	Taxon A Description	Chicken-Minohiki and Onagadori and Ohiki and Shokoku and Uzurao-white with a few pigmented feathers	Taxon B Description

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	GO - Biological Process	

(<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/>

