GEPHE SUMMARY

Gephebase Gene

EDN3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EDN3^#gephebase-summary-title)

GepheID

GP00000258

Entry Status

Published

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

WT

Trait State in Taxon B

Silky/Silkie pigmentation (Fibromelanosis) - dominant

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?

No

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

several breeds including Ayam Cemani ; Black H'Mong ; Silky/Silkie

GENOTYPIC CHANGE

Generic Gene Name

EDN3

Synonyms

ET3; ET-3; WS4B; HSCR4; PPET3

String

9606.ENSP00000337128 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000337128)

Sequence Similarities

Belongs to the endothelin/sarafotoxin family.

GO - Molecular Function

GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)
GO:0031708 : endothelin B receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031708)

UniProtKB Homo sapiens

P14138 (http://www.uniprot.org/uniprot/P14138)

GenebankID or UniProtKB

BAE45237 (https://www.ncbi.nlm.nih.gov/nuccore/BAE45237)

GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179)

GO - Biological Process

GO:0007275 : multicellular organism development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)
GO:0030182 : neuron differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030182)
GO:0001755 : neural crest cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0001755)
GO:0006874 : cellular calcium ion homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006874)
GO:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0043406 : positive regulation of MAP kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0043406)
GO:0007267 : cell-cell signaling (https://www.ebi.ac.uk/QuickGO/term/GO:0007267)
GO:0045597 : positive regulation of cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0045597)
GO:0010460 : positive regulation of heart rate
(https://www.ebi.ac.uk/QuickGO/term/GO:0010460)
GO:0048070 : regulation of developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048070)
GO:0030593 : neutrophil chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0030593)
GO:0010468 : regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010468)
GO:0008015 : blood circulation (https://www.ebi.ac.uk/QuickGO/term/GO:0008015)
GO:0007166 : cell surface receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007166)
GO:0010961 : cellular magnesium ion homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0010961)
GO:0048016 : inositol phosphate-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0048016)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0030072 : peptide hormone secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0030072)
GO:0046887 : positive regulation of hormone secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0046887)
GO:0002690 : positive regulation of leukocyte chemotaxis
(https://www.ebi.ac.uk/QuickGO/term/GO:0002690)
GO:0045840 : positive regulation of mitotic nuclear division
(https://www.ebi.ac.uk/QuickGO/term/GO:0045840)
GO:1901381 : positive regulation of potassium ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:1901381)
GO:0003100 : regulation of systemic arterial blood pressure by endothelin
(https://www.ebi.ac.uk/QuickGO/term/GO:0003100)
GO:0019229 : regulation of vasoconstriction
(https://www.ebi.ac.uk/QuickGO/term/GO:0019229)
GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)
GO:0014826 : vein smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:0014826)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005623 : cell (https://www.ebi.ac.uk/QuickGO/term/GO:0005623)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

Complex Change (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Complex Change^#gephebase-summary-title)

Molecular Details of the Mutation

Large duplication

Experimental Evidence

Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)

Main Reference

A complex genomic rearrangement involving the endothelin 3 locus causes dermal hyperpigmentation in the chicken. (2011) (https://pubmed.ncbi.nlm.nih.gov/22216010)

Authors

Dorshorst B; Molin AM; Rubin CJ; Johansson AM; StrÃ¶mstedt L; Pham MH; Chen CF; HallbÃ¤Ã¤k F; Ashwell C; Andersson L

Abstract

Dermal hyperpigmentation or Fibromelanosis (FM) is one of the few examples of skin pigmentation phenotypes in the chicken, where most other pigmentation variants influence feather color and patterning. The Silkie chicken is the most widespread and well-studied breed displaying this phenotype. The presence of the dominant FM allele results in extensive pigmentation of the dermal layer of skin and the majority of internal connective tissue. Here we identify the causal mutation of FM as an inverted duplication and junction of two genomic regions separated by more than 400 kb in wild-type individuals. One of these duplicated regions contains endothelin 3 (EDN3), a gene with a known role in promoting melanoblast proliferation. We show that EDN3 expression is increased in the developing Silkie embryo during the time in which melanoblasts are migrating, and elevated levels of expression are maintained in the adult skin tissue. We have examined four different chicken breeds from both Asia and Europe displaying dermal hyperpigmentation and conclude that the same structural variant underlies this phenotype in all chicken breeds. This complex genomic rearrangement causing a specific monogenic trait in the chicken illustrates how novel mutations with major phenotypic effects have been reused during

breed formation in domestic animals.

Additional References

Characterization of genetic diversity and gene mapping in two Swedish local chicken breeds. (2015) (https://pubmed.ncbi.nlm.nih.gov/25741364)
Identification and functional characterization of copy number variations in diverse chicken breeds. (2014) (https://pubmed.ncbi.nlm.nih.gov/25344733)
The origin and evolution of fibromelanosis in domesticated chickens: Genomic comparison of Indonesian Cemani and Chinese Silkie breeds. (2017) (https://pubmed.ncbi.nlm.nih.gov/28379963)

RELATED GEPHE

Related Genes

14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, Endothelin receptor B2, 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

No matches found.

EXTERNAL LINKS

COMMENTS

Copy Number Variation in different breeds ; https://omia.org/OMIA001671/9031/