GEPHE SUMMARY

Gephebase Gene

GDF7 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^GDF7^#gephebase-summary-title)

GepheID

GP00002185

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

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

Trait State in Taxon A

Chicken breeds with normal feathering

Trait State in Taxon B

Naked neck fowls with reduced body feathering : ~20% in heterozygotes and - 40% in homozygotes (well visible in neck) ; increased thermotolerance in hot climates

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?

No

GENOTYPIC CHANGE

Generic Gene Name

Gdf7

Synonyms

BMP12; Gdf-7

String

10090.ENSMUSP00000038301 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000038301)

Sequence Similarities

Belongs to the TGF-beta family.

GO - Molecular Function

GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)

GO - Biological Process

UniProtKB Mus musculus

P43029 (http://www.uniprot.org/uniprot/P43029)

GenebankID or UniProtKB

()

GO:0045893 : positive regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0060571 : morphogenesis of an epithelial fold
(https://www.ebi.ac.uk/QuickGO/term/GO:0060571)
GO:0010628 : positive regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0030509 : BMP signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0030509)
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0010862)
GO:0060395 : SMAD protein signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0060395)
GO:0045165 : cell fate commitment (https://www.ebi.ac.uk/QuickGO/term/GO:0045165)
GO:0045666 : positive regulation of neuron differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0045666)
GO:0007411 : axon guidance (https://www.ebi.ac.uk/QuickGO/term/GO:0007411)
GO:0030855 : epithelial cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030855)
GO:0022612 : gland morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0022612)
GO:0030901 : midbrain development
(https://www.ebi.ac.uk/QuickGO/term/GO:0030901)
GO:0048608 : reproductive structure development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048608)
GO:0032924 : activin receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0032924)
GO:0060389 : pathway-restricted SMAD protein phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0060389)
GO:0048754 : branching morphogenesis of an epithelial tube
(https://www.ebi.ac.uk/QuickGO/term/GO:0048754)
GO:0021527 : spinal cord association neuron differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0021527)
GO:0021915 : neural tube development
(https://www.ebi.ac.uk/QuickGO/term/GO:0021915)
GO:0048853 : forebrain morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048853)
GO:2001051 : positive regulation of tendon cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:2001051)
GO:0021509 : roof plate formation (https://www.ebi.ac.uk/QuickGO/term/GO:0021509)

GO - Cellular Component

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

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

Aberration Type

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

Insertion Size

100-1000 kb

Molecular Details of the Mutation

a large insertion approximately 260kb downstream from the BMP12 gene (now known as GFD7) increasing the expression of this gene in embryonic skin

Experimental Evidence

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

Main Reference

Cryptic patterning of avian skin confers a developmental facility for loss of neck feathering. (2011) (https://pubmed.ncbi.nlm.nih.gov/21423653)

Authors

Mou C; Pitel F; Gourichon D; Vignoles F; Tzika A; Tato P; Yu L; Burt DW; Bed'hom B; Tixier-Boichard M; Painter KJ; Headon DJ

Abstract

Vertebrate skin is characterized by its patterned array of appendages, whether feathers, hairs, or scales. In avian skin the distribution of feathers occurs on two distinct spatial levels. Grouping of feathers within discrete tracts, with bare skin lying between the tracts, is termed the macropattern, while the smaller scale periodic spacing between individual feathers is referred to as the micropattern. The degree of integration between the patterning mechanisms that operate on these two scales during development and the mechanisms underlying the remarkable evolvability of skin macropatterns are unknown. A striking example of macropattern variation is the convergent loss of neck feathering in multiple species, a trait associated with heat tolerance in both wild and domestic birds. In chicken, a mutation called Naked neck is characterized by a reduction of body feathering and completely bare neck. Here we perform genetic fine mapping of the causative region and identify a large insertion associated with the Naked neck trait. A strong candidate gene in the critical interval, BMP12/GDF7, displays markedly elevated expression in Naked neck embryonic skin due to a cis-regulatory effect of the causative mutation. BMP family members inhibit embryonic feather formation by acting in a reaction-diffusion mechanism, and we find that selective production of retinoic acid by neck skin potentiates BMP signaling, making neck skin more sensitive than body skin to suppression of feather development. This selective production of retinoic acid by neck skin constitutes a cryptic pattern as its effects on feathering are not revealed until gross BMP levels are altered. This developmental modularity of neck and body skin allows simple quantitative changes in BMP levels to produce a sparsely feathered or bare neck while maintaining robust feather patterning on the body.

Additional References

RELATED GEPHE

Related Genes

14 (ABCA1, CDKN2A, CYP19A1, Endothelin receptor B2, MC1R, PMEL17, SLC45A2=MATP, SOX10, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1), FGF20, HOXC8 - uncertain, KRT6A, KRT75L4) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Feathers/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Inserted region is the copy of an other chromosomal section that may have brought in Wnt11 enhancers (also expressed in the neck) ; https://omia.org/OMIA000701/9031/