GEPHE SUMMARY

Gephebase Gene

Hoxb8 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Hoxb8^#gephebase-summary-title)

GepheID

GP00002193

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

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

Trait State in Taxon A

WT chickens

Trait State in Taxon B

Chickens with muffs and beards ; incompletely dominant autosomal Mb mutation ; several breeds including Huiyang Bearded

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

Hoxb8

Synonyms

Hox-2.4; Hoxb-8

String

10090.ENSMUSP00000052496 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000052496)

Sequence Similarities

Belongs to the Antp homeobox family.

GO - Molecular Function

GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0001227 : DNA-binding transcription repressor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001227)

GO - Biological Process

UniProtKB Mus musculus

P09632 (http://www.uniprot.org/uniprot/P09632)

GenebankID or UniProtKB

()

GO:0009952 : anterior/posterior pattern specification
(https://www.ebi.ac.uk/QuickGO/term/GO:0009952)
GO:0048704 : embryonic skeletal system morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048704)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0007625 : grooming behavior (https://www.ebi.ac.uk/QuickGO/term/GO:0007625)
GO:0008344 : adult locomotory behavior
(https://www.ebi.ac.uk/QuickGO/term/GO:0008344)
GO:0019233 : sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0019233)
GO:0048705 : skeletal system morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048705)
GO:0045638 : negative regulation of myeloid cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0045638)
GO:0021516 : dorsal spinal cord development
(https://www.ebi.ac.uk/QuickGO/term/GO:0021516)

GO - Cellular Component

GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)

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

10-100 kb

Molecular Details of the Mutation

Complex Structural Variation with 3 CNVs including a 14.8kb copy of hoxb7 + hoxb8 ; results in ectopic (unrepressed) expression of hoxb8 in feathers especially in the facial region

Experimental Evidence

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

Main Reference

A Complex Structural Variation on Chromosome 27 Leads to the Ectopic Expression of HOXB8 and the Muffs and Beard Phenotype in Chickens. (2016)
(https://pubmed.ncbi.nlm.nih.gov/27253709)

Authors

Guo Y; Gu X; Sheng Z; Wang Y; Luo C; Liu R; Qu H; Shu D; Wen J; Crooijmans RP; Carlborg Ã–; Zhao Y; Hu X; Li N

Abstract

Muffs and beard (Mb) is a phenotype in chickens where groups of elongated feathers gather from both sides of the face (muffs) and below the beak (beard). It is an autosomal, incomplete dominant phenotype encoded by the Muffs and beard (Mb) locus. Here we use genome-wide association (GWA) analysis, linkage analysis, Identity-by-Descent (IBD) mapping, array-CGH, genome re-sequencing and expression analysis to show that the Mb allele causing the Mb phenotype is a derived allele where a complex structural variation (SV) on GGA27 leads to an altered expression of the gene HOXB8. This Mb allele was shown to be completely associated with the Mb phenotype in nine other independent Mb chicken breeds. The Mb allele differs from the wild-type mb allele by three duplications, one in tandem and two that are translocated to that of the tandem repeat around 1.70 Mb on GGA27. The duplications contain total seven annotated genes and their expression was tested during distinct stages of Mb morphogenesis. A continuous high ectopic expression of HOXB8 was found in the facial skin of Mb chickens, strongly suggesting that HOXB8 directs this regional feather-development. In conclusion, our results provide an interesting example of how genomic structural rearrangements alter the regulation of genes leading to novel phenotypes. Further, it again illustrates the value of utilizing derived phenotypes in domestic animals to dissect the genetic basis of developmental traits, herein providing novel insights into the likely role of HOXB8 in feather development and differentiation.

Additional References

RELATED GEPHE

Related Genes

17 (ABCA1, CDKN2A, CYP19A1, Endothelin receptor B2, MC1R, PMEL17, SLC45A2=MATP, SOX10, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1), FGF20, GDF7, HOXC8 - uncertain, KRT6A, KRT75L4, PDSS2, Prolactin receptor) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Feather/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@CNV @GainOfFunction with ectopic expression ; https://omia.org/OMIA000668/9031/