

GEPHE SUMMARY

	Gephebase Gene		GepheID
Hoxb8 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~Hoxb8~#gephebase-summary-title)		GP00002193	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~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+TaxonomicStatus=~Domesticated~#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		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)	
	Common Name		Common Name
chicken		chicken	
	Synonyms		Synonyms
Gallus gallus domesticus; chicken; bantam; chickens		Gallus gallus domesticus; chicken; bantam; chickens	
	Rank		Rank
species		species	
	Lineage		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		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		Parent
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)		Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)		9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Hoxb8		P09632 (http://www.uniprot.org/uniprot/P09632)	
	Synonyms		GenebankID or UniProtKB
Hox-2.4; Hoxb-8		0	
	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		

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](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

Molecular Type

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

Aberration Type

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

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](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=))

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 Å-; 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](https://www.gephebase.org/search-criteria?/or+Taxon ID=))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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

