

GEPHE SUMMARY

	Gephebase Gene		GepheID
SOX5 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~SOX5~#gephebase-summary-title)		GP00001070	
	Entry Status	Courtier	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)		Trait	
Bird head comb (reduced comb and wattles) (https://www.gephebase.org/search-criteria?/and+Trait=~Bird head comb (reduced comb and wattles)~#gephebase-summary-title)			
	Trait State in Taxon A		
Gallus gallus		Trait State in Taxon B	
Gallus gallus - Pea Comb			
	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		Yes	
			Taxon B Description
		Gallus gallus - Pea Comb	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Sox5		P35710 (http://www.uniprot.org/uniprot/P35710)	
	Synonyms		GenebankID or UniProtKB
AI528773; A730017D01Rik; Sox-5		NP_001004385 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001004385)	
	String		
10090.ENSMUSP00000047567 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000047567)			
	Sequence Similarities		
-			
	GO - Molecular Function		
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)			
GO:0046982 : protein heterodimerization activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0046982>)
GO:0044212 : transcription regulatory region DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044212>)
GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0045892 : negative regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0001701 : in utero embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001701>)
GO:0061036 : positive regulation of cartilage development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061036>)
GO:0051216 : cartilage development (<https://www.ebi.ac.uk/QuickGO/term/GO:0051216>)
GO:0045165 : cell fate commitment (<https://www.ebi.ac.uk/QuickGO/term/GO:0045165>)
GO:0032332 : positive regulation of chondrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032332>)
GO:0071560 : cellular response to transforming growth factor beta stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071560>)
GO:0048709 : oligodendrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048709>)
GO:0055059 : asymmetric neuroblast division
(<https://www.ebi.ac.uk/QuickGO/term/GO:0055059>)
GO:0021953 : central nervous system neuron differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0021953>)
GO:2000741 : positive regulation of mesenchymal stem cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000741>)
GO:0060164 : regulation of timing of neuron differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060164>)

GO - Cellular Component

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0044798 : nuclear transcription factor complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044798>)

	Presumptive Null
No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)	
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)	Molecular Type
Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)	Aberration Type
10-100 kb	Insertion Size
Copy Number Variation in Intron 1 - about 30 copies of a 3 kb sequence - corresponding to about 85 kb of extra sequence	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)	Experimental Evidence
Copy number variation in intron 1 of SOX5 causes the Pea-comb phenotype in chickens. (2009) (https://pubmed.ncbi.nlm.nih.gov/19521496)	Main Reference
Wright D; Boije H; Meadows JR; Bed'hom B; Gourichon D; Vieaud A; Tixier-Boichard M; Rubin CJ; Imsland F; Hallbäck F; Andersson L	Authors
Pea-comb is a dominant mutation in chickens that drastically reduces the size of the comb and wattles. It is an adaptive trait in cold climates as it reduces heat loss and makes the chicken less susceptible to frost lesions. Here we report that Pea-comb is caused by a massive amplification of a duplicated sequence located near evolutionary conserved non-coding sequences in intron 1 of the gene encoding the SOX5 transcription factor. This must be the causative mutation since all other polymorphisms associated with the Pea-comb allele were excluded by genetic analysis. SOX5 controls cell fate and differentiation and is essential for skeletal development, chondrocyte differentiation, and extracellular matrix production. Immunostaining in early embryos demonstrated that Pea-comb is associated with ectopic expression of SOX5 in mesenchymal cells located just beneath the surface ectoderm where the comb and wattles will subsequently develop. The results imply that the duplication expansion interferes with the regulation of SOX5 expression during the differentiation of cells crucial for the development of comb and wattles. The study provides novel insight into the nature of mutations that contribute to phenotypic evolution and is the first description of a spontaneous and fully viable mutation in this developmentally important gene.	Abstract
	Additional References

RELATED GEPHE

	Related Genes
2 (EOMES (eomesodermin), MNR2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Bird head comb/and+groupHaplotypes=true#gephebase-summary-title)	
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@CNV <https://omia.org/OMIA000782/9031/>