

GEPHE SUMMARY

SOX10 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~SOX10~#gephebase-summary-title)	Gephebase Gene	GP00001069	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)		Trait Category
Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+feathers^#gephebase-summary-title)		Trait
Gallus gallus	Trait State in Taxon A	
Gallus gallus - Dark Brown mutants and Light Yellow mutants	Trait State in Taxon B	
Taxon A	Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)	Taxonomic Status	
Taxon A	Taxon B	
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)	Latin Name
chicken	chicken	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Gallus gallus domesticus; chicken; bantam; chickens	Synonyms
species	species	Rank
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	Lineage
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)	Parent
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)	NCBI Taxonomy ID
No	Yes	is Taxon A an Intraspecies?
		Taxon B Description
	Gallus gallus - Dark Brown mutants and Light Yellow mutants	

GENOTYPIC CHANGE

Sox10	Generic Gene Name	Q04888 (http://www.uniprot.org/uniprot/Q04888)	UniProtKB Mus musculus
	Synonyms		GenebankID or UniProtKB
gt; Dom; Sox21; Sox-10		AAD38050 (https://www.ncbi.nlm.nih.gov/nucore/AAD38050)	
	String		
10090.ENSMUSP00000039466 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000039466)			
	Sequence Similarities		
-			
	GO - Molecular Function		
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)			
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)			

GO:0003700 : DNA-binding transcription factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003700>)
GO:0008134 : transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008134>)
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:0003682 : chromatin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003682>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
GO:0000980 : RNA polymerase II distal enhancer sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000980>)
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-specific binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003705>)
GO:1990841 : promoter-specific chromatin binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:1990841>)

GO - Biological Process

GO:0007417 : central nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007417>)
GO:0043066 : negative regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)
GO:0007422 : peripheral nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007422>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0006355 : regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006355>)
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
GO:0045892 : negative regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0001755 : neural crest cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001755>)
GO:0045893 : positive regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0090090 : negative regulation of canonical Wnt signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090090>)
GO:0001701 : in utero embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001701>)
GO:0010628 : positive regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0048546 : digestive tract morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048546>)
GO:0002009 : morphogenesis of an epithelium
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002009>)
GO:0048484 : enteric nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048484>)
GO:0048469 : cell maturation (<https://www.ebi.ac.uk/QuickGO/term/GO:0048469>)
GO:0032808 : lacrimal gland development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032808>)
GO:0048589 : developmental growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048589>)
GO:0071393 : cellular response to progesterone stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071393>)
GO:0022010 : central nervous system myelination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0022010>)
GO:0061138 : morphogenesis of a branching epithelium
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061138>)
GO:0010626 : negative regulation of Schwann cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010626>)
GO:0014003 : oligodendrocyte development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014003>)
GO:0048709 : oligodendrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048709>)
GO:0014015 : positive regulation of gliogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014015>)
GO:0031643 : positive regulation of myelination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031643>)
GO:0002052 : positive regulation of neuroblast proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002052>)
GO:0006368 : transcription elongation from RNA polymerase II promoter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006368>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0000785 : chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000785>)
GO:0031315 : extrinsic component of mitochondrial outer membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031315>)

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>)

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

1-10 kb

8.3kb deletion upstream of the SOX10 transcription start site

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

The Dark brown plumage color in chickens is caused by an 8.3-kb deletion upstream of SOX10. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21210960>)

Gunnarsson U; Kerje S; Bed'hom B; Sahlqvist AS; Ekwall O; Tixier-Boichard M; KÄmpe O; Andersson L

The Dark brown (DB) mutation in chickens reduces expression of black eumelanin and enhances expression of red pheomelanin, but only in certain parts of the plumage. Here, we present genetic evidence that an 8.3-kb deletion upstream of the SOX10 transcription start site is the causal mutation underlying the DB phenotype. The SOX10 transcription factor has a well-established role in melanocyte biology and is essential for melanocyte migration and survival. Previous studies have demonstrated that the mouse homolog of a highly conserved element within the deleted region is a SOX10 enhancer. The mechanism of action of this mutation remains to be established, but one possible scenario is that the deletion leads to reduced SOX10 expression which in turn down-regulates expression of key enzymes in pigment synthesis such as tyrosinase. Lower tyrosinase activity leads to a shift toward a more pheomelanistic (reddish) plumage color, which is the characteristic feature of the DB phenotype.

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A Deletion Upstream of SOX10 Causes Light Yellow Plumage Colour in Chicken. (2022) (<https://pubmed.ncbi.nlm.nih.gov/35205371>)

RELATED GEPHE

14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, GRAMD3, MC1R, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, SLCO1B3, 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>)

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism <https://omia.org/OMIA001569/9031/>