

GEPHE SUMMARY

	Gephebase Gene	GephelD
PDSS2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^PDSS2^#gephebase-summary-title)	GP00002254	Main curator
Published	Entry Status	Martin

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait		
Feather (https://www.gephebase.org/search-criteria?/and+Trait=^Feather^#gephebase-summary-title)	Trait State in Taxon A		
WT Feathering	Trait State in Taxon B		
Silky/Silkie feathering (recessive)	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	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)	
chicken	Common Name	chicken	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms	Gallus gallus domesticus; chicken; bantam; chickens	Synonyms
species	Rank	species	Rank
	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	
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)	Parent	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)	NCBI Taxonomy ID	9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

-	Generic Gene Name	UniProtKB
-	Q86YH6NULL (http://www.uniprot.org/uniprot/Q86YH6NULL)	GenebankID or UniProtKB
-	Synonyms	
-	0	
-	String	
-	Sequence Similarities	
-	GO - Molecular Function	
-	GO - Biological Process	
-	GO - Cellular Component	
-		Presumptive Null
No (https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^No^#gephebase-summary-title)		Molecular Type

Cis-regulatory ([#gephebase-summary-title](https://www.gephebase.org/search-criteria/?and+Molecular%20Type=%Cis-regulatory))

Aberration Type

SNP ([#gephebase-summary-title](https://www.gephebase.org/search-criteria/?and+Aberration%20Type=%SNP))

Molecular Details of the Mutation

Only variant explaining homozygous phenotype is c.-103C>G and shown to reduce PDSS2 promoter activity in vitro

Experimental Evidence

Linkage Mapping ([#gephebase-summary-title](https://www.gephebase.org/search-criteria/?and+Experimental%20Evidence=%Linkage%20Mapping))

Main Reference

A cis-regulatory mutation of PDSS2 causes silky-feather in chickens. (2014) (<https://pubmed.ncbi.nlm.nih.gov/25166907/>)

Authors

Feng C; Gao Y; Dorshorst B; Song C; Gu X; Li Q; Li J; Liu T; Rubin CJ; Zhao Y; Wang Y; Fei J; Li H; Chen K; Qu H; Shu D; Ashwell C; Da Y; Andersson L; Hu X; Li N

Abstract

Silky-feather has been selected and fixed in some breeds due to its unique appearance. This phenotype is caused by a single recessive gene (*hookless*, *h*). Here we map the silky-feather locus to chromosome 3 by linkage analysis and subsequently fine-map it to an 18.9 kb interval using the identical by descent (IBD) method. Further analysis reveals that a C to G transversion located upstream of the prenyl (decaprenyl) diphosphate synthase, subunit 2 (PDSS2) gene is causing silky-feather. All silky-feather birds are homozygous for the G allele. The silky-feather mutation significantly decreases the expression of PDSS2 during feather development in vivo. Consistent with the regulatory effect, the C to G transversion is shown to remarkably reduce PDSS2 promoter activity in vitro. We report a new example of feather structure variation associated with a spontaneous mutation and provide new insight into the PDSS2 function.

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, Hoxb8, Prolactin receptor) ([#gephebase-summary-title](https://www.gephebase.org/search-criteria/?or+TaxonID=%9031^/and+Trait=Feather/and+groupHaplotypes=true))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000913/9031/>