

GEPHE SUMMARY

KRT6A (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~KRT6A^#gephebase-summary-title)	Gephebase Gene	GP00000527	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Feathers (https://www.gephebase.org/search-criteria?/and+Trait=~Feathers^#gephebase-summary-title)		Trait	
Gallus gallus		Trait State in Taxon A	
Gallus gallus - Frizzle Feather		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Gallusgallus^#gephebase-summary-title)		Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Gallusgallus^#gephebase-summary-title)	
chicken		chicken	
Gallus gallus domesticus; chicken; bantam; chickens		Gallus gallus domesticus; chicken; bantam; chickens	
species		species	
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)		Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030)	
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)	
No		Yes	
		Taxon B Description	
		Gallus gallus - Frizzle Feather	

GENOTYPIC CHANGE

Krt6a		Generic Gene Name		UniProtKB Mus musculus
K6A; MK6; MK6a; CK-6A; K6[a]; Krt2-; mK6[a]; Krt2-6; mK6[a]; Krt2-6a; Krt2-6c; 2310016L08Rik; Ker2; Krt6		Synonyms		GenebankID or UniProtKB
10090.ENSMUSP00000023788 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023788)		String		AEX59147.1 (https://www.ncbi.nlm.nih.gov/nuccore/AEX59147.1)
Belongs to the intermediate filament family.		Sequence Similarities		
-		GO - Molecular Function		
GO:0050830 : defense response to Gram-positive bacterium		GO - Biological Process		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0050830>)
GO:0002009 : morphogenesis of an epithelium
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002009>)
GO:0042060 : wound healing (<https://www.ebi.ac.uk/QuickGO/term/GO:0042060>)
GO:0045109 : intermediate filament organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045109>)
GO:0016055 : Wnt signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016055>)
GO:0061844 : antimicrobial humoral immune response mediated by antimicrobial peptide
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061844>)
GO:0031424 : keratinization (<https://www.ebi.ac.uk/QuickGO/term/GO:0031424>)
GO - Cellular Component
GO:0045095 : keratin filament (<https://www.ebi.ac.uk/QuickGO/term/GO:0045095>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	Molecular Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title)	Aberration Type
10-99 bp	Deletion Size
69bp deletion partially overlapping with exon 5	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	Experimental Evidence
The chicken frizzle feather is due to an Î±-keratin (KRT75) mutation that causes a defective rachis. (2012) (https://pubmed.ncbi.nlm.nih.gov/22829773)	Main Reference
Ng CS; Wu P; Foley J; Foley A; McDonald ML; Juan WT; Huang CJ; Lai YT; Lo WS; Chen CF; Leal SM; Zhang H; Widelitz RB; Patel PI; Li WH; Chuong CM	Authors

Abstract

Feathers have complex forms and are an excellent model to study the development and evolution of morphologies. Existing chicken feather mutants are especially useful for identifying genetic determinants of feather formation. This study focused on the gene F, underlying the frizzle feather trait that has a characteristic curled feather rachis and barbs in domestic chickens. Our developmental biology studies identified defects in feather medulla formation, and physical studies revealed that the frizzle feather curls in a stepwise manner. The frizzle gene is transmitted in an autosomal incomplete dominant mode. A whole-genome linkage scan of five pedigrees with 2678 SNPs revealed association of the frizzle locus with a keratin gene-enriched region within the linkage group E22C19W28...E50C23. Sequence analyses of the keratin gene cluster identified a 69 bp in-frame deletion in a conserved region of KRT75, an Î±-keratin gene. Retroviral-mediated expression of the mutated F cDNA in the wild-type rectrix qualitatively changed the bending of the rachis with some features of frizzle feathers including irregular kinks, severe bending near their distal ends, and substantially higher variations among samples in comparison to normal feathers. These results confirmed KRT75 as the F gene. This study demonstrates the potential of our approach for identifying genetic determinants of feather forms.

Additional References

RELATED GEPHE

14 (ABCA1, CDKN2A, CYP19A1, Endothelin receptor B2, MC1R, PMEL17, SLC45A2=MATP, SOX10, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1), FGF20, GDF7, HOXC8 - uncertain, KRT75L4) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9031~/and+Trait=Feathers/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000394/9031/> @Parallelism