

GEPHE SUMMARY

KRT71 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~KRT71~#gephebase-summary-title)	Gephebase Gene	GP00001730	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)	Trait Category		
Hair type (curly and hairless) (https://www.gephebase.org/search-criteria?/and+Trait=~Hairtype (curly and hairless)~#gephebase-summary-title)	Trait		
cattle - wild hair	Trait State in Taxon A		
a few of the Hereford cattle - curly hair or hairless	Trait State in Taxon B		
Taxon A	Ancestral State	Taxon B	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated~#gephebase-summary-title)	Taxonomic Status		
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Bos taurus~#gephebase-summary-title)	Latin Name	Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Bos taurus~#gephebase-summary-title)	Latin Name
cattle	Common Name	cattle	Common Name
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	Synonyms	Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos	Lineage
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)	Parent	Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)	Parent
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)	NCBI Taxonomy ID	9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
		Hereford cattle	Taxon B Description

GENOTYPIC CHANGE

Krt71	Generic Gene Name	Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)	UniProtKB Mus musculus
Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g	Synonyms	0	GenebankID or UniProtKB
10090.ENSMUSP00000023710 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710)	String		
Belongs to the intermediate filament family.	Sequence Similarities		
GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)	GO - Molecular Function		
GO:0031069 : hair follicle morphogenesis	GO - Biological Process		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031069>)
GO:0045109 : intermediate filament organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045109>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0045095 : keratin filament (<https://www.ebi.ac.uk/QuickGO/term/GO:0045095>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes~#gephebase-summary-title))

Molecular Type

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

Aberration Type

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

eight base pair deletion mutation in exon one of the keratin 71 (KRT71) c.334delTGTGCCCA p.Met93AsnfsX14

Experimental Evidence

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

Main Reference

The naked truth: Sphynx and Devon Rex cat breed mutations in KRT71. (2010) (<https://pubmed.ncbi.nlm.nih.gov/20953787>)

Authors

Gandolfi B; Outerbridge CA; Beresford LG; Myers JA; Pimentel M; Alhaddad H; Grahn JC; Grahn RA; Lyons LA

Abstract

Hair is a unique structure, characteristic of mammals, controlling body homeostasis, as well as cell and tissue integration. Previous studies in dog, mouse, and rat have identified polymorphisms in Keratin 71 (KRT71) as responsible for the curly/wavy phenotypes. The coding sequence and the 3' UTR of KRT71 were directly sequenced in randomly bred and pedigreed domestic cats with different pelage mutations, including hairless varieties. A SNP altering a splice site was identified in the Sphynx breed and suggested to be the hairless (hr) allele, and a complex sequence alteration, also causing a splice variation, was identified in the Devon Rex breed and suggested to be the curly (re) allele. The polymorphisms were genotyped in approximately 200 cats. All the Devon Rex were homozygous for the complex alterations and most of the Sphynx were either homozygous for the hr allele or compound heterozygotes with the Devon-associated re allele, suggesting that the phenotypes are a result of the identified SNPs. Two Sphynx carrying the proposed hr mutation did not carry the Devon-associated alteration. No other causative mutations for eight different rexoid and hairless cat phenotypes were identified. The allelic series KRT71(+) > KRT71(hr) > KRT71(re) is suggested.

Additional References

RELATED GEPHE

Related Genes

1 (KRT27) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9913~/and+Trait=Hair type/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9913~/and+Trait=Hair+type/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

In afflicted Herefords the hair coat can be very short; fine; kinky; curly; or appear frosted and the tail switch can be underdeveloped Reference Gandolfi et al. only mentions this case in one sentence. The actual main reference for this case is a poster presentation - see <https://portal.nifa.usda.gov/web/crisprojectpages/0213533-identification-of-the-causal-mutation-for-hypotrichosis-in-hereford-cattle.html> <https://omia.org/OMIA002114/9913/>