

## GEPHE SUMMARY

KRT71 ( <a href="https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=KRT71">#gephebase-summary-title)</a>	Gephebase Gene	GP00001730	GephelD
Published	Entry Status	Courtier	Main curator

## PHENOTYPIC CHANGE

Trait Category	
Morphology ( <a href="https://www.gephebase.org/search-criteria/?and+Trait+Category=Morphology">#gephebase-summary-title)</a>	Trait

Hair type (curly and hairless) ( <a 297="" 324"="" 47="" 487="" data-label="Table" href="https://www.gephebase.org/search-criteria/?and+Trait=Hair+type+(curly+and+hairless)#gephebase-summary-title)&lt;/a&gt;&lt;/td&gt;&lt;td&gt;Trait State in Taxon A&lt;/td&gt;&lt;/tr&gt; &lt;tr&gt; &lt;td&gt;cattle - wild hair&lt;/td&gt;&lt;td&gt;Trait State in Taxon B&lt;/td&gt;&lt;/tr&gt; &lt;/table&gt; &lt;/div&gt; &lt;div data-bbox="> <table> <tr> <td>a few of the Hereford cattle - curly hair or hairless</td><td>Ancestral State</td></tr> </table> </a>	a few of the Hereford cattle - curly hair or hairless	Ancestral State
a few of the Hereford cattle - curly hair or hairless	Ancestral State	

Taxon A		Taxonomic Status	
Bos taurus	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>	Bos taurus	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>

Taxon A		Latin Name	
Bos taurus	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>	Bos taurus	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>

Taxon A		Common Name	
Bos taurus	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>	cattle	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>

Taxon A		Synonyms	
Bos taurus	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>	Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=Bos+taurus">#gephebase-summary-title)</a>

Taxon A		Rank	
Bos	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903</a> )	species	Rank

Taxon A		Lineage	
Bos	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903</a> )	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	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

Taxon A		Parent	
Bos (oxen, cattle) - (Rank: genus)	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903</a> )	Bos (oxen, cattle) - (Rank: genus)	Parent

Taxon A		NCBI Taxonomy ID	
Bos	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903</a> )	9903	NCBI Taxonomy ID

Taxon A		is Taxon A an Infraspecies?	
No		Yes	is Taxon B an Infraspecies?

Taxon A		Taxon B	
		Hereford cattle	Taxon B Description

## GENOTYPIC CHANGE

Generic Gene Name	
Krt71	Q9RoH5 ( <a href="http://www.uniprot.org/uniprot/Q9RoH5">http://www.uniprot.org/uniprot/Q9RoH5</a> )

Synonyms	
Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g	0

String	
10090.ENSMUSP00000023710 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710</a> )	UniProtKB Mus musculus

Sequence Similarities	
Belongs to the intermediate filament family.	GenebankID or UniProtKB

GO - Molecular Function	
GO:0005198 : structural molecule activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005198">https://www.ebi.ac.uk/QuickGO/term/GO:0005198</a> )	GenebankID or UniProtKB

GO - Biological Process	
GO:0031069 : hair follicle morphogenesis	UniProtKB Mus musculus

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

Molecular Type

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

Aberration Type

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

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

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

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/>