GEPHE SUMMARY

Gephebase Gene

KRT71 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^KRT71^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001730

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Hair type (curly and hairless) (https://www.gephebase.org/search-criteria?/and+Trait=^Hair type (curly and hairless)^#gephebase-summary-title)

Trait State in Taxon A

cattle - wild hair

Trait State in Taxon B

a few of the Hereford cattle - curly hair or hairless

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)

Common Name

cattle

Synonyms

Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

Rank

species

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

Parent

Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID

9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)

Common Name

cattle

Synonyms

Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

Rank

species

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

Parent

Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID

9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Hereford cattle

GENOTYPIC CHANGE

Generic Gene Name

Krt71

Synonyms

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

String

10090.ENSMUSP00000023710 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710)

Sequence Similarities

Belongs to the intermediate filament family.

GO - Molecular Function

GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)

GO - Biological Process

GO:0031069 : hair follicle morphogenesis

UniProtKB Mus musculus

Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)

GenebankID or UniProtKB

()

(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)

Molecular Type

Coding (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)

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)

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)

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/