GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00000525

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

Felis catus

Trait State in Taxon B

Felis catus - Devon Rex

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

domestic cat

Synonyms

Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.

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; Carnivora; Feliformia; Felidae; Felinae; Felis

Parent

Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)

NCBI Taxonomy ID

9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

domestic cat

Synonyms

Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.

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; Carnivora; Feliformia; Felidae; Felinae; Felis

Parent

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NCBI Taxonomy ID

9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Felis catus - Devon Rex and hairless (Sphynx and Kohana breeds) - probable allelic series is KRT71+ > KRT71hr > KRT71re

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

UniProtKB Mus musculus

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

GenebankID or UniProtKB

ADM23868 (https://www.ncbi.nlm.nih.gov/nuccore/ADM23868)

GO:0031069 : hair follicle morphogenesis (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)

Mutation #1

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

SNP Coding Change

-

Molecular Details of the Mutation

c.816+1G>A at position 1 of intron 4 - disrupts the highly conserved â€œGâ€ needed for the 5â€² splice site recognition @Splicing

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

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

Mutation #2

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Indel Size

10-99 bp

Molecular Details of the Mutation

81-bp deletion plus two insertions of 8 and 1 bp starting at the 5â€² end of intron 6 and involving exon 7

Experimental Evidence

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

Main Reference

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^KRT71^/and+Taxon ID=^9685^/or+Gene Gephebase=^KRT71^/and+Taxon ID=^9685^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Splicing @AllelicSeries https://omia.org/OMIA001581/9685/