

GEPHE SUMMARY

<div>KRT71 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~KRT71~#gephebase-summary-title)</div> <div>Published</div>	Gephebase Gene	GP00000525	GepheID
	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

		Trait Category	
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~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+TaxonomicStatus=~Domesticated~#gephebase-summary-title)			
Taxon A		Taxon B	
Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Felis catus~#gephebase-summary-title)		Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Felis catus~#gephebase-summary-title)	
Common Name		Common Name	
domestic cat		domestic cat	
Synonyms		Synonyms	
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.		Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	
Rank		Rank	
species		species	
Lineage		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		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		Parent	
Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)		Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)		9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		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		Q9RoH5 (http://www.uniprot.org/uniprot/Q9RoH5)	
		Synonyms	
Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g		GenebankID or UniProtKB	
		ADM23868 (https://www.ncbi.nlm.nih.gov/nuccore/ADM23868)	
		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
(<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 $\hat{\alpha}$ G $\hat{\alpha}$ needed for the 5 $\hat{\alpha}$ 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 5 $\hat{\alpha}$ 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	(<a #gephebase-summary-title"="" +krt71+"="" and+taxon+id="+9685" href="https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=" or+gene+gephebase="+KRT71">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/>