

## GEPHE SUMMARY

KRT71 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+KRT71+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+KRT71+Gephebase-summary-title</a> )	Gephebase Gene	GP00002232	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+Gephebase-summary-title</a> )	Trait Category		
Hair type (curly) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Hair+type+curly+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Hair+type+curly+Gephebase-summary-title</a> )	Trait		
WT hair	Trait State in Taxon A		
Curly coat in Selkirk rex	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+Gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Felis catus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+Gephebase-summary-title</a> )	Latin Name	Felis catus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+Gephebase-summary-title</a> )	Latin Name
domestic cat	Common Name	domestic cat	Common Name
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	Synonyms	Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	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; Carnivora; Feliformia; Felidae; Felinae; Felis	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	Lineage
Felis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682</a> )	Parent	Felis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682</a> )	Parent
9685 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685</a> )	NCBI Taxonomy ID	9685 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

Krt71	Generic Gene Name	Q9R0H5 ( <a href="http://www.uniprot.org/uniprot/Q9R0H5">http://www.uniprot.org/uniprot/Q9R0H5</a> )	UniProtKB Mus musculus
Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g	Synonyms	()	GenebankID or UniProtKB
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> )	String		
Belongs to the intermediate filament family.	Sequence Similarities		
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> )	GO - Molecular Function		
GO:0031069 : hair follicle morphogenesis ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031069">https://www.ebi.ac.uk/QuickGO/term/GO:0031069</a> )	GO - Biological Process		
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

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

SNP Coding Change

-

Molecular Details of the Mutation

c.445-1G>C in splice site likely disrupts the highly conserved acceptor splicing site of intron one. Sequence of the complete RNA transcript revealed that an alternative downstream acceptor was employed

Experimental Evidence

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

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

Main Reference

A splice variant in KRT71 is associated with curly coat phenotype of Selkirk Rex cats. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23770706>)

Authors

Gandolfi B; Alhaddad H; Joslin SE; Khan R; Filler S; Brem G; Lyons LA

Abstract

One of the salient features of the domestic cat is the aesthetics of its fur. The Selkirk Rex breed is defined by an autosomal dominant woolly rexoid hair (ADWH) abnormality that is characterized by tightly curled hair shafts. A genome-wide case - control association study was conducted using 9 curly coated Selkirk Rex and 29 controls, including straight-coated Selkirk Rex, British Shorthair and Persian, to localize the Selkirk autosomal dominant rexoid locus (SADRE). Although the control cats were from different breed lineages, they share recent breeding histories and were validated as controls by Bayesian clustering, multi-dimensional scaling and genomic inflation. A significant association was found on cat chromosome B4 (Praw = 2.87 Å–10(-11)), and a unique haplotype spanning ~600 Kb was found in all the curly coated cats. Direct sequencing of four candidate genes revealed a splice site variant within the KRT71 gene associated with the hair abnormality in Selkirk Rex.

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](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

@AllelicSeries @Splicing <https://omia.org/OMIA001712/9685/>