

GEPHE SUMMARY

	Gephebase Gene	GephelD
FGF5 (https://www.gephebase.org/search-criteria/?and+Gene Gephebase="^FGF5">#gephebase-summary-title)	GP00002180	
	Entry Status	Main curator
Published	Martin	

PHENOTYPIC CHANGE

	Trait Category	
Morphology (https://www.gephebase.org/search-criteria/?and+Trait Category="Morphology">#gephebase-summary-title)		Trait
Hair length (https://www.gephebase.org/search-criteria/?and+Trait=^Hair length:#gephebase-summary-title)	Trait State in Taxon A	
Wild llama (Lama guanicoe) with WT hair and short fleece	Trait State in Taxon B	
Domesticated llama and alpacas with long (angora) hair ; introgression from llamas to alpacas	Ancestral State	
Taxon A		Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic Status="Domesticated">#gephebase-summary-title)		

Taxon A	Latin Name	
Lama guanicoe (#gephebase-summary-title")		
guanaco	Common Name	
Lama guanicoe guanaco; guanaco	Synonyms	
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; Artiodactyla; Tylopoda; Camelidae; Lama	Lineage	
Lama () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9839)	Parent	
9840 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9840)	NCBI Taxonomy ID	
No	is Taxon A an Infraspecies?	

Taxon B #1	Latin Name	
Lama glama (#gephebase-summary-title")		
llama	Common Name	
Camelus glama; Lama guanicoe glama; llama; Lama glama (Linnaeus, 1758); Llama glama	Synonyms	
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; Artiodactyla; Tylopoda; Camelidae; Lama	Lineage	
Lama () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9839)	Parent	
9844 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9844)	NCBI Taxonomy ID	
No	is Taxon B an Infraspecies?	

Taxon B #2	Latin Name	
Vicugna pacos (#gephebase-summary-title")		
alpaca	Common Name	
Lama guanicoe pacos; Lama pacos; alpaca	Synonyms	
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; Artiodactyla; Tylopoda; Camelidae; Vicugna	Lineage	
Vicugna () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30539)	Parent	

30538

<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30538>

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Fgf5	Generic Gene Name	P15656 (http://www.uniprot.org/uniprot/P15656)	UniProtKB Mus musculus
go; Fgf-5; HBGF-5; angora	Synonyms	ABB87177 (https://www.ncbi.nlm.nih.gov/nuccore/ABB87177)	GenebankID or UniProtKB
10090.ENSMSUPO0000031280 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMSUPO0000031280)	String		
	Sequence Similarities		
Belongs to the heparin-binding growth factors family.	GO - Molecular Function		
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)			
GO:0005104 : fibroblast growth factor receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005104)			
	GO - Biological Process		
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)			
GO:0008284 : positive regulation of cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008284)			
GO:0051781 : positive regulation of cell division (https://www.ebi.ac.uk/QuickGO/term/GO:0051781)			
GO:0008543 : fibroblast growth factor receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0008543)			
GO:0010001 : glial cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0010001)			
GO:0023019 : signal transduction involved in regulation of gene expression (https://www.ebi.ac.uk/QuickGO/term/GO:0023019)			
	GO - Cellular Component		
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)			Presumptive Null
Yes (https://www.gephebase.org/search-criteria/?and+Presumptive Null=%27Yes%23gephebase-summary-title)			Molecular Type
Coding (https://www.gephebase.org/search-criteria/?and+Molecular Type=%27Coding%23gephebase-summary-title)			Aberration Type
SNP (https://www.gephebase.org/search-criteria/?and+Aberration Type=%27SNP%23gephebase-summary-title)			SNP Coding Change
Nonsense			Molecular Details of the Mutation
transition C>T at position 499 downstream of the ATG codon with transcriptional readthrough ; difference in post-transcriptional readthrough may underlie the different fleece types of the alpaca suri and huacaya breeds ; derived allele was introgressed from lamas to alpacas			Experimental Evidence
Candidate Gene (https://www.gephebase.org/search-criteria/?and+Experimental Evidence=%27Candidate Gene%23gephebase-summary-title)			

Taxon A	Taxon B	Position
Codon	TGA	167
Amino-acid	STP	167

Main Reference
Molecular characterization of the llama FGF5 gene and identification of putative loss of function mutations. (2017) (https://pubmed.ncbi.nlm.nih.gov/29024003)
Authors

Daverio MS; Vidal-Rioja L; Frank EN; Di Rocco F

Abstract

Llama, the most numerous domestic camelid in Argentina, has good fiber-production ability. Although a few genes related to other productive traits have been characterized, the molecular genetic basis of fiber growth control in camelids is still poorly understood. Fibroblast growth factor 5 (FGF5) is a secreted signaling protein that controls hair growth in humans and other mammals. Mutations in the FGF5 gene have been associated with long-hair phenotypes in several species. Here, we sequenced the llama FGF5 gene, which consists of three exons encoding 813 Å bp. cDNA analysis from hair follicles revealed the expression of two FGF5 alternative spliced transcripts, in one of which exon 2 is absent. DNA variation analysis showed four polymorphisms in the coding region: a synonymous SNP (c.210A>G), a single base deletion (c.348delA), a 12-bp insertion (c.351_352insCATATAACATAG) and a non-sense mutation (c.499C>T). The deletion was always found together with the insertion forming a haplotype and producing a putative truncated protein of 123 amino acids. The c.499C>T mutation also leads to a premature stop codon at position 168. In both cases, critical functional domains of FGF5, including one heparin binding site, are lost. All animals analyzed were homozygous for one of the deleterious mutations or compound heterozygous for both (i.e. c.348delA, c.351_352insCATATAACATAG/c.499T). Sequencing of guanaco samples showed that the FGF5 gene encodes a full-length 270-amino acid protein. These results suggest that FGF5 is likely functional in short-haired wild species and non-functional in the domestic fiber-producing species, the llama.

RELATED GEPHE

No matches found.

Related Genes

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=%FGF5%/and+Taxon ID=%9840%/or+Gene Gephebase=%FGF5%/and+Taxon ID=%9844%/or+Gene Gephebase=%FGF5%/and+Taxon ID=%30538%#gephebase-summary-title>)

Related Haplotypes

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000439/9844/> @AllelicSeries @Parallelism @Introgression