

GEPHE SUMMARY

	Gephebase Gene	GephelD
FGF5 (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=^FGF5^#gephebase-summary-title)	GP00002179	Main curator
Published	Entry Status	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 (<i>Lama guanicoe</i>) with WT hair and short fleece	Trait State in Taxon B		
Domesticated llama (<i>Llama glama</i>) with long (angora) hair	Ancestral State		
Taxon A		Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic Status=^Domesticated^#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Lama guanicoe (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Lama+guanicoe^#gephebase-summary-title)		Lama glama (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Lama+glama^#gephebase-summary-title)	
guanaco	Common Name		Common Name
Lama guanicoe guanaco; guanaco	Synonyms		Synonyms
species	Rank	Camelus glama; Lama guanicoe glama; llama; Lama glama (Linnaeus, 1758); Llama glama	Rank
	Lineage	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; Tylopoda; Camelidae; Lama		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	
Lama () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9839)	Parent	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	9844 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9844)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
	No		

GENOTYPIC CHANGE

Fgf5	Generic Gene Name	UniProtKB Mus musculus
go: Fgf-5; HBGF-5; angora	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000031280 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031280)	String	ABB87177 (https://www.ncbi.nlm.nih.gov/nuccore/ABB87177)
Belongs to the heparin-binding growth factors family.	Sequence Similarities	
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 - Molecular Function	
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 - Biological Process	

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 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^Yes))

Molecular Type

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

Aberration Type

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

Indel Size

10-99 bp

Molecular Details of the Mutation

a single base deletion (c.348delA) + a 12-bp insertion (c.351_352insCATATAACATAG)

Experimental Evidence

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

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

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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.

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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=^FGF5^/and+Taxon+ID=^9840^/or+Gene+Gephebase=^FGF5^/and+Taxon+ID=^9844^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

[@AllelicSeries @Parallelism](https://omia.org/OMIA000439/9844)