

GEPHE SUMMARY

FGF5 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~FGF5~#gephebase-summary-title)	Gephebase Gene	GP00002179	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)		Trait Category	
Hair length (https://www.gephebase.org/search-criteria?/and+Trait=~Hairlength~#gephebase-summary-title)		Trait	
Wild llama (Lama guanicoe) with WT hair and short fleece		Trait State in Taxon A	
Domesticated llama (Llama glama) with long (angora) hair		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
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)	
Common Name		Common Name	
guanaco		llama	
Synonyms		Synonyms	
Lama guanicoe guanaco; guanaco		Camelus glama; Lama guanicoe glama; llama; Lama glama (Linnaeus, 1758); Llama glama	
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; 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	
Parent		Parent	
Lama () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9839)		Lama () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9839)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9840 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9840)		9844 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9844)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

Fgf5		Generic Gene Name	
go; Fgf-5; HBGF-5; angora		P15656 (http://www.uniprot.org/uniprot/P15656)	
String		GenebankID or UniProtKB	
10090.ENSMUSP00000031280 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031280)		ABB87177 (https://www.ncbi.nlm.nih.gov/nucleotide/ABB87177)	
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=~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

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

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 ([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))

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 813Å 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](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

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