

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

	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	
	Trait
Hair length (https://www.gephebase.org/search-criteria?/and+Trait=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 lamas to alpacas	
	Ancestral State
Taxon A	
	Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Domesticated^#gephebase-summary-title)	

Taxon B #2	
Vicugna pacos (<a +vicugna+pacos+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Vicugna+pacos+"#gephebase-summary-title)	Latin Name
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

NCBI Taxonomy ID

30538
([https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30538](https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30538))
is Taxon B an Intraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

UniProtKB Mus musculus

Fgf5

P15656 (<http://www.uniprot.org/uniprot/P15656>)

Synonyms

GenebankID or UniProtKB

go; Fgf-5; HBGF-5; angora

ABB87177 (<https://www.ncbi.nlm.nih.gov/nucore/ABB87177>)

String

10090.ENSMUSP00000031280
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031280)

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

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

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 llamas to alpacas

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	CGA	TGA	167
Amino-acid	Arg	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 813Â 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

Evidence of post-transcriptional readthrough regulation in FGF5 gene of alpaca. (2018) (<https://pubmed.ncbi.nlm.nih.gov/29307854>)

RELATED GEPHE

No matches found.

Related Genes

Related Haplotypes

1 ([#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^"/or+Gene Gephebase="FGF5^"/and+Taxon ID="30538^">#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=))

EXTERNAL LINKS

COMMENTS

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