

GEPHE SUMMARY

	Gephebase Gene		GepheID
FGF5 (<a +fgf5+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?and+Gene=Gephebase=">https://www.gephebase.org/search-criteria?and+Gene=Gephebase="+FGF5+"#gephebase-summary-title)		GP00001370	
	Entry Status	Prigent	Main curator
Published			

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)		Trait Category	
Hair length (https://www.gephebase.org/search-criteria?/and+Trait=~Hair+length~#gephebase-summary-title)		Trait	
donkey ; short coat (in Berry Black and Pyrenean and Provence and Normand and Cotentin and Bourbonnais)		Trait State in Taxon A	
Baudet du Poitou ; long coat		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Equus asinus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+asinus~#gephebase-summary-title)		Equus asinus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+asinus~#gephebase-summary-title)	
Common Name		Common Name	
ass		ass	
Synonyms		Synonyms	
ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey		ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey	
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; Perissodactyla; Equidae; Equus; Asinus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Perissodactyla; Equidae; Equus; Asinus	
Parent		Parent	
Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)		Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)		9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
Yes		Yes	
Taxon A Description		Taxon B Description	
donkey ; short coat (in Berry Black and Pyrenean and Provence and Normand and Cotentin and Bourbonnais)		Baudet du Poitou ; long coat	

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	KJ725178 (https://www.ncbi.nlm.nih.gov/nucore/KJ725178)
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 ([#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

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

c.245G>A p.W82* nonsense mutation leading to stop codon at position 82

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Trp	STP	82

Main Reference

Two recessive mutations in FGF5 are associated with the long-hair phenotype in donkeys. (2014) (<https://pubmed.ncbi.nlm.nih.gov/25927731>)

Authors

Legrand R; Tiret L; Abitbol M

Abstract

Seven donkey breeds are recognized by the French studbook. Individuals from the Pyrenean, Provence, Berry Black, Normand, Cotentin and Bourbonnais breeds are characterized by a short coat, while those from the Poitou breed (Baudet du Poitou) are characterized by a long-hair phenotype. We hypothesized that loss-of-function mutations in the FGF5 (fibroblast growth factor 5) gene, which are associated with a long-hair phenotype in several mammalian species, may account for the special coat feature of Poitou donkeys. To the best of our knowledge, mutations in FGF5 have never been described in Equidae.

We sequenced the FGF5 gene from 35 long-haired Poitou donkeys, as well as from a panel of 67 short-haired donkeys from the six other French breeds and 131 short-haired ponies and horses.

We identified a recessive c.433_434delAT frameshift deletion in FGF5, present in Poitou and three other donkey breeds and a recessive nonsense c.245G>A substitution, present in Poitou and four other donkey breeds. The frameshift deletion was associated with the long-hair phenotype in Poitou donkeys when present in two copies (nâ€™=â€™31) or combined with the nonsense mutation (nâ€™=â€™4). The frameshift deletion led to a stop codon at position 159 whereas the nonsense mutation led to a stop codon at position 82 in the FGF5 protein. In silico, the two truncated FGF5 proteins were predicted to lack the critical Î² strands involved in the interaction between FGF5 and its receptor, a mandatory step to inhibit hair growth.

Our results highlight the allelic heterogeneity of the long-hair phenotype in donkeys and enlarge the panel of recessive FGF5 loss-of-function alleles described in mammals. Thanks to the DNA test developed in this study, breeders of non-Poitou breeds will have the opportunity to identify long-hair carriers in their breeding stocks.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~FGF5^/and+Taxon ID=~9793^/or+Gene Gephebase=~FGF5^/and+Taxon ID=~9793))

EXTERNAL LINKS

COMMENTS

Recessive ; @AllelicSeries ;protein is predicted to lack the critical beta strands involved in the interaction with its receptor ; only found as heterozygous but involved in the long haired phenotype when combined with a c.433_434delAT allele ; possibly originate from @Introgression from a long-haired Spanish breed ; <https://omia.org/OMIA000439/9793/>

