

## GEPHE SUMMARY

<p>FGF5 (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~FGF5~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~FGF5~#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001369</p> <p>Prigent</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title</a>)</p> <p>Hair length (<a href="https://www.gephebase.org/search-criteria?/and+Trait=~Hair+length~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Hair+length~#gephebase-summary-title</a>)</p> <p>donkey ; short coat (in Berry Black and Pyrenean and Provence and Normand and Cotentin and Bourbonnais)</p> <p>Baudet du Poitou ; long coat</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Equus asinus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+asinus~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+asinus~#gephebase-summary-title</a>)</p> <p>ass</p> <p>ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey</p> <p>species</p> <p>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</p> <p>Asinus () - (Rank: subgenus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508</a>)</p> <p>9793 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>Yes</p> <p>donkey ; short coat (in Berry Black and Pyrenean and Provence and Normand and Cotentin and Bourbonnais)</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p> <p>Taxon A Description</p>	<p>Equus asinus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+asinus~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+asinus~#gephebase-summary-title</a>)</p> <p>ass</p> <p>ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey</p> <p>species</p> <p>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</p> <p>Asinus () - (Rank: subgenus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508</a>)</p> <p>9793 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Baudet du Poitou ; long coat</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>
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## GENOTYPIC CHANGE

<p>Fgf5</p> <p>go; Fgf-5; HBGF-5; angora</p> <p>10090.ENSMUSP00000031280 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031280">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031280</a>)</p> <p>Belongs to the heparin-binding growth factors family.</p> <p>GO:0008083 : growth factor activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008083">https://www.ebi.ac.uk/QuickGO/term/GO:0008083</a>)</p> <p>GO:0005104 : fibroblast growth factor receptor binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005104">https://www.ebi.ac.uk/QuickGO/term/GO:0005104</a>)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P15656 (<a href="http://www.uniprot.org/uniprot/P15656">http://www.uniprot.org/uniprot/P15656</a>)</p> <p>KJ725177 (<a href="https://www.ncbi.nlm.nih.gov/nucleotide/KJ725177">https://www.ncbi.nlm.nih.gov/nucleotide/KJ725177</a>)</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
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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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

c.433\_434delAT frameshift deletion leading to a stop codon at position 159

Experimental Evidence

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

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; Turet 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 (n=31) or combined with the nonsense mutation (n=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  $\beta^2$  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+Taxon+ID+9793+Gene+Gephebase+FGF5+Taxon+ID+9793))

EXTERNAL LINKS

COMMENTS

Recessive ;@AllelicSeries; protein is predicted to lack the critical beta strands involved in the interaction with its receptor ; <https://omia.org/OMIA000439/9793/>

