

GEPHE SUMMARY

	Gephebase Gene	GephelD
FGF5 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^FGF5^#gephebase-summary-title)	GP00002442	Main curator
Published	Entry Status	Courtier

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
normal hair length	Trait State in Taxon B
abnormally long hair - moja recessive mutation	Ancestral State
Taxon A	Taxonomic Status

Taxon A	Latin Name	Taxon B	Latin Name
Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Mus+musculus^#gephebase-summary-title)	Common Name	Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Mus+musculus^#gephebase-summary-title)	Common Name
house mouse	Synonyms	house mouse	Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	Rank	house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	Rank
species	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; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus	Parent	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus	Parent
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)	NCBI Taxonomy ID	Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)	NCBI Taxonomy ID
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)	is Taxon A an Infraspecies?	10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)	is Taxon B an Infraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Fgf5	Synonyms	GenebankID or UniProtKB
go: Fgf-5; HBGF-5; angora	String	0
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

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

Deletion Size

1-10 kb

Molecular Details of the Mutation

Deletion of a 9.3-kb region in the Fgf5 gene including exon 3 and its 5' and 3' flanking sequences. The genomic deletion site also shows insertion of a 498-bp early transposon element long terminal repeat.

Experimental Evidence

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

Main Reference

Retrotransposon-mediated Fgf5(go-Utr) mutant mice with long pelage hair. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21512271>)

Authors

Mizuno S; Iijima S; Okano T; Kajiwara N; Kunita S; Sugiyama F; Yagami K

Abstract

We found 6 spontaneous mutant mice with long pelage hair in our ICR breeding colony. The abnormal trait was restricted to long hair in these mice, which we named moja. They were fertile and showed the same growth and behavior as wild-type mice. To investigate the manner of the genetic inheritance of the moja allele, offspring were bred by mating the moja mice; all offspring had long pelage hair. Furthermore, we performed a reciprocal cross between moja mice and wild-type ICR mice with normal hair. All offspring exhibited normal hair suggesting an autosomal recessive inheritance of the trait. The moja/moja hair phenotype was maintained in skin grafted onto nude mice, suggesting that circulating or diffusible humoral factors regulating the hair cycle are not involved in the abnormal trait. The phenotype of moja/moja mice is similar to that of Fgf5-deficient mice. Therefore, we examined the expression of Fgf5 by RT-PCR in moja/moja mice. As expected, no Fgf5 expression was found in moja/moja mouse skin. PCR and DNA sequence analyses were performed to investigate the structure of the Fgf5 gene. We found a deletion of a 9.3-kb region in the Fgf5 gene including exon 3 and its 5' and 3' flanking sequences. Interestingly, the genomic deletion site showed insertion of a 498-bp early transposon element long terminal repeat. Taken together, these results suggest that the long hair mutation of moja/moja mice is caused by disruption of Fgf5 mediated by insertion of a retrotransposon.

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=^10090/or+Gene+Gephebase=^FGF5/and+Taxon+ID=^10090))

EXTERNAL LINKS

COMMENTS

@TE - These were spontaneous mutant mice with long pelage hair in a breeding colony.