

GEPHE SUMMARY

FGF5 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~FGF5~#gephebase-summary-title)	Gephebase Gene	GP00002176	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	
WT hair		Trait State in Taxon A	
recessive long hair in some Afghan Hound and Eurasier dogs		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	
Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canislupus~#gephebase-summary-title)		Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canislupusfamiliaris~#gephebase-summary-title)	
gray wolf		dog	
gray wolf; grey wolf; Canis lupus Linnaeus, 1758		Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	
species		subspecies	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus	
Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611)		Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)	
9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)		9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

Fgf5		P15656 (http://www.uniprot.org/uniprot/P15656)	
go; Fgf-5; HBGF-5; angora		ABB87177 (https://www.ncbi.nlm.nih.gov/nuccore/ABB87177)	
10090.ENSMUSP00000031280 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031280)			
Belongs to the heparin-binding growth factors family.			
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: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

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title>)

Molecular Type

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title>)

Aberration Type

Insertion (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~Insertion~#gephebase-summary-title>)

Insertion Size

1-9 bp

Molecular Details of the Mutation

c.559_560dupGG p.R188Afs*73

Experimental Evidence

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

Main Reference

Allelic heterogeneity of FGF5 mutations causes the long-hair phenotype in dogs. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23384345>)

Authors

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Abstract

Hitherto, the only known mutant gene leading to the long-hair phenotype in mammals is the fibroblast growth factor 5 (FGF5). In many dog breeds, the previously discovered FGF5:p.Cys95Phe mutation appeared completely concordant with the long-hair phenotype, but for some breeds, the long-hair phenotype could not be resolved. First, we studied the role of the FGF5:p.Cys95Phe and FGF5:g.145_150dupACCAGC mutations in 268 dogs descending from 27 breeds and seven wolves. As these mutations did not explain all the long-hair phenotypes, all exons and their neighbouring regions of FGF5 were re-sequenced. We detected three novel mutations in the coding sequence and one novel non-coding splice-site mutation in FGF5 associated with the long-hair phenotype. The FGF5:p.Ala193Val polymorphism was perfectly consistent with long hair in Akitas and probably in Siberian huskies, too. Dogs of the long-hair breed Samoyed were either homozygous or compound heterozygous for the FGF5:p.Ala193Val or the FGF5:p.Cys95Phe polymorphisms respectively. The two newly detected polymorphisms FGF5:c.559_560dupGG and FGF5:g.8193T>A and the known mutation FGF5:p.Cys95Phe explained the long-hair phenotype of all Afghan hounds analysed. An FGF5:c.556_571del16 mutation was found in one longhaired Eurasier. All long-hair-associated mutations follow a recessive mode of inheritance, and allelic heterogeneity was a common finding in breeds other than Akita.

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Additional References

Coat variation in the domestic dog is governed by variants in three genes. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19713490>)

RELATED GEPHE

Related Genes

1 (R-spondin-2 (RSPO2)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9612~/and+Trait=Hair length/or+Taxon ID=~9615~/and+Trait=Hair length/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

5 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~FGF5~/and+Taxon ID=~9612~/or+Gene Gephebase=~FGF5~/and+Taxon ID=~9615~#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

Allelic Heterogeneity @AllelicSeries <https://omia.org/OMIA000439/9615/>