GEPHE SUMMARY

Gephebase Gene

FGF5 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^FGF5^#gephebase-summary-title)

GepheID

GP00002174

Entry Status

Published

Main curator

Martin

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

WT hair

Trait State in Taxon B

recessive long hair in Akita ;Samoyed ; Siberian Huskie

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus^#gephebase-summary-title)

Common Name

gray wolf

Synonyms

gray wolf; grey wolf; Canis lupus Linnaeus, 1758

Rank

species

Lineage

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

Parent

Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611)

NCBI Taxonomy ID

9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title)

Common Name

dog

Synonyms

Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank

subspecies

Lineage

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

Parent

Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

NCBI Taxonomy ID

9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Fgf5

Synonyms

go; Fgf-5; HBGF-5; angora

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

UniProtKB Mus musculus

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

GenebankID or UniProtKB

ABB87177 (https://www.ncbi.nlm.nih.gov/nuccore/ABB87177)

(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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

g.4528639C>T c.578C>T p.A193V

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ala	Val	193

Main Reference

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

Authors

Dierks C; MÃ¶mke S; Philipp U; Distl O

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/