

GEPHE SUMMARY

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|--|----------------|--------------|
| | Gephebase Gene | GephelD |
| FGF5 (#gephebase-summary-title) | GP00002177 | Main curator |
| Published | Entry Status | Martin |
| | | |

PHENOTYPIC CHANGE

| | Trait Category | | |
|---|---|---------|-----------------------------|
| Morphology (#gephebase-summary-title) | Trait | | |
| Hair length (#gephebase-summary-title) | Trait State in Taxon A | | |
| WT hair | Trait State in Taxon B | | |
| recessive long hair in one Eurasier dog | Ancestral State | | |
| Taxon A | Taxonomic Status | | |
| Domesticated (#gephebase-summary-title) | | | |
| Taxon A | Latin Name | Taxon B | Latin Name |
| Canis lupus (#gephebase-summary-title) | Canis lupus familiaris (#gephebase-summary-title) | | |
| gray wolf | Common Name | | Common Name |
| gray wolf; grey wolf; Canis lupus Linnaeus, 1758 | Synonyms | | Synonyms |
| species | Rank | | Rank |
| 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 | Lineage | | Lineage |
| Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611) | Parent | | Parent |
| 9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612) | NCBI Taxonomy ID | | NCBI Taxonomy ID |
| No | is Taxon A an Infraspecies? | | is Taxon B an Infraspecies? |
| | No | | |

GENOTYPIC CHANGE

| | | |
|---|-------------------------|--|
| Fgf5 | Generic Gene Name | UniProtKB Mus musculus |
| go: Fgf-5; HBGF-5; angora | Synonyms | GenebankID or UniProtKB |
| 10090.ENSMUSP00000031280 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031280) | String | ABB87177 (https://www.ncbi.nlm.nih.gov/nuccore/ABB87177) |
| Belongs to the heparin-binding growth factors family. | Sequence Similarities | |
| GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083) | GO - Molecular Function | |
| 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 |
| No (#gephebase-summary-title) | Molecular Type |
| Coding (#gephebase-summary-title) | Aberration Type |
| Deletion (#gephebase-summary-title) | Deletion Size |
| 1-9 bp | Molecular Details of the Mutation |
| c.556_571del16 p.A186Tfs*69 | 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 |
| Dierks C; MÄ¶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. | |
| Â© 2013 The Authors, Animal Genetics Â© 2013 Stichting International Foundation for Animal Genetics. | 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

| | |
|--|--------------------|
| 1 (R-spondin-2 (RSPO2)) (#gephebase-summary-title) | Related Genes |
| 5 (#gephebase-summary-title) | Related Haplotypes |

EXTERNAL LINKS

COMMENTS

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