

GEPHE SUMMARY

	Gephebase Gene	GephelD
FGF5 (https://www.gephebase.org/search-criteria/?and+Gene Gephebase="#gephebase-summary-title")	GP00000310	
	Entry Status	Main curator
Published	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
various long-haired breeds including Afghan Hound; Border Collie; Cocker Spaniel; Collie; Corgi; Dachshund; German Shepherd Dog; Golden Retriever; Pomeranian; Samoyed	Ancestral State
Taxon A	Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic Status="Domesticated">#gephebase-summary-title")	

Taxon A		Taxon B	
	Latin Name		Latin Name
Canis lupus (#gephebase-summary-title")	Common Name	Canis lupus familiaris (#gephebase-summary-title")	Common Name
gray wolf	Synonyms	dog	Synonyms
gray wolf; grey wolf; Canis lupus Linnaeus, 1758	Rank	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Rank
species	Lineage	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	Parent	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 () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611)	NCBI Taxonomy ID	Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)	NCBI Taxonomy ID
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)	
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Fgf5	P15656 (http://www.uniprot.org/uniprot/P15656)	
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:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)	GO - Biological Process	

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Cys95Phe

Experimental Evidence

Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=^Association+Mapping^#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	Phe	95

Main Reference

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

Authors

Cadieu E; Neff MW; Quignon P; Walsh K; Chase K; Parker HG; Vonholdt BM; Rhue A; Boyko A; Byers A; Wong A; Mosher DS; Elkahloun AG; Spady TC; AndrÃ© C; Lark KG; Cargill M; Bustamante CD; Wayne RK; Ostrander EA

Abstract

Coat color and type are essential characteristics of domestic dog breeds. Although the genetic basis of coat color has been well characterized, relatively little is known about the genes influencing coat growth pattern, length, and curl. We performed genome-wide association studies of more than 1000 dogs from 80 domestic breeds to identify genes associated with canine fur phenotypes. Taking advantage of both inter- and intrabreed variability, we identified distinct mutations in three genes, RSPO2, FGF5, and KRT71 (encoding R-spondin-2, fibroblast growth factor-5, and keratin-71, respectively), that together account for most coat phenotypes in purebred dogs in the United States. Thus, an array of varied and seemingly complex phenotypes can be reduced to the combinatorial effects of only a few genes.

Additional References

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/>

