

GEPHE SUMMARY

KRT71 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~KRT71^#gephebase-summary-title) Published	Gephebase Gene	GP00000524	GepheID
	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Hair type (curly) (https://www.gephebase.org/search-criteria?/and+Trait=~Hair type (curly)^#gephebase-summary-title)		Trait	
Canis familiaris - various breeds		Trait State in Taxon A	
Canis familiaris - various breeds		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=~Canis lupus^#gephebase-summary-title)		Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Canis lupus familiaris^#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

Krt71		Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)	
Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g		ADA57168 (https://www.ncbi.nlm.nih.gov/nuccore/ADA57168)	
10090.ENSMUSP00000023710 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710)			
Belongs to the intermediate filament family.			
GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)			
GO:0031069 : hair follicle morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0031069)			
GO:0045109 : intermediate filament organization			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0045109>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)

GO:0045095 : keratin filament (<https://www.ebi.ac.uk/QuickGO/term/GO:0045095>)

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

c.451A>T in exon 2 - Arg151Trp

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	Arg	Trp	151

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; Andr   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

No matches found.

Related Haplotypes

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~KRT71^/and+Taxon ID=~9612^/or+Gene Gephebase=~KRT71^/and+Taxon ID=~9615^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

@AllelicSeries <https://omia.org/OMIA000245/9615/>