

GEPHE SUMMARY

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

PHENOTYPIC CHANGE

Morphology (<https://www.gephebase.org/search-criteria?/and+Trait>
Category=^Morphology^#gephebase-summary-title)

Hair type (curly) (<https://www.gephebase.org/search-criteria?/and+Trait=^Hair+type>
(curly)^#gephebase-summary-title)

Canis familiaris - various breeds

Taxon A

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

Taxon A

Canis lupus

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

gray wolf

gray wolf; grey wolf; Canis lupus Linnaeus, 1758

species

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 () - (Rank: genus)

(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611>)

9612

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

is Taxon A an Infraspecies?

No

GENOTYPIC CHANGE

Krt71

Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g

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

Sequence Similarities

GO - Molecular Function

GO - Biological Process

UniProtKB Mus musculus

Q9RoH5 (<http://www.uniprot.org/uniprot/Q9RoH5>)

GenebankID or UniProtKB

ADA57168 (<https://www.ncbi.nlm.nih.gov/nuccore/ADA57168>)

(<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; 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

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/>