GEPHE SUMMARY

Gephebase Gene

KRT71 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^KRT71^#gephebase-summary-title)

GepheID

GP00000524

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

Trait State in Taxon A

Canis familiaris - various breeds

Trait State in Taxon B

Canis familiaris - various breeds

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

Krt71

Synonyms

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

String

10090.ENSMUSP00000023710 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710)

Sequence Similarities

Belongs to the intermediate filament family.

GO - Molecular Function

GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)

GO - Biological Process

GO:0031069 : hair follicle morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0031069)
GO:0045109 : intermediate filament organization

UniProtKB Mus musculus

Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)

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/