GEPHE SUMMARY

Gephebase Gene
R-spondin-2 (RSPO2) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^R-spondin-2 (RSPO2)^#gephebase-summary-title)

GepheID
GP00000945

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 length (moustache / eyebrows) (https://www.gephebase.org/search-criteria?/and+Trait=^Hair length (moustache / eyebrows)^#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
Rspo2

Synonyms
ftls; AA673245; D430027K22; 2610028F08Rik

String
10090.ENSMUSP00000067325 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000067325)

Sequence Similarities
Belongs to the R-spondin family.

GO - Molecular Function
GO:0008201 : heparin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008201)
GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)
GO:0005109 : frizzled binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005109)

GO - Biological Process
GO:0001649 : osteoblast differentiation

UniProtKB Mus musculus
Q8BFU0 (http://www.uniprot.org/uniprot/Q8BFU0)

GenebankID or UniProtKB
XP_005627928 (https://www.ncbi.nlm.nih.gov/nuccore/XP_005627928)

(https://www.ebi.ac.uk/QuickGO/term/GO:0001649)
GO:0090263 : positive regulation of canonical Wnt signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0090263)
GO:0030282 : bone mineralization (https://www.ebi.ac.uk/QuickGO/term/GO:0030282)
GO:0035116 : embryonic hindlimb morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0035116)
GO:0016055 : Wnt signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0016055)
GO:0071542 : dopaminergic neuron differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0071542)
GO:0035115 : embryonic forelimb morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0035115)
GO:0060441 : epithelial tube branching involved in lung morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0060441)
GO:0060437 : lung growth (https://www.ebi.ac.uk/QuickGO/term/GO:0060437)
GO:0042489 : negative regulation of odontogenesis of dentin-containing tooth
(https://www.ebi.ac.uk/QuickGO/term/GO:0042489)
GO:0060535 : trachea cartilage morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0060535)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)

Aberration Type

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

Insertion Size

100-999 bp

Molecular Details of the Mutation

a 167bp insertion within the 3'UTR at position which leads to a threefold increase in transcription of RSPO2 in muzzle skin of dogs with furnishings

Experimental Evidence

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

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 (FGF5) (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

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA001531/9615/