

GEPHE SUMMARY

	Gephebase Gene		GepheID
R-spondin-2 (RSPO2) (https://www.gephebase.org/search-criteria?/and+Gene)		GP00000945	
Gephebase="R-spondin-2 (RSPO2)"#gephebase-summary-title)			Main curator
	Entry Status	Martin	
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait)			
Category="Morphology"#gephebase-summary-title)	Trait		
Hair length (moustache / eyebrows) (<a (moustache="" eyebrows)"#gephebase-summary-title"="" hair="" href="https://www.gephebase.org/search-criteria?/and+Trait=" length="">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		Taxon B	
	Latin Name		Latin Name
Canis lupus		Canis lupus familiaris	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Canis lupus"#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Canis lupus familiaris"#gephebase-summary-title)	
	Common Name		Common Name
gray wolf		dog	
	Synonyms		Synonyms
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	
	Rank		Rank
species		subspecies	
	Lineage		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		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		Parent
Canis () - (Rank: genus)		Canis lupus (gray wolf) - (Rank: species)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9612		9615	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)		(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

	Generic Gene Name		UniProtKB Mus musculus
Rspo2		Q8BFU0 (http://www.uniprot.org/uniprot/Q8BFU0)	
	Synonyms		GenebankID or UniProtKB
ftls; AA673245; D430027K22; 2610028F08Rik		XP_005627928 (https://www.ncbi.nlm.nih.gov/nuccore/XP_005627928)	
	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			

(<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>)

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)	Presumptive Null
	Molecular Type
	Aberration Type
	Insertion Size
	Insertion Size
100-999 bp	
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	Molecular Details of the Mutation
	Experimental Evidence
	Experimental Evidence
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	Experimental Evidence
Coat variation in the domestic dog is governed by variants in three genes. (2009) (https://pubmed.ncbi.nlm.nih.gov/19713490)	Main Reference
	Main Reference
	Main Reference
Cadieu E; Neff MW; Quignon P; Walsh K; Chase K; Parker HG; Vonholdt BM; Rhue A; Boyko A; Byers A; Wong A; Mosher DS; Elkahoul AG; Spady TC; Andr�� C; Lark KG; Cargill M; Bustamante CD; Wayne RK; Ostrander EA	Authors
	Authors
	Authors
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.	Abstract
	Abstract
	Abstract
	Additional References
	Additional References
	Additional References

RELATED GEPHE

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 Genes
	Related Genes
	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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

