

GPR22 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~GPR22~#gephebase-summary-title)	Gephebase Gene	GP00002389	GepheID
Published	Entry Status	Courtier	Main curator

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)		Trait Category	
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title)		Trait	
white coat		Trait State in Taxon A	
red coat		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris~#gephebase-summary-title)	Latin Name	Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris~#gephebase-summary-title)	Latin Name
dog	Common Name	dog	Common Name
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms
subspecies	Rank	subspecies	Rank
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	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	Lineage
Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)	Parent	Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)	Parent
9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615)	NCBI Taxonomy ID	9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615)	NCBI Taxonomy ID
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
Yes		Yes	
Poodle breed	Taxon A Description	Poodle breed	Taxon B Description

Generic Gene Name	UniProtKB Homo sapiens
GPR22	Q99680 (http://www.uniprot.org/uniprot/Q99680)
Synonyms	GenebankID or UniProtKB
-	A0A8C0RPZ3 (https://www.ncbi.nlm.nih.gov/nuccore/A0A8C0RPZ3)
String	
9606.ENSP00000302676 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000302676)	
Sequence Similarities	
Belongs to the G-protein coupled receptor 1 family.	
GO - Molecular Function	
GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)	
GO:0042277 : peptide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042277)	
GO - Biological Process	

GO:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)
GO:0030030 : cell projection organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030030>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#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

10-100 kb

Molecular Details of the Mutation

A SNNL1 retrocopy element is inserted within the intron of COG5 and 2.8kbp upstream of and in the same orientation as GPR22. It is likely disrupting regulation of the CPR22 gene, resulting in higher gene expression and atypical expression in the skin.

Experimental Evidence

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

Main Reference

An SNN retrocopy insertion upstream of GPR22 is associated with dark red coat color in Poodles. (2022) (<https://pubmed.ncbi.nlm.nih.gov/36047852>)

Authors

Batcher K; Varney S; Affolter VK; Friedenberg SG; Bannasch D

Abstract

Pigment production and distribution is controlled through multiple genes, resulting in a wide range of coat color phenotypes in dogs. Dogs that produce only the pheomelanin pigment vary in intensity from white to deep red. The Poodle breed has a wide range of officially recognized coat colors, including the pheomelanin-based white, cream, apricot and red coat colors, which are not fully explained by the previously identified genetic variants involved in pigment intensity. Here, a genome-wide association study for pheomelanin intensity was performed in Poodles which identified an association on canine chromosome 18. Whole genome sequencing data revealed an SNN retrocopy insertion (SNNL1) in apricot and red Poodles within the associated region on chromosome 18. While equal numbers of melanocytes were observed in all Poodle skin hair bulbs, higher melanin content was observed in the darker Poodles. Several genes involved in melanogenesis were also identified as highly overexpressed in red Poodle skin. The most differentially expressed gene however was GPR22, which was highly expressed in red Poodle skin while unexpressed in white Poodle skin (log2 fold-change in expression 6.1, $P < 0.001$). GPR22 is an orphan G-protein coupled receptor normally expressed exclusively in the brain and heart. The SNNL1 retrocopy inserted 2.8kb upstream of GPR22 and is likely disrupting regulation of the gene, resulting in atypical expression in the skin. Thus, we identify the SNNL1 insertion as a candidate variant for the CFA18 pheomelanin intensity locus in red Poodles.

© Published by Oxford University Press 2022. This work is written by a US Government employee and is in the public domain in the US.

Additional References

RELATED GEPHE

Related Genes

12 (Agouti (ASIP), MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@TE