

GEPHE SUMMARY

	Gephebase Gene	GepheID
beta-defensin 103 (CBD103) (https://www.gephebase.org/search-criteria?/and+Gene)	GP00000141	
Gephebase="beta-defensin 103 (CBD103)"#gephebase-summary-title)		Main curator
	Entry Status	Martin
Published		

PHENOTYPIC CHANGE

Trait Category	
Morphology, Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~/and+Trait+Category=~Physiology~#gephebase-summary-title)	Trait
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title)	Trait State in Taxon A
Canis familiaris "yellow" breeds	Trait State in Taxon B
Canis familiaris "black" breeds; autosomal dominant	Ancestral State
Taxon A	Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title)	

Taxon A	Taxon B #1
Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus~#gephebase-summary-title)	Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris~#gephebase-summary-title)
gray wolf	dog
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
species	subspecies
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
Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611)	Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)
9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)	9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615)
No	No

Taxon B #2	
Canis latrans	Latin Name
(<a +canis+latrans`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis latrans`#gephebase-summary-title)	
coyote	Common Name
coyote	Synonyms
species	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	Lineage
Canis () - (Rank: genus)	Parent
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611)	
	NCBI Taxonomy ID

9614
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9614)
is Taxon B an Intraspecies?
No

Taxon B #3	
Canis lupus	Latin Name
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="+Canis lupus^#gephebase-summary-title)	
gray wolf	Common Name
gray wolf; grey wolf; Canis lupus Linnaeus, 1758	Synonyms
species	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	Lineage
Canis () - (Rank: genus)	Parent
(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)	
Yes	is Taxon B an Intraspecies?
Yellowstone Black Wolf	Taxon B Description

GENOTYPIC CHANGE

CBD103	Generic Gene Name	UniProtKB Canis lupus familiaris
-	Synonyms	GenebankID or UniProtKB
9615.ENSACFP00000036690	String	
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9615.ENSACFP00000036690)		
-	Sequence Similarities	
GO:0031731 : CCR6 chemokine receptor binding	GO - Molecular Function	
(https://www.ebi.ac.uk/QuickGO/term/GO:0031731)		
GO:0042056 : chemoattractant activity		
(https://www.ebi.ac.uk/QuickGO/term/GO:0042056)		
GO:0060326 : cell chemotaxis	GO - Biological Process	
(https://www.ebi.ac.uk/QuickGO/term/GO:0060326)		
GO:0006935 : chemotaxis		
(https://www.ebi.ac.uk/QuickGO/term/GO:0006935)		
GO:0042742 : defense response to bacterium		
(https://www.ebi.ac.uk/QuickGO/term/GO:0042742)		
GO:0005615 : extracellular space	GO - Cellular Component	
(https://www.ebi.ac.uk/QuickGO/term/GO:0005615)		
No	(<a +no^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No^#gephebase-summary-title)	Presumptive Null
Coding	(<a +coding^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding^#gephebase-summary-title)	Molecular Type
Deletion	(<a +deletion^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+Deletion^#gephebase-summary-title)	Aberration Type
1-9 bp		Deletion Size
3bp in-frame deletion in exon 2 = deletion of Gly23		Molecular Details of the Mutation
Linkage Mapping	(<a +linkage="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=" mapping^#gephebase-summary-title"="">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Linkage Mapping^#gephebase-summary-title)	Experimental Evidence
A -defensin mutation causes black coat color in domestic dogs. (2007)	(https://pubmed.ncbi.nlm.nih.gov/17947548)	Main Reference
Candille SI; Kaelin CB; Cattanach BM; Yu B; Thompson DA; Nix MA; Kerns JA; Schmutz SM; Millhauser GL; Barsh GS		Authors
		Abstract

Genetic analysis of mammalian color variation has provided fundamental insight into human biology and disease. In most vertebrates, two key genes, Agouti and Melanocortin 1 receptor (Mc1r), encode a ligand-receptor system that controls pigment type-switching, but in domestic dogs, a third gene is implicated, the K locus, whose genetic characteristics predict a previously unrecognized component of the melanocortin pathway. We identify the K locus as beta-defensin 103 (CBD103) and show that its protein product binds with high affinity to the Mc1r and has a simple and strong effect on pigment type-switching in domestic dogs and transgenic mice. These results expand the functional role of beta-defensins, a protein family previously implicated in innate immunity, and identify an additional class of ligands for signaling through melanocortin receptors.

Additional References

Molecular and evolutionary history of melanism in North American gray wolves. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19197024>)
Natural Selection and Origin of a Melanistic Allele in North American Gray Wolves. (2018) (<https://pubmed.ncbi.nlm.nih.gov/29688543>)

RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), RALY (hnRNP associated with lethal yellow)) (<https://www.gephebase.org/search-criteria?/or+TaxonID=~9612~/and+Trait=Coloration/or+TaxonID=~9615~/and+Trait=Coloration/or+TaxonID=~9614~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Same allele introgressed in wild Canis spp. (Wolves in North-America and Italy ; Coyotes) ; the "domesticated" melanic allele may have originated from the wild by introgression as well ; evidence of selection with possible effect on viral immunity ; @fitness @Introgression @Dominance @BalancingSelection