

GEPHE SUMMARY

|   |                                 |   |
|---|---------------------------------|---|
| <p style="text-align: right;">Gephebase Gene</p> <p>beta-defensin 103 (CBD103) (<a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a><br/>Gephebase="beta-defensin 103 (CBD103)"#gephebase-summary-title)</p> <p style="text-align: right;">Entry Status</p> <p>Published</p> | <p>GP00000141</p> <p>Martin</p> | <p style="text-align: right;">GepheID</p> <p>Main curator</p> |
|---|---------------------------------|---|

PHENOTYPIC CHANGE

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| <p>Morphology, Physiology (<a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a><br/>Category="Morphology"/and+Trait Category="Physiology"#gephebase-summary-title)</p> <p style="text-align: right;">Trait</p> <p>Coloration (coat) (<a coloration"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="Coloration</a><br/>(coat)#gephebase-summary-title)</p> <p style="text-align: right;">Trait State in Taxon A</p> <p>Canis familiaris "yellow" breeds</p> <p style="text-align: right;">Trait State in Taxon B</p> <p>Canis familiaris "black" breeds; autosomal dominant</p> <p style="text-align: right;">Ancestral State</p> <p>Taxon A</p> <p style="text-align: right;">Taxonomic Status</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a><br/>Status="Domesticated"#gephebase-summary-title)</p> |  |
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Taxon A

|  |  |
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| <p>Canis lupus<br/>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms="Canis lupus"#gephebase-summary-title)</p> <p style="text-align: right;">Common Name</p> <p>gray wolf</p> <p style="text-align: right;">Synonyms</p> <p>gray wolf; grey wolf; Canis lupus Linnaeus, 1758</p> <p style="text-align: right;">Rank</p> <p>species</p> <p style="text-align: right;">Lineage</p> <p>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</p> <p style="text-align: right;">Parent</p> <p>Canis () - (Rank: genus)<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611</a>)</p> <p style="text-align: right;">NCBI Taxonomy ID</p> <p>9612<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a>)</p> <p style="text-align: right;">is Taxon A an Intraspecies?</p> <p>No</p> |  |
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Taxon B #1

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| <p>Canis lupus familiaris<br/>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms="Canis lupus familiaris"#gephebase-summary-title)</p> <p style="text-align: right;">Common Name</p> <p>dog</p> <p style="text-align: right;">Synonyms</p> <p>Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758</p> <p style="text-align: right;">Rank</p> <p>subspecies</p> <p style="text-align: right;">Lineage</p> <p>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</p> <p style="text-align: right;">Parent</p> <p>Canis lupus (gray wolf) - (Rank: species)<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a>)</p> <p style="text-align: right;">NCBI Taxonomy ID</p> <p>9615<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615</a>)</p> <p style="text-align: right;">is Taxon B an Intraspecies?</p> <p>No</p> |  |
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Taxon B #2

|   |  |
|---|--|
| <p>Canis latrans<br/>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms="Canis latrans"#gephebase-summary-title)</p> <p style="text-align: right;">Common Name</p> <p>coyote</p> <p style="text-align: right;">Synonyms</p> <p>coyote</p> <p style="text-align: right;">Rank</p> <p>species</p> <p style="text-align: right;">Lineage</p> <p>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</p> <p style="text-align: right;">Parent</p> <p>Canis () - (Rank: genus)<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611</a>)</p> <p style="text-align: right;">NCBI Taxonomy ID</p> |  |
|---|--|

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

| Taxon B #3  |                             |
|---|-----------------------------|
| Canis lupus<br>(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Canis lupus"#gephebase-summary-title)  | Latin Name                  |
| 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)<br>(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611 )  | Parent<br>NCBI Taxonomy ID  |
| 9612<br>(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612 )  | is Taxon B an Intraspecies? |
| Yes   | Taxon B Description         |
| Yellowstone Black Wolf  |                             |

## GENOTYPIC CHANGE

|  |                         |                                   |
|--|-------------------------|-----------------------------------|
| CBD103   | Generic Gene Name       | UniProtKB Canis lupus familiaris  |
| -  | Synonyms                | GenebankID or UniProtKB           |
| 9615.ENSCAFP00000036690<br>(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9615.ENSCAFP00000036690 )  | String                  |                                   |
| -  | Sequence Similarities   |                                   |
| GO:0031731 : CCR6 chemokine receptor binding<br>(https://www.ebi.ac.uk/QuickGO/term/GO:0031731)<br>GO:0042056 : chemoattractant activity<br>(https://www.ebi.ac.uk/QuickGO/term/GO:0042056)  | GO - Molecular Function |                                   |
| GO:0060326 : cell chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0060326)<br>GO:0006935 : chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0006935)<br>GO:0042742 : defense response to bacterium<br>(https://www.ebi.ac.uk/QuickGO/term/GO:0042742) | GO - Biological Process |                                   |
| GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)   | GO - Cellular Component |                                   |
| No (https://www.gephebase.org/search-criteria?/and+Presumptive Null="No"#gephebase-summary-title)  |                         | Presumptive Null                  |
| Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type="Coding"#gephebase-summary-title)  |                         | Molecular Type                    |
| Deletion (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 (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 (Mclr), 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 Mclr 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