

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+MC1R+Gephebase-summary-title)	Gephebase Gene	GP00002303	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

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		
WT coat	Trait State in Taxon A		
Grizzle coat in Afghan Hound and Saluki	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 (https://www.gephebase.org/search-criteria?/and+Taxon+Synonyms+Canis+lupus+Gephebase-summary-title)	Latin Name	Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+Synonyms+Canis+lupus+familiaris+Gephebase-summary-title)	Latin Name
gray wolf	Common Name	dog	Common Name
gray wolf; grey wolf; Canis lupus Linnaeus, 1758	Synonyms	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758;	Synonyms
species	Rank	Canis lupus familiaris Linnaeus, 1758	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	subspecies	Lineage
Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611)	Parent	Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)	Parent
9612 (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)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	()	GenebankID or UniProtKB
9606.ENSP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605)	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)	GO - Molecular Function		
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)			
GO:0004980 : melanocyte-stimulating hormone receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004980)			

GO:0031625 : ubiquitin protein ligase binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031625>)

GO - Biological Process

GO:0007275 : multicellular organism development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)
GO:0051897 : positive regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
GO:0019233 : sensory perception of pain
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019233>)
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007189>)
GO:0035556 : intracellular signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide second messenger (<https://www.ebi.ac.uk/QuickGO/term/GO:0007187>)
GO:0032720 : negative regulation of tumor necrosis factor production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032720>)
GO:0010739 : positive regulation of protein kinase A signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010739>)
GO:0090037 : positive regulation of protein kinase C signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090037>)
GO:0009650 : UV protection (<https://www.ebi.ac.uk/QuickGO/term/GO:0009650>)
GO:0070914 : UV-damage excision repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070914>)

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

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.233G>T p.G78V

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Val	78

Main Reference

A new mutation in MC1R explains a coat color phenotype in 2 "old" breeds: Saluki and Afghan hound. (2010 Sep-Oct) (<https://pubmed.ncbi.nlm.nih.gov/20525767>)

Authors

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Abstract

Melanocortin 1 Receptor (MC1R) has been studied in a wide variety of domestic animals (Klungland et al. 1995; Marklund et al. 1996; VÃ¥ge et al. 1997; Kijas et al. 1998; Newton et al. 2000; VÃ¥ge et al. 2003), and also several wild animals (Robbins et al. 1993; Ritland et al. 2001; Eizirik et al. 2003; Nachman et al. 2003; McRobie et al. 2009) in relation to coat color variation. A variety of phenotypic changes have been reported including coat colors from pure black to pure red, as well as some phenotypes with hairs with red and black bands. One phenotype, called grizzle in Salukis and domino in Afghan Hounds, appears to be unique to these 2 old dog breeds. This pattern is characterized by a pale face with a widow's peak above the eyes. The body hairs on the dorsal surface of Salukis and Afghan Hounds have both phaeomelanin and eumelanin portions, even though they had an a(t)/a(t) genotype at ASIP. In addition, all had at least one copy of a newly identified mutation in MC1R, g.233G>T, resulting in p.Gly78Val. This new allele, that we suggest be designated as E(G), is dominant to the E and e (p.Arg306ter) alleles at MC1R but recessive to the E(M) (p.Met264Val) allele. The K(B) allele (p.Gly23del) at DEF103 and the a(y) allele (p.Ala82Ser and p.Arg83His) of ASIP are epistatic to grizzle and domino.

Additional References

RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3, FGF4, FGF19, ORAOV1, Kit, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103), RALY (hnRNP associated with lethal yellow)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=^9612^/and+Trait=Coloration/or+Taxon+ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001495/9615/@Parallelism>