

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000569	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)	Trait Category		
	Trait		
	Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)^#gephebase-summary-title)		
Trait State in Taxon A			
Vulpes lagopus ; red fox			
Trait State in Taxon B			
Vulpes lagopus ; silver coats in arctic fox including blue variant			
Ancestral State			
Unknown			
Taxonomic Status			
Interspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Interspecific^#gephebase-summary-title)			

Taxon A		Taxon B	
Vulpes vulpes (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Vulpesvulpes^#gephebase-summary-title)	Latin Name	Vulpes lagopus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Vulpeslagopus^#gephebase-summary-title)	Latin Name
	Common Name	Arctic fox	Common Name
	Synonyms		Synonyms
red fox		Alopex lagopus; Arctic fox	
Canis vulpes; red fox; silver fox	Rank		Rank
species		species	
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; Vulpes	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; Vulpes	Lineage
	Parent		Parent
	Vulpes () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9625)	Vulpes () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9625)	
9627 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9627)	NCBI Taxonomy ID	494514 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=494514)	NCBI Taxonomy ID
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
	Synonyms	CAH10741 (https://www.ncbi.nlm.nih.gov/nuccore/CAH10741)	GenebankID or UniProtKB
	String		
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)			
Sequence Similarities			
Belongs to the G-protein coupled receptor 1 family.			
GO - Molecular Function			
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)			
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			

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>)

Mutation #1

Presumptive Null

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

G5C

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	GGC	TGC	13
Amino-acid	Gly	Cys	5

Main Reference

Two cysteine substitutions in the MC1R generate the blue variant of the Arctic fox (*Alopex lagopus*) and prevent expression of the white winter coat. (2005) (<https://pubmed.ncbi.nlm.nih.gov/15982782>)

Authors

VÃge DI; Fuglei E; Snipstad K; Beheim J; Landsem VM; Klungland H

Abstract

We have characterized two mutations in the MC1R gene of the blue variant of the arctic fox (*Alopex lagopus*) that both incorporate a novel cysteine residue into the receptor. A family study in farmed arctic foxes verified that the dominant expression of the blue color phenotype cosegregates completely with the allele harboring these two mutations. Additionally to the altered pigment synthesis, the blue fox allele suppresses the seasonal change in coat color found in the native arctic fox. Consequently, these findings suggest that the MC1R/agouti regulatory system is involved in the seasonal changes of coat color found in arctic fox.

Additional References

Genetic variations of the coding region of the melanocortin receptor 1 (MC1R) gene in the fox. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27072328>)

Mutation #2

Presumptive Null

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

F280C

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

	Taxon A	Taxon B	Position
Codon	TTC	TGC	839
Amino-acid	Phe	Cys	280

Experimental Evidence

Main Reference

Two cysteine substitutions in the MC1R generate the blue variant of the Arctic fox (*Alopex lagopus*) and prevent expression of the white winter coat. (2005) (<https://pubmed.ncbi.nlm.nih.gov/15982782>)

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Additional References

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RELATED GEPHE

Related Genes

2 (Agouti, Kit (type III receptor protein-tyrosine kinase)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9627~/and+Trait=Coloration/or+Taxon ID=~494514~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~MC1R~/and+Taxon ID=~9627~/or+Gene Gephebase=~MC1R~/and+Taxon ID=~494514~#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS