

## GEPHE SUMMARY

MC1R ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+^MC1R^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+^MC1R^#gephebase-summary-title</a> )	Gephebase Gene	GP00000604	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category+^Morphology+^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category+^Morphology+^#gephebase-summary-title</a> )	Trait Category		
Coloration (feathers) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+^Coloration+(feathers)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+^Coloration+(feathers)^#gephebase-summary-title</a> )	Trait		
Falco rusticolus	Trait State in Taxon A		
Falco rusticolus	Trait State in Taxon B		
Data not curated	Ancestral State		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+^Intraspecific+^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+^Intraspecific+^#gephebase-summary-title</a> )	Taxonomic Status		

Taxon A		Taxon B	
	Latin Name		Latin Name
Falco rusticolus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Falco+rusticolus+^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Falco+rusticolus+^#gephebase-summary-title</a> )	Falco rusticolus	Falco rusticolus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Falco+rusticolus+^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Falco+rusticolus+^#gephebase-summary-title</a> )	Falco rusticolus
gyrfalcon	Common Name	gyrfalcon	Common Name
gyrfalcon	Synonyms	gyrfalcon	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Falconiformes; Falconidae; Falco	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Falconiformes; Falconidae; Falco	Lineage
Falco (falcons) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8952">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8952</a> )	Parent	Falco (falcons) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8952">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8952</a> )	Parent
120794 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=120794">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=120794</a> )	NCBI Taxonomy ID	120794 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=120794">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=120794</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 ( <a href="http://www.uniprot.org/uniprot/Q01726">http://www.uniprot.org/uniprot/Q01726</a> )	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	AFF57733 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AFF57733">https://www.ncbi.nlm.nih.gov/nuccore/AFF57733</a> )	GenebankID or UniProtKB
9606.ENSP00000451605 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605</a> )	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008528">https://www.ebi.ac.uk/QuickGO/term/GO:0008528</a> )	GO - Molecular Function		
GO:0004977 : melanocortin receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004977">https://www.ebi.ac.uk/QuickGO/term/GO:0004977</a> )			
GO:0004980 : melanocyte-stimulating hormone receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004980">https://www.ebi.ac.uk/QuickGO/term/GO:0004980</a> )			

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

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

Val128Ile

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

-

-

-

Main Reference

Genetics of plumage color in the Gyrfalcon (*Falco rusticolus*): analysis of the melanocortin-1 receptor gene. (2012 May-Jun) (<https://pubmed.ncbi.nlm.nih.gov/22504110>)

Authors

Johnson JA; Ambers AD; Burnham KK

Abstract

Genetic variation at the melanocortin-1 receptor (MC1R) gene is correlated with melanin color variation in a few reported vertebrates. In Gyrfalcon (*Falco rusticolus*), plumage color variation exists throughout their arctic and subarctic circumpolar distribution, from white to gray and almost black. Multiple color variants do exist within the majority of populations; however, a few areas (e.g., northern Greenland and Iceland) possess a single color variant. Here, we show that the white/melanin color pattern observed in Gyrfalcons is explained by allelic variation at MC1R. Six nucleotide substitutions in MC1R resulted in 9 alleles that differed in geographic frequency with at least 2 MC1R alleles observed in almost all sampled populations in Greenland, Iceland, Canada, and Alaska. In north Greenland, where white Gyrfalcons predominate, a single MC1R allele was observed at high frequency (>98%), whereas in Iceland, where only gray Gyrfalcons are known to breed, 7 alleles were observed. Of the 6 nucleotide substitutions, 3 resulted in amino acid substitutions, one of which (Val(128)Ile) was perfectly associated with the white/melanin polymorphism. Furthermore, the degree of melanism was correlated with number of MC1R variant alleles, with silver Gyrfalcons all heterozygous and the majority of dark gray individuals homozygous (Ile(128)). These results provide strong support that MC1R is associated with plumage color in this species.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS