GEPHE SUMMARY

Gephebase Gene

MC1R (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MC1R^#gephebase-summary-title)

GepheID

GP00001353

Entry Status

Published

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title)

Trait State in Taxon A

Goat ; Murciano-Granadina ; red

Trait State in Taxon B

Goat ; Murciano-Granadina and Maltese ; black

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)

Common Name

goat

Synonyms

Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent

Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID

9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Goat ; Murciano-Granadina ; red

Taxon B

Latin Name

Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)

Common Name

goat

Synonyms

Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent

Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID

9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Goat ; Murciano-Granadina and Maltese ; black

GENOTYPIC CHANGE

Generic Gene Name

Mc1r

Synonyms

e; Tob; Mcr1; Mshra; Msh-r

String

10090.ENSMUSP00000095929 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000095929)

UniProtKB Mus musculus

Q01727 (http://www.uniprot.org/uniprot/Q01727)

GenebankID or UniProtKB

()

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family.

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:0042562 : hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042562)

GO - Biological Process

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: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: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:0070914 : UV-damage excision repair
(https://www.ebi.ac.uk/QuickGO/term/GO:0070914)
GO:2000253 : positive regulation of feeding behavior
(https://www.ebi.ac.uk/QuickGO/term/GO:2000253)
GO:0060259 : regulation of feeding behavior
(https://www.ebi.ac.uk/QuickGO/term/GO:0060259)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)

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

c.801C>G p.Cys267Trp

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

Missense and nonsense mutations in melanocortin 1 receptor (MC1R) gene of different goat breeds: association with red and black coat colour phenotypes but with unexpected evidences. (2009) (https://pubmed.ncbi.nlm.nih.gov/19706191)

Authors

Fontanesi L; Beretti F; Riggio V; Dall'Olio S; GonzÃ¡lez EG; Finocchiaro R; Davoli R; Russo V; Portolano B

Abstract

Agouti and Extension loci control the relative amount of eumelanin and pheomelanin production in melanocytes that, in turn, affects pigmentation of skin and hair. The Extension locus encodes the melanocortin 1 receptor (MC1R) whose permanent activation, caused by functional mutations, results in black coat colour, whereas other inactivating mutations cause red coat colour in different mammals.

The whole coding region of the MC1R gene was sequenced in goats of six different breeds showing different coat colours (Girgentana, white cream with usually small red spots in the face; Maltese, white with black cheeks and ears; Derivata di Siria, solid red; Murciano-Granadina, solid black or solid brown; Camosciata delle Alpi, brown with black stripes; Saanen, white; F1 goats and the parental animals). Five single nucleotide polymorphisms (SNPs) were identified: one nonsense mutation (p.Q225X), three missense mutations (p.A81V, p.F250V, and p.C267W), and one silent mutation. The stop codon at position 225 should cause the production of a shorter MC1R protein whose functionality may be altered. These SNPs were investigated in a larger sample of animals belonging to the six breeds. The Girgentana breed was almost fixed for the p.225X allele. However, there was not complete association between the presence of red spots in the face and the presence of this allele in homozygous condition. The same allele was identified in the Derivata di Siria breed. However, its frequency was only 33%, despite the fact that these animals are completely red. The p.267W allele was present in all Murciano-Granadina black goats, whereas it was never identified in the brown ones. Moreover, the same substitution was present in almost all Maltese goats providing evidence of association between this mutation and black coat colour.

According to the results obtained in the investigated goat breeds, MC1R mutations may determine eumelanic and pheomelanic phenotypes. However, they are probably not the only factors. In particular, the surprising not complete association of the nonsense mutation (p.Q225X) with red coat colour raises a few hypotheses on the determination of pheomelanic phenotypes in goats that should be further investigated.

Additional References

RELATED GEPHE

Related Genes

3 (Agouti (ASIP), EDNRA, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+TaxonID=^9925^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

mutation is dominant