

GEPHE SUMMARY

|                                                                                                                                                                                                                |                |            |              |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------|--------------|
| MC1R ( <a href="https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title</a> ) | Gephebase Gene | GP00000592 | GepheID      |
|                                                                                                                                                                                                                | Entry Status   | Martin     | Main curator |
|                                                                                                                                                                                                                | Published      |            |              |

PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                                                   |  |                                                                                                                                                                                                                                                                                                                                                   |  |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|
| Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title</a> )                                                                                                                  |  | Trait Category                                                                                                                                                                                                                                                                                                                                    |  |
| Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)^#gephebase-summary-title</a> )                                                                                                               |  | Trait                                                                                                                                                                                                                                                                                                                                             |  |
| Ovis aries                                                                                                                                                                                                                                                                                                                                        |  | Trait State in Taxon A                                                                                                                                                                                                                                                                                                                            |  |
| Ovis aries                                                                                                                                                                                                                                                                                                                                        |  | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                            |  |
| Data not curated                                                                                                                                                                                                                                                                                                                                  |  | Ancestral State                                                                                                                                                                                                                                                                                                                                   |  |
| Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title</a> )                                                                                                        |  | Taxonomic Status                                                                                                                                                                                                                                                                                                                                  |  |
| Taxon A                                                                                                                                                                                                                                                                                                                                           |  | Taxon B                                                                                                                                                                                                                                                                                                                                           |  |
| Ovis aries<br>( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Ovis+aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Ovis+aries^#gephebase-summary-title</a> )                                                                                                     |  | Ovis aries<br>( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Ovis+aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Ovis+aries^#gephebase-summary-title</a> )                                                                                                     |  |
| sheep                                                                                                                                                                                                                                                                                                                                             |  | sheep                                                                                                                                                                                                                                                                                                                                             |  |
| Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758                                                                                                                                                                                                                           |  | Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758                                                                                                                                                                                                                           |  |
| 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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis |  | cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis |  |
| Ovis () - (Rank: genus)<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )                                                                                                                                                                    |  | Ovis () - (Rank: genus)<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )                                                                                                                                                                    |  |
| 9940<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )                                                                                                                                                                                       |  | 9940<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )                                                                                                                                                                                       |  |
| No                                                                                                                                                                                                                                                                                                                                                |  | No                                                                                                                                                                                                                                                                                                                                                |  |

GENOTYPIC CHANGE

|                                                                                                                                                                                                                                 |                   |                                                                                                                        |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------|------------------------------------------------------------------------------------------------------------------------|
| MC1R                                                                                                                                                                                                                            | Generic Gene Name | UniProtKB Homo sapiens                                                                                                 |
|                                                                                                                                                                                                                                 | Synonyms          | Q01726 ( <a href="http://www.uniprot.org/uniprot/Q01726">http://www.uniprot.org/uniprot/Q01726</a> )                   |
|                                                                                                                                                                                                                                 | String            | ABF71418 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/ABF71418">https://www.ncbi.nlm.nih.gov/nuccore/ABF71418</a> ) |
| 9606.ENSP00000451605<br>( <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> ) |                   |                                                                                                                        |
| Belongs to the G-protein coupled receptor 1 family.                                                                                                                                                                             |                   |                                                                                                                        |
| GO:0008528 : G protein-coupled peptide receptor activity<br>( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008528">https://www.ebi.ac.uk/QuickGO/term/GO:0008528</a> )                                                       |                   |                                                                                                                        |
| GO:0004977 : melanocortin receptor activity<br>( <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<br>( <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>)

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

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

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

Nonsynonymous

M73K

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

|            | Taxon A | Taxon B | Position |
|------------|---------|---------|----------|
| Codon      | -       | -       | -        |
| Amino-acid | -       | -       | -        |

Molecular and pharmacological characterization of dominant black coat color in sheep. (1999) (<https://pubmed.ncbi.nlm.nih.gov/9892731>)

VÃ¥ge DI; Klungland H; Lu D; Cone RD

Dominant black coat color in sheep is predicted to be caused by an allele ED at the extension locus. Recent studies have shown that this gene encodes the melanocyte stimulating hormone receptor (MC1-R). In mouse and fox, naturally occurring mutations in the coding region of MC1-R produce a constitutively activated receptor that switches the synthesis from phaeomelanin to eumelanin within the melanocyte, explaining the black coat color observed phenotypically. In the sheep, we have identified a Met-->Lys mutation in position 73 (M73K) together with a Asp --> Asn change at position 121 (D121N) showing complete cosegregation with dominant black coat color in a family lineage. Only the M73K mutation showed constitutive activation when introduced into the corresponding mouse receptor (mMC1-R) for pharmacological analysis; however, the position corresponding to D121 in the mouse receptor is required for high affinity ligand binding. The pharmacological profile of the M73K change is unique compared to the constitutively active E92K mutation in the sombre mouse and C123R mutation in the Alaska silver fox, indicating that the M73K change activates the receptor via a mechanism distinct from these previously characterized mutations.

Epistatic Interaction of the Melanocortin 1 Receptor and Agouti Signaling Protein Genes Modulates Wool Color in the Brazilian Creole Sheep. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27288530>)

Main Reference

Authors

Abstract

Additional References

RELATED GEPHE

EXTERNAL LINKS

COMMENTS