GEPHE SUMMARY

Gephebase Gene

PMEL17 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^PMEL17^#gephebase-summary-title)

GepheID

GP00002027

Entry Status

Published

Main curator

Courtier

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

Domesticated cattle

Trait State in Taxon B

Domesticated cattle with coat colour dilution

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

cattle

Synonyms

Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent

Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID

9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Holstein

Taxon B

Latin Name

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

Common Name

cattle

Synonyms

Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent

Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID

9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Charolais

GENOTYPIC CHANGE

Generic Gene Name

PMEL

Synonyms

SILV; PMEL17; RPE1

String

9913.ENSBTAP00000005250 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9913.ENSBTAP00000005250)

Sequence Similarities

Belongs to the PMEL/NMB family.

GO - Molecular Function

-

GO - Biological Process

GO:0042438 : melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)

UniProtKB Bos taurus

Q06154 (http://www.uniprot.org/uniprot/Q06154)

GenebankID or UniProtKB

()

GO:0032438 : melanosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032438)

GO - Cellular Component

GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)
GO:0005789 : endoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005789)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0005771 : multivesicular body (https://www.ebi.ac.uk/QuickGO/term/GO:0005771)

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

g.57669926G>A c.64G>A p.Gly22Arg

Experimental Evidence

Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)

	Taxon A	Taxon B	Position
Codon	GGR	AGR	57669926
Amino-acid	Gly	Arg	22

Main Reference

Genetic effects on coat colour in cattle: dilution of eumelanin and phaeomelanin pigments in an F2-Backcross Charolais x Holstein population. (2007)
(https://pubmed.ncbi.nlm.nih.gov/17705851)

Authors

GutiÃ©rrez-Gil B; Wiener P; Williams JL

Abstract

In cattle, the gene coding for the melanocortin receptor 1 (MC1R) is known to be the main regulator of the switch between the two coat colour pigments: eumelanin (black pigment) and phaeomelanin (red pigment). Some breeds, such as Charolais and Simmental, exhibit a lightening of the original pigment over the whole body. The dilution mutation in Charolais (Dc) is responsible for the white coat colour of this breed. Using an F2-Backcross Charolais x Holstein population which includes animals with both pigment backgrounds, we present a linkage mapping study of the Charolais dilution locus.

A Charolais x Holstein crossbred population was investigated for genetic effects on coat colour dilution. Three different traits representing the dilution of the phaeomelanin, eumelanin, and non-pigment-specific dilution were defined. Highly significant genome-wide associations were detected on chromosome 5 for the three traits analysed in the marker interval [ETH10-DIK5248]. The SILV gene was examined as the strongest positional and functional candidate gene. A previously reported non-synonymous mutation in exon 1 of this gene, SILV c.64A>G, was associated with the coat colour dilution phenotype in this resource population. Although some discrepancies were identified between this mutation and the dilution phenotype, no convincing recombination events were found between the SILV c.64A>G mutation and the Dc locus. Further analysis identified a region on chromosome 28 influencing the variation in pigment intensity for a given coat colour category.

The present study has identified a region on bovine chromosome 5 that harbours the major locus responsible for the dilution of the eumelanin and phaeomelanin seen in Charolais crossbred cattle. In this study, no convincing evidence was found to exclude SILV c.64A>G as the causative mutation for the Charolais dilution phenotype, although other genetic effects may influence the coat colour variation in the population studied. A region on chromosome 28 influences the intensity of pigment within coat colour categories, and therefore may include a modifier of the Dc locus. A candidate gene for this effect, LYST, was identified.

Additional References

RELATED GEPHE

Related Genes

11 (Agouti, coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, SLC45A2=MATP, Twist2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^9913^/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^9913^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS