

GEPHE SUMMARY

Gephebase Gene
coatomer protein complex subunit alpha (COPA)

Entry Status
Published

GepheID
GP00001329

Main curator
Prigent

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
Holstein dairy cattle-wild type

Trait State in Taxon B
Holstein dairy cattle-Dominant Red

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Bos taurus

Common Name
cattle

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

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; Bovinae; Bos

Parent
Bos (oxen, cattle) - (Rank: genus)

NCBI Taxonomy ID
9913

is Taxon A an Intraspecies?
Yes

Taxon A Description
Holstein dairy cattle-wild type

Taxon B

Latin Name
Bos taurus

Common Name
cattle

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

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; Bovinae; Bos

Parent
Bos (oxen, cattle) - (Rank: genus)

NCBI Taxonomy ID
9913

is Taxon B an Intraspecies?
Yes

Taxon B Description
Holstein dairy cattle-Dominant Red

GENOTYPIC CHANGE

Generic Gene Name
COPA

Synonyms
AILJK; HEP-COP; alpha-COP

String
9606.ENSP00000357048

Sequence Similarities
-

GO - Molecular Function
GO:0005198 : structural molecule activity
GO:0005179 : hormone activity

GO - Biological Process
GO:0006886 : intracellular protein transport
GO:0006888 : ER to Golgi vesicle-mediated transport
GO:0006891 : intra-Golgi vesicle-mediated transport
GO:0030157 : pancreatic juice secretion

UniProtKB Homo sapiens
P53621

GenebankID or UniProtKB

GO:0006890 : retrograde vesicle-mediated transport, Golgi to ER

GO - Cellular Component

- GO:0005737 : cytoplasm
- GO:0005829 : cytosol
- GO:0000139 : Golgi membrane
- GO:0016020 : membrane
- GO:0070062 : extracellular exosome
- GO:0005615 : extracellular space
- GO:0005789 : endoplasmic reticulum membrane
- GO:0030133 : transport vesicle
- GO:0030126 : COPI vesicle coat

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.C>T p.Arg160Cys

Experimental Evidence

Linkage Mapping

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

Main Reference

Dominant Red Coat Color in Holstein Cattle Is Associated with a Missense Mutation in the Coatomeer Protein Complex, Subunit Alpha (COPA) Gene. (2015)

Authors

Dorshorst B; Henegar C; Liao X; SÄillman AlmÄn M; Rubin CJ; Ito S; Wakamatsu K; Stothard P; Van Doormaal B; Plastow G; Barsh GS; Andersson L

Abstract

Coat color in Holstein dairy cattle is primarily controlled by the melanocortin 1 receptor (MC1R) gene, a central determinant of black (eumelanin) vs. red/brown pheomelanin synthesis across animal species. The major MC1R alleles in Holsteins are Dominant Black (MC1RD) and Recessive Red (MC1Re). A novel form of dominant red coat color was first observed in an animal born in 1980. The mutation underlying this phenotype was named Dominant Red and is epistatic to the constitutively activated MC1RD. Here we show that a missense mutation in the coatomeer protein complex, subunit alpha (COPA), a gene with previously no known role in pigmentation synthesis, is completely associated with Dominant Red in Holstein dairy cattle. The mutation results in an arginine to cysteine substitution at an amino acid residue completely conserved across eukaryotes. Despite this high level of conservation we show that both heterozygotes and homozygotes are healthy and viable. Analysis of hair pigment composition shows that the Dominant Red phenotype is similar to the MC1R Recessive Red phenotype, although less effective at reducing eumelanin synthesis. RNA-seq data similarly show that Dominant Red animals achieve predominantly pheomelanin synthesis by downregulating genes normally required for eumelanin synthesis. COPA is a component of the coat protein I seven subunit complex that is involved with retrograde and cis-Golgi intracellular coated vesicle transport of both protein and RNA cargo. This suggests that Dominant Red may be caused by aberrant MC1R protein or mRNA trafficking within the highly compartmentalized melanocyte, mimicking the effect of the Recessive Red loss of function MC1R allele.

Additional References

RELATED GEPHE

Related Genes

10 (Agouti, Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PMEL17, SLC45A2=MATP, Twist2, tyrosinase-related protein 1 (TYRP1))

Related Haplotypes

No matches found.

COMMENTS

@Epistasis - Probably hypomorphic as homozygous are viable and healthy - <https://omia.org/OMIA001529/9913/>

