GEPHE SUMMARY

Gephebase Gene
SLC45A2=MATP

GephelD
GP00002036

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat; albinism)

Trait State in Taxon A
Bos taurus

Trait State in Taxon B
Bos taurus - oculocutaneous albinism

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 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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

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

NCBI Taxonomy ID
9913

is Taxon A an Infraspecies?
No

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 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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

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

NCBI Taxonomy ID
9913

is Taxon B an Infraspecies?
Yes

Taxon B Description
Braunvieh cattle

GENOTYPIC CHANGE

Generic Gene Name
SLC45A2

Synonyms
1A1; AIM1; MATP; OCA4; SHEP5

String
9606.ENSP00000296589

Sequence Similarities
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.

GO - Molecular Function
GO:0008506 : sucrose:proton symporter activity

GO - Biological Process
GO:0042438 : melanin biosynthetic process
GO:0048066 : developmental pigmentation
GO:0007601 : visual perception
GO:0050896 : response to stimulus

UniProtKB Homo sapiens
Q9UMX9

GenebankID or UniProtKB

GO:0015770 : sucrose transport

GO - Cellular Component
GO:0016021 : integral component of membrane
GO:0033162 : melanosome membrane

Presumptive Null
No

Molecular Type
Coding

Aberration Type
SNP

SNP Coding Change
Nonsynonymous

Molecular Details of the Mutation
exact causing mutation(s) unknown - two possible amino acid changes

Experimental Evidence
Linkage Mapping

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

Main Reference
Detection of two non-synonymous SNPs in SLC45A2 on BTA20 as candidate causal mutations for oculocutaneous albinism in Braunvieh cattle. (2017)

Authors
Rothammer S; Kunz E; Seichter D; Krebs S; Wassertheurer M; Fries R; Brem G; Medugorac I

Abstract
Cases of albinism have been reported in several species including cattle. So far, research has identified many genes that are involved in this eye-catching phenotype. Thus, when two paternal Braunvieh half-sibs with oculocutaneous albinism were detected on a private farm, we were interested in knowing whether their phenotype was caused by an already known gene/mutation.

Analysis of genotyping data (50K) of the two albino individuals, their mothers and five other relatives identified a 47.61-Mb candidate haplotype on Bos taurus chromosome BTA20. Subsequent comparisons of the sequence of this haplotype with sequence data from four Braunvieh sires and the Aurochs genome identified two possible candidate causal mutations at positions 39,829,806Â bp (G/A; R45Q) and 39,864,148Â bp (C/T; T444I) that were absent in 1682 animals from various bovine breeds included in the 1000 bull genomes project. Both polymorphisms represent coding variants in the SLC45A2 gene, for which the human equivalent harbors numerous variants associated with oculocutaneous albinism type 4. We demonstrate an association of R45Q and T444I with the albino phenotype by targeted genotyping.

Although the candidate gene SLC45A2 is known to be involved in albinism in different species, to date in cattle only mutations in the TYR and MITF genes were reported to be associated with albinism or albinism-like phenotypes. Thus, our study extends the list of genes that are associated with bovine albinism. However, further research and more samples from related animals are needed to elucidate if only one of these two single nucleotide polymorphisms or the combination of both is the actual causal variant.

Additional References

RELATED GEPHE

Related Genes
10 (Agouti, coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, PMEL17, Twist2, tyrosinase-related protein 1 (TYRP1))
Related Haplotypes
No matches found.

COMMENTS

https://omia.org/OMIA001821/9913/