

GEPHE SUMMARY

SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] SLC45A2=MATP [^] #gephebase-summary-title)	Gephebase Gene	GP00002036	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category= [^] Morphology [^] #gephebase-summary-title)	Trait Category		
Coloration (coat; albinism) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Coloration">https://www.gephebase.org/search-criteria?/and+Trait=[^]Coloration (coat; albinism) [^] #gephebase-summary-title)	Trait		
Bos taurus	Trait State in Taxon A		
Bos taurus - oculocutaneous albinism	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status= [^] Domesticated [^] #gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Bos taurus [^] #gephebase-summary-title)	Latin Name	Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Bos taurus [^] #gephebase-summary-title)	Latin Name
cattle	Common Name	cattle	Common Name
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	Synonyms	Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	Synonyms
species	Rank	species	Rank
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	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	Lineage
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)	Parent	Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)	Parent
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)	NCBI Taxonomy ID	9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
		Braunvieh cattle	Taxon B Description

GENOTYPIC CHANGE

SLC45A2	Generic Gene Name	Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)	UniProtKB Homo sapiens
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	0	GenebankID or UniProtKB
9606.ENSPP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000296589)	String		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.	Sequence Similarities		
GO:0008506 : sucrose:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)	GO - Molecular Function		
	GO - Biological Process		

GO:0042438 : melanin biosynthetic process
 (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
 GO:0048066 : developmental pigmentation
 (https://www.ebi.ac.uk/QuickGO/term/GO:0048066)
 GO:0007601 : visual perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007601)
 GO:0050896 : response to stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:0050896)
 GO:0015770 : sucrose transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015770)
 GO - Cellular Component

GO:0016021 : integral component of membrane
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
 GO:0033162 : melanosome membrane
 (https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title) Presumptive Null
 Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title) Molecular Type
 SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title) Aberration Type
 Nonsynonymous SNP Coding Change
 exact causing mutation(s) unknown - two possible amino acid changes Molecular Details of the Mutation
 Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title) Experimental Evidence

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

Detection of two non-synonymous SNPs in SLC45A2 on BTA20 as candidate causal mutations for oculocutaneous albinism in Braunvieh cattle. (2017)
 (https://pubmed.ncbi.nlm.nih.gov/28982372) Main Reference

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

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. Abstract

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 39,829,806 Å bp (G/A; R45Q) and 39,864,148 Å 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

11 (Agouti, coatmer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PMEL17, 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 Genes

No matches found. Related Haplotypes

EXTERNAL LINKS

COMMENTS

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

