

GEPHE SUMMARY

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

PHENOTYPIC CHANGE

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

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
SLC45A2		
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	GenebankID or UniProtKB
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589)	String	0
	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 (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)		
	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>)

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=%27No%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27No%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27SNP%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27SNP%27#gephebase-summary-title))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

exact causing mutation(s) unknown - two possible amino acid changes

Experimental Evidence

Linkage Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Linkage Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Linkage%20Mapping%27#gephebase-summary-title))

	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)
 (<https://pubmed.ncbi.nlm.nih.gov/28982372>)

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 39,829,806 Å bp (G/A; R45Q) and 39,864,148 Å bp (C/T; T441I) 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 T441I 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

11 (Agouti, coatomer 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+TaxonID=%279913%27/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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

