GEPHE SUMMARY

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

GepheID
GP00002025

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Milk protein content (casein) (https://www.gephebase.org/search-criteria?/and+Trait=^Milk protein content (casein)^#gephebase-summary-title)

Trait State in Taxon A
Bos taurus - normal milk

Trait State in Taxon B
Bos taurus - A milk - mostly lacks a form of Î²-casein proteins called A1 and instead has mostly the A2 form - allele A2

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?
No

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?
No

GENOTYPIC CHANGE

Generic Gene Name
CSN2

Synonyms
-

String
9913.ENSBTAP00000003409 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9913.ENSBTAP00000003409)

Sequence Similarities
Belongs to the beta-casein family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0016209 : antioxidant activity (https://www.ebi.ac.uk/QuickGO/term/GO:0016209)
GO:0008191 : metalloendopeptidase inhibitor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008191)

UniProtKB Bos taurus
P02666 (http://www.uniprot.org/uniprot/P02666)

GenebankID or UniProtKB
()

GO:0019870 : potassium channel inhibitor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0019870)
GO:0004869 : cysteine-type endopeptidase inhibitor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004869)

GO - Biological Process

GO:0050728 : negative regulation of inflammatory response
(https://www.ebi.ac.uk/QuickGO/term/GO:0050728)
GO:0032355 : response to estradiol (https://www.ebi.ac.uk/QuickGO/term/GO:0032355)
GO:0008217 : regulation of blood pressure
(https://www.ebi.ac.uk/QuickGO/term/GO:0008217)
GO:0009408 : response to heat (https://www.ebi.ac.uk/QuickGO/term/GO:0009408)
GO:2000117 : negative regulation of cysteine-type endopeptidase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:2000117)
GO:0043124 : negative regulation of I-kappaB kinase/NF-kappaB signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0043124)
GO:1903720 : negative regulation of I-kappaB phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:1903720)
GO:1903488 : negative regulation of lactation
(https://www.ebi.ac.uk/QuickGO/term/GO:1903488)
GO:0010804 : negative regulation of tumor necrosis factor-mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0010804)
GO:1903496 : response to 11-deoxycorticosterone
(https://www.ebi.ac.uk/QuickGO/term/GO:1903496)
GO:1903494 : response to dehydroepiandrosterone
(https://www.ebi.ac.uk/QuickGO/term/GO:1903494)
GO:0032570 : response to progesterone
(https://www.ebi.ac.uk/QuickGO/term/GO:0032570)

GO - Cellular Component

GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)
GO:0005796 : Golgi lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0005796)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

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.87181619A>C c.245A>C p.H82P CAT>CCT

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	CAT	CCT	87181619
Amino-acid	His	Pro	82

Main Reference

DNA-based identification of novel bovine casein gene variants. (2013) (https://pubmed.ncbi.nlm.nih.gov/23102962)

Authors

Gallinat JL; Qanbari S; DrÃ¶gemÃ¼ller C; Pimentel EC; Thaller G; Tetens J

Abstract

In cattle, at least 39 variants of the 4 casein proteins (Î±(S1)-, Î²-, Î±(S2)- and Îº-casein) have been described to date. Many of these variants are known to affect milk-production traits, cheese-processing properties, and the nutritive value of milk. They also provide valuable information for phylogenetic studies. So far, the majority of studies exploring the genetic variability of bovine caseins considered European taurine cattle breeds and were carried out at the protein level by electrophoretic techniques. This only allows the identification of variants that, due to amino acid exchanges, differ in their electric charge, molecular weight, or isoelectric point. In this study, the open reading frames of the casein genes CSN1S1, CSN2, CSN1S2, and CSN3 of 356 animals belonging to 14 taurine and 3 indicine cattle breeds were sequenced. With this approach, we identified 23 alleles, including 5 new DNA sequence variants, with a predicted effect on the protein sequence. The new variants were only found in indicine breeds and in one local Iranian breed, which has been phenotypically classified as a taurine breed. A multidimensional scaling approach based on available SNP chip data, however, revealed an admixture of taurine and indicine populations in this breed as well as in the local Iranian breed Golpayegani. Specific indicine casein alleles were also identified in a few European taurine breeds, indicating the introgression of indicine breeds into these populations. This study shows the existence of substantial undiscovered genetic variability of bovine casein loci, especially in indicine cattle breeds. The identification of new variants is a valuable tool for phylogenetic studies and investigations into the evolution of the milk protein genes.

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA002033/9913/