GEPHE SUMMARY

Gephebase Gene

HMX1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^HMX1^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002191

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

Organ size (ear; cropped) (https://www.gephebase.org/search-criteria?/and+Trait=^Organ size (ear; cropped)^#gephebase-summary-title)

Trait State in Taxon A

Normal ears in swiss Highland cattle

Trait State in Taxon B

Cropped ears with a notch ; autosomal co-dominant

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

HMX1

Synonyms

H6; NKX5-3

String

9606.ENSP00000383516 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000383516)

Sequence Similarities

Belongs to the HMX homeobox family.

GO - Molecular Function

GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001227 : DNA-binding transcription repressor activity, RNA polymerase II-specific

UniProtKB Homo sapiens

Q9NP08 (http://www.uniprot.org/uniprot/Q9NP08)

GenebankID or UniProtKB

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(https://www.ebi.ac.uk/QuickGO/term/GO:0001227)

GO - Biological Process

GO:0007275 : multicellular organism development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0045892 : negative regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045892)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0000790 : nuclear chromatin (https://www.ebi.ac.uk/QuickGO/term/GO:0000790)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)

Aberration Type

Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)

Insertion Size

10-99 bp

Molecular Details of the Mutation

76bp duplication in an ultra-conserved enhancer located 148 kb apart of the coding region of HMX1

Experimental Evidence

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

Main Reference

A non-coding genomic duplication at the HMX1 locus is associated with crop ears in highland cattle. (2013) (https://pubmed.ncbi.nlm.nih.gov/24194898)

Authors

Koch CT; Bruggmann R; Tetens J; DrÃ¶gemÃ¼ller C

Abstract

Highland cattle with congenital crop ears have notches of variable size on the tips of both ears. In some cases, cartilage deformation can be seen and occasionally the external ears are shortened. We collected 40 cases and 80 controls across Switzerland. Pedigree data analysis confirmed a monogenic autosomal dominant mode of inheritance with variable expressivity. All affected animals could be traced back to a single common ancestor. A genome-wide association study was performed and the causative mutation was mapped to a 4 Mb interval on bovine chromosome 6. The H6 family homeobox 1 (HMX1) gene was selected as a positional and functional candidate gene. By whole genome re-sequencing of an affected Highland cattle, we detected 6 non-synonymous coding sequence variants and two variants in an ultra-conserved element at the HMX1 locus with respect to the reference genome. Of these 8 variants, only a non-coding 76 bp genomic duplication (g.106720058_106720133dup) located in the conserved region was perfectly associated with crop ears. The identified copy number variation probably results in HMX1 misregulation and possible gain-of-function. Our findings confirm the role of HMX1 during the development of the external ear. As it is sometimes difficult to phenotypically diagnose Highland cattle with slight ear notches, genetic testing can now be used to improve selection against this undesired trait.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Probably not a selected variant but included as an oddity ; https://omia.org/OMIA000317/9913/ ;