

GEPHE SUMMARY

Gephebase Gene
GH

Entry Status
Published

GepheID
GP00002037

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Body size (dwarfism)

Trait State in Taxon A
Bos taurus

Trait State in Taxon B
Bos taurus - miniature

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 Taurus

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 Intraspecies?
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 Taurus

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 Intraspecies?
Yes

Taxon B Description
Brahman cattle

GENOTYPIC CHANGE

Generic Gene Name
GH1

Synonyms
GH; GHN; GH-N; GHB5; IGHD2; hGH-N; IGHD1A; IGHD1B

String
9606.ENSP00000312673

Sequence Similarities
Belongs to the somatotropin/prolactin family.

GO - Molecular Function
GO:0046872 : metal ion binding
GO:0008083 : growth factor activity
GO:0005179 : hormone activity
GO:0005148 : prolactin receptor binding
GO:0005131 : growth hormone receptor binding

GO - Biological Process
GO:0043406 : positive regulation of MAP kinase activity

UniProtKB Homo sapiens
P01241

GenebankID or UniProtKB

GO:0050731 : positive regulation of peptidyl-tyrosine phosphorylation
 GO:0043568 : positive regulation of insulin-like growth factor receptor signaling pathway
 GO:0032355 : response to estradiol
 GO:0040018 : positive regulation of multicellular organism growth
 GO:0010828 : positive regulation of glucose transmembrane transport
 GO:0060396 : growth hormone receptor signaling pathway
 GO:0060397 : JAK-STAT cascade involved in growth hormone signaling pathway
 GO:0046427 : positive regulation of JAK-STAT cascade
 GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
 GO:0014068 : positive regulation of phosphatidylinositol 3-kinase signaling
 GO:0048513 : animal organ development
 GO:0031667 : response to nutrient levels
 GO:0070977 : bone maturation
 GO:0010536 : positive regulation of activation of Janus kinase activity
 GO:0045927 : positive regulation of growth

GO - Cellular Component

GO:0005576 : extracellular region
 GO:0005615 : extracellular space
 GO:0070195 : growth hormone receptor complex
 GO:0031904 : endosome lumen

Presumptive Null

Unknown

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

g.48768780C>T c.593C>T p.T198M

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	ACG	ATG	48768780
Amino-acid	Thr	Met	198

Main Reference

A miniature condition in Brahman cattle is associated with a single nucleotide mutation within the growth hormone gene. (2009)

Authors

McCormack BL; Chase CC; Olson TA; Elsasser TH; Hammond AC; Welsh TH; Jiang H; Randel RD; Okamura CA; Lucy MC

Abstract

Miniature Brahman cattle at the USDA ARS Subtropical Agriculture Research Station in Brooksville, FL have normal proportioned growth but are approximately 70% of mature height and weight when compared with Brahman cattle in the same herd. Pedigree analyses suggest that the condition is inherited through a recessive allele. The miniature Brahman cattle in the Brooksville herd have been used for studies of growth and reproduction, but the underlying causative mutation is unknown. Presumably, the miniature condition could arise from a mutation in the GH gene. The objective, therefore, was to clone the GH cDNA from Brooksville miniature Brahman cattle, compare its sequence to normal Brahman cattle, and test the biological activity of the native GH protein. Messenger RNA was isolated from the pituitary, and a cDNA for the protein coding region of the GH gene was amplified by reverse-transcription polymerase chain reaction (PCR) from each of 2 miniature Brahman bulls. The cDNA were cloned into plasmid vectors, and top and bottom strands were sequenced by automated DNA sequencing. The sequence of both cDNA clones derived from miniature cattle differed from *Bos indicus* GH (GenBank AF034386) at base number 641 because there was a cytosine (C) instead of a thymine (T). The C to T change encoded a mutation (threonine to methionine) at amino acid 200 (T200M mutation). The mutation was confirmed by sequencing of an additional 2 miniature cattle and comparing their sequence to 2 normal cattle. The threonine is located in the fourth alpha helix of GH and is 1 of 8 amino acids that participate in binding of GH to the GH receptor. Twelve miniature Brahman and 9 normal Brahman cattle were tested by using a restriction fragment length polymorphism analysis that employed the BsmBI restriction enzyme (specific for the mutated nucleotide). The 12 miniature Brahman cattle were homozygous for the mutation (-/-). Seven of the normal Brahman cattle were homozygous for the wild-type allele (+/+), and 2 were heterozygous (+/-). When tested in a cell-based bioassay, GH isolated from the pituitary of -/- cattle (n=4) had approximately 60% activity when compared with GH isolated from the pituitary of +/+ cattle (n=5). In summary, miniature Brahman cattle were homozygous for a single nucleotide polymorphism that encodes a mutation in an amino acid involved in binding of GH to the GH receptor. Cattle of normal size had at least 1 copy of the normal GH allele. Threonine 200 in bovine GH is required for normal growth in cattle.

Additional References

RELATED GEPHE

Related Genes

2 (LCORL, PLAG1)

Related Haplotypes

No matches found.

COMMENTS

