

GEPHE SUMMARY

<p>GH (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="GH"#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002037</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

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

<p>GH1</p> <p>GH; GHN; GH-N; GHB5; IGHD2; hGH-N; IGHD1A; IGHD1B</p> <p>9606.ENSPO0000312673 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000312673)</p> <p>Belongs to the somatotropin/prolactin family.</p> <p>GO - Molecular Function</p> <p>GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872) GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083) GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179) GO:0005148 : prolactin receptor binding</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p>	<p>P01241 (http://www.uniprot.org/uniprot/P01241)</p> <p>0</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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(<https://www.ebi.ac.uk/QuickGO/term/GO:0005148>)
 GO:0005131 : growth hormone receptor binding
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0005131>)

GO - Biological Process

GO:0043406 : positive regulation of MAP kinase activity
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043406>)
 GO:0050731 : positive regulation of peptidyl-tyrosine phosphorylation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0050731>)
 GO:0043568 : positive regulation of insulin-like growth factor receptor signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043568>)
 GO:0032355 : response to estradiol (<https://www.ebi.ac.uk/QuickGO/term/GO:0032355>)
 GO:0040018 : positive regulation of multicellular organism growth
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0040018>)
 GO:0010828 : positive regulation of glucose transmembrane transport
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0010828>)
 GO:0060396 : growth hormone receptor signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0060396>)
 GO:0060397 : JAK-STAT cascade involved in growth hormone signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0060397>)
 GO:0046427 : positive regulation of JAK-STAT cascade
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0046427>)
 GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0042531>)
 GO:0014068 : positive regulation of phosphatidylinositol 3-kinase signaling
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0014068>)
 GO:0048513 : animal organ development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048513>)
 GO:0031667 : response to nutrient levels
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0031667>)
 GO:0070977 : bone maturation (<https://www.ebi.ac.uk/QuickGO/term/GO:0070977>)
 GO:0010536 : positive regulation of activation of Janus kinase activity
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0010536>)
 GO:0045927 : positive regulation of growth
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0045927>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
 GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)
 GO:0070195 : growth hormone receptor complex
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0070195>)
 GO:0031904 : endosome lumen (<https://www.ebi.ac.uk/QuickGO/term/GO:0031904>)

Presumptive Null

Unknown ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Unknown))

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

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

Experimental Evidence

Candidate Gene ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+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) (<https://pubmed.ncbi.nlm.nih.gov/19524387>)

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

5 (aggrecan, LCORL, PLAG1, PRKG2, RNF11) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=^9913^/and+Trait=Body+size/and+groupHaplotypes=true#gephebase-summary-title))

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

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