

GEPHE SUMMARY

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

PHENOTYPIC CHANGE

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

GENOTYPIC CHANGE

GH1		Generic Gene Name		UniProtKB Homo sapiens
GH; GHN; GH-N; GHB5; IGHD2; hGH-N; IGHD1A; IGHD1B		Synonyms		GenebankID or UniProtKB
9606.ENSPP00000312673 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000312673">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000312673</a> )		String		0
Belongs to the somatotropin/prolactin family.		Sequence Similarities		
GO:0046872 : metal ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a> )		GO - Molecular Function		
GO:0008083 : growth factor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008083">https://www.ebi.ac.uk/QuickGO/term/GO:0008083</a> )				
GO:0005179 : hormone activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005179">https://www.ebi.ac.uk/QuickGO/term/GO:0005179</a> )				
GO:0005148 : prolactin receptor binding				

(<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>)

Unknown ( <a href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Unknown~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Unknown~#gephebase-summary-title</a> )	Presumptive Null
	Molecular Type
	Aberration Type
	SNP Coding Change
Coding ( <a href="https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title</a> )	SNP Coding Change
SNP ( <a href="https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title</a> )	SNP Coding Change
Nonsynonymous	Molecular Details of the Mutation
g.48768780C>T c.593C>T p.T198M	
Candidate Gene ( <a href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title</a> )	Experimental Evidence

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

A miniature condition in Brahman cattle is associated with a single nucleotide mutation within the growth hormone gene. (2009) ( <a href="https://pubmed.ncbi.nlm.nih.gov/19524387">https://pubmed.ncbi.nlm.nih.gov/19524387</a> )	Main Reference
	Authors
	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))

No matches found.

Related Genes

Related Haplotypes

EXTERNAL LINKS

COMMENTS

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