

## GEPHE SUMMARY

PLAG1 ( <a href="https://www.gephebase.org/search-criteria?/and+GeneGephebase=~PLAG1^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+GeneGephebase=~PLAG1^#gephebase-summary-title</a> )	Gephebase Gene	GP00001569	GepheID
Published	Entry Status	Prigent	Main curator

## 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 (height; weight) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=~Body size (height; weight)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Body size (height; weight)^#gephebase-summary-title</a> )	Trait		
Cattle Jersey breed - small stature	Trait State in Taxon A		
Cattle Holstein-Friesian breed - higher stature	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
	Latin Name		Latin Name
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	Common Name	cattle	Common Name
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	Synonyms	Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	Synonyms
species	Rank	species	Rank
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	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	Lineage
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> )	Parent	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> )	Parent
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> )	NCBI Taxonomy ID	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> )	NCBI Taxonomy ID
Yes	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
Cattle Jersey breed - small stature	Taxon A Description	Cattle Holstein-Friesian breed - higher stature	Taxon B Description

## GENOTYPIC CHANGE

PLAG1	Generic Gene Name	E1BEA5 ( <a href="http://www.uniprot.org/uniprot/E1BEA5">http://www.uniprot.org/uniprot/E1BEA5</a> )	UniProtKB Bos taurus
-	Synonyms	0	GenebankID or UniProtKB
9913.ENSBTAP00000005251 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9913.ENSBTAP00000005251">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9913.ENSBTAP00000005251</a> )	String		
-	Sequence Similarities		
GO:0003700 : DNA-binding transcription factor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003700">https://www.ebi.ac.uk/QuickGO/term/GO:0003700</a> )	GO - Molecular Function		
GO:0003677 : DNA binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003677">https://www.ebi.ac.uk/QuickGO/term/GO:0003677</a> )			
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA			

polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001077>)  
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)

GO - Biological Process

GO:0006355 : regulation of transcription, DNA-templated  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006355>)  
GO:0010629 : negative regulation of gene expression  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010629>)  
GO:0035264 : multicellular organism growth  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035264>)  
GO:0060736 : prostate gland growth (<https://www.ebi.ac.uk/QuickGO/term/GO:0060736>)  
GO:0006351 : transcription, DNA-templated  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006351>)  
GO:0022612 : gland morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0022612>)  
GO:0060252 : positive regulation of glial cell proliferation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060252>)

GO - Cellular Component

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)  
GO:0016607 : nuclear speck (<https://www.ebi.ac.uk/QuickGO/term/GO:0016607>)

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

a (CCG)<sub>n</sub> trinucleotide repeat with 9> 11 copies and a SNP A>G immediately upstream of PLAG1 transcriptional start site

Experimental Evidence

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

Main Reference

Variants modulating the expression of a chromosome domain encompassing PLAG1 influence bovine stature. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21516082>)

Authors

Karim L; Takeda H; Lin L; Druet T; Arias JA; Baurain D; Cambisano N; Davis SR; Farnir F; Grisart B; Harris BL; Keehan MD; Littlejohn MD; Spelman RJ; Georges M; Coppieters W

Abstract

We report mapping of a quantitative trait locus (QTL) with a major effect on bovine stature to a  $\approx 1/4780$ -kb interval using a Hidden Markov Model-based approach that simultaneously exploits linkage and linkage disequilibrium. We re-sequenced the interval in six sires with known QTL genotype and identified 13 clustered candidate quantitative trait nucleotides (QTNs) out of >9,572 discovered variants. We eliminated five candidate QTNs by studying the phenotypic effect of a recombinant haplotype identified in a breed diversity panel. We show that the QTL influences fetal expression of seven of the nine genes mapping to the  $\approx 1/4780$ -kb interval. We further show that two of the eight candidate QTNs, mapping to the PLAG1-CHCHD7 intergenic region, influence bidirectional promoter strength and affect binding of nuclear factors. By performing expression QTL analyses, we identified a splice site variant in CHCHD7 and exploited this naturally occurring null allele to exclude CHCHD7 as single causative gene.

Additional References

## RELATED GEPHE

Related Genes

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

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

Non-null mutation.