

GEPHE SUMMARY

Gephebase Gene
Prolactin

Entry Status
Published

GepheID
GP00002032

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Hair length (long)

Trait State in Taxon A
Bos taurus

Trait State in Taxon B
Bos taurus: hairy; lactation failure and thermoregulatory dysfunction

Ancestral State
Taxon A

Taxonomic Status
Domesticated

	Taxon A	Taxon B
Latin Name	<i>Bos taurus</i>	<i>Bos taurus</i>
Common Name	cattle	cattle
Synonyms	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
Rank	species	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	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)	Bos (oxen, cattle) - (Rank: genus)
NCBI Taxonomy ID	9913	9913
is Taxon A an Intraspecies?	No	No

GENOTYPIC CHANGE

Generic Gene Name
PRL

Synonyms
GHA1; Prol

String
9913.ENSBTAP00000020313

Sequence Similarities
Belongs to the somatotropin/prolactin family.

GO - Molecular Function
GO:0005179 : hormone activity
GO:0005148 : prolactin receptor binding

GO - Biological Process
GO:0043066 : negative regulation of apoptotic process
GO:0010629 : negative regulation of gene expression
GO:0045807 : positive regulation of endocytosis
GO:0008284 : positive regulation of cell proliferation
GO:0010628 : positive regulation of gene expression
GO:0051092 : positive regulation of NF-kappaB transcription factor activity

UniProtKB Bos taurus
P01239

GenebankID or UniProtKB

GO:0010751 : negative regulation of nitric oxide mediated signal transduction
 GO:0007565 : female pregnancy
 GO:1901224 : positive regulation of NIK/NF-kappaB signaling
 GO:0030072 : peptide hormone secretion
 GO:0023019 : signal transduction involved in regulation of gene expression
 GO:0009612 : response to mechanical stimulus
 GO:0032094 : response to food
 GO:0046427 : positive regulation of JAK-STAT cascade
 GO:0048571 : long-day photoperiodism
 GO:0045429 : positive regulation of nitric oxide biosynthetic process
 GO:0030879 : mammary gland development
 GO:0007595 : lactation
 GO:1903489 : positive regulation of lactation
 GO:0031667 : response to nutrient levels
 GO:0009058 : biosynthetic process
 GO:0001825 : blastocyst formation
 GO:0045723 : positive regulation of fatty acid biosynthetic process
 GO:1903538 : regulation of meiotic cell cycle process involved in oocyte maturation
 GO:0043207 : response to external biotic stimulus
 GO:1903576 : response to L-arginine

GO - Cellular Component

GO:0005829 : cytosol
 GO:0005615 : extracellular space
 GO:0005788 : endoplasmic reticulum lumen
 GO:0005789 : endoplasmic reticulum membrane

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

g.35105313A>C - c.661A>C - p.C221G

Experimental Evidence

Linkage Mapping

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	Gly	221

Main Reference

Functionally reciprocal mutations of the prolactin signalling pathway define hairy and slick cattle. (2014)

Authors

Littlejohn MD; Henty KM; Tiplady K; Johnson T; Harland C; Lopdell T; Sherlock RG; Li W; Lukefahr SD; Shanks BC; Garrick DJ; Snell RG; Spelman RJ; Davis SR

Abstract

Lactation, hair development and homeothermy are characteristic evolutionary features that define mammals from other vertebrate species. Here we describe the discovery of two autosomal dominant mutations with antagonistic, pleiotropic effects on all three of these biological processes, mediated through the prolactin signalling pathway. Most conspicuously, mutations in prolactin (PRL) and its receptor (PRLR) have an impact on thermoregulation and hair morphology phenotypes, giving prominence to this pathway outside of its classical roles in lactation.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1

COMMENTS

<https://omia.org/OMIA000441/9913/>

