

## GEPHE SUMMARY

**Gephebase Gene**  
Prolactin

**Entry Status**  
Published

**GepheID**  
GP00002033

**Main curator**  
Courtier

## PHENOTYPIC CHANGE

**Trait Category**  
Morphology

**Trait**  
Hair length (short)

**Trait State in Taxon A**  
Bos taurus

**Trait State in Taxon B**  
Bos taurus: slick; short hair

**Ancestral State**  
Taxon A

**Taxonomic Status**  
Domesticated

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

	Taxon A	Taxon B
<b>Latin Name</b>	<i>Bos taurus</i>	<i>Bos taurus</i>
<b>Common Name</b>	cattle	cattle
<b>Synonyms</b>	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
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<b>Parent</b>	Bos (oxen, cattle) - (Rank: genus)	Bos (oxen, cattle) - (Rank: genus)
<b>NCBI Taxonomy ID</b>	9913	9913
<b>is Taxon B an Intraspecies?</b>	Yes	Yes
<b>Taxon B Description</b>		Senepol

## 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

**UniProtKB** Bos taurus  
P01239

**GenebankID or UniProtKB**

GO:0010628 : positive regulation of gene expression  
GO:0051092 : positive regulation of NF-kappaB transcription factor activity  
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

Yes

#### Molecular Type

Coding

#### Aberration Type

Deletion

#### Deletion Size

1-9 bp

#### Molecular Details of the Mutation

1-bp deletion in exon 10 that introduces a premature stop codon (p.Leu462\*) and loss of 120 C-terminal amino acids from the long isoform of the receptor (ss1067289408; chr20:39136558GC>G)

#### Experimental Evidence

##### Linkage Mapping

##### 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

@Parallelism - <https://omia.org/OMIA000441/9913/>

