GEPHE SUMMARY

Gephebase Gene
Prolactin (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Prolactin^#gephebase-summary-title)

GepheID
GP00002032

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Hair length (long) (https://www.gephebase.org/search-criteria?/and+Trait=^Hair length (long)^#gephebase-summary-title)

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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)

Common Name
cattle

Synonyms
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

Rank
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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)

Common Name
cattle

Synonyms
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

Rank
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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
PRL

Synonyms
GHA1; Prol

String
9913.ENSBTAP00000020313 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9913.ENSBTAP00000020313)

Sequence Similarities
Belongs to the somatotropin/prolactin family.

GO - Molecular Function
GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179)
GO:0005148 : prolactin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005148)

GO - Biological Process
GO:0043066 : negative regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043066)

UniProtKB Bos taurus
P01239 (http://www.uniprot.org/uniprot/P01239)

GenebankID or UniProtKB
()

GO:0010629 : negative regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010629)
GO:0045807 : positive regulation of endocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0045807)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0010628 : positive regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0051092 : positive regulation of NF-kappaB transcription factor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0051092)
GO:0010751 : negative regulation of nitric oxide mediated signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0010751)
GO:0007565 : female pregnancy (https://www.ebi.ac.uk/QuickGO/term/GO:0007565)
GO:1901224 : positive regulation of NIK/NF-kappaB signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:1901224)
GO:0030072 : peptide hormone secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0030072)
GO:0023019 : signal transduction involved in regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0023019)
GO:0009612 : response to mechanical stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0009612)
GO:0032094 : response to food (https://www.ebi.ac.uk/QuickGO/term/GO:0032094)
GO:0046427 : positive regulation of JAK-STAT cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0046427)
GO:0048571 : long-day photoperiodism
(https://www.ebi.ac.uk/QuickGO/term/GO:0048571)
GO:0045429 : positive regulation of nitric oxide biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045429)
GO:0030879 : mammary gland development
(https://www.ebi.ac.uk/QuickGO/term/GO:0030879)
GO:0007595 : lactation (https://www.ebi.ac.uk/QuickGO/term/GO:0007595)
GO:1903489 : positive regulation of lactation
(https://www.ebi.ac.uk/QuickGO/term/GO:1903489)
GO:0031667 : response to nutrient levels
(https://www.ebi.ac.uk/QuickGO/term/GO:0031667)
GO:0009058 : biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0009058)
GO:0001825 : blastocyst formation (https://www.ebi.ac.uk/QuickGO/term/GO:0001825)
GO:0045723 : positive regulation of fatty acid biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045723)
GO:1903538 : regulation of meiotic cell cycle process involved in oocyte maturation
(https://www.ebi.ac.uk/QuickGO/term/GO:1903538)
GO:0043207 : response to external biotic stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0043207)
GO:1903576 : response to L-arginine (https://www.ebi.ac.uk/QuickGO/term/GO:1903576)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
GO:0005788 : endoplasmic reticulum lumen
(https://www.ebi.ac.uk/QuickGO/term/GO:0005788)
GO:0005789 : endoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005789)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

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

Experimental Evidence

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

	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) (https://pubmed.ncbi.nlm.nih.gov/25519203)

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 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Prolactin^/and+Taxon ID=^9913^/or+Gene Gephebase=^Prolactin^/and+Taxon ID=^9913^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

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