

GEPHE SUMMARY

<p>Prolactin (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~Prolactin^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002033</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology^#gephebase-summary-title)</p> <p>Hair length (short) (https://www.gephebase.org/search-criteria?/and+Trait=~Hair+length+(short)^#gephebase-summary-title)</p> <p>Bos taurus</p> <p>Bos taurus: slick; short hair</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated^#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Bos taurus</p> <p>(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+taurus^#gephebase-summary-title)</p> <p>Common Name</p> <p>cattle</p> <p>Synonyms</p> <p>Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Bos (oxen, cattle) - (Rank: genus)</p> <p>(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)</p> <p>NCBI Taxonomy ID</p> <p>9913</p> <p>(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Bos taurus</p> <p>(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+taurus^#gephebase-summary-title)</p> <p>Common Name</p> <p>cattle</p> <p>Synonyms</p> <p>Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Bos (oxen, cattle) - (Rank: genus)</p> <p>(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)</p> <p>NCBI Taxonomy ID</p> <p>9913</p> <p>(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Taxon B Description</p> <p>Senepol</p>
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GENOTYPIC CHANGE

<p>PRL</p> <p>GHA1; Prol</p> <p>9913.ENSBTAP00000020313</p> <p>(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9913.ENSBTAP00000020313)</p> <p>Belongs to the somatotropin/prolactin family.</p> <p>GO - Molecular Function</p> <p>GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179)</p> <p>GO:0005148 : prolactin receptor binding</p> <p>(https://www.ebi.ac.uk/QuickGO/term/GO:0005148)</p> <p>GO - Biological Process</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p>	<p>P01239 (http://www.uniprot.org/uniprot/P01239)</p> <p>0</p>	<p>UniProtKB Bos taurus</p> <p>GenebankID or UniProtKB</p>
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GO:0043066 : negative regulation of apoptotic process
<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>
 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

Yes (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title>)

Molecular Type

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

Aberration Type

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

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; chr20:39136558G>G)

Experimental Evidence

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

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 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=))

EXTERNAL LINKS

COMMENTS

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