

GEPHE SUMMARY

Gephebase Gene		GepheID	
Prolactin receptor (https://www.gephebase.org/search-criteria?/and+GeneGephebase=Prolactin+receptor#gephebase-summary-title)		GP00002282	
Entry Status		Martin	
Published		Main curator	

PHENOTYPIC CHANGE

Trait #1		Trait Category	
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=Morphology#gephebase-summary-title)		Trait	
Hair (slick ; short ; thermotolerance) (https://www.gephebase.org/search-criteria?/and+Trait=Hair+(slick+;+short+;+thermotolerance)#gephebase-summary-title)		Trait State in Taxon A	
WT hair		Trait State in Taxon B	
Slick hair (dominant) - Carora ; Romosinuano ; Senepol ; Limonero			

Trait #2		Trait Category	
Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=Physiology#gephebase-summary-title)		Trait	
Thermoregulation (tolerance to warm climate) (https://www.gephebase.org/search-criteria?/and+Trait=Thermoregulation+(tolerance+to+warm+climate)#gephebase-summary-title)		Trait State in Taxon A	
Non thermotolerant		Trait State in Taxon B	
Thermotolerant breed			

Ancestral State			
Taxon A		Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=Domesticated#gephebase-summary-title)			
Taxon A		Taxon B	
Latin Name		Latin Name	
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos+taurus#gephebase-summary-title)		Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos+taurus#gephebase-summary-title)	
Common Name		Common Name	
cattle		cattle	
Synonyms		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		Rank	
species		species	
Lineage		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		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		Parent	
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)		Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)		9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

PRLR	Generic Gene Name	P16471 (http://www.uniprot.org/uniprot/P16471)	UniProtKB Homo sapiens
	Synonyms	AAA51417 (https://www.ncbi.nlm.nih.gov/nucore/AAA51417)	GenebankID or UniProtKB
	String		
	Sequence Similarities		
HPRL; MFAB; hPRLr; RI-PRLR			
9606.ENSPO0000371432 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000371432)			
Belongs to the type I cytokine receptor family. Type 1 subfamily.			
GO - Molecular Function			
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)			
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)			
GO:0017046 : peptide hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017046)			
GO:0019955 : cytokine binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019955)			
GO:0004896 : cytokine receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004896)			
GO:0042978 : ornithine decarboxylase activator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042978)			
GO:0004925 : prolactin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004925)			
GO - Biological Process			
GO:0043066 : negative regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043066)			
GO:0007171 : activation of transmembrane receptor protein tyrosine kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0007171)			
GO:0007166 : cell surface receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0007166)			
GO:0120162 : positive regulation of cold-induced thermogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0120162)			
GO:0042976 : activation of Janus kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042976)			
GO:0060397 : JAK-STAT cascade involved in growth hormone signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0060397)			
GO:0042110 : T cell activation (https://www.ebi.ac.uk/QuickGO/term/GO:0042110)			
GO:0007566 : embryo implantation (https://www.ebi.ac.uk/QuickGO/term/GO:0007566)			
GO:0007595 : lactation (https://www.ebi.ac.uk/QuickGO/term/GO:0007595)			
GO:0006694 : steroid biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006694)			
GO - Cellular Component			
GO:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)			
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)			
GO:0043235 : receptor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0043235)			
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)			
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)			
GO:0009897 : external side of plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0009897)			
GO:0031904 : endosome lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0031904)			
Yes (#gephebase-summary-title)			Presumptive Null
Coding (#gephebase-summary-title)			Molecular Type
Deletion (#gephebase-summary-title)			Aberration Type
1-9 bp			Deletion Size
g.39136559delC p.Leu462*			Molecular Details of the Mutation
-			Experimental Evidence
Functionally reciprocal mutations of the prolactin signalling pathway define hairy and slick cattle. (2014) (https://pubmed.ncbi.nlm.nih.gov/25519203)			Main Reference
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			Authors
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.			Abstract
			Additional References

RELATED GEPHE

Related Genes

3 (KRT27, KRT71, Prolactin) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Hair/or+Taxon ID=^9913^/and+Trait=Thermoregulation/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

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

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001372/9913/> @AllelicSeries @Pleiotropy