

GEPHE SUMMARY

Agouti (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^Agouti^#gephebase-summary-title)	Gephebase Gene	GP00000059	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

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

GENOTYPIC CHANGE

Asip	Generic Gene Name	UniProtKB Mus musculus
As; ASP; A<y>; ASIP; a	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)	String	CAA68003 (https://www.ncbi.nlm.nih.gov/nuccore/CAA68003)
-	Sequence Similarities	
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779) GO:0031781 : type 3 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)	GO - Molecular Function	

GO:0031782 : type 4 melanocortin receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031782>)

GO - Biological Process

GO:0008343 : adult feeding behavior
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008343>)
GO:0006091 : generation of precursor metabolites and energy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006091>)
GO:0071514 : genetic imprinting (<https://www.ebi.ac.uk/QuickGO/term/GO:0071514>)
GO:0009755 : hormone-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
GO:0032438 : melanosome organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
GO:0032402 : melanosome transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032402>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0048023 : positive regulation of melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
GO:0040030 : regulation of molecular function, epigenetic
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040030>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005623 : cell (<https://www.ebi.ac.uk/QuickGO/term/GO:0005623>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Cis-regulatory~#gephebase-summary-title)	Molecular Type
Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Insertion~#gephebase-summary-title)	Aberration Type
1-10 kb	Insertion Size
LINE element insertion in 5' region of gene; over expression of alternative splice forms	Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	Experimental Evidence
The insertion of a full-length Bos taurus LINE element is responsible for a transcriptional deregulation of the Normande Agouti gene. (2006) (https://pubmed.ncbi.nlm.nih.gov/16827753)	Main Reference
Girardot M; Guibert S; Laforet MP; Gallard Y; Larroque H; Oulmouden A	Authors
Mammalian pigmentation is controlled by the concerted action of Tyr, Tyrp1 and Dct producing eumelanin and/or pheomelanin in melanocytes. The ratio of these two pigments is determined by the agonist alpha-melanocyte stimulating hormone and the antagonist Agouti protein acting on the Mc1r. Here we show that the Agouti gene is over-expressed in Normande breed compared with Prim'Holstein breed. The Normande cattle have a characteristic coat color phenotype with a variable presence of black (eumelanin) hair over a red/brown background. We have found a previously undescribed full-length L1-BT element inserted in the 5'-genomic sequence of the Agouti gene in Normande cattle which promotes the over-expression of alternative transcripts. The variable expression of the alternative transcript directed by the long interspersed nuclear element promoter may be the origin of the brindle coat color pattern of the Normande breed. This new bovine Agouti allele isolated in Normande breed has been named Abr. Finally, as ectopic over-expression of Agouti in Ay mice is responsible for the obesity syndrome, we discuss the possible consequences of Abr for meat and milk production in cattle.	Abstract
	Additional References

RELATED GEPHE

11 (coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PMEL17, SLC45A2=MATP, Twist2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9913~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@TE Cluster of paralogous transcription factors <https://omia.org/OMIA000201/9913/>

