

GEPHE SUMMARY

EDNRA (<a +ednra+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+EDNRA+"#gephebase-summary-title)	Gephebase Gene	GP00002170	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)	Trait Category		
Coloration (coat ; white-spotting) (<a +coloration+(coat+;+white-spotting)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(coat+;+white-spotting)+"#gephebase-summary-title)	Trait		
Goats with plain color	Trait State in Taxon A		
White-spotted	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Capra hircus (<a +capra+hircus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Capra+hircus+"#gephebase-summary-title)	Latin Name	Capra hircus (<a +capra+hircus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Capra+hircus+"#gephebase-summary-title)	Latin Name
goat	Common Name	goat	Common Name
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species	Synonyms	Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species	Synonyms
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; Caprinae; Capra	Lineage	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; Caprinae; Capra	Lineage
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)	Parent	Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)	Parent
9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)	NCBI Taxonomy ID	9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)	NCBI Taxonomy ID
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
Yes		Yes	
Boer goat	Taxon A Description	Boer goat	Taxon B Description

GENOTYPIC CHANGE

Ednra	Generic Gene Name	Q61614 (http://www.uniprot.org/uniprot/Q61614)	UniProtKB Mus musculus
ETa; ET-AR; AEA001; Gpcr10; Mhdaaea1	Synonyms	0	GenebankID or UniProtKB
10090.ENSMUSP00000034029 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000034029)	String		
Belongs to the G-protein coupled receptor 1 family. Endothelin receptor subfamily.	Sequence Similarities		
GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)	GO - Molecular Function		
GO:0004962 : endothelin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004962)			

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)

GO:0007507 : heart development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007507>)

GO:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)

GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)

GO:0008284 : positive regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)

GO:0001701 : in utero embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001701>)

GO:0043084 : penile erection (<https://www.ebi.ac.uk/QuickGO/term/GO:0043084>)

GO:0007204 : positive regulation of cytosolic calcium ion concentration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007204>)

GO:0071260 : cellular response to mechanical stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071260>)

GO:0032496 : response to lipopolysaccharide
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032496>)

GO:0050729 : positive regulation of inflammatory response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050729>)

GO:0008217 : regulation of blood pressure
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008217>)

GO:0042310 : vasoconstriction (<https://www.ebi.ac.uk/QuickGO/term/GO:0042310>)

GO:0001666 : response to hypoxia (<https://www.ebi.ac.uk/QuickGO/term/GO:0001666>)

GO:0048484 : enteric nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048484>)

GO:0001934 : positive regulation of protein phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001934>)

GO:0050678 : regulation of epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050678>)

GO:0019233 : sensory perception of pain
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019233>)

GO:0018108 : peptidyl-tyrosine phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018108>)

GO:0007193 : adenylyate cyclase-inhibiting G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007193>)

GO:0051482 : positive regulation of cytosolic calcium ion concentration involved in phospholipase C-activating G protein-coupled signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051482>)

GO:0007266 : Rho protein signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007266>)

GO:0001821 : histamine secretion (<https://www.ebi.ac.uk/QuickGO/term/GO:0001821>)

GO:0043278 : response to morphine (<https://www.ebi.ac.uk/QuickGO/term/GO:0043278>)

GO:0001569 : branching involved in blood vessel morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001569>)

GO:0014824 : artery smooth muscle contraction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014824>)

GO:0048144 : fibroblast proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0048144>)

GO:0003094 : glomerular filtration (<https://www.ebi.ac.uk/QuickGO/term/GO:0003094>)

GO:0060322 : head development (<https://www.ebi.ac.uk/QuickGO/term/GO:0060322>)

GO:0014032 : neural crest cell development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014032>)

GO:0051928 : positive regulation of calcium ion transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051928>)

GO:0090184 : positive regulation of kidney development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090184>)

GO:0090023 : positive regulation of neutrophil chemotaxis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090023>)

GO:0042482 : positive regulation of odontogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042482>)

GO:0051281 : positive regulation of release of sequestered calcium ion into cytosol
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051281>)

GO:0007205 : protein kinase C-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007205>)

GO:0010827 : regulation of glucose transmembrane transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010827>)

GO:0007585 : respiratory gaseous exchange by respiratory system
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007585>)

GO:0048659 : smooth muscle cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048659>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)

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

GO:0030315 : T-tubule (<https://www.ebi.ac.uk/QuickGO/term/GO:0030315>)

GO:0031965 : nuclear membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0031965>)

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

Presumptive Null

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

Molecular Type

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

Aberration Type

>1 Mb

Insertion Size

1â€‰Mb copy number variant (CNV) harboring 5 genes including EDNRA ; The duplicated EDNRA copies contain a missense variant (p.Tyr129His) predicted to increase the affinity of the encoded mutant receptor for endothelin 3. Menzi et al. proposed a hypothesis whereby ectopic overexpression of a mutant EDNRA scavenges EDN3 required for EDNRB signaling and normal melanocyte development and thus likely leads to an absence of melanocytes in the non-pigmented body areas of Boer goats (thanks to OMIA for the summary)

Molecular Details of the Mutation

Experimental Evidence

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

Main Reference

Genomic amplification of the caprine EDNRA locus might lead to a dose dependent loss of pigmentation. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27329507>)

Authors

Menzi F; Keller I; Reber I; Beck J; Brenig B; SchÃ¼tz E; Leeb T; DrÃ¶gemÃ¼ller C

Abstract

The South African Boer goat displays a characteristic white spotting phenotype, in which the pigment is limited to the head. Exploiting the existing phenotype variation within the breed, we mapped the locus causing this white spotting phenotype to chromosome 17 by genome wide association. Subsequent whole genome sequencing identified a 1â€‰Mb copy number variant (CNV) harboring 5 genes including EDNRA. The analysis of 358 Boer goats revealed 3 alleles with one, two, and three copies of this CNV. The copy number is correlated with the degree of white spotting in goats. We propose a hypothesis that ectopic overexpression of a mutant EDNRA scavenges EDN3 required for EDNRB signaling and normal melanocyte development and thus likely lead to an absence of melanocytes in the non-pigmented body areas of Boer goats. Our findings demonstrate the value of domestic animals as reservoir of unique mutants and for identifying a precisely defined functional CNV.

Additional References

RELATED GEPHE

3 (Agouti (ASIP), MC1R, tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9925^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

The causal role of EDNRA is speculative but testable and reasonable ; @Duplication @CNV <https://omia.org/OMIA002164/9925/> ; white-spotting phenotype without adverse health consequences.