GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00002170

Entry Status

Published

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

Coloration (coat ; white-spotting) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat ; white-spotting)^#gephebase-summary-title)

Trait State in Taxon A

Goats with plain color

Trait State in Taxon B

White-spotted

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)

Common Name

goat

Synonyms

Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent

Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID

9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Boer goat

Taxon B

Latin Name

Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Capra hircus^#gephebase-summary-title)

Common Name

goat

Synonyms

Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent

Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID

9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Boer goat

GENOTYPIC CHANGE

Generic Gene Name

Ednra

Synonyms

ETa; ET-AR; AEA001; Gpcr10; Mhdaaea1

String

10090.ENSMUSP00000034029 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000034029)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family. Endothelin receptor subfamily. EDNRA sub-subfamily.

GO - Molecular Function

GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)
GO:0004962 : endothelin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004962)

UniProtKB Mus musculus

Q61614 (http://www.uniprot.org/uniprot/Q61614)

GenebankID or UniProtKB

()

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 : adenylate 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)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Insertion Size

>1 Mb

Molecular Details of the Mutation

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)

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

Related Genes

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 Haplotypes

No matches found.

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.