GEPHE SUMMARY

Gephebase Gene
EDN3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EDN3^#gephebase-summary-title)

GepheID
GP00002182

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait #1

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

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

Trait State in Taxon A
Normal pigmentation of coat and eyes

Trait State in Taxon B
Domesticated llama and alpacas with white fleece ; introgression from alpacas to lamas

Trait #2

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

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

Trait State in Taxon A
Normal pigmentation of coat and eyes

Trait State in Taxon B
Domesticated llama and alpacas with blue eyes ; introgression from alpacas to lamas

Trait #3

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

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

Trait State in Taxon A
Normal pigmentation of coat and eyes

Trait State in Taxon B
Leucism with Deafness

Ancestral State
Taxon A

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

Taxon A

Latin Name
Vicugna vicugna (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Vicugna vicugna^#gephebase-summary-title)

Common Name
vicugna

Synonyms
Camelus vicugna; Lama vicugna; vicugna; vicuna; Vicugna vicugna (Molina, 1782)

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; Artiodactyla; Tylopoda; Camelidae; Vicugna

Parent
Vicugna () - (Rank: genus)

Taxon B #1

Latin Name
Lama glama (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Lama glama^#gephebase-summary-title)

Common Name
llama

Synonyms
Camelus glama; Lama guanicoe glama; llama; Lama glama (Linnaeus, 1758); Llama glama

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; Artiodactyla; Tylopoda; Camelidae; Lama

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30539)

NCBI Taxonomy ID

9843

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9843)

is Taxon A an Infraspecies?

No

Parent

Lama () - (Rank: genus)

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9839)

NCBI Taxonomy ID

9844

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9844)

is Taxon B an Infraspecies?

No

Taxon B #2

Latin Name

Vicugna pacos

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

Common Name

alpaca

Synonyms

Lama guanicoe pacos; Lama pacos; alpaca

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; Artiodactyla; Tylopoda; Camelidae; Vicugna

Parent

Vicugna () - (Rank: genus)

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30539)

NCBI Taxonomy ID

30538

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30538)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

EDN3

Synonyms

ET3; ET-3; WS4B; HSCR4; PPET3

String

9606.ENSP00000337128

(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000337128)

Sequence Similarities

Belongs to the endothelin/sarafotoxin family.

GO - Molecular Function

GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)
GO:0031708 : endothelin B receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031708)
GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179)

GO - Biological Process

GO:0007275 : multicellular organism development (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)
GO:0030182 : neuron differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030182)
GO:0001755 : neural crest cell migration (https://www.ebi.ac.uk/QuickGO/term/GO:0001755)
GO:0006874 : cellular calcium ion homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0006874)
GO:0007186 : G protein-coupled receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0008284 : positive regulation of cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0043406 : positive regulation of MAP kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0043406)
GO:0007267 : cell-cell signaling (https://www.ebi.ac.uk/QuickGO/term/GO:0007267)
GO:0045597 : positive regulation of cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0045597)
GO:0010460 : positive regulation of heart rate (https://www.ebi.ac.uk/QuickGO/term/GO:0010460)

UniProtKB Homo sapiens

P14138 (http://www.uniprot.org/uniprot/P14138)

GenebankID or UniProtKB

()

GO:0048070 : regulation of developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048070)
GO:0030593 : neutrophil chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0030593)
GO:0010468 : regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010468)
GO:0008015 : blood circulation (https://www.ebi.ac.uk/QuickGO/term/GO:0008015)
GO:0007166 : cell surface receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007166)
GO:0010961 : cellular magnesium ion homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0010961)
GO:0048016 : inositol phosphate-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0048016)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0030072 : peptide hormone secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0030072)
GO:0046887 : positive regulation of hormone secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0046887)
GO:0002690 : positive regulation of leukocyte chemotaxis
(https://www.ebi.ac.uk/QuickGO/term/GO:0002690)
GO:0045840 : positive regulation of mitotic nuclear division
(https://www.ebi.ac.uk/QuickGO/term/GO:0045840)
GO:1901381 : positive regulation of potassium ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:1901381)
GO:0003100 : regulation of systemic arterial blood pressure by endothelin
(https://www.ebi.ac.uk/QuickGO/term/GO:0003100)
GO:0019229 : regulation of vasoconstriction
(https://www.ebi.ac.uk/QuickGO/term/GO:0019229)
GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)
GO:0014826 : vein smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:0014826)

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)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

Introgression from alpacas to lamas and clear correlation wit phenotype but no information on the mutation(s)

Experimental Evidence

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

Main Reference

Genomic analysis of the domestication and post-Spanish conquest evolution of the llama and alpaca. (2020) (https://pubmed.ncbi.nlm.nih.gov/32616020)

Authors

Fan R; Gu Z; Guang X; MarÃn JC; Varas V; GonzÃlez BA; Wheeler JC; Hu Y; Li E; Sun X; Yang X; Zhang C; Gao W; He J; Munch K; Corbett-Detig R; Barbato M; Pan S; Zhan X; Bruford MW; Dong C

Abstract

Despite their regional economic importance and being increasingly reared globally, the origins and evolution of the llama and alpaca remain poorly understood. Here we report reference genomes for the llama, and for the guanaco and vicuÃ±a (their putative wild progenitors), compare these with the published alpaca genome, and resequence seven individuals of all four species to better understand domestication and introgression between the llama and alpaca.

Phylogenomic analysis confirms that the llama was domesticated from the guanaco and the alpaca from the vicuÃ±a. Introgression was much higher in the alpaca genome (36%) than the llama (5%) and could be dated close to the time of the Spanish conquest, approximately 500â€‰years ago. Introgression patterns are at their most variable on the X-chromosome of the alpaca, featuring 53 genes known to have deleterious X-linked phenotypes in humans. Strong genome-wide introgression signatures include olfactory receptor complexes into both species, hypertension resistance into alpaca, and fleece/fiber traits into llama. Genomic signatures of domestication in the llama include male reproductive traits, while in alpaca feature fleece characteristics, olfaction-related and hypoxia adaptation traits. Expression analysis of the introgressed region that is syntenic to human HSA4q21, a gene cluster previously associated with hypertension in humans under hypoxic conditions, shows a previously undocumented role for PRDM8 downregulation as a potential transcriptional regulation mechanism, analogous to that previously reported at high altitude for hypoxia-inducible factor 1Î±.

The unprecedented introgression signatures within both domestic camelid genomes may reflect post-conquest changes in agriculture and the breakdown of traditional management practices.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism @Introgression @Pleiotropy