GEPHE SUMMARY

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

GepheID
GP00000454

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

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

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

Trait State in Taxon A
Other mammals

Trait State in Taxon B
Lama spp.

Ancestral State
Data not curated

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

Taxon A

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

Common Name
mammals

Synonyms
mammals

Rank
class

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota

Parent
Amniota (amniotes) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32524)

NCBI Taxonomy ID
40674 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 40674)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
-

Synonyms
-

Rank
genus

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; Tylopoda; Camelidae

Parent
Camelidae () - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9835)

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

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBA1

Synonyms
HBH; ECYT7; HBA-T3; METHBA

String
9606.ENSP00000322421 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000322421)

UniProtKB Homo sapiens
P69905 (http://www.uniprot.org/uniprot/P69905)

Sequence Similarities
Belongs to the globin family.

GenebankID or UniProtKB
()

GO - Molecular Function
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)
GO:0005506 : iron ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005506)
GO:0005344 : oxygen carrier activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005344)
GO:0043177 : organic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043177)
GO:0019825 : oxygen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019825)

GO - Biological Process
GO:0006898 : receptor-mediated endocytosis

(https://www.ebi.ac.uk/QuickGO/term/GO:0006898)
GO:0042542 : response to hydrogen peroxide
(https://www.ebi.ac.uk/QuickGO/term/GO:0042542)
GO:0015701 : bicarbonate transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015701)
GO:0098869 : cellular oxidant detoxification
(https://www.ebi.ac.uk/QuickGO/term/GO:0098869)
GO:0042744 : hydrogen peroxide catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042744)
GO:0015671 : oxygen transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015671)
GO:0010942 : positive regulation of cell death
(https://www.ebi.ac.uk/QuickGO/term/GO:0010942)
GO:0051291 : protein heterooligomerization
(https://www.ebi.ac.uk/QuickGO/term/GO:0051291)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
GO:0072562 : blood microparticle (https://www.ebi.ac.uk/QuickGO/term/GO:0072562)
GO:0071682 : endocytic vesicle lumen
(https://www.ebi.ac.uk/QuickGO/term/GO:0071682)
GO:0022627 : cytosolic small ribosomal subunit
(https://www.ebi.ac.uk/QuickGO/term/GO:0022627)
GO:0031838 : haptoglobin-hemoglobin complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0031838)
GO:0005833 : hemoglobin complex (https://www.ebi.ac.uk/QuickGO/term/GO:0005833)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Asp122His

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Interaction of allosteric effectors with alpha-globin chains and high altitude respiration of mammals. The primary structure of two tylopoda hemoglobins with high oxygen affinity: vicuna (Lama vicugna) and alpaca (Lama pacos). (1986) (https://pubmed.ncbi.nlm.nih.gov/3964445)

Authors

Kleinschmidt T; MÃ¤rz J; JÃ¼rgens KD; Braunitzer G

Abstract

Polyacrylamide gel electrophoresis and ion-exchange chromatography revealed one hemoglobin component for vicuna (Lama vicugna) and alpaca (Lama pacos). Following chain separation by chromatography on carboxymethyl-cellulose, the amino-acid sequences were elucidated for the alpha- and beta-chains of both hemoglobins using automatic Edman degradation of the chains and the tryptic peptides. Vicuna and alpaca have identical beta-chains showing no substitutions to llama (Lama glama) either. In the alpha-chains alpaca differs from llama by the exchange of one amino-acid residue: alpha 122(H5)Asp----His. The same substitution is present in vicuna too, but in addition we found two more exchanges: alpha 10(A8)Ile----Val and alpha 130(H13)Ala----Thr. The close relationship between llama and alpaca suggests that they both originate from the wild guanaco, and there is no domesticated form of vicuna. The sequence data show that the higher oxygen affinity in vicuna compared to llama and alpaca must be due to the alpha-chains as the beta-chains are identical. The significance of the substitutions in alpha 122(H5), an alpha 1/beta 1-contact, and alpha 130(H13) is discussed.

Additional References

RELATED GEPHE

Related Genes

7 (BHLHE41, EGLN1, EPAS1, PPAR-alpha, hemoglobin; HBA and HBB, hemoglobin; HBA-T1 and T2 paralogues, hemoglobin; HBB-T1 and T2 paralogues) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^40674^/and+Trait=Hypoxia response/or+Taxon ID=^9839^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS