

## GEPHE SUMMARY

hemoglobin; HBA ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> )	Gephebase Gene	GP00000454	GepheID
Gephebase="hemoglobin; HBA"#gephebase-summary-title)			Main curator
Published	Entry Status	Martin	

## PHENOTYPIC CHANGE

Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> )	Trait Category		
Category="Physiology"#gephebase-summary-title)			
Hypoxia response ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=" hypoxia"="">https://www.gephebase.org/search-criteria?/and+Trait="Hypoxia</a> )	Trait		
response"#gephebase-summary-title)			
Other mammals	Trait State in Taxon A		
Lama spp.	Trait State in Taxon B		
Data not curated	Ancestral State		
Interspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> )	Taxonomic Status		
Status="Interspecific"#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Mammalia	Lama		
( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> )	( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> )		
Synonyms="Mammalia"#gephebase-summary-title)	Synonyms="Lama"#gephebase-summary-title)		
mammals	Common Name	-	Common Name
mammals	Synonyms	-	Synonyms
class	Rank	genus	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;	Lineage
Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;		Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;	
Dipnotetrapodomorpha; Tetrapoda; Amniota	Parent	Laurasiatheria; Cetartiodactyla; Tylopoda; Camelidae	Parent
Amniota (amniotes) - (Rank: no rank)	NCBI Taxonomy ID	Camelidae () - (Rank: family)	NCBI Taxonomy ID
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32524">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32524</a> )	40674	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9835">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9835</a> )	9839
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=40674">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=40674</a> )	is Taxon A an Intraspecies?	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9839">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9839</a> )	is Taxon B an Intraspecies?
No		No	

## GENOTYPIC CHANGE

HBA1	Generic Gene Name	P69905 ( <a href="http://www.uniprot.org/uniprot/P69905">http://www.uniprot.org/uniprot/P69905</a> )	UniProtKB Homo sapiens
HBH; ECTY7; HBA-T3; METHBA	Synonyms	()	GenebankID or UniProtKB
9606.ENSPP00000322421	String		
( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000322421">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000322421</a> )			
Belongs to the globin family.	Sequence Similarities		
GO:0020037 : heme binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0020037">https://www.ebi.ac.uk/QuickGO/term/GO:0020037</a> )	GO - Molecular Function		
GO:0005506 : iron ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005506">https://www.ebi.ac.uk/QuickGO/term/GO:0005506</a> )			
GO:0005344 : oxygen carrier activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005344">https://www.ebi.ac.uk/QuickGO/term/GO:0005344</a> )			
GO:0043177 : organic acid binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043177">https://www.ebi.ac.uk/QuickGO/term/GO:0043177</a> )			
GO:0019825 : oxygen binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0019825">https://www.ebi.ac.uk/QuickGO/term/GO:0019825</a> )	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; MÄrz J; JÄrgens 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