

GEPHE SUMMARY

hemoglobin; HBA2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~hemoglobin; HBA2^#gephebase-summary-title)	Gephebase Gene	GP00000462	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Physiology^#gephebase-summary-title)	Trait Category		
Hypoxia response (https://www.gephebase.org/search-criteria?/and+Trait=~Hypoxia+response^#gephebase-summary-title)	Trait		
Anser anser	Trait State in Taxon A		
Anser indicus - high altitude	Trait State in Taxon B		
Taxon A	Ancestral State		
Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Interspecific^#gephebase-summary-title)	Taxonomic Status		

Taxon A		Taxon B	
	Latin Name		Latin Name
Anser anser (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Anser+anser^#gephebase-summary-title)	Anser indicus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Anser+indicus^#gephebase-summary-title)		
domestic goose	bar-headed goose		
domestic goose; graylag goose; greylag goose; Anser anser (Linnaeus, 1758)	Eulabeia indica; Eulabia indica; bar-headed goose; Anser indicus (Latham, 1790)		
species	species		
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Anseriformes; Anatidae; Anserinae; Anser	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Anseriformes; Anatidae; Anserinae; Anser		
Anser (geese) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8842)	Anser (geese) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8842)		
8843 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8843)	8846 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8846)		
No	No	is Taxon A an Intraspecies?	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

HBA1	Generic Gene Name	P69905 (http://www.uniprot.org/uniprot/P69905)	UniProtKB Homo sapiens
HBH; ECT7; HBA-T3; METHBA	Synonyms	GQ271003.1 (https://www.ncbi.nlm.nih.gov/nucleotide/GQ271003.1)	GenebankID or UniProtKB
9606.ENSPO0000322421 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000322421)	String		
Belongs to the globin family.	Sequence Similarities		
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)	GO - Molecular Function		
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

Pro119Ala

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

Adaptation of bird hemoglobins to high altitudes: demonstration of molecular mechanism by protein engineering. (1991) (https://pubmed.ncbi.nlm.nih.gov/1862080)

Authors

Jessen TH; Weber RE; Fermi G; Tame J; Braunitzer G

Abstract

Of two closely related species of geese, one, the greylag goose, lives in the Indian plains all year round, while the other, the bar-headed goose, lives at the Tibetan lakes and migrates across the Himalayas to winter in India. Another species, the Andean goose, lives in the High Andes all year round. Possession of a Hb with high oxygen affinity helps to adapt bar-headed and Andean geese to high altitudes. The Hb amino acid sequences of the bar-headed and the greylag geese differ by four substitutions, of which only one is unique among bird sequences: Pro-119 alpha (H2)----Ala. Perutz proposed that the two-carbon gap left by this substitution at the alpha 1 beta 1 contact raises the oxygen affinity, because it relaxes the tension in the deoxy or T structure [Perutz, M. F. (1983) Mol. Biol. Evol. 1, 1-28]. It was later found that the Hb of the Andean goose has a gap in the same position, due to the complementary substitution Leu-55 beta (D6)----Ser. We have tested Perutz's hypothesis by introducing each of these substitutions into human globin synthesized in *Escherichia coli*. The reconstituted Hbs combine cooperatively with oxygen. Their oxygen affinities exceed that of normal human Hb by an even larger factor than that found between the high-flying geese and the greylag goose. The mutant Hb Met-55 beta (D6)----Ser was crystallized. Its structure is the same as that of HbA, except in the immediate environment of the gap left by the substitution of the serine for the methionine side chain, which evidently causes the increased oxygen affinity of this Hb.

Additional References

Parallel evolution in the major haemoglobin genes of eight species of Andean waterfowl. (2009) (https://pubmed.ncbi.nlm.nih.gov/19754505)

Phylogenetic and structural analysis of the HbA (alphaA/betaA) and HbD (alphaD/betaA) hemoglobin genes in two high-altitude waterfowl from the Himalayas and the Andes: Bar-headed goose (*Anser indicus*) and Andean goose (*Chloephaga melanoptera*). (2010) (https://pubmed.ncbi.nlm.nih.gov/20434566)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Needs curation