

GEPHE SUMMARY

hemoglobin; HBB (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~hemoglobin; HBB~#gephebase-summary-title)	Gephebase Gene	GP00000473	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Physiology~#gephebase-summary-title)		Trait Category	
Hypoxia response (https://www.gephebase.org/search-criteria?/and+Trait=~Hypoxia response~#gephebase-summary-title)		Trait	
Neochen jubata		Trait State in Taxon A	
Chloephaga melanoptera - high altitude		Trait State in Taxon B	
Taxon A		Ancestral State	
Interspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Interspecific~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Neochen jubata (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Neochen jubata~#gephebase-summary-title)		Chloephaga melanoptera (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Chloephaga melanoptera~#gephebase-summary-title)	
Orinoco goose		Andean goose	
Neochen jubatus; Orinoco goose; Neochen jubata (Spix, 1825)		Andean goose	
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; Tadorinae; Neochen		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; Tadorinae; Chloephaga	
Neochen () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8881)		Chloephaga () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8859)	
8882 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8882)		8860 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8860)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

HBB	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	P68871 (http://www.uniprot.org/uniprot/P68871)
	String	GenebankID or UniProtKB
ECYT6; CD113t-C; beta-globin		GQ271792.1 (https://www.ncbi.nlm.nih.gov/nuccore/GQ271792.1)
9606.ENSPP00000333994 (http://string-db.org/newstring_cg/show_network_section.pl?identifier=9606.ENSPP00000333994)		
Belongs to the globin family.		
GO - Molecular Function		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)		
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)		
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:0031721 : hemoglobin alpha binding		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031721>)
GO:0030492 : hemoglobin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0030492>)
GO - Biological Process

GO:0006898 : receptor-mediated endocytosis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006898>)
GO:0007596 : blood coagulation (<https://www.ebi.ac.uk/QuickGO/term/GO:0007596>)
GO:0008217 : regulation of blood pressure
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008217>)
GO:0042542 : response to hydrogen peroxide
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042542>)
GO:0043312 : neutrophil degranulation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043312>)
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:0030185 : nitric oxide transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0030185>)
GO:0070527 : platelet aggregation (<https://www.ebi.ac.uk/QuickGO/term/GO:0070527>)
GO:0045429 : positive regulation of nitric oxide biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045429>)
GO:0050880 : regulation of blood vessel size
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050880>)
GO:0070293 : renal absorption (<https://www.ebi.ac.uk/QuickGO/term/GO:0070293>)

GO - Cellular Component

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
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: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>)
GO:1904813 : ficolin-1-rich granule lumen
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904813>)
GO:1904724 : tertiary granule lumen (<https://www.ebi.ac.uk/QuickGO/term/GO:1904724>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title)	Aberration Type
	SNP Coding Change
Nonsynonymous	
	Molecular Details of the Mutation
Leu55Ser	
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	Experimental Evidence

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

Adaptation of bird hemoglobins to high altitudes: demonstration of molecular mechanism by protein engineering. (1991) (https://pubmed.ncbi.nlm.nih.gov/1862080)	Main Reference
Jessen TH; Weber RE; Fermi G; Tame J; Braunitzer G	Authors

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.

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
1 (hemoglobin; HBA2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~8882~/and+Trait=Hypoxia response/or+Taxon ID=~8860~/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)	
	Related Haplotypes
2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~hemoglobin; HBB~/and+Taxon ID=~8882~/or+Gene Gephebase=~hemoglobin; HBB~/and+Taxon ID=~8860~#gephebase-summary-title)	

EXTERNAL LINKS

COMMENTS

Needs curation