GEPHE SUMMARY

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

GepheID
GP00000473

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
Neochen jubata

Trait State in Taxon B
Chloephaga melanoptera - high altitude

Ancestral State
Taxon A

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

Taxon A

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

Common Name
Orinoco goose

Synonyms
Neochen jubatus; Orinoco goose; Neochen jubata (Spix, 1825)

Rank
species

Lineage
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; Tadorninae; Neochen

Parent
Neochen () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8881)

NCBI Taxonomy ID
8882 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8882)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
Andean goose

Synonyms
Andean goose

Rank
species

Lineage
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; Tadorninae; Chloephaga

Parent
Chloephaga () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8859)

NCBI Taxonomy ID
8860 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8860)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBB

Synonyms
ECYT6; CD113t-C; beta-globin

String
9606.ENSP00000333994 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000333994)

Sequence Similarities
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

UniProtKB Homo sapiens
P68871 (http://www.uniprot.org/uniprot/P68871)

GenebankID or UniProtKB
GQ271792.1 (https://www.ncbi.nlm.nih.gov/nuccore/GQ271792.1)

(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)

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

Leu55Ser

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

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