

## GEPHE SUMMARY

hemoglobin; HBB ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~hemoglobin;+HBB^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~hemoglobin;+HBB^#gephebase-summary-title</a> )	Gephebase Gene	GP00000474	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

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

## GENOTYPIC CHANGE

HBB	Generic Gene Name	P68871 ( <a href="http://www.uniprot.org/uniprot/P68871">http://www.uniprot.org/uniprot/P68871</a> )	UniProtKB Homo sapiens
ECYT6; CD113t-C; beta-globin	Synonyms	()	GenebankID or UniProtKB
9606.ENSPP00000333994 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000333994">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000333994</a> )	String		
Belongs to the globin family.	Sequence Similarities		
GO:0046872 : metal ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a> )	GO - Molecular Function		
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: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: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>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ala86Ser

Experimental Evidence

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

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

Main Reference

Convergent Evolution of Hemoglobin Function in High-Altitude Andean Waterfowl Involves Limited Parallelism at the Molecular Sequence Level. (2015)  
 (<https://pubmed.ncbi.nlm.nih.gov/26637114>)

Authors

Natarajan C; Projecto-Garcia J; Moriyama H; Weber RE; Muñoz-Fuentes V; Green AJ; Kopuchian C; Tubaro PL; Alza L; Bulgarella M; Smith MM; Wilson RE; Fago A; McCracken KG; Storz JF

Abstract

A fundamental question in evolutionary genetics concerns the extent to which adaptive phenotypic convergence is attributable to convergent or parallel changes at the molecular sequence level. Here we report a comparative analysis of hemoglobin (Hb) function in eight phylogenetically replicated pairs of high- and low-altitude waterfowl taxa to test for convergence in the oxygenation properties of Hb, and to assess the extent to which convergence in biochemical phenotype is attributable to repeated amino acid replacements. Functional experiments on native Hb variants and protein engineering experiments based on site-directed mutagenesis revealed the phenotypic effects of specific amino acid replacements that were responsible for convergent increases in Hb-O<sub>2</sub> affinity in multiple high-altitude taxa. In six of the eight taxon pairs, high-altitude taxa evolved derived increases in Hb-O<sub>2</sub> affinity that were caused by a combination of unique replacements, parallel replacements (involving identical-by-state variants with independent mutational origins in different lineages), and collateral replacements (involving shared, identical-by-descent variants derived via introgressive hybridization). In genome scans of nucleotide differentiation involving high- and low-altitude populations of three separate species, function-altering amino acid polymorphisms in the globin genes emerged as highly significant outliers, providing independent evidence for adaptive divergence in Hb function. The

experimental results demonstrate that convergent changes in protein function can occur through multiple historical paths, and can involve multiple possible mutations. Most cases of convergence in Hb function did not involve parallel substitutions and most parallel substitutions did not affect Hb-O<sub>2</sub> affinity, indicating that the repeatability of phenotypic evolution does not require parallelism at the molecular level.

Additional References

## RELATED GEPHE

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](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 Genes

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>)

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

Needs curation