

## GEPHE SUMMARY

hemoglobin; HBA2 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~hemoglobin;+HBA2^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~hemoglobin;+HBA2^#gephebase-summary-title</a> )	Gephebase Gene	GP00000463	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		
Lophonetta specularioides- low-altitude	Trait State in Taxon A		
Lophonetta specularioides- High-altitude	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific^#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Lophonetta specularioides ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Lophonetta+specularioides^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Lophonetta+specularioides^#gephebase-summary-title</a> )	Latin Name	Lophonetta specularioides ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Lophonetta+specularioides^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Lophonetta+specularioides^#gephebase-summary-title</a> )	Latin Name
crested duck	Common Name	crested duck	Common Name
Anas specularioides; Lophonetta specularioides; crested duck; Lophonetta specularioides (King, 1828)	Synonyms	Anas specularioides; Lophonetta specularioides; crested duck; Lophonetta specularioides (King, 1828)	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; Anatinae; Lophonetta	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; Anatinae; Lophonetta	Lineage
Lophonetta () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75872">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75872</a> )	Parent	Lophonetta () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75872">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75872</a> )	Parent
75873 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75873">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75873</a> )	NCBI Taxonomy ID	75873 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75873">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75873</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## 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; ECYT7; HBA-T3; METHBA	Synonyms	ACT80420 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/ACT80420">https://www.ncbi.nlm.nih.gov/nuccore/ACT80420</a> )	GenebankID or UniProtKB
9606.ENSPP00000322421 ( <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> )	String		
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: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>)

- 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
- Nonsynonymous SNP Coding Change
- Ala5Thr Molecular Details of the Mutation
- 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	-	-	-

- Parallel evolution in the major haemoglobin genes of eight species of Andean waterfowl. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19754505>) Main Reference
- McCracken KG; Barger CP; Bulgarella M; Johnson KP; Sonsthagen SA; Trucco J; Valqui TH; Wilson RE; Winker K; Sorenson MD Authors
- Theory predicts that parallel evolution should be common when the number of beneficial mutations is limited by selective constraints on protein structure. However, confirmation is scarce in natural populations. Here we studied the major haemoglobin genes of eight Andean duck lineages and compared them to 115 other waterfowl species, including the bar-headed goose (*Anser indicus*) and Abyssinian blue-winged goose (*Cyanochen cyanopterus*), two additional species living at high altitude. One to five amino acid replacements were significantly overrepresented or derived in each highland population, and parallel substitutions were more common than in simulated sequences evolved under a neutral model. Two substitutions evolved in parallel in the alpha A subunit of two (Ala-alpha 8) and five (Thr-alpha 77) taxa, and five identical beta A subunit substitutions were observed in two (Ser-beta 4, Glu-beta 94, Met-beta 133) or three (Ser-beta 13, Ser-beta 116) taxa. Substitutions at adjacent sites within the same functional protein region were also observed. Five such replacements were in exterior, solvent-accessible positions on the A helix and AB corner of the alpha A subunit. Five others were in close proximity to inositolpentaphosphate binding sites, and two pairs of independent replacements occurred at two different alpha(1)beta(1) intersubunit contacts. More than half of the substitutions in highland lineages resulted in the acquisition of serine or threonine (18 gains vs. 2 losses), both of which possess a hydroxyl group that can hydrogen bond to a variety of polar substrates. The patterns of parallel evolution observed in these waterfowl suggest that adaptation to high-altitude hypoxia has resulted from selection on unique but overlapping sets of one to five amino acid substitutions in each lineage. Abstract
- 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>) Additional References

RELATED GEPHE

- 1 (hemoglobin; HBB) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~75873~/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title>) Related Genes
- No matches found. Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

Needs curation