

GEPHE SUMMARY

hemoglobin; HBA2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~hemoglobin; HBA2^#gephebase-summary-title)	Gephebase Gene	GP00000460	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
Anas versicolor	Trait State in Taxon A
Anas puna	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	Latin Name	Taxon B	Latin Name
Anas versicolor (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Anas+versicolor^#gephebase-summary-title)	Anas puna (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Anas+puna^#gephebase-summary-title)		
-	Common Name	-	Common Name
-	Synonyms	-	Synonyms
species	Rank	Anas versicolor puna	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; Anas	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; Anas	Lineage
Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)	Parent	Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)	Parent
75863 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75863)	NCBI Taxonomy ID	75855 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75855)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

HBA1	Generic Gene Name	P69905 (http://www.uniprot.org/uniprot/P69905)	UniProtKB Homo sapiens
HBH; ECYT7; HBA-T3; METHBA	Synonyms	ACT80680 (https://www.ncbi.nlm.nih.gov/nuccore/ACT80680)	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

Ala77Thr

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

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

Authors

McCracken KG; Barger CP; Bulgarella M; Johnson KP; Sonsthagen SA; Trucco J; Valqui TH; Wilson RE; Winker K; Sorenson MD

Abstract

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.

Additional References

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)

RELATED GEPHE

Related Genes

1 (hemoglobin; HBB) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^75863^/and+Trait=Hypoxia response/or+Taxon ID=^75855^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephabase=^hemoglobin; HBA2^/and+Taxon ID=^75863^/or+Gene Gephabase=^hemoglobin; HBA2^/and+Taxon ID=^75855^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

Needs curation