

## GEPHE SUMMARY

	Gephebase Gene	GephelD
hemoglobin; HBA2 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a>	GP00000459	
Gephebase=^hemoglobin; HBA2" #gephebase-summary-title)		Main curator
	Entry Status	Martin
Published		

## PHENOTYPIC CHANGE

	Trait Category	
Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a>	Trait	
Category="Physiology" #gephebase-summary-title)		
Hypoxia response ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Hypoxia">https://www.gephebase.org/search-criteria?/and+Trait=^Hypoxia</a>	Trait State in Taxon A	
response" #gephebase-summary-title)		
Anas flavirostris flavirostris	Trait State in Taxon B	
Anas flavirostris oxyptera	Ancestral State	
Taxon A	Taxonomic Status	
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a>		
Status="Intraspecific" #gephebase-summary-title)		
Taxon A	Latin Name	Taxon B
Anas flavirostris	( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Anas+flavirostris">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Anas+flavirostris" #gephebase-summary-title)</a>	Anas flavirostris
Yellow-billed teal	Common Name	
Yellow-billed teal; speckled teal; Anas flavirostris Vieillot, 1816	Synonyms	
species	Rank	
	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		
	Parent	
Anas (ducks) - (Rank: genus)		
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835</a> )		
75845	NCBI Taxonomy ID	
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75845">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75845</a> )		
Yes	is Taxon A an Infraspecies?	
Anas flavirostris flavirostris	Taxon A Description	
		Taxon B Description
		Anas flavirostris oxyptera

## GENOTYPIC CHANGE

HBAD	Generic Gene Name	UniProtKB Gallus gallus
HBA2; HBAD	Synonyms	GenebankID or UniProtKB
9031.ENSGALP00000012054	String	0
( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9031.ENSGALP00000012054">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9031.ENSGALP00000012054</a> )	Sequence Similarities	
Belongs to the globin family.	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: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 (<https://www.ebi.ac.uk/QuickGO/term/GO:0019825>)  
GO - Biological Process

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:0051291 : protein heterooligomerization

(<https://www.ebi.ac.uk/QuickGO/term/GO:0051291>)

GO - Cellular Component

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=%27No%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27No%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27SNP%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27SNP%27#gephebase-summary-title))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ser130Phe

Experimental Evidence

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Candidate Gene%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Candidate%20Gene%27#gephebase-summary-title))

	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

Related Genes

3 (EGLN1, EPAS1, hemoglobin; HBB) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=%2775845%27/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon%20ID=%2775845%27/and+Trait=Hypoxia%20response/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=%27hemoglobin;%20HBA2%27/and+Taxon ID=%2775845%27/or+Gene Gephebase=%27hemoglobin;%20HBA2%27/and+Taxon ID=%2775845%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene%20Gephebase=%27hemoglobin;%20HBA2%27/and+Taxon%20ID=%2775845%27/or+Gene%20Gephebase=%27hemoglobin;%20HBA2%27/and+Taxon%20ID=%2775845%27#gephebase-summary-title))

## EXTERNAL LINKS

## COMMENTS

Needs curation

