## GEPHE SUMMARY

Gephebase Gene

hemoglobin; HBA2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^hemoglobin; HBA2^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000464

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

Merganetta armata - low altitude

Trait State in Taxon B

Merganetta armata - high-altitude

Ancestral State

Taxon A

Taxonomic Status

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

### Taxon A

Latin Name

Merganetta armata (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Merganetta armata^#gephebase-summary-title)

Common Name

-

Synonyms

-

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; Merganetta

Parent

Merganetta () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 658918 )

NCBI Taxonomy ID

658919 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 658919 )

is Taxon A an Infraspecies?

No

### Taxon B

Latin Name

Merganetta armata (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Merganetta armata^#gephebase-summary-title)

Common Name

-

Synonyms

-

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; Merganetta

Parent

Merganetta () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 658918 )

NCBI Taxonomy ID

658919 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 658919 )

is Taxon B an Infraspecies?

No

## GENOTYPIC CHANGE

Generic Gene Name

HBA1

Synonyms

HBH; ECYT7; HBA-T3; METHBA

String

9606.ENSP00000322421 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000322421 )

Sequence Similarities

Belongs to the globin family.

GO - Molecular Function

GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)
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

UniProtKB Homo sapiens

P69905 (http://www.uniprot.org/uniprot/P69905)

GenebankID or UniProtKB

()

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

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; MuÃ±oz-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-O2 affinity in multiple high-altitude taxa. In six of the eight taxon pairs, high-altitude taxa evolved derived increases in Hb-O2 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-O2 affinity, indicating that the repeatability of phenotypic evolution does not require parallelism at the molecular level.

Additional References

## RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

Needs curation