GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00000465

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

Neochen jubata

Trait State in Taxon B

Chloephaga melanoptera - high altitude

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

Orinoco goose

Synonyms

Neochen jubatus; Orinoco goose; Neochen jubata (Spix, 1825)

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; Neochen

Parent

Neochen () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8881)

NCBI Taxonomy ID

8882 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8882)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

Andean goose

Synonyms

Andean goose

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; Chloephaga

Parent

Chloephaga () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8859)

NCBI Taxonomy ID

8860 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8860)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

HBAD

Synonyms

HBA2; HBAD

String

9031.ENSGALP00000012054 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9031.ENSGALP00000012054)

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 Gallus gallus

P02001 (http://www.uniprot.org/uniprot/P02001)

GenebankID or UniProtKB

()

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)

Mutation #1

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

Ile10Leu + Val96Ala

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	Ile	Leu	10

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

Mutation #2

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

Ile10Leu + Val96Ala

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	Val	Ala	96

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

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Additional References

RELATED GEPHE

Related Genes

1 (hemoglobin; HBB) (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 Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Needs curation @SeveralMutationsWithEffect