GEPHE SUMMARY

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

GepheID
GP00000472

Entry Status
Published

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
Lophonetta specularioides- low-altitude

Trait State in Taxon B
Lophonetta specularioides- 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
Lophonetta specularioides (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Lophonetta specularioides^#gephebase-summary-title)

Common Name
crested duck

Synonyms
Anas specularioides; Lophonetta specularoides; crested duck; Lophonetta specularioides (King, 1828)

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; Anatinae; Lophonetta

Parent
Lophonetta () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75872)

NCBI Taxonomy ID
75873 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75873)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Lophonetta specularioides (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Lophonetta specularioides^#gephebase-summary-title)

Common Name
crested duck

Synonyms
Anas specularioides; Lophonetta specularoides; crested duck; Lophonetta specularioides (King, 1828)

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; Anatinae; Lophonetta

Parent
Lophonetta () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75872)

NCBI Taxonomy ID
75873 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75873)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBB

Synonyms
ECYT6; CD113t-C; beta-globin

String
9606.ENSP00000333994 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000333994)

Sequence Similarities
Belongs to the globin family.

GO - Molecular Function
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)
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)

UniProtKB Homo sapiens
P68871 (http://www.uniprot.org/uniprot/P68871)

GenebankID or UniProtKB
()

GO:0031721 : hemoglobin alpha binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031721)
GO:0030492 : hemoglobin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030492)

GO - Biological Process

GO:0006898 : receptor-mediated endocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006898)
GO:0007596 : blood coagulation (https://www.ebi.ac.uk/QuickGO/term/GO:0007596)
GO:0008217 : regulation of blood pressure
(https://www.ebi.ac.uk/QuickGO/term/GO:0008217)
GO:0042542 : response to hydrogen peroxide
(https://www.ebi.ac.uk/QuickGO/term/GO:0042542)
GO:0043312 : neutrophil degranulation
(https://www.ebi.ac.uk/QuickGO/term/GO:0043312)
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:0030185 : nitric oxide transport (https://www.ebi.ac.uk/QuickGO/term/GO:0030185)
GO:0070527 : platelet aggregation (https://www.ebi.ac.uk/QuickGO/term/GO:0070527)
GO:0045429 : positive regulation of nitric oxide biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045429)
GO:0050880 : regulation of blood vessel size
(https://www.ebi.ac.uk/QuickGO/term/GO:0050880)
GO:0070293 : renal absorption (https://www.ebi.ac.uk/QuickGO/term/GO:0070293)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
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: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)
GO:1904813 : ficolin-1-rich granule lumen
(https://www.ebi.ac.uk/QuickGO/term/GO:1904813)
GO:1904724 : tertiary granule lumen (https://www.ebi.ac.uk/QuickGO/term/GO:1904724)

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

Asp94Glu

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

1 (hemoglobin; HBA2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^75873^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^hemoglobin; HBB^/and+Taxon ID=^75873^/or+Gene Gephebase=^hemoglobin; HBB^/and+Taxon ID=^75873^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

Needs curation