

GEPHE SUMMARY

Gephebase Gene
hemoglobin; HBB

Entry Status
Published

GepheID
GP00000470

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Hypoxia response

Trait State in Taxon A
Anas spp.; other crested ducks

Trait State in Taxon B
Lophonetta specularioides

Ancestral State
Taxon A

Taxonomic Status
Interspecific

Taxon A

Latin Name
Anatidae

Common Name
waterfowl

Synonyms
waterfowl

Rank
family

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

Parent
Anseriformes () - (Rank: order)

NCBI Taxonomy ID
8830

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Lophonetta specularioides

Common Name
crested duck

Synonyms
Anas specularioides; Lophonetta specularioides; 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)

NCBI Taxonomy ID
75873

is Taxon B an Intraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBB

Synonyms
ECYT6; CD113t-C; beta-globin

String
9606.ENSP00000333994

Sequence Similarities
Belongs to the globin family.

GO - Molecular Function
GO:0046872 : metal ion binding
GO:0020037 : heme binding
GO:0005344 : oxygen carrier activity
GO:0043177 : organic acid binding
GO:0019825 : oxygen binding
GO:0031721 : hemoglobin alpha binding
GO:0030492 : hemoglobin binding

GO - Biological Process

UniProtKB Homo sapiens
P68871

GenebankID or UniProtKB
ACT80420

GO:0006898 : receptor-mediated endocytosis
 GO:0007596 : blood coagulation
 GO:0008217 : regulation of blood pressure
 GO:0042542 : response to hydrogen peroxide
 GO:0045312 : neutrophil degranulation
 GO:0015701 : bicarbonate transport
 GO:0098869 : cellular oxidant detoxification
 GO:0042744 : hydrogen peroxide catabolic process
 GO:0015671 : oxygen transport
 GO:0010942 : positive regulation of cell death
 GO:0051291 : protein heterooligomerization
 GO:0030185 : nitric oxide transport
 GO:0070527 : platelet aggregation
 GO:0045429 : positive regulation of nitric oxide biosynthetic process
 GO:0050880 : regulation of blood vessel size
 GO:0070293 : renal absorption

GO - Cellular Component

GO:0005829 : cytosol
 GO:0070062 : extracellular exosome
 GO:0005576 : extracellular region
 GO:0005615 : extracellular space
 GO:0072562 : blood microparticle
 GO:0071682 : endocytic vesicle lumen
 GO:0031838 : haptoglobin-hemoglobin complex
 GO:0005833 : hemoglobin complex
 GO:1904813 : ficolin-1-rich granule lumen
 GO:1904724 : tertiary granule lumen

Mutation #1

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Thr4Ser

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Thr	Ser	4

Main Reference

Parallel evolution in the major haemoglobin genes of eight species of Andean waterfowl. (2009)

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

Mutation #2

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Asp94Glu

Experimental Evidence

[Candidate Gene](#)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Asp	Glu	94

Main Reference

[Parallel evolution in the major haemoglobin genes of eight species of Andean waterfowl. \(2009\)](#)

Authors

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Additional References

RELATED GEPHE

Related Genes

4 ([EGLN1](#), [EPAS1](#), [EGNL1](#), [hemoglobin; HBA2](#))

Related Haplotypes

8

EXTERNAL LINKS

COMMENTS

Needs curation @SeveralMutationsWithEffect