

GEPHE SUMMARY

Gephebase Gene
EPAS1

Entry Status
Published

GepheID
GP00002131

Main curator
Courtier

PHENOTYPIC CHANGE

Trait #1
Trait Category
Physiology
Trait
High-altitude adaptation
Trait State in Taxon A
low altitude
Trait State in Taxon B
high altitude

Trait #2
Trait Category
Physiology
Trait
Hypoxia response
Trait State in Taxon A
low altitude
Trait State in Taxon B
high altitude

Ancestral State
Taxon A

Taxonomic Status
Intraspecific

Taxon A

Latin Name
Anas cyanoptera

Common Name
cinnamon teal

Synonyms
cinnamon teal; *Anas cyanoptera* Vieillot, 1816

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; Anas

Parent
Anas (ducks) - (Rank: genus)

NCBI Taxonomy ID
75840

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Anas cyanoptera

Common Name
cinnamon teal

Synonyms
cinnamon teal; *Anas cyanoptera* Vieillot, 1816

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; Anas

Parent
Anas (ducks) - (Rank: genus)

NCBI Taxonomy ID
75840

is Taxon B an Intraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
EPAS1

UniProtKB Homo sapiens
Q99814

Synonyms

HLF; MOP2; ECVT4; HIF2A; PASD2; bHLHe73; BHLHE73

GenebankID or UniProtKB

String

9606.ENSP00000263734

Sequence Similarities

-

GO - Molecular Function

GO:0046982 : protein heterodimerization activity
GO:0043565 : sequence-specific DNA binding
GO:0008134 : transcription factor binding
GO:0003677 : DNA binding
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific
GO:0035035 : histone acetyltransferase binding

GO - Biological Process

GO:0007165 : signal transduction
GO:0045944 : positive regulation of transcription by RNA polymerase II
GO:0006357 : regulation of transcription by RNA polymerase II
GO:0043687 : post-translational protein modification
GO:0030324 : lung development
GO:0071456 : cellular response to hypoxia
GO:0061418 : regulation of transcription from RNA polymerase II promoter in response to hypoxia
GO:0001666 : response to hypoxia
GO:0001525 : angiogenesis
GO:0001974 : blood vessel remodeling
GO:0048469 : cell maturation
GO:0001892 : embryonic placenta development
GO:0030218 : erythrocyte differentiation
GO:0055072 : iron ion homeostasis
GO:0007005 : mitochondrion organization
GO:0048625 : myoblast fate commitment
GO:0042415 : norepinephrine metabolic process
GO:0120162 : positive regulation of cold-induced thermogenesis
GO:0002027 : regulation of heart rate
GO:0043619 : regulation of transcription from RNA polymerase II promoter in response to oxidative stress
GO:0043129 : surfactant homeostasis
GO:0006366 : transcription by RNA polymerase II
GO:0007601 : visual perception

GO - Cellular Component

GO:0005829 : cytosol
GO:0005654 : nucleoplasm
GO:0005634 : nucleus
GO:0005667 : transcription factor complex
GO:0016607 : nuclear speck

Presumptive Null

Unknown

Molecular Type

Unknown

Aberration Type

Unknown

Molecular Details of the Mutation

several candidate coding mutations

Experimental Evidence

Candidate Gene

Main Reference

Convergent evolution on the hypoxia-inducible factor (HIF) pathway genes EGLN1 and EPAS1 in high-altitude ducks. (2019)

Authors

Graham AM; McCracken KG

Abstract

During periods of reduced O supply, the most profound changes in gene expression are mediated by hypoxia-inducible factor (HIF) transcription factors that play a key role in cellular responses to low-O tension. Using target-enrichment sequencing, we tested whether variation in 26 genes in the HIF signaling pathway was associated with high altitude and therefore corresponding O availability in three duck species that colonized the Andes from ancestral low-altitude habitats in South America. We found strong support for convergent evolution in the case of two of the three duck species with the same genes (EGLN1, EPAS1), and even the same exons (exon 12, EPAS1), exhibiting extreme outliers with a high probability of directional selection in the high-altitude populations. These results mirror patterns of adaptation seen in human populations, which showed mutations in EPAS1, and transcriptional regulation differences in EGLN1, causing changes in downstream target transactivation, associated with a blunted hypoxic response.

Additional References

Related Genes

2 (EGNL1, hemoglobin; HBA2)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Genomic region identified as outlier of high Fst