GEPHE SUMMARY

Gephebase Gene
EPAS1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EPAS1^#gephebase-summary-title)
Entry Status
Published

GepheID
GP00002131
Main curator
Courtier

PHENOTYPIC CHANGE

Trait #1
Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)
Trait
High-altitude adaptation (https://www.gephebase.org/search-criteria?/and+Trait=^High-altitude adaptation^#gephebase-summary-title)
Trait State in Taxon A
low altitude
Trait State in Taxon B
high altitude

Trait #2
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
low altitude
Trait State in Taxon B
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
Anas cyanoptera (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas cyanoptera^#gephebase-summary-title)
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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)
NCBI Taxonomy ID
75840 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75840)
is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Anas cyanoptera (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas cyanoptera^#gephebase-summary-title)
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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)
NCBI Taxonomy ID
75840 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75840)
is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name

EPAS1

Synonyms

HLF; MOP2; ECYT4; HIF2A; PASD2; bHLHe73; BHLHE73

String

9606.ENSP00000263734 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000263734)

Sequence Similarities

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GO - Molecular Function

GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0008134 : transcription factor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008134)
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)
GO:0035035 : histone acetyltransferase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0035035)

GO - Biological Process

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

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0005667 : transcription factor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0005667)
GO:0016607 : nuclear speck (https://www.ebi.ac.uk/QuickGO/term/GO:0016607)

UniProtKB Homo sapiens

Q99814 (http://www.uniprot.org/uniprot/Q99814)

GenebankID or UniProtKB

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Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

Unknown (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Unknown^#gephebase-summary-title)

Aberration Type

Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)

Molecular Details of the Mutation

several candidate coding mutations

Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)

Main Reference

Convergent evolution on the hypoxia-inducible factor (HIF) pathway genes EGLN1 and EPAS1 in high-altitude ducks. (2019) (https://pubmed.ncbi.nlm.nih.gov/30631144)

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 GEPHE

Related Genes

2 (EGNL1, hemoglobin; HBA2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^75840^/and+Trait=High-altitude adaptation/or+Taxon ID=^75840^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Genomic region identified as outlier of high Fst