GEPHE SUMMARY

Gephebase Gene

EGNL1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EGNL1^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002134

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

EGLN1

Synonyms

HPH2; PHD2; SM20; ECYT3; HALAH; HPH-2; HIFPH2; ZMYND6; C1orf12; HIF-PH2; PNAS-118; PNAS-137

String

9606.ENSP00000355601 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000355601)

Sequence Similarities

-

GO - Molecular Function

GO:0016706 : oxidoreductase activity, acting on paired donors, with incorporation or reduction of molecular oxygen, 2-oxoglutarate as one donor, and incorporation of one atom each of oxygen into both donors (https://www.ebi.ac.uk/QuickGO/term/GO:0016706)
GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)
GO:0008198 : ferrous iron binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008198)
GO:0031418 : L-ascorbic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031418)
GO:0031545 : peptidyl-proline 4-dioxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0031545)
GO:0031543 : peptidyl-proline dioxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0031543)

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:1901214 : regulation of neuron death (https://www.ebi.ac.uk/QuickGO/term/GO:1901214)
GO:0006879 : cellular iron ion homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0006879)
GO:0055008 : cardiac muscle tissue morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0055008)
GO:0060347 : heart trabecula formation (https://www.ebi.ac.uk/QuickGO/term/GO:0060347)
GO:0060711 : labyrinthine layer development (https://www.ebi.ac.uk/QuickGO/term/GO:0060711)
GO:0051344 : negative regulation of cyclic-nucleotide phosphodiesterase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0051344)
GO:0043433 : negative regulation of DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0043433)
GO:0032364 : oxygen homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0032364)
GO:0018401 : peptidyl-proline hydroxylation to 4-hydroxy-L-proline (https://www.ebi.ac.uk/QuickGO/term/GO:0018401)
GO:0045765 : regulation of angiogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0045765)
GO:0099175 : regulation of postsynapse organization (https://www.ebi.ac.uk/QuickGO/term/GO:0099175)
GO:0099576 : regulation of protein catabolic process at postsynapse, modulating synaptic transmission (https://www.ebi.ac.uk/QuickGO/term/GO:0099576)
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:0071731 : response to nitric oxide (https://www.ebi.ac.uk/QuickGO/term/GO:0071731)
GO:0060412 : ventricular septum morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0060412)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0098978 : glutamatergic synapse (https://www.ebi.ac.uk/QuickGO/term/GO:0098978)
GO:0014069 : postsynaptic density (https://www.ebi.ac.uk/QuickGO/term/GO:0014069)

UniProtKB Homo sapiens

Q9GZT9 (http://www.uniprot.org/uniprot/Q9GZT9)

GenebankID or UniProtKB

()

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

probably cis-regulatory because no nonsynonymous mutations in the coding exon with high Fst

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 (EPAS1, 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