

GEPHE SUMMARY

EGLN1 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=EGLN1#gephebase-summary-title)	Gephebase Gene	GP00002136	GepheID
Published	Entry Status	Courtier	Main curator

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	

Taxon A	Ancestral State
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=Intraspecific#gephebase-summary-title)	Taxonomic Status

Taxon A	Latin Name	Common Name	Synonyms	Rank	Lineage	Parent	NCBI Taxonomy ID	is Taxon A an Intraspecies?	
Anas georgica (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Anas+georgica#gephebase-summary-title)		yellow-billed pintail	yellow-billed pintail; Anas georgica Gmelin, 1789	species	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	Anas (ducks) - (Rank: genus)	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8835)	75847 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75847)	No

Taxon B	Latin Name	Common Name	Synonyms	Rank	Lineage	Parent	NCBI Taxonomy ID	is Taxon B an Intraspecies?	
Anas georgica (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Anas+georgica#gephebase-summary-title)		yellow-billed pintail	yellow-billed pintail; Anas georgica Gmelin, 1789	species	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	Anas (ducks) - (Rank: genus)	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8835)	75847 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=75847)	No

GENOTYPIC CHANGE

Gene Name	Generic Gene Name	UniProtKB Homo sapiens
EGLN1	Q9GZT9 (http://www.uniprot.org/uniprot/Q9GZT9)	GenebankID or UniProtKB
HPH2; PHD2; SM20; ECTY3; HALAH; HPH-2; HIFPH2; ZMYND6; C1orf12; HIF-PH2; PNAS-118; PNAS-137	Synonyms ()	
9606.ENSPP00000355601 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000355601)	String	
-	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)		
Unknown (https://www.gephebase.org/search-criteria?and+Presumptive Null=^Unknown^#gephebase-summary-title)		Presumptive Null
Unknown (https://www.gephebase.org/search-criteria?and+Molecular Type=^Unknown^#gephebase-summary-title)		Molecular Type
Unknown (https://www.gephebase.org/search-criteria?and+Aberration Type=^Unknown^#gephebase-summary-title)		Aberration Type
probably cis-regulatory because no nonsynonymous mutations in the coding exon with high Fst		Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria?and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)		Experimental Evidence
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)		Main Reference
Graham AM; McCracken KG		Authors
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		Abstract

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; HBB) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^75847^/and+Trait=High-altitude adaptation/or+Taxon ID=^75847^/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