

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

EGLN1	Generic Gene Name	UniProtKB Homo sapiens
	Q9GZT9 (http://www.uniprot.org/uniprot/Q9GZT9)	
	Synonyms	GenebankID or UniProtKB
HPH2; PHD2; SM20; ECTY3; HALAH; HPH-2; HIFPH2; ZMYND6; C1orf12; HIF-PH2; PNAS-118; PNAS-137	()	
	String	
9606.ENSPP00000355601		
http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000355601		
9606.ENSPP00000355601	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)		
		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
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; 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