

GEPHE SUMMARY

EPAS1 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^EPAS1^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^EPAS1^#gephebase-summary-title</a> )	Gephebase Gene	GP00002133	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

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

Trait #2	Trait Category
Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title</a> )	Trait
Hypoxia response ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Hypoxia response^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Hypoxia response^#gephebase-summary-title</a> )	Trait State in Taxon A
low altitude	Trait State in Taxon B
high altitude	

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

	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

EPAS1	Generic Gene Name	Q99814 ( <a href="http://www.uniprot.org/uniprot/Q99814">http://www.uniprot.org/uniprot/Q99814</a> )	UniProtKB Homo sapiens
	Synonyms	0	GenebankID or UniProtKB
HLF; MOP2; ECYT4; HIF2A; PASD2; BHLHE73; BHLHE73	String		
9606.ENSP00000263734			
<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000263734">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000263734</a>			
9606.ENSP00000263734	Sequence Similarities		
-			
	GO - Molecular Function		
GO:0046982 : protein heterodimerization activity			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046982">https://www.ebi.ac.uk/QuickGO/term/GO:0046982</a>			
GO:0043565 : sequence-specific DNA binding			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043565">https://www.ebi.ac.uk/QuickGO/term/GO:0043565</a>			
GO:0008134 : transcription factor binding			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008134">https://www.ebi.ac.uk/QuickGO/term/GO:0008134</a>			
GO:0003677 : DNA binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003677">https://www.ebi.ac.uk/QuickGO/term/GO:0003677</a> )			
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0000981">https://www.ebi.ac.uk/QuickGO/term/GO:0000981</a>			
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001077">https://www.ebi.ac.uk/QuickGO/term/GO:0001077</a> )			
GO:0035035 : histone acetyltransferase binding			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0035035">https://www.ebi.ac.uk/QuickGO/term/GO:0035035</a>			
	GO - Biological Process		
GO:0007165 : signal transduction ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007165">https://www.ebi.ac.uk/QuickGO/term/GO:0007165</a> )			
GO:0045944 : positive regulation of transcription by RNA polymerase II			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0045944">https://www.ebi.ac.uk/QuickGO/term/GO:0045944</a>			
GO:0006357 : regulation of transcription by RNA polymerase II			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0006357">https://www.ebi.ac.uk/QuickGO/term/GO:0006357</a>			
GO:0043687 : post-translational protein modification			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043687">https://www.ebi.ac.uk/QuickGO/term/GO:0043687</a>			
GO:0030324 : lung development ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0030324">https://www.ebi.ac.uk/QuickGO/term/GO:0030324</a> )			
GO:0071456 : cellular response to hypoxia			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0071456">https://www.ebi.ac.uk/QuickGO/term/GO:0071456</a>			
GO:0061418 : regulation of transcription from RNA polymerase II promoter in response to hypoxia ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0061418">https://www.ebi.ac.uk/QuickGO/term/GO:0061418</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0061418">https://www.ebi.ac.uk/QuickGO/term/GO:0061418</a>			
GO:0001666 : response to hypoxia ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001666">https://www.ebi.ac.uk/QuickGO/term/GO:0001666</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001666">https://www.ebi.ac.uk/QuickGO/term/GO:0001666</a>			
GO:0001525 : angiogenesis ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001525">https://www.ebi.ac.uk/QuickGO/term/GO:0001525</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001525">https://www.ebi.ac.uk/QuickGO/term/GO:0001525</a>			
GO:0001974 : blood vessel remodeling			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001974">https://www.ebi.ac.uk/QuickGO/term/GO:0001974</a>			
GO:0048469 : cell maturation ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0048469">https://www.ebi.ac.uk/QuickGO/term/GO:0048469</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0048469">https://www.ebi.ac.uk/QuickGO/term/GO:0048469</a>			
GO:0001892 : embryonic placenta development			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001892">https://www.ebi.ac.uk/QuickGO/term/GO:0001892</a>			
GO:0030218 : erythrocyte differentiation			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0030218">https://www.ebi.ac.uk/QuickGO/term/GO:0030218</a>			
GO:0055072 : iron ion homeostasis ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0055072">https://www.ebi.ac.uk/QuickGO/term/GO:0055072</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0055072">https://www.ebi.ac.uk/QuickGO/term/GO:0055072</a>			
GO:0007005 : mitochondrion organization			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007005">https://www.ebi.ac.uk/QuickGO/term/GO:0007005</a>			
GO:0048625 : myoblast fate commitment			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0048625">https://www.ebi.ac.uk/QuickGO/term/GO:0048625</a>			
GO:0042415 : norepinephrine metabolic process			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042415">https://www.ebi.ac.uk/QuickGO/term/GO:0042415</a>			
GO:0120162 : positive regulation of cold-induced thermogenesis			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0120162">https://www.ebi.ac.uk/QuickGO/term/GO:0120162</a>			
GO:0002027 : regulation of heart rate			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0002027">https://www.ebi.ac.uk/QuickGO/term/GO:0002027</a>			
GO:0043619 : regulation of transcription from RNA polymerase II promoter in response to oxidative stress ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043619">https://www.ebi.ac.uk/QuickGO/term/GO:0043619</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043619">https://www.ebi.ac.uk/QuickGO/term/GO:0043619</a>			
GO:0043129 : surfactant homeostasis ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043129">https://www.ebi.ac.uk/QuickGO/term/GO:0043129</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043129">https://www.ebi.ac.uk/QuickGO/term/GO:0043129</a>			
GO:0006366 : transcription by RNA polymerase II			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0006366">https://www.ebi.ac.uk/QuickGO/term/GO:0006366</a>			
GO:0007601 : visual perception ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0007601">https://www.ebi.ac.uk/QuickGO/term/GO:0007601</a> )			
	GO - Cellular Component		
GO:0005829 : cytosol ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005829">https://www.ebi.ac.uk/QuickGO/term/GO:0005829</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005829">https://www.ebi.ac.uk/QuickGO/term/GO:0005829</a>			
GO:0005654 : nucleoplasm ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005654">https://www.ebi.ac.uk/QuickGO/term/GO:0005654</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005654">https://www.ebi.ac.uk/QuickGO/term/GO:0005654</a>			
GO:0005634 : nucleus ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005634">https://www.ebi.ac.uk/QuickGO/term/GO:0005634</a> )			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005634">https://www.ebi.ac.uk/QuickGO/term/GO:0005634</a>			
GO:0005667 : transcription factor complex			
<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005667">https://www.ebi.ac.uk/QuickGO/term/GO:0005667</a>			
GO:0016607 : nuclear speck ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0016607">https://www.ebi.ac.uk/QuickGO/term/GO:0016607</a> )			
Unknown ( <a href="https://www.gephebase.org/search-criteria/?and+Presumptive+Null+^Unknown^#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Presumptive+Null+^Unknown^#gephebase-summary-title</a> )			Presumptive Null
			Molecular Type
Unknown ( <a href="https://www.gephebase.org/search-criteria/?and+Molecular+Type+^Unknown^#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Molecular+Type+^Unknown^#gephebase-summary-title</a> )			Aberration Type
Unknown ( <a href="https://www.gephebase.org/search-criteria/?and+Aberration+Type+^Unknown^#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Aberration+Type+^Unknown^#gephebase-summary-title</a> )			Molecular Details of the Mutation
several candidate coding mutations			

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 (EGLN1, 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