

GEPHE SUMMARY

	Gephebase Gene		GepheID
EPAS1 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^EPAS1^#gephebase-summary-title)		GP00000274	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Physiology^#gephebase-summary-title)			
	Trait		
Hypoxia response (https://www.gephebase.org/search-criteria?/and+Trait=^Hypoxiaresponse^#gephebase-summary-title)			
	Trait State in Taxon A		
Homo sapiens - lowlands			
	Trait State in Taxon B		
Homo sapiens - Tibetan highlands			
	Ancestral State		
Data not curated			
	Taxonomic Status		
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Intraspecific^#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	
	Common Name		Common Name
human		human	
	Synonyms		Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens		human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	
	Rank		Rank
species		species	
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo	
	Parent		Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
EPAS1		Q99814 (http://www.uniprot.org/uniprot/Q99814)	
	Synonyms		GenebankID or UniProtKB
HLF; MOP2; ECYT4; HIF2A; PASD2; bHLHe73; BHLHE73		U51626 (https://www.ncbi.nlm.nih.gov/nuccore/U51626)	
	String		
9606.ENSPO0000263734 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000263734)			
	Sequence Similarities		
-			
	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>)

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)	Presumptive Null
	Molecular Type
	Aberration Type
Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)	Molecular Details of the Mutation
unknown	Experimental Evidence
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	Main Reference
Genetic evidence for high-altitude adaptation in Tibet. (2010) (https://pubmed.ncbi.nlm.nih.gov/20466884)	Authors
Simonson TS; Yang Y; Huff CD; Yun H; Qin G; Witherspoon DJ; Bai Z; Lorenzo FR; Xing J; Jorde LB; Prchal JT; Ge R	Abstract

Tibetans have lived at very high altitudes for thousands of years, and they have a distinctive suite of physiological traits that enable them to tolerate environmental hypoxia. These phenotypes are clearly the result of adaptation to this environment, but their genetic basis remains unknown. We report genome-wide scans that reveal positive selection in several regions that contain genes whose products are likely involved in high-altitude adaptation. Positively selected haplotypes of EGLN1 and PPARA were significantly associated with the decreased hemoglobin phenotype that is unique to this highland population. Identification of these genes provides support for previously hypothesized mechanisms of high-altitude adaptation and illuminates the complexity of hypoxia-response pathways in humans.

A genome-wide search for signals of high-altitude adaptation in Tibetans. (2011) (https://pubmed.ncbi.nlm.nih.gov/20961960)	Additional References
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RELATED GEPHE

4 (BHLHE41, EGLN1, PPAR-alpha, hemoglobin; HBA and HBB) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title>)

No matches found.

Related Genes

Related Haplotypes

EXTERNAL LINKS

COMMENTS