

GEPHE SUMMARY

EGLN1 (<a +egln1+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+EGLN1+"#gephebase-summary-title)		Gephebase Gene	GP00000261	GepheID
Published		Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Physiology (<a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology+"#gephebase-summary-title)		Trait Category	
Hypoxia response (<a +hypoxia+response+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Hypoxia+response+"#gephebase-summary-title)		Trait	
Homo sapiens - lowlands		Trait State in Taxon A	
Homo sapiens - Tibetan highlands		Trait State in Taxon B	
Taxon A		Ancestral State	
Intraspecific (<a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Intraspecific+"#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)		Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)	
human		human	
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	
species		species	
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	
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)	
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)	
No		No	

GENOTYPIC CHANGE

EGLN1		Generic Gene Name	UniProtKB Homo sapiens
HPH2; PHD2; SM20; ECYT3; HALAH; HPH-2; HIFPH2; ZMYND6; C1orf12; HIF-PH2; PNAS-118; PNAS-137		Synonyms	GenebankID or UniProtKB
9606.ENSPO0000355601 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000355601)		String	
-		Sequence Similarities	
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 - Molecular Function	

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>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No^#gephebase-summary-title)	Presumptive Null
	Molecular Type
	Aberration Type
	SNP Coding Change
	SNP
Nonsynonymous	SNP Coding Change
	Molecular Details of the Mutation
possibly D4E	Experimental Evidence
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping^#gephebase-summary-title)	

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Genetic evidence for high-altitude adaptation in Tibet. (2010) (https://pubmed.ncbi.nlm.nih.gov/20466884)	Main Reference
	Authors
	Abstract
Simonson TS; Yang Y; Huff CD; Yun H; Qin G; Witherspoon DJ; Bai Z; Lorenzo FR; Xing J; Jorde LB; Prchal JT; Ge R	
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.	
Identifying signatures of natural selection in Tibetan and Andean populations using dense genome scan data. (2010) (https://pubmed.ncbi.nlm.nih.gov/20838600)	Additional References

A genome-wide search for signals of high-altitude adaptation in Tibetans. (2011) (<https://pubmed.ncbi.nlm.nih.gov/20961960>)
Identification of a Tibetan-specific mutation in the hypoxic gene EGLN1 and its contribution to high-altitude adaptation. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23666208>)

RELATED GEPHE

	Related Genes
4 (BHLHE41, EPAS1, 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)	
	Related Haplotypes
1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^EGLN1^/and+Taxon ID=^9606^/or+Gene Gephebase=^EGLN1^/and+Taxon ID=^9606^#gephebase-summary-title)	

EXTERNAL LINKS

COMMENTS