

GEPHE SUMMARY

Gephebase Gene		GepheID
PPAR-alpha (#gephebase-summary-title)	GP00000917	
Entry Status		Main curator
Published		

PHENOTYPIC CHANGE

Trait Category			
Physiology (#gephebase-summary-title)			
Trait			
Hypoxia response (#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 (#gephebase-summary-title)			
Taxon A		Taxon B	
Latin Name		Latin Name	
Homo sapiens (#gephebase-summary-title)		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
PPARA		Q07869 (http://www.uniprot.org/uniprot/Q07869)
Synonyms		GenebankID or UniProtKB
PPAR; NR1C1; hPPAR; PPARalpha		CH471138 (https://www.ncbi.nlm.nih.gov/nucore/CH471138)
String		
9606.ENSPP00000262735 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000262735)		
Sequence Similarities		
Belongs to the nuclear hormone receptor family. NR1 subfamily.		
GO - Molecular Function		
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)		
GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)		

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:0008270 : zinc ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008270>)
GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
GO:0008289 : lipid binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008289>)
GO:0044877 : protein-containing complex binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044877>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
GO:0004879 : nuclear receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004879>)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001077>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
GO:0001227 : DNA-binding transcription repressor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001227>)
GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001078>)
GO:0008144 : drug binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008144>)
GO:0005504 : fatty acid binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005504>)
GO:0038023 : signaling receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038023>)
GO:0003707 : steroid hormone receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003707>)
GO:0019904 : protein domain specific binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019904>)
GO:0000976 : transcription regulatory region sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000976>)
GO:0097371 : MDM2/MDM4 family protein binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097371>)
GO:0051525 : NFAT protein binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0051525>)
GO:0030374 : nuclear receptor transcription coactivator activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030374>)
GO:0019902 : phosphatase binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019902>)
GO:0001103 : RNA polymerase II repressing transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001103>)
GO:0001223 : transcription coactivator binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001223>)
GO:0031624 : ubiquitin conjugating enzyme binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031624>)

GO - Biological Process

GO:0007275 : multicellular organism development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
GO:0009755 : hormone-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
GO:0007507 : heart development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007507>)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0045892 : negative regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0045893 : positive regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0042157 : lipoprotein metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042157>)
GO:0050728 : negative regulation of inflammatory response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050728>)
GO:1901215 : negative regulation of neuron death
(<https://www.ebi.ac.uk/QuickGO/term/GO:1901215>)
GO:0006367 : transcription initiation from RNA polymerase II promoter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006367>)
GO:0032922 : circadian regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032922>)
GO:0035095 : behavioral response to nicotine
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035095>)
GO:0042752 : regulation of circadian rhythm
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042752>)
GO:0046321 : positive regulation of fatty acid oxidation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046321>)
GO:0008544 : epidermis development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008544>)
GO:0001666 : response to hypoxia (<https://www.ebi.ac.uk/QuickGO/term/GO:0001666>)
GO:0006631 : fatty acid metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006631>)
GO:0006629 : lipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006629>)
GO:0042060 : wound healing (<https://www.ebi.ac.uk/QuickGO/term/GO:0042060>)
GO:0032099 : negative regulation of appetite
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032099>)
GO:0061052 : negative regulation of cell growth involved in cardiac muscle cell development (<https://www.ebi.ac.uk/QuickGO/term/GO:0061052>)
GO:0033993 : response to lipid (<https://www.ebi.ac.uk/QuickGO/term/GO:0033993>)
GO:0070166 : enamel mineralization (<https://www.ebi.ac.uk/QuickGO/term/GO:0070166>)
GO:0015908 : fatty acid transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015908>)
GO:0045776 : negative regulation of blood pressure
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045776>)
GO:0010887 : negative regulation of cholesterol storage
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010887>)
GO:0045820 : negative regulation of glycolytic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045820>)
GO:1903038 : negative regulation of leukocyte cell-cell adhesion
(<https://www.ebi.ac.uk/QuickGO/term/GO:1903038>)
GO:0010745 : negative regulation of macrophage derived foam cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010745>)
GO:1902894 : negative regulation of pri-miRNA transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902894>)
GO:0032091 : negative regulation of protein binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032091>)
GO:0010871 : negative regulation of receptor biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010871>)
GO:0010891 : negative regulation of sequestering of triglyceride
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010891>)
GO:0032000 : positive regulation of fatty acid beta-oxidation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032000>)
GO:0045722 : positive regulation of gluconeogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045722>)
GO:0072366 : regulation of cellular ketone metabolic process by positive regulation of transcription from RNA polymerase II promoter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0072366>)
GO:0072363 : regulation of glycolytic process by positive regulation of transcription from RNA polymerase II promoter (<https://www.ebi.ac.uk/QuickGO/term/GO:0072363>)
GO:0019216 : regulation of lipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019216>)
GO:0072369 : regulation of lipid transport by positive regulation of transcription from RNA polymerase II promoter (<https://www.ebi.ac.uk/QuickGO/term/GO:0072369>)
GO:0032868 : response to insulin (<https://www.ebi.ac.uk/QuickGO/term/GO:0032868>)
GO - Cellular Component
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:0090575 : RNA polymerase II transcription factor complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090575>)

	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
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.	
	Additional References
Identifying signatures of natural selection in Tibetan and Andean populations using dense genome scan data. (2010) (https://pubmed.ncbi.nlm.nih.gov/20838600)	

RELATED GEPHE

	Related Genes
4 (BHLHE41, EGLN1, EPAS1, 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.

EXTERNAL LINKS

COMMENTS