GEPHE SUMMARY

Gephebase Gene

EGLN1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EGLN1^#gephebase-summary-title)

GepheID

GP00000262

Entry Status

Published

Main curator

Martin

PHENOTYPIC CHANGE

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

Homo sapiens - lowlands

Trait State in Taxon B

Homo sapiens - Andean highlands

Ancestral State

Data not curated

Taxonomic Status

Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name

Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name

human

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

Rank

species

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

Parent

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name

human

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

Rank

species

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

Parent

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

EGLN1

Synonyms

HPH2; PHD2; SM20; ECYT3; HALAH; HPH-2; HIFPH2; ZMYND6; C1orf12; HIF-PH2; PNAS-118; PNAS-137

String

9606.ENSP00000355601 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000355601)

UniProtKB Homo sapiens

Q9GZT9 (http://www.uniprot.org/uniprot/Q9GZT9)

Sequence Similarities

-

GenebankID or UniProtKB

AF277176 (https://www.ncbi.nlm.nih.gov/nuccore/AF277176)

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

unknown

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)

Main Reference

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

Authors

Bigham A; Bauchet M; Pinto D; Mao X; Akey JM; Mei R; Scherer SW; Julian CG; Wilson MJ; LÃ³pez HerrÃiez D; Brutsaert T; Parra EJ; Moore LG; Shriver MD

Abstract

High-altitude hypoxia (reduced inspired oxygen tension due to decreased barometric pressure) exerts severe physiological stress on the human body. Two high-altitude regions where humans have lived for millennia are the Andean Altiplano and the Tibetan Plateau. Populations living in these regions exhibit unique circulatory, respiratory, and hematological adaptations to life at high altitude. Although these responses have been well characterized physiologically, their underlying genetic basis remains unknown. We performed a genome scan to identify genes showing evidence of adaptation to hypoxia. We looked across each chromosome to identify genomic regions with previously unknown function with respect to altitude phenotypes. In addition, groups of genes functioning in oxygen metabolism and sensing were examined to test the hypothesis that particular pathways have been involved in genetic adaptation to altitude. Applying four population genetic statistics commonly used for detecting signatures of natural selection, we identified selection-nominated candidate genes and gene regions in these two populations (Andeans and Tibetans) separately. The Tibetan and Andean patterns of genetic adaptation are largely distinct from one another, with both populations showing evidence of positive natural selection in different genes or gene regions. Interestingly, one gene previously known to be important in cellular oxygen sensing, EGLN1 (also known as PHD2), shows evidence of positive selection in both Tibetans and Andeans. However, the pattern of variation for this gene differs between the two populations. Our results indicate that several key HIF-regulatory and targeted genes are responsible for adaptation to high altitude in Andeans and Tibetans, and several different chromosomal regions are implicated in the putative response to selection. These data suggest a genetic role in high-altitude adaption and provide a basis for future genotype/phenotype association studies necessary to confirm the role of selection-nominated candidate genes and gene regions in adaptation to altitude.

Additional References

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