GEPHE SUMMARY

Gephebase Gene
alcohol dehydrogenase (ADH1B) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^alcohol dehydrogenase (ADH1B)^#gephebase-summary-title)

GepheID
GP00000077

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
Xenobiotic resistance (alcohol) (https://www.gephebase.org/search-criteria?/and+Trait=^Xenobiotic resistance (alcohol)^#gephebase-summary-title)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens - incresaed metabolism of alcohol

Ancestral State
Taxon A

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
ADH1B

Synonyms
ADH2; HEL-S-117

String
9606.ENSP00000306606 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000306606)

Sequence Similarities
Belongs to the zinc-containing alcohol dehydrogenase family.

GO - Molecular Function
GO:0004024 : alcohol dehydrogenase activity, zinc-dependent (https://www.ebi.ac.uk/QuickGO/term/GO:0004024)
GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)

GO - Biological Process

UniProtKB Homo sapiens
P00325 (http://www.uniprot.org/uniprot/P00325)

GenebankID or UniProtKB
X15452 (https://www.ncbi.nlm.nih.gov/nuccore/X15452)

GO:0006069 : ethanol oxidation (https://www.ebi.ac.uk/QuickGO/term/GO:0006069)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Arg47His; the derived allele results in 100-fold enzymatic rate increase and shows signs of positive selection in correlation with history of rice domestication

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	-	-	-

Main Reference

Geographically separate increases in the frequency of the derived ADH1B*47His allele in eastern and western Asia. (2007) (https://pubmed.ncbi.nlm.nih.gov/17847010)

Authors

Li H; Mukherjee N; Soundararajan U; Tarnok Z; Barta C; Khaliq S; Mohyuddin A; Kajuna SL; Mehdi SQ; Kidd JR; Kidd KK

Abstract

The ADH1B Arg47His polymorphism has been convincingly associated with alcoholism in numerous studies of several populations in Asia and Europe. In a review of literature from the past 30 years, we have identified studies that report allele frequencies of this polymorphism for 131 population samples from many different parts of the world. The derived ADH1B*47His allele reaches high frequencies only in western and eastern Asia. To pursue this pattern, we report here new frequency data for 37 populations. Most of our data are from South and Southeast Asia and confirm that there is a low frequency of this allele in the region between eastern and western Asia. The distribution suggests that the derived allele increased in frequency independently in western and eastern Asia after humans had spread across Eurasia.

Additional References

Geographically separate increases in the frequency of the derived ADH1B*47His allele in eastern and western Asia. (2007) (https://pubmed.ncbi.nlm.nih.gov/17847010)

RELATED GEPHE

Related Genes

4 (CYP2C9, CYP4F2, MDR1, Vkorc1) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Xenobiotic resistance/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS