

GEPHE SUMMARY

		Gephebase Gene		GepheID
alcohol dehydrogenase (ADH1B) (https://www.gephebase.org/search-criteria?/and+Gene)			GP00000077	
Gephebase= [^] alcohol dehydrogenase (ADH1B) [^] #gephebase-summary-title				Main curator
Published		Entry Status	Martin	

PHENOTYPIC CHANGE

		Trait Category	
Physiology (https://www.gephebase.org/search-criteria?/and+Trait)		Category= [^] Physiology [^] #gephebase-summary-title	
		Trait	
Xenobiotic resistance (alcohol) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Xenobiotic resistance (alcohol)<sup>^</sup>#gephebase-summary-title">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 - increased 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		Taxon B	
		Latin Name	Latin Name
Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Homo sapiens<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Homo sapiens[^]#gephebase-summary-title)		Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Homo sapiens<sup>^</sup>#gephebase-summary-title">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 sapien; 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 sapien; 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
ADH1B		P00325 (http://www.uniprot.org/uniprot/P00325)	GenebankID or UniProtKB
		Synonyms	
ADH2; HEL-S-117		X15452 (https://www.ncbi.nlm.nih.gov/nucleotide/X15452)	
		String	
9606.ENSPO0000306606 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000306606)			
		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	

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