

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
			Entry Status	Martin	
Published					

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			
Homo sapiens				Homo sapiens	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Homo sapiens [^] #gephebase-summary-title)				(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Homo sapiens [^] #gephebase-summary-title)	
		Common Name			
human				human	
		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			
species				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				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)				Homo () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)				(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
		NCBI Taxonomy ID			
9606				9606	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)				(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
				is Taxon A an Intraspecies?	
No				is Taxon B an Intraspecies?	
				No	

GENOTYPIC CHANGE

			Generic Gene Name		UniProtKB Homo sapiens	
ADH1B					P00325 (http://www.uniprot.org/uniprot/P00325)	
			Synonyms		GenebankID or UniProtKB	
ADH2; HEL-S-117					X15452 (https://www.ncbi.nlm.nih.gov/nucleotide/X15452)	
			String			
9606.ENSPP00000306606						
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSPP00000306606)						
			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 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No))

Molecular Type

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

Aberration Type

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

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

	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