

GEPHE SUMMARY

	Gephebase Gene	GephelD
ABO histo blood group glycosyltransferase (https://www.gepheebase.org/search-criteria?/and+Gene Gephebase=^ABO histo blood group glycosyltransferase^#gepheebase-summary-title)	GP00000017	Main curator
	Entry Status	Martin
Published		

PHENOTYPIC CHANGE

	Trait Category		
Physiology (https://www.gepheebase.org/search-criteria?/and+Trait Category=^Physiology^#gepheebase-summary-title)	Trait		
ABO antigen blood type (https://www.gepheebase.org/search-criteria?/and+Trait=^ABO antigen blood type^#gepheebase-summary-title)	Trait State in Taxon A		
Homo sapiens; other primates - blood group A	Trait State in Taxon B		
Homo sapiens; other primates -blood group B	Ancestral State		
Data not curated	Taxonomic Status		
Intraspecific (https://www.gepheebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gepheebase-summary-title)			
Taxon A		Taxon B	
Homininae (https://www.gepheebase.org/search-criteria?/and+Taxon and Synonyms=^Homininae^#gepheebase-summary-title)	Latin Name	Homininae (https://www.gepheebase.org/search-criteria?/and+Taxon and Synonyms=^Homininae^#gepheebase-summary-title)	Latin Name
-	Common Name	-	Common Name
Homo/Pan/Gorilla group	Synonyms	Homo/Pan/Gorilla group	Synonyms
subfamily	Rank	subfamily	Rank
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	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	Lineage
Hominidae (great apes) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9604)	Parent	Hominidae (great apes) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9604)	Parent
207598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 207598)	NCBI Taxonomy ID	207598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 207598)	NCBI Taxonomy ID
	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
No		No	

GENOTYPIC CHANGE

ABO	Generic Gene Name	UniProtKB Homo sapiens
GTB; NAGAT; A3GALNT; A3GALT1	Synonyms	GenebankID or UniProtKB
-	String	AAC50121 (https://www.ncbi.nlm.nih.gov/nuccore/AAC50121)
	Sequence Similarities	
Belongs to the glycosyltransferase 6 family.		
	GO - Molecular Function	
GO:0003823 : antigen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003823)		
GO:0004381 : fucosylgalactoside 3-alpha-galactosyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004381)		
GO:0004380 : glycoprotein-fucosylgalactoside alpha-N-acetylglactosaminyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004380)		
GO:0030145 : manganese ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030145)		
GO:0000166 : nucleotide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000166)		

GO:0016757 : transferase activity, transferring glycosyl groups
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016757>)

GO - Biological Process

GO:0005975 : carbohydrate metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005975>)

GO:0030259 : lipid glycosylation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030259>)

GO:0006486 : protein glycosylation (<https://www.ebi.ac.uk/QuickGO/term/GO:0006486>)

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)

GO:0005794 : Golgi apparatus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005794>)

GO:0032580 : Golgi cisterna membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0032580>)

GO:0031982 : vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0031982>)

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

Gly268 (group A) <-> Ala (group B)

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=%Candidate+Gene%#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Sugar-nucleotide donor specificity of histo-blood group A and B transferases is based on amino acid substitutions. (1990) (<https://pubmed.ncbi.nlm.nih.gov/2121736>)

Main Reference

Yamamoto F; Hakomori S

Authors

Four amino acid substitutions (aa 176, 235, 266, and 268) have been found between the coding regions of cDNAs for histo-blood group A and B transferases (Yamamoto F., Clausen, H., White, T., Marken, J., and Hakomori, S. (1990) *Nature* 345, 229-233). Here we establish the basis of differential affinity of these glycosyltransferases to nucleotide-sugar (UDP-GalNAc or UDP-Gal). On the basis of gene reconstruction experiments and studies of expression in DNA transfected HeLa cells, the third as well as the fourth aa substitutions (leucine and glycine in A and methionine and alanine in B), which were calculated to modify flexibility of the protein, were found to be crucial in determining nucleotide-sugar specificity. The second substitution (glycine in A and serine in B) also affects the specificity. We have also created new enzymes which catalyze the transfer of both GalNAc and Gal, and may provide an explanation of the rare *cis*-AB phenotype.

Abstract

The ABO blood group is a trans-species polymorphism in primates. (2012) (<https://pubmed.ncbi.nlm.nih.gov/23091028>)

Additional References

RELATED GEPHE

	Related Genes
1 (FUT2) (https://www.gephebase.org/search-criteria?/or+Taxon+ID=%207598%and+Trait=ABO+antigen+blood+type/and+groupHaplotypes=true%#gephebase-summary-title)	Related Haplotypes
7 (https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=%ABO+histo+blood+group+glycosyltransferase%and+Taxon+ID=%207598%or+Gene+Gephebase=%ABO+histo+blood+group+glycosyltransferase%and+Taxon+ID=%207598%#gephebase-summary-title)	

EXTERNAL LINKS

COMMENTS

