GEPHE SUMMARY

Gephebase Gene

ABO histo blood group glycosyltransferase (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^ABO histo blood group glycosyltransferase^#gephebase-summary-title)

GepheID

GP00000018

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

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

Taxon A

Latin Name

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

Common Name

-

Synonyms

Homo/Pan/Gorilla group

Rank

subfamily

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

Parent

Hominidae (great apes) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9604)

NCBI Taxonomy ID

207598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 207598)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

-

Synonyms

Homo/Pan/Gorilla group

Rank

subfamily

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

Parent

Hominidae (great apes) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9604)

NCBI Taxonomy ID

207598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 207598)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

ABO

Synonyms

GTB; NAGAT; A3GALNT; A3GALT1

String

-

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-acetylgalactosaminyltransferase 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)

UniProtKB Homo sapiens

P16442 (http://www.uniprot.org/uniprot/P16442)

GenebankID or UniProtKB

AAC50121 (https://www.ncbi.nlm.nih.gov/nuccore/AAC50121)

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

Leu266 (group A) <-> Met (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	-	-	-

Main Reference

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)

Authors

Yamamoto F; Hakomori S

Abstract

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.

Additional References

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

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