

GEPHE SUMMARY

<p>ABO histo blood group glycosyltransferase (<a +abo+histo+blood+group+glycosyltransferase+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+ABO+histo+blood+group+glycosyltransferase+"#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000018</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
---	---	---------------------------------	------------------------------------

PHENOTYPIC CHANGE

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

GENOTYPIC CHANGE

<p>ABO</p> <p>GTB; NAGAT; A3GALNT; A3GALT1</p> <p>-</p> <p>Belongs to the glycosyltransferase 6 family.</p> <p>GO:0003823 : antigen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003823)</p> <p>GO:0004381 : fucosylgalactoside 3-alpha-galactosyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004381)</p> <p>GO:0004380 : glycoprotein-fucosylgalactoside alpha-N-acetylgalactosaminyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004380)</p> <p>GO:0030145 : manganese ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030145)</p> <p>GO:0000166 : nucleotide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000166)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P16442 (http://www.uniprot.org/uniprot/P16442)</p> <p>AAC50121 (https://www.ncbi.nlm.nih.gov/nucleotide/AAC50121)</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
---	--	---	--

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](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

Molecular Type

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

Aberration Type

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

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](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=))

	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](https://www.gephebase.org/search-criteria?/or+Taxon ID=))

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](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=))

EXTERNAL LINKS

COMMENTS

