

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>GP00000019</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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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</p> <p>Homo sapiens (alleles O01; O02; O09)</p> <p>Taxon A</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>Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</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; Homininae; Homo</p> <p>Parent</p> <p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</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; Homininae; Homo</p> <p>Parent</p> <p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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GENOTYPIC CHANGE

<p>ABO</p> <p>GTB; NAGAT; A3GALNT; A3GALT1</p> <p>-</p> <p>Belongs to the glycosyltransferase 6 family.</p> <p>GO - Molecular Function</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>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p>	<p>UniProtKB Homo sapiens</p> <p>P16442 (http://www.uniprot.org/uniprot/P16442)</p> <p>X84746 (https://www.ncbi.nlm.nih.gov/nuccore/X84746)</p> <p>GenebankID or UniProtKB</p>
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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>)

Yes (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gepbase-summary-title>)

Presumptive Null

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gepbase-summary-title>)

Molecular Type

Deletion (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gepbase-summary-title>)

Aberration Type

1-9 bp

Deletion Size

1bp deletion (258G) resulting in frameshift

Molecular Details of the Mutation

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

Experimental Evidence

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

Evolutionary dynamics of the human ABO gene. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18629539>)

Additional References

RELATED GEPHE

1 (FUT2) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9606^/and+Trait=ABO antigen blood type/and+groupHaplotypes=true#gepbase-summary-title>)

Related Genes

3 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~ABO histo blood group glycosyltransferase^/and+Taxon ID=~9606^/or+Gene Gephebase=~ABO histo blood group glycosyltransferase^/and+Taxon ID=~9606^#gepbase-summary-title>)

Related Haplotypes

EXTERNAL LINKS

COMMENTS