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

GP00000020

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

Trait State in Taxon B

Homo sapiens - allele O03

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

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

Common Name

human

Synonyms

human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank

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

Parent

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

human

Synonyms

human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank

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

Parent

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

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)

UniProtKB Homo sapiens

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

GenebankID or UniProtKB

X84748 (https://www.ncbi.nlm.nih.gov/nuccore/X84748)

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

Arg176+Gly268 (group A) <-> Gly176+Arg268 (group O03 = weak AB allele rather than complete loss-of-function)
two nucleotide substitutions at nt. 526 and nt. 802. The authors have not examined whether both amino acid substitutions are necessary to disrupt the enzymatic activity or if one of them is sufficient. Nevertheless they assume that the single amino acid substitution at aa. 268 (glycine+arginine) may be crucial for their nullifying effect.

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	802
Amino-acid	Gly	Arg	268

Main Reference

Molecular genetic analysis of the ABO blood group system: 4. Another type of O allele. (1993) (https://pubmed.ncbi.nlm.nih.gov/8484251)

Authors

Yamamoto F; McNeill PD; Yamamoto M; Hakomori S; Bromilow IM; Duguid JK

Abstract

We have encountered an allele which seems to be another type of O allele at the human histo-blood group ABO locus. We have determined the nucleotide sequence of this allele over the coding region in the last two coding exons. This allele does not possess the single-nucleotide deletion found common among all the O alleles previously analyzed. Compared with A1 allele, this allele has three nucleotide substitutions resulting in two amino acid substitutions. The introduction of these amino acid substitutions into the A1 transferase expression construct apparently abolished the enzymatic activity of A1 transferase.

Additional References

Blood grouping discrepancies between ABO genotype and phenotype caused by O alleles. (2008) (https://pubmed.ncbi.nlm.nih.gov/18832934)

RELATED GEPHE

Related Genes

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

Related Haplotypes

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^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS