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)

Entry Status

Published

GepheID

GP00000022

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 O52

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

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#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

Nonsense

Molecular Details of the Mutation

1bp change resulting in stop codon

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

New and unusual O alleles at the ABO locus are implicated in unexpected blood group phenotypes. (2005) (https://pubmed.ncbi.nlm.nih.gov/15647021)

Authors

Hosseini-Maaf B; Irshaid NM; Hellberg A; Wagner T; Levene C; Hustinx H; Steffensen R; Chester MA; Olsson ML

Abstract

In the ABO blood group system mutations in the A gene may lead to weak A subgroups owing to a dysfunctional 3-alpha-N-acetylgalactosaminyltransferase.

Blood and DNA were investigated to correlate weak A phenotypes with genotype, and an overrepresentation of the infrequent O2 allele was observed. Consequently, 57 available O2 alleles were examined in detail.

Two new O2 alleles were identified having mutations resulting in Gly229Asp with or without Arg217Cys. A recently described O2 variant (488C>T; Thr163Met) was also found. Surprisingly, both the original and the variant O2 alleles were associated with either O or Aweak phenotypes. Three novel O alleles surfaced in six other samples with suspected A subgroups. These were A1-like alleles having nonsense mutations causing premature truncation at codons 56, 107, or 181. A second example of the rare O3 allele was also identified. A newly described O1 allele having 768C>A was found to be the third most frequent O allele among Swedish donors. Of the five novel O alleles, three were incorrectly interpreted as A1 following routine ABO genotyping.

Apparent O alleles lacking 261delG may cause weak A expression on red blood cells and/or inhibit anti-A production. A hypothesis that exchange of genetic material between principally dissimilar O alleles during mitosis ("autologous chimerism") restores glycosyltransferase activity in some cells would explain this interesting phenomenon.

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