GEPHE SUMMARY

Gephebase Gene
ABO histo blood group glycosyltransferase

GephelD
GP00000020

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
ABO antigen blood type

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens - allele O03

Ancestral State
Data not curated

Taxonomic Status
Intraspecific

Taxon A

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens

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)

NCBI Taxonomy 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
GO:0004381 : fucosylgalactoside 3-alpha-galactosyltransferase activity
GO:0004380 : glycoprotein-fucosylgalactoside alpha-N-acetylgalactosaminyltransferase activity
GO:0030145 : manganese ion binding
GO:0000166 : nucleotide binding
GO:0016757 : transferase activity, transferring glycosyl groups

UniProtKB Homo sapiens
P16442

GenebankID or UniProtKB
X84748

GO - Biological Process
GO:0005975 : carbohydrate metabolic process
GO:0030259 : lipid glycosylation
GO:0006486 : protein glycosylation

GO - Cellular Component
GO:0016021 : integral component of membrane
GO:0005576 : extracellular region
GO:0005794 : Golgi apparatus
GO:0032580 : Golgi cisterna membrane
GO:0031982 : vesicle

Presumptive Null
No

Molecular Type
Coding

Aberration Type
SNP

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

	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)

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)

RELATED GEPHE

Related Genes
1 (FUT2)
Related Haplotypes
3

EXTERNAL LINKS

COMMENTS