

GEPHE SUMMARY

	Gephebase Gene	GepheID
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)	GP00000021	
	Martin	Main curator
Published	Entry Status	

PHENOTYPIC CHANGE

	Trait Category		
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)	Trait		
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)	Trait State in Taxon A		
Homo sapiens	Trait State in Taxon B		
Homo sapiens -allele O51	Ancestral State		
Taxon A	Taxonomic Status		
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)			
	Taxon A		Taxon B
	Latin Name		Latin Name
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)	Common Name		Common Name
human	Synonyms		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		Rank
species	Lineage		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		Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	NCBI Taxonomy ID		NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No			No

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
ABO	Synonyms	P16442 (http://www.uniprot.org/uniprot/P16442)
GTB; NAGAT; A3GALNT; A3GALT1	String	GenebankID or UniProtKB X84748 (https://www.ncbi.nlm.nih.gov/nuccore/X84748)
-	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)		

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~#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	Molecular Type
Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Insertion~#gephebase-summary-title)	Aberration Type
1-9 bp	Insertion Size
1bp insertion resulting in premature stop codon	Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	Experimental Evidence
New and unusual O alleles at the ABO locus are implicated in unexpected blood group phenotypes. (2005) (https://pubmed.ncbi.nlm.nih.gov/15647021)	Main Reference
Hosseini-Maaf B; Irshaid NM; Hellberg A; Wagner T; Levene C; Hustinx H; Steffensen R; Chester MA; Olsson ML	Authors
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.	Abstract

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.

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

RELATED GEPHE

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 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~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

