GEPHE SUMMARY

Gephebase Gene

FUT2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^FUT2^#gephebase-summary-title)

Entry Status

Published

GephelD

GP00000379

Main curator

Courtier

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

Ancestral State

Taxon A

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?

Yes

Taxon B Description

human - Samoan population

GENOTYPIC CHANGE

Generic Gene Name

FUT2

Synonyms

SE; Se2; sej; SEC2; B12QTL1

String

9606.ENSP00000375748 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000375748)

Sequence Similarities

Belongs to the glycosyltransferase 11 family.

GO - Molecular Function

GO:0008417 : fucosyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008417)

UniProtKB Homo sapiens

Q10981 (http://www.uniprot.org/uniprot/Q10981)

GenebankID or UniProtKB

U17894 (https://www.ncbi.nlm.nih.gov/nuccore/U17894)

GO:0008107 : galactoside 2-alpha-L-fucosyltransferase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0008107)

GO - Biological Process

GO:0005975 : carbohydrate metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0005975)
GO:0006486 : protein glycosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0006486)
GO:0036065 : fucosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0036065)
GO:0042355 : L-fucose catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042355)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
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)

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

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

Deletion Size

1-10 kb

Molecular Details of the Mutation

9.3 kb deletion mediated by recombination between Alu sequences

Experimental Evidence

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

Main Reference

Sequence and expression of a candidate for the human Secretor blood group alpha(1,2)fucosyltransferase gene (FUT2). Homozygosity for an enzyme-inactivating nonsense mutation commonly correlates with the non-secretor phenotype. (1995) (https://pubmed.ncbi.nlm.nih.gov/7876235)

Authors

Kelly RJ; Rouquier S; Giorgi D; Lennon GG; Lowe JB

Abstract

Synthesis of soluble A, B, H, and Lewis b blood group antigens in humans is determined by the Secretor (Se) (FUT2) blood group locus. Genetic, biochemical, and molecular analyses indicate that this locus corresponds to an alpha(1,2)fucosyltransferase gene distinct from the genetically-linked H blood group alpha(1,2)fucosyltransferase locus. The accompanying paper (Rouquier, S., Lowe, J. B., Kelly, R. J., Fertitta, A. L., Lennon, G. G., and Giorgi, D. (1995) J. Biol. Chem. 270, 4632-4639) describes the molecular cloning and mapping of two human DNA segments that are physically linked to, and cross-hybridize with, the H locus. We present here an analysis of these two new DNA segments. One of these, termed Sec1, is a pseudogene, because translational frameshifts and termination codons interrupt potential open reading frames that would otherwise share primary sequence similarity with the H alpha(1,2)fucosyltransferase. The other DNA segment, termed Sec2, predicts a 332-amino acid-long polypeptide, and a longer isoform, that share 68% sequence identity with the COOH-terminal 292 residues of the human H blood group alpha(1,2)fucosyltransferase. Sec2 encodes an alpha(1,2)fucosyltransferase with catalytic properties that mirror those ascribed to the Secretor locus-encoded alpha(1,2)fucosyltransferase. Approximately 20% of randomly-selected individuals were found to be apparently homozygous for an enzyme-inactivating nonsense allele (Trp143-->ter) at this locus, in correspondence to the frequency of the non-secretor phenotype in most human populations. Furthermore, each of six unrelated non-secretor individuals are also apparently homozygous for this null allele. These results indicate that Sec2 corresponds to the human Secretor blood group locus (FUT2) and indicate that homozygosity for a common nonsense allele is responsible for the nonsecretor phenotype in many non-secretor individuals.

Additional References

Two distinct Alu-mediated deletions of the human ABO-secretor (FUT2) locus in Samoan and Bangladeshi populations. (2000) (https://pubmed.ncbi.nlm.nih.gov/10980544)

RELATED GEPHE

Related Genes

1 (ABO histo blood group glycosyltransferase) (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=^FUT2^/and+Taxon ID=^9606^/or+Gene Gephebase=^FUT2^/and+Taxon ID=^9606^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

The frequency of the se(del2) in the Samoan population is 0.104