GEPHE SUMMARY

Gephebase Gene
FUT2

GephelD
GP00000382

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

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
FUT2

Synonyms
SE; Se2; sej; SEC2; B12QTL1

String
9606.ENSP00000375748

Sequence Similarities
Belongs to the glycosyltransferase 11 family.

GO - Molecular Function
GO:0008417 : fucosyltransferase activity
GO:0008107 : galactoside 2-alpha-L-fucosyltransferase activity

GO - Biological Process
GO:0005975 : carbohydrate metabolic process
GO:0006486 : protein glycosylation
GO:0036065 : fucosylation
GO:0042355 : L-fucose catabolic process

UniProtKB Homo sapiens
Q10981

GenebankID or UniProtKB
U17894

GO - Cellular Component
GO:0016021 : integral component of membrane
GO:0070062 : extracellular exosome
GO:0005794 : Golgi apparatus
GO:0032580 : Golgi cisterna membrane

Presumptive Null
No

Molecular Type
Coding

Aberration Type
SNP

SNP Coding Change
Nonsynonymous

Molecular Details of the Mutation
Ile129Phe

Experimental Evidence
Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

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)

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
A natural history of FUT2 polymorphism in humans. (2009)

RELATED GEPHE

Related Genes
1 (ABO histo blood group glycosyltransferase)
Related Haplotypes
3

COMMENTS