Names for H (ISBT 018) Blood Group Alleles

General description:	The H blood group system consists of one antigen, H, that is carried on glycolipids and glycoproteins on the RBC membrane, where it is synthesised by the fucosyltransferase product of the *FUT1* gene; as well as on glycoproteins on epithelial cells and in body fluids, where it is synthesised by the fucosyltransferase product of the *FUT2* gene. In group O individuals, H antigen is the terminal antigen however, in group A and B individuals, the H antigen serves as the precursor structure for A and B blood-group-specific glycosyltransferases. Thus, group O people will test strongly H+ whereas groups A, B and AB will express very little H antigen. Mutations that negatively affect the α2FucT1 enzyme activity (encoded by *FUT1*) will result in reduced or absent H production (and a concomitant decrease in A and/or B antigens in individuals where those enzymes are encoded). Total absence of H, A and B antigens is called the O_h or Bombay phenotype. Weak expression is referred to as the paraBombay phenotype. The enzymes α2FucT1and α2FucT2 are single pass type II membrane glycoproteins in the Golgi. The α2FucT1 protein consists of 365 amino acids and is encoded by *FUT1* or *H*, if analysis is to predict a blood group antigen. The *FUT2* gene produces two transcripts; one of 343 amino acids and another more abundant form of 332 amino acids. The longer transcript encodes a protein with approximately one fourth the enzymatic activity and the shorter form is considered to be the active enzyme. Thus, the numbers given below are counted from the second initiating codon. The α2FucT2 protein is encoded by *FUT2* or *Se*, if analysis is to predict a blood group antigen.
Gene name:	*FUT1*
Number of exons:	4
Initiation codon:	Beginning of exon 4
Stop codon:	Within exon 4
Entrez Gene ID:	2523
LRG sequence:	NG_007510.1 (genomic) NM_000148.3 (transcript)
Reference allele:	*FUT1*01* (shaded) Acceptable: *H* if inferred by hemagglutination/inhibition

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
H+	*FUT1*01*			
H+	*FUT1*02*	c.35C>T	4	p.Ala12Val
Weak phenotypes†				
H+weak	*FUT1*01W.01*	c.293C>T	4	p.Thr98Met
H+weak	*FUT1*01W.02*	c.328G>A	4	p.Ala110Thr
H+weak	*FUT1*01W.03*	c.349C>T	4	p.His117Tyr
H+weak	*FUT1*01W.04*	c.442G>T	4	p.Asp148Tyr
H+weak	*FUT1*01W.05.01*	c.460T>C	4	p.Tyr154His
H+weak	*FUT1*01W.05.02*	c.460T>C; c.1042G>A	4	p.Tyr154His; p.Glu348Lys
H+weak	*FUT1*01W.07*	c.491T>A	4	p.Leu164His
H+weak	*FUT1*01W.08*	c.522C>A	4	p.Phe174Leu
H+weak	*FUT1*01W.09*	c.658C>T	4	p.Arg220Cys
H+weak	*FUT1*01W.10*	c.659G>A	4	p.Arg220His
H+weak	*FUT1*01W.11*	c.661C>T	4	p.Arg221Cys
H+weak	*FUT1*01W.12*	c.682A>G	4	p.Met228Val
H+weak	*FUT1*01W.13*	c.689A>C	4	p.Gln230Pro
H+weak	*FUT1*01W.14*	c.721T>C	4	p.Tyr241His
H+weak	*FUT1*01W.15*	c.801G>C	4	p.Trp267Cys
H+weak	*FUT1*01W.16*	c.801G>T	4	p.Trp267Cys
H+weak	*FUT1*01W.17*	c.832G>A	4	p.Asp278Asn
H+weak	*FUT1*01W.18*	c.903_904insAAC	4	p.Asn301_His302insAsn
H+weak	*FUT1*01W.19*	c.917C>T	4	p.Thr306Ile
H+weak	*FUT1*01W.20*	c.990delG	4	p.Pro331Glnfs*6
H+weak	*FUT1*01W.21*	c.235G>C	4	p.Gly79Arg
H+weak	*FUT1*01W.22*	c.991C>A	4	p.Pro331Thr
H+weak	*FUT1*01W.23*	c.424C>T	4	p.Arg142Trp
H+weak	*FUT1*01W.24*	c.649G>T	4	p.Val217Phe
H+weak	*FUT1*01W.25 Identical to FUT*01W.21?*	c.235G>C	4	p.Gly79Arg
H+weak	*FUT1*01W.26*	c.545G>A	4	p.Arg182His

H+weak	*FUT1*01W.27*	c.958G>A	4	p.Gly320Arg
H+weak	*FUT1*01W.28*	c.896A>C	4	p.Gln299Pro
H+weak	*FUT1*01W.29*	c.655G>C	4	p.Val219Leu
H+weak	*FUT1*02W.01*	c.269G>T	4	p.Gly90Val
H+weak	*FUT1*02W.02*	c.371T>G	4	p.Phe124Cys
H+weak	*FUT1*02W.03*	c.682A>G	4	p.Met228Val
H+weak	*FUT1*02W.04*	c.980A>C	4	p.Asn327Thr
H+weak	*FUT1*02W.05*	c.748C>T	4	p.Arg250Trp
Null alleles				
H–	*FUT1*01N.01*	c.422G>A	4	p.Trp141Ter
H–	*FUT1*01N.02*	c.461A>G	4	p.Tyr154Cys
H–	*FUT1*01N.03*	c.462C>A	4	p.Tyr154Ter
H–	*FUT1*01N.04*	c.513G>C	4	p.Trp171Cys
H–	*FUT1*01N.05*	c.538C>T	4	p.Gln180Ter
H–	*FUT1*01N.06*	c.551_552delAG	4	p.Glu184Valfs*85
H–	*FUT1*01N.07*	c.586C>T	4	p.Gln196Ter
H–	*FUT1*01N.08*	c.695G>A	4	p.Trp232Ter
H–	*FUT1*01N.09*	c.725T>G	4	p.Leu242Arg
H–	*FUT1*01N.10*	c.776T>A	4	p.Val259Glu
H–	*FUT1*01N.11*	c.785G>A; c.786C>A	4	p.Ser262Lys
H–	*FUT1*01N.12*	c.826C>T	4	p.Gln276Ter
H–	*FUT1*01N.13*	c.881_882delTT	4	p.Phe294Cysfs*40
H–	*FUT1*01N.14*	c.944C>T	4	p.Ala315Val
H–	*FUT1*01N.15*	c.948C>G	4	p.Tyr316Ter
H–	*FUT1*01N.16*	c.980A>C	4	p.Asn327Thr
H–	*FUT1*01N.17*	c.1047G>C	4	p.Trp349Cys
H–	*FUT1*01N.18*	c.684G>A	4	p.Met228Ile
H–	*FUT1*01N.19*	c.694T>C	4	p.Trp232Arg
H–	*FUT1*01N.20*	c.768delC	4	p.Val257Phefs*23
H–	*FUT1*02N.01*	c.423G>A	4	p.Trp141Ter

† Note that H expression will be masked if a functional *A* or *B* allele is also inherited. Also, that H antigen may be weakly detectable on RBCs where *FUT*01N* homozygosity occurs, due to the adsorption of soluble H antigen synthesized by *FUT2*.

Gene name: *FUT2*
Number of exons: 2
Initiation codon: Beginning of exon 2
Stop codon: Within exon 2
Entrez Gene ID: 2524
LRG sequence: NG_007511.1 (genomic)
NM_000511.5 (transcript)
Reference allele: *FUT2*01* (shaded)
Acceptable: *Se* if inferred by hemagglutination/inhibition

Phenotype (saliva) †	Allele name	Nucleotide change	Exon	Predicted amino acid change
H+	*FUT2*01*			
H+	*FUT2*02*	c.4G>A	2	p.Ala2Thr
H+	*FUT2*03.01*	c.40A>G	2	p.Ile14Val
H+	*FUT2*03.02*	c.40A>G; c.113C>T	2	p.Ile14Val; p.Ala38Val
H+	*FUT2*03.03*	c.40A>G; c.481G>A	2	p.Ile14Val; p.Asp161Asn
H+	*FUT2*04*	c.379C>T	2	p.Arg127Cys
H+	*FUT2*05*	c.400G>A	2	p.Val134Ile
H+	*FUT2*06*	c.481G>A	2	p.Asp161Asn
H+	*FUT2*07*	c.665G>A	2	p.Arg222His
H+	*FUT2*08*	c.685G>A	2	p.Val229Met
H+	*FUT2*09*	c.716G>A	2	p.Arg239Gln
H+	*FUT2*10*	c.747_748insGTG	2	p.249_250insVal
Weak phenotypes				
H+w	*FUT2*01W.01*	c.278C>T	2	p.Ala93Val
H+w	*FUT2*01W.02.01*	c.385A>T	2	p.Ile129Phe
H+w	*FUT2*01W.02.02*	c.385A>T; c.617T>G	2	p.Ile129Phe, p.Val206Gly
H+w	*FUT2*01W.02.03*	c.385A>T; c.841G>A	2	p.Ile129Phe, p.Gly281Arg
H+w	*FUT2*01W.03*	c.853G>A	2	p.Ala285Thr
Null phenotypes – Nucleotide polymorphisms				
H–	*FUT2*01N.01*	c.244G>A; c.385A>T	2	p.Ala82Thr; p.Ile129Phe
H–	*FUT2*01N.02*	c.428G>A;	2	p.Trp143Ter;

		c.739A>G (after termination?)		p.Gly247Ser (after termination?)
H–	*FUT2*01N.03*	c.569G>A	2	p.Arg190His
H–	*FUT2*01N.04*	c.571C>T	2	p.Arg191Ter
H–	*FUT2*01N.05*	c.628C>T	2	p.Arg210Ter
H–	*FUT2*01N.06*	c.658C>T	2	p.Arg220Ter
H–	*FUT2*01N.07*	c.664C>T	2	p.Arg222Cys
H–	*FUT2*01N.08*	c.685_686delGT	2	p.Val229Glyfs*4
H–	*FUT2*01N.09*	c.688_690delGTC	2	p.Val230del
H–	*FUT2*01N.10*	c.400G>A; c.760G>A	2	p.Val134Ile; p.Asp254Asn
H–	*FUT2*01N.11*	c.778delC	2	p.Pro260Leufs*16
H–	*FUT2*01N.12*	c.849G>A	2	p.Trp283Ter
H–	*FUT2*01N.13*	c.868 G>A	2	p.Gly290Arg
H–	*FUT2*01N.14*	c.950C>T	2	p.Pro317Leu
H–	*FUT2*01N.15*	c.302C>T	2	p.Pro101Leu
H–	*FUT2*01N.16*	c.960A>G (wrong position?)	2	p.Gly247Ser (wrong position?)
H–	*FUT2*01N.17*	c.412G>A	2	p.Gly138Ser
H–	*FUT2*01N.18*	c.818C>A	2	p.Thr273Asn
Null phenotypes – Gene deletions				
H–	*FUT2*0N.01*	Gene deletion		p.0
H–	*FUT2*0N.02*	Coding region deleted		p.0
H–	*FUT2*0N.03*	Fusion gene 1 between *FUT2* and *Sec1*		-
H–	*FUT2*0N.04*	Fusion gene 2 between *FUT2* and *Sec1*		-

† Saliva phenotype is shown here to represent secreted H antigen in all body fluids