

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 α 2FucT1 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 α 2FucT1 and α 2FucT2 are single pass type II membrane glycoproteins in the Golgi. The α 2FucT1 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 α 2FucT2 protein is encoded by *FUT2* or *Se*, if analysis is to predict a blood group antigen.

| | |
|-------------------|--|
| Gene name: | <i>FUT1</i> |
| 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: | <i>FUT1*01</i> (shaded) Acceptable: <i>H</i> if inferred by hemagglutination/inhibition |

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

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|--------------|--------------------|--------------------|---|------------------|
| H+weak | <i>FUT1*01W.27</i> | c.958G>A | 4 | p.Gly320Arg |
| H+weak | <i>FUT1*01W.28</i> | c.896A>C | 4 | p.Gln299Pro |
| H+weak | <i>FUT1*01W.29</i> | c.655G>C | 4 | p.Val219Leu |
| H+weak | <i>FUT1*02W.01</i> | c.269G>T | 4 | p.Gly90Val |
| H+weak | <i>FUT1*02W.02</i> | c.371T>G | 4 | p.Phe124Cys |
| H+weak | <i>FUT1*02W.03</i> | c.682A>G | 4 | p.Met228Val |
| H+weak | <i>FUT1*02W.04</i> | c.980A>C | 4 | p.Asn327Thr |
| H+weak | <i>FUT1*02W.05</i> | c.748C>T | 4 | p.Arg250Trp |
| Null alleles | | | | |
| H- | <i>FUT1*01N.01</i> | c.422G>A | 4 | p.Trp141Ter |
| H- | <i>FUT1*01N.02</i> | c.461A>G | 4 | p.Tyr154Cys |
| H- | <i>FUT1*01N.03</i> | c.462C>A | 4 | p.Tyr154Ter |
| H- | <i>FUT1*01N.04</i> | c.513G>C | 4 | p.Trp171Cys |
| H- | <i>FUT1*01N.05</i> | c.538C>T | 4 | p.Gln180Ter |
| H- | <i>FUT1*01N.06</i> | c.551_552delAG | 4 | p.Glu184Valfs*85 |
| H- | <i>FUT1*01N.07</i> | c.586C>T | 4 | p.Gln196Ter |
| H- | <i>FUT1*01N.08</i> | c.695G>A | 4 | p.Trp232Ter |
| H- | <i>FUT1*01N.09</i> | c.725T>G | 4 | p.Leu242Arg |
| H- | <i>FUT1*01N.10</i> | c.776T>A | 4 | p.Val259Glu |
| H- | <i>FUT1*01N.11</i> | c.785G>A; c.786C>A | 4 | p.Ser262Lys |
| H- | <i>FUT1*01N.12</i> | c.826C>T | 4 | p.Gln276Ter |
| H- | <i>FUT1*01N.13</i> | c.881_882delTT | 4 | p.Phe294Alafs*40 |
| H- | <i>FUT1*01N.14</i> | c.944C>T | 4 | p.Ala315Val |
| H- | <i>FUT1*01N.15</i> | c.948C>G | 4 | p.Tyr316Ter |
| H- | <i>FUT1*01N.16</i> | c.980A>C | 4 | p.Asn327Thr |
| H- | <i>FUT1*01N.17</i> | c.1047G>C | 4 | p.Trp349Cys |
| H- | <i>FUT1*01N.18</i> | c.684G>A | 4 | p.Met228Ile |
| H- | <i>FUT1*01N.19</i> | c.694T>C | 4 | p.Trp232Arg |
| H- | <i>FUT1*01N.20</i> | c.768delC | 4 | p.Val257Phefs*23 |
| H- | <i>FUT1*02N.01</i> | 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*.

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

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

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