

Names for KEL (ISBT 006) Blood Group Alleles

General description: The Kell blood group system consists of 32 antigens carried on a type II glycoprotein (aka CD238) of 732 amino acids. The Kell glycoprotein is a zinc-dependent metalloproteinase that has been shown to have endothelin-3-converting activity.

Gene name: *KEL*
 Number of exons: 19
 Initiation codon: Within exon 1
 Stop codon: Within exon 19
 Entrez Gene ID: 3792
 LRG sequence: NG_007492.1 (genomic)
 NM_000420.2 (transcript)
 Reference allele: *KEL*02* (shaded)
 Acceptable: *k* if inferred by haemagglutination

| Reference allele <i>KEL*02</i> encodes KEL2, KEL4, KEL5, KEL7, KEL11, KEL12, KEL14, KEL18, KEL19, KEL22, KEL26, KEL27, KEL29, KEL30, KEL32, KEL33, KEL34, KEL35, KEL36, KEL37, KEL38 | | | | |
|--|---------------------|--------------------|------|-----------------------------|
| Phenotype | Allele name | Nucleotide change† | Exon | Predicted amino acid change |
| KEL:1,-2 or K+ k- | <i>KEL*01.01</i> | c.578C>T | 6 | p.Thr193Met |
| KEL:1weak | <i>KEL*01.02</i> | c.577A>T | 6 | p.Thr193Ser |
| KEL:2 or k+ | <i>KEL*02</i> | | | |
| KEL:3,-4,-21 or Kp(a+b-c-) | <i>KEL*02.03</i> | c.841C>T | 8 | p.Arg281Trp |
| KEL:6,-7 or Js(a+b-) | <i>KEL*02.06</i> | c.1790T>C | 17 | p.Leu597Pro |
| KEL:10 | <i>KEL*02.10</i> | c.1481A>T | 13 | p.Glu494Val |
| KEL: -12 | <i>KEL*02.-12</i> | c.1643A>G | 15 | p.His548Arg |
| KEL:-14,-24 | <i>KEL*02.-14.1</i> | c.538C>T | 6 | p.Arg180Cys |
| KEL:-14 | <i>KEL*02.-14.2</i> | c.539G>A | 6 | p.Arg180His |
| KEL: -11,17 | <i>KEL*02.17</i> | c.905T>C | 8 | p.Val302Ala |
| KEL:-18 | <i>KEL*02.-18.1</i> | c.388C>T | 4 | p.Arg130Trp |
| KEL:-18 | <i>KEL*02.-18.2</i> | c.389G>A | 4 | p.Arg130Gln |
| KEL:-19 | <i>KEL*02.-19</i> | c.1475G>A | 13 | p.Arg492Gln |
| KEL:-3,-4,21 or Kp(a-b-c+) | <i>KEL*02.21</i> | c.842G>A | 8 | p.Arg281Gln |
| KEL:-22 | <i>KEL*02.-22</i> | c.965C>T | 9 | p.Ala322Val |

Names for KEL (ISBT 006) blood group alleles v4.0 160701

| | | | | |
|-----------------|---|--------------------------|-----------|--------------------------------------|
| KEL:23 | <i>KEL*02.23</i> | c.1145A>G | 10 | p.Gln382Arg |
| KEL: -14,24 | <i>KEL*02.24</i> | c.539G>C | 6 | p.Arg180Pro |
| KEL:25,-28 | <i>KEL*02.25</i> | c.743G>A | 8 | p.Arg248Gln |
| KEL:-26 | <i>KEL*02.-26</i> | c.1217G>A | 11 | p.Arg406Gln |
| KEL:-27 | <i>KEL*02.-27</i> | c.745G>A | 8 | p.Glu249Lys |
| KEL:-25,28 | <i>KEL*02.28</i> | c.742C>T | 8 | p.Arg248Trp |
| KEL:-29 | <i>KEL*02.-29</i> | c.1868G>A | 17 | p.Arg623Lys |
| KEL:-30 | <i>KEL*02.-30</i> | c.913G>A | 8 | p.Asp305Asn |
| KEL:31,-38 | <i>KEL*02.31</i> | c.875G>A | 8 | p.Arg292Gln |
| KEL:-32 | <i>KEL*02.-32</i> | c.1271C>T | 11 | p.Ala424Val |
| KEL:-33 | <i>KEL*02.-33</i> | c.1283G>T | 11 | p.Arg428Leu |
| KEL:-34 | <i>KEL*02.-34</i> | c.758A>G | 8 | p.Tyr253Cys |
| KEL:-35 [1] | <i>KEL*02.-35</i> | c.780G>T; c.2024G>A | 8 18 | p.Leu260Phe; p.Arg675Gln |
| KEL:-36 [2] | <i>KEL*02.-36</i> | c.1391C>T | 12 | p.Thr464Ile |
| KEL:-37 | <i>KEL*02.-37</i> | c.877C>T | 8 | p.Arg293Trp |
| Null phenotypes | | | | |
| K ₀ | <i>KEL*01N.01</i> | c.578C>T; c.1678C>G | 6 15 | p.Thr193Met; p.Pro560Ala |
| K ₀ | <i>KEL*01N.02</i> | c.244T>C; c.578C>T | 4 6 | p.Cys82Arg; p.Thr193Met |
| K ₀ | <i>KEL*02N.01</i> | c.223+1G>C | Intron 3 | p.Arg75fs*?; Alternative splicing |
| K ₀ | <i>KEL*02N.02</i> Identical to <i>KEL*02N.26?</i> | c.382C>T; c.1790C (?) | 4 (17) | p.Arg128Ter |
| K ₀ | <i>KEL*02N.03</i> | c.246T>A | 4 | p.Cys82Ter |
| K ₀ | <i>KEL*02N.04</i> | c.1042C>T | 9 | p.Gln348Ter |
| K ₀ | <i>KEL*02N.05</i> | c.2027G>A | 18 | p.Ser676Asn |
| K ₀ | <i>KEL*02N.06</i> | c.223+1G>A | Intron 3 | p.Arg75fs; Alternative splicing |
| K ₀ | <i>KEL*02N.07</i> | c.574C>T | 6 | p.Arg192Ter |
| K ₀ | <i>KEL*02N.08</i> | c.526-2A>G | Intron 5 | Alternative splicing |
| K ₀ | <i>KEL*02N.09</i> | c.1377G>A | 12 | p.Trp459Ter |

Names for KEL (ISBT 006) blood group alleles v4.0 160701

| | | | | |
|--|---|------------------------|----------|-----------------------------|
| K ₀ | <i>KEL*02N.10</i> | c.1420C>T | 13 | p.Gln474Ter |
| K ₀ | <i>KEL*02N.11</i> | c.903delG | 8 | p.Val302Serfs*28 |
| K ₀ | <i>KEL*02N.12</i> | c.924+1G>A | Intron 8 | Alternative splicing |
| K ₀ | <i>KEL*02N.13</i> | c.924+1G>T | Intron 8 | Alternative splicing |
| K ₀ | <i>KEL*02N.14</i> | c.948G>A | 9 | p.Trp316Ter |
| K ₀ | <i>KEL*02N.15</i> | c.1216C>T | 11 | p.Arg406Ter |
| K ₀ | <i>KEL*02N.16</i> | c.1477C>T | 13 | p.Gln493Ter |
| K ₀ | <i>KEL*02N.17</i> | c.1546C>T | 14 | p.Arg516Ter |
| K ₀ | <i>KEL*02N.19</i> | c.2023C>T | 18 | p.Arg675Ter |
| K ₀ | <i>KEL*02N.20</i> | c.1596G>A | 15 | p.Trp532Ter |
| K ₀ | <i>KEL*02N.21</i> | c.1947C>G | 18 | p.Tyr649Ter |
| K ₀ | <i>KEL*02N.22</i> | c.736-1G>C | Intron 7 | Alternative splicing |
| K ₀ | <i>KEL*02N.23</i> | c.184_185insT | 3 | p.Ser62Phefs*17 |
| K ₀ | <i>KEL*02N.24</i> | c.715G>T | 7 | p.Glu239Ter |
| K ₀ | <i>KEL*02N.25</i> | c.1975delG | 19 | p.Glu659Argfs*22 |
| K ₀ | <i>KEL*02N.26</i> Identical to <i>KEL*02N.02?</i> | c.382C>T | 4 | p.Arg128Ter |
| K ₀ | <i>KEL*02N.27</i> | c.730delG | 7 | p.Ala244Profs*8 |
| K ₀ | <i>KEL*02N.28</i> | c.230G>T | 4 | p.Cys77Phe |
| K ₀ | <i>KEL*02N.29</i> | c.1664G>A | 15 | p.Gly555Glu |
| Mod phenotypes Classification of a mod phenotype may depend on the reagents used. | | | | |
| Kmod; KEL:1weak | <i>KEL*01M.01</i> | c.578C>G | 6 | p.Thr193Arg |
| Kmod | <i>KEL*02M.01</i> | c.1088G>A | 10 | p.Ser363Asn |
| Kmod | <i>KEL*02M.02</i> | c.2030A>G | 18 | p.Tyr677Cys |
| Kmod KEL:-13 | <i>KEL*02M.03</i> | c.986T>C | 9 | p.Leu329Pro |
| Kmod | <i>KEL*02M.04</i> | c.2107G>A | 19 | p.Gly703Arg |
| Kmod | <i>KEL*02M.05</i> (Only a silent mutation?) | c.1719C>T | 16 | p.Gly573= |
| Kmod | <i>KEL*02M.06</i> | c.306C>A; c.1298C>T | 4 11 | p.Asp102Glu; p.Pro433Leu |

Names for KEL (ISBT 006) blood group alleles v4.0 160701

| | | | | |
|------|-------------------|-----------|----|-------------|
| Kmod | <i>KEL*02M.07</i> | c.1763A>G | 16 | p.Tyr588Cys |
| Kmod | <i>KEL*02M.08</i> | c.1490A>T | 13 | p.Asp497Val |
| Kmod | <i>KEL*02M.09</i> | c.1757T>G | 16 | p.Ile586Ser |
| Kmod | <i>KEL*02M.10</i> | c.787G>A | 8 | p.Gly263Arg |
| Kmod | <i>KEL*02M.11</i> | c.1268C>T | 11 | p.Ala423Val |

† Nucleotide 1 is the first nucleotide of the translation-initiating codon, which is 120 bp downstream of the traditional position for the first nucleotide in early reports.

1. Karamatic Crew V, et al. KELP (KEL35): a new high incidence antigen in the Kell blood group system defined by two homozygous missense mutations in KEL. *Transfus Med* 2010; 20(Suppl.1):30.
2. Karamatic Crew V, et al. KETI, a novel high incidence antigen in the Kell blood group system: a serological and molecular study. *Vox Sanguinis* 2011;101(Suppl 1):19.