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 *KEL*02* 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−	*KEL*01.01*	c.578C>T	6	p.Thr193Met
KEL:1weak	*KEL*01.02*	c.577A>T	6	p.Thr193Ser
KEL:2 or k+	*KEL*02*			
KEL:3,−4,−21 or Kp(a+b–c–)	*KEL*02.03*	c.841C>T	8	p.Arg281Trp
KEL:6,−7 or Js(a+b–)	*KEL*02.06*	c.1790T>C	17	p.Leu597Pro
KEL:10	*KEL*02.10*	c.1481A>T	13	p.Glu494Val
KEL: −12	*KEL*02.−12*	c.1643A>G	15	p.His548Arg
KEL:−14,−24	*KEL*02.−14.1*	c.538C>T	6	p.Arg180Cys
KEL:−14	*KEL*02.−14.2*	c.539G>A	6	p.Arg180His
KEL: −11,17	*KEL*02.17*	c.905T>C	8	p.Val302Ala
KEL:−18	*KEL*02.−18.1*	c.388C>T	4	p.Arg130Trp
KEL:−18	*KEL*02.−18.2*	c.389G>A	4	p.Arg130Gln
KEL:−19	*KEL*02.−19*	c.1475G>A	13	p.Arg492Gln
KEL:−3,−4,21 or Kp(a–b–c+)	*KEL*02.21*	c.842G>A	8	p.Arg281Gln
KEL:−22	*KEL*02.−22*	c.965C>T	9	p.Ala322Val

KEL:23	*KEL*02.23*	c.1145A>G	10	p.Gln382Arg
KEL: −14,24	*KEL*02.24*	c.539G>C	6	p.Arg180Pro
KEL:25,−28	*KEL*02.25*	c.743G>A	8	p.Arg248Gln
KEL:−26	*KEL*02.−26*	c.1217G>A	11	p.Arg406Gln
KEL:−27	*KEL*02.−27*	c.745G>A	8	p.Glu249Lys
KEL:−25,28	*KEL*02.28*	c.742C>T	8	p.Arg248Trp
KEL:−29	*KEL*02.−29*	c.1868G>A	17	p.Arg623Lys
KEL:−30	*KEL*02.−30*	c.913G>A	8	p.Asp305Asn
KEL:31,–38	*KEL*02.31*	c.875G>A	8	p.Arg292Gln
KEL:−32	*KEL*02.−32*	c.1271C>T	11	p.Ala424Val
KEL:−33	*KEL*02.−33*	c.1283G>T	11	p.Arg428Leu
KEL:−34	*KEL*02.−34*	c.758A>G	8	p.Tyr253Cys
KEL:−35 [1]	*KEL*02.−35*	c.780G>T; c.2024G>A	8 18	p.Leu260Phe; p.Arg675Gln
KEL:–36 [2]	*KEL*02.–36*	c.1391C>T	12	p.Thr464Ile
KEL:−37	*KEL*02.–37*	c.877C>T	8	p.Arg293Trp
Null phenotypes				
K_0	*KEL*01N.01*	c.578C>T; c.1678C>G	6 15	p.Thr193Met; p.Pro560Ala
K_0	*KEL*01N.02*	c.244T>C; c.578C>T	4 6	p.Cys82Arg; p.Thr193Met
K_0	*KEL*02N.01*	c.223+1G>C	Intron 3	p.Arg75fs*?; Alternative splicing
K_0	*KEL*02N.02* Identical to *KEL*02N.26*?	c.382C>T; c.1790C (?)	4 (17)	p.Arg128Ter
K_0	*KEL*02N.03*	c.246T>A	4	p.Cys82Ter
K_0	*KEL*02N.04*	c.1042C>T	9	p.Gln348Ter
K_0	*KEL*02N.05*	c.2027G>A	18	p.Ser676Asn
K_0	*KEL*02N.06*	c.223+1G>A	Intron 3	p.Arg75fs; Alternative splicing
K_0	*KEL*02N.07*	c.574C>T	6	p.Arg192Ter
K_0	*KEL*02N.08*	c.526-2A>G	Intron 5	Alternative splicing
K_0	*KEL*02N.09*	c.1377G>A	12	p.Trp459Ter

K_0	*KEL*02N.10*	c.1420C>T	13	p.Gln474Ter
K_0	*KEL*02N.11*	c.903delG	8	p.Val302Serfs*28
K_0	*KEL*02N.12*	c.924+1G>A	Intron 8	Alternative splicing
K_0	*KEL*02N.13*	c.924+1G>T	Intron 8	Alternative splicing
K_0	*KEL*02N.14*	c.948G>A	9	p.Trp316Ter
K_0	*KEL*02N.15*	c.1216C>T	11	p.Arg406Ter
K_0	*KEL*02N.16*	c.1477C>T	13	p.Gln493Ter
K_0	*KEL*02N.17*	c.1546C>T	14	p.Arg516Ter
K_0	*KEL*02N.19*	c.2023C>T	18	p.Arg675Ter
K_0	*KEL*02N.20*	c.1596G>A	15	p.Trp532Ter
K_0	*KEL*02N.21*	c.1947C>G	18	p.Tyr649Ter
K_0	*KEL*02N.22*	c.736-1G>C	Intron 7	Alternative splicing
K_0	*KEL*02N.*23	c.184_185insT	3	p.Ser62Phefs*17
K_0	*KEL*02N.24*	c.715G>T	7	p.Glu239Ter
K_0	*KEL*02N.25*	c.1975delG	19	p.Glu659Argfs*22
K_0	*KEL*02N.26* Identical to KEL*02N.02?	c.382C>T	4	p.Arg128Ter
K_0	*KEL*02N.27*	c.730delG	7	p.Ala244Profs*8
K_0	*KEL*02N.28*	c.230G>T	4	p.Cys77Phe
K_0	*KEL*02N.29*	c.1664G>A	15	p.Gly555Glu
Mod phenotypes Classification of a mod phenotype may depend on the reagents used.				
Kmod; KEL:1weak	*KEL*01M.01*	c.578C>G	6	p.Thr193Arg
Kmod	*KEL*02M.01*	c.1088G>A	10	p.Ser363Asn
Kmod	*KEL*02M.02*	c.2030A>G	18	p.Tyr677Cys
Kmod KEL:−13	*KEL*02M.03*	c.986T>C	9	p.Leu329Pro
Kmod	*KEL*02M.04*	c.2107G>A	19	p.Gly703Arg
Kmod	*KEL*02M.05 (Only a silent mutation?)*	c.1719C>T	16	p.Gly573=
Kmod	*KEL*02M.06*	c.306C>A; c.1298C>T	4 11	p.Asp102Glu; p.Pro433Leu

Kmod	*KEL*02M.07*	c.1763A>G	16	p.Tyr588Cys
Kmod	*KEL*02M.08*	c.1490A>T	13	p.Asp497Val
Kmod	*KEL*02M.09*	c.1757T>G	16	p.Ile586Ser
Kmod	*KEL*02M.10*	c.787G>A	8	p.Gly263Arg
Kmod	*KEL*02M.11*	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.