Names for Kx (ISBT 019) Blood Group Alleles

Intro

General description:	The Kx blood group system contains one antigen carried on a multipass protein of 444 amino acids, which is linked to the Kell glycoprotein through a disulphide bond.
Gene name:	*XK*
Number of exons:	3
Initiation codon:	Within exon 1
Stop codon:	Within exon 3
Entrez Gene ID:	7504
LRG:	LRG_812
LRG sequence:	NG_007473.3 (genomic) NM_021083.4 (transcript) NP_066569.1
Reference allele:	*XK*01 (shaded)*
Reference allele *XK*01* encodes:	*Kx+*
Antithetical antigens:	The Kx system consists of one antigen, Kx (XK:1 or ISBT 019 001). No antithetical antigen has been reported

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID:	Accession number	rs number
XK:1 or Kx+	XK*01				(1), PMID: 8004674	NG_007473.2	
XK:1 or Kx+	*XK*01.02*	c.121T>G	1	p.(Leu41Val)	(46), PMID: in submission	KY926705	
Null phenotypes							
XK:-1 kx-	*XK*N.01*	Deletion of *XK* gene	1 to 3	No protein present	See *XK*N.01* series below for references		
XK:-1 kx-	*XK*N.02*	del Exon 1	1	p.(Met1_Arg82del)	(2), PMID: 11761473	In Submission	
XK:-1 kx-	*XK*N.03*	del Promoter + Exon 1	1	p.(Met1_Arg82del)	(3), PMID: 15504163	AY655133 AY655134 AY655135	
XK:-1 kx-	*XK*N.04*	del Exon 2	2	p.(Arg82_Ser170del)	(4), PMID: 12899725		
XK:-1 kx-	*XK*N.05*	del Intron 2 + Exon 3	3	p.(Ser170_Ala444del)			
XK:-1 kx-	*XK*N.06*	del -272 to 119	1 + 2	p.(Met1fs*45)			
XK:-1 kx-	*XK*N.07*	c.172delG	1 + 2	p.(Val58Tyrfs*72)	(5), PMID: 16314760		
XK:-1 kx-	*XK*N.08*	c.269delA	2	p.(Tyr90Serfs*40)			
XK:-1 kx-	*XK*N.09*	c.268delT	2	p.(Tyr90Thrfs*40)	(6), PMID: 8619554		
XK:-1 kx-	*XK*N.10*	c.450_451insC	2 + 3	p.(Gln151Profs*48)	(7), PMID: 10930599 (8), PMID: 16344536	KY939773.1	
XK:-1 kx-	*XK*N.11*	c.686_687delTT	3	p.(Phe229Tyrfs*36)	(9), PMID: 11761473		
XK:-1 kx-	*XK*N.12*	c.771delG	3	p.(Trp257Cysfs*11)	(9), PMID: 11761473		
XK:-1 kx-	*XK*N.13*	c.856_860delCTCTA	3	p.(Leu286Tyrfs*16)	(9), PMID: 11761473 (10), PMID: 23943810		
XK:-1 kx-	*XK*N.14*	c.938_951del	3	p.(Asn313Thrfs*24)	(9), PMID: 11761473		
XK:-1 kx-	*XK*N.15*	c.1013delT	3	p.(Phe338Serfs*70)	(11), PMID: 10426139		rs1602159120

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID:	Accession number	rs number
XK:-1 kx-	*XK*N.16*	c.107G>A	1	p.(Trp36*)	(9), PMID: 11761473		
XK:-1 kx-	*XK*N.17*	c.397C>T	2	p.(Arg133*)	(9), PMID: 11761473 (12), PMID: 18167163 (13), PMID: 17870653		
XK:-1 kx-	*XK*N.18*	c.463C>T	2	p.(Gln155*)	(9), PMID: 11761473		
XK:-1 kx-	*XK*N.19*	c.707G>A	3	p.(Trp236*)	(9), PMID: 11761473		
XK:-1 kx-	*XK*N.20*	c.895C>T	3	p.(Gln299*)	(14), PMID: 11261514		rs104894954
XK:-1 kx-	*XK*N.21*	c.941G>A	3	p.(Trp314*)	(15), PMID: 11703337		rs104894953
XK:-1 kx-	*XK*N.22*	c.245+1G>C	1	Aberrant splicing	(16), PMID: 11961232 (17), PMID: 11099667		
XK:-1 kx-	*XK*N.23*	c.246-1G>A	1	Aberrant splicing	(18), PMID: 19040496		
XK:-1 kx-	*XK*N.24*	c.508+1G>A	2	Aberrant splicing	(1), PMID: 8004674 (16), PMID: 11961232		rs1602145991
XK:-1 kx-	*XK*N.25*	c.508+5G>A	2	Aberrant splicing	(19), PMID: 8916972 (20), PMID: 17302777		
XK:-1 kx-	*XK*N.26*	c.509-1G>A	2	Aberrant splicing	(1), PMID: 8004674		rs1602158863
XK:-1 kx-	*XK*N.27*	c.664C>G	3	p.Arg222Gly	(16), PMID: 11961232 (20), PMID: 17302777		
XK:-1 kx-	*XK*N.28*	c.880T>C	3	p.(Cys294Arg)	(9), PMID: 11761473		rs28933690
XK:-1 kx-	*XK*N.29*	c.979G>A	3	p.Glu327Lys	(21), PMID: 12823753		
XK:-1 kx-	*XK*N.30*	c.1124G>C	3	p.(Arg375Pro)	(22), PMID: 9268240		
XK:-1 kx-	*XK*N.31*	c.1134C>G	3	p.(Asn378Lys)	(22), PMID: 9268240		
XK:-1 kx-	*XK*N.32*	c.962A>G	3	p.(Tyr321Cys)	(23), PMID: 21145924		
XK:-1 kx-	*XK*N.33*	c.509-13C>G	3	Aberrant splicing	(23), PMID: 21145924		
XK:-1 kx-	*XK*N.34*	c.523insA	3	p.(Ile175Asnfs*24)	(24), PMID: 21463873		
XK:-1 kx-	*XK*N.35*	c.509-2A>G	3	Aberrant splicing	(24), PMID: 21463873	MH729875	

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID:	Accession number	rs number
XK:-1 kx-	*XK*N.36*	c.229delC	1 + 2	p.(Leu80Phefs*50)	(25), PMID: 24529944		
XK:-1 kx-	*XK*N.37*	c.154C>T	1	p.(Gln52*)	(26), PMID: 24635891		
XK:-1 kx-	*XK*N.38*	669-673del5ins13(delTGTAGinsGGTCCTCTTTACC)	3	p.(Val225Leufs*12)	(27), doi: 10.1111/ncn3.12042 (28), doi 10.1111/ncn3.12256		
XK:-1 kx-	*XK*N.39*	c.195-198delCCGC	1	p.(Pro67Serfs*62)	(29), PMID: 28555782	LT838808.1	
XK:-1 kx-	*XK*N.40*	c.640_645del TGGAGG	3	p.(Trp213_Arg214del)	(30), doi: 10.1093/ajcp/140.suppl1.063		
XK:-1 kx-	*XK*N.41*	c.475delA	2 + 3	p.(Ser159Valfs*15)	(31), Abstract	MH730936	
XK:-1 kx-	*XK*N.42*	c.1015A>T	3	p.(Lys339*)	(31), Abstract	MH727540	
XK:-1 kx-	*XK*N.43*	c.452A>C	2	p.(Gln151Pro)	(31), Abstract	MH727541	
XK:-1 kx-	*XK*N.44*	c.82insC	1	p.(Tyr28Leufs*58)	(32), PMID: 33190237	LC543982	
XK:-1 kx-	*XK*N.45*	c.435delC	2	p.(Leu146Trpfs*7)	(33), Genbank submission number only	MK071977.1	
XK:-1 kx-	*XK*N.46*	c.642G>A	3	p.(Trp214*)	(34) PMID: 29524658		
XK:-1 kx-	*XK*N.47*	c.942G>A	3	p.(Trp314*)	(35) Abstract, (46) PMID: in submission	KM821167.1	
XK:-1 kx-	*XK*N.48*	c.577A>T	3	p.(Lys193*)	(46) PMID: in submission		
Null phenotypes in the *XK*N.01* .series from *XK*N.01.001* to *099*: represents *XK* allele deleted but precise breakpoints unknown							
Phenotype	**Allele name**	**Size of deletion** (chromosomal location)	**Exon Intron**	**Predicted amino acid change**	**(Reference No.) PMID:**	**Accession number**	**rs number**
XK:-1 kx-	*XK*N.01.001*	del Xp22.3 – 21.1	1 to 3	No protein present	(36), PMID: 6510024		

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID:	Accession number	rs number
XK:-1 kx-	*XK*N.01.002*	del Xp21.3 – 21.1 5-10mbp	1 to 3	No protein present	(37), PMID: 4039107		
XK:-1 kx-	*XK*N.01.003*	del Xp21.2-21.1	1 to 3	No protein present	(38), PMID: 3358422		
XK:-1 kx-	*XK*N.01.004*	del Xp21.2	1 to 3	No protein present	(38) PMID: 3358422		
XK:-1 kx-	*XK*N.01.005*	del Xp21.2-21.1	1 to 3	No protein present	(1), PMID: 8004674 (2), PMID: 11761473		
XK:-1 kx-	*XK*N.01.006*	del Xp21.2-21.1	1 to 3	No protein present	(39), PMID: 3334897		
XK:-1 kx-	*XK*N.01.007*	del Xp21.2-21.1	1 to 3	No protein present	(40), PMID: 3417309		
XK:-1 kx-	*XK*N.01.008*	del Xp21.2-21.1	1 to 3	No protein present	(41), PMID: 10651848		
XK:-1 kx-	*XK*N.01.009*	del Xp21.2-21.1	1 to 3	No protein present	(41), PMID: 10651848		
XK:-1 kx-	*XK*N.01.010*	del Xp21.2-21.1	1 to 3	No protein present	(41), PMID: 10651848		
XK:-1 kx-	*XK*N.01.011*	del Xp21.2-21.1	1 to 3	No protein present	(42), PMID: 24446915		
Null phenotypes in the *XK*N.01.* series from *XK*N.01.100* onward: *XK* allele deleted and precise breakpoints mapped							
XK:-1 kx-	*XK*N.01.100*	del 1.12mbp (telomeric *LOC441488* to centromeric *XK* Intron 2)	1 to 2	No protein present	(43), PMID: 17300882		
XK:-1 kx-	*XK*N.01.101*	del 5.65mbp (telomeric *TCTE1L* to centromeric *DMD*)	1 to 3	No protein present	(43), PMID: 17300882		
XK:-1 kx-	*XK*N.01.102*	del 0.59mpb (telomeric *LANCL3* to centromeric *DYNLT3*)	1 to 3	No protein present	(44), PMID: 22383943		

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID:	Accession number	rs number
XK:-1 kx-	*XK*N.01.103*	del 1.94mbp (telomeric *CXorf22* to centromeric *DYNLT3*)	1 to 3	No protein present	(44), PMID: 22383943		
XK:-1 kx-	*XK*N.01.104*	del 5.71mbp (telomeric *DMD* to centromeric *DYNLT3*)	1 to 3	No protein present	(44), PMID: 22383943		
XK:-1 kx-	*XK*N.01.105*	del 0.151mpb (telomeric *LANCL3* to centromeric *CYBB*)	1 to 3	No protein present	(45), PMID: 28555782	LT838809	

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	Track of changes		from version	to version
1	**Version**		**v4.0 29th April 2019**	**v5.0 30-JUN-2021**
2	Author	created:	Eileen Roulis, v4.0	Catherine Hyland, and Nysa McGowan Dec to Feb 2020
3	Reviewer	reviewed:	Catherine Hyland	Eileen Roulis, February 2021, Christoph Gassner, June 2021
4	General		Last word version published on ISBT website	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	Intro	Reference allele line moved from Allele Table to Intro:		Reference allele *XK*01* (shaded) Reference allele *XK*01* encodes:Kx+
6	Allele Table			Table columns "(Reference No.) PMID", "Accession number" and "rs-number" added, content added.
7	Allele Table	Text change: Line moved to Intro:	Reference allele *XK*01* (shaded) Reference allele *XK*01* encodes: Kx+	see above
8	Allele Table	Text change:		Nucleotide changes described according to HUGO Nomenclature for all entries: From *XK*N.07* nuceotide change 172delG becomes c.172delG. Predicted amino acid change V58Y +fs 129X becomes p.(Val58Tyrfs*72). Corresponding conversions occur for all entries. Brackets indicate predicted amino acid change.
9	Allele Table	Antigen/allele added:	n.a.	*XK*N.44* , *XK*N.45* , *XK*N.46* and *XK*N.47* added as described in references 32 to 35
10	References	All references new:	n.a.	All references (1) to (4+D45) provided in Reference format for the first time.
11	Allele Table	Two alleles added	n.a.	Added alleles *XK*01.01, XK*N.48*
12	References	One reference added+B2	n.a.	Added reference 46 to *XK*01.01, XK*N.47* and *XK*N.48*
13	**End Version**		**v4.0 29th April 2019**	**v5.0 30-JUN-2021**