Names for Transcription Factor KLF1 Alleles

General Description	*KLF1* encodes EKLF (erythroid Krüppel-like factor 1) which does not represent a blood group system. Variation in this gene is associated with In(Lu), a phenotype inherited in a dominant fashion. The In(Lu) phenotype is characterized by reduced expression of antigens of the Lutheran system and for P1, Inb, and AnWj antigens.
General Description (from GeneCards)	This gene encodes a hematopoietic-specific transcription factor that induces high-level expression of adult beta-globin and other erythroid genes. The zinc-finger protein binds to the DNA sequence CCACACCCT found in the beta hemoglobin promoter. Heterozygous loss-of-function mutations in this gene result in the dominant In(Lu) blood phenotype.
Gene name:	*KLF1* , also known as *EKLF, EKLF/KLF1*
RefGene Name	LRG_825
Number of exons:	3
Initiation codon:	Within exon 1
Stop codon:	Within exon 3
Entrez Gene ID:	10661
LRG sequence (Genomic)	NG_013087.1
LRG sequence (Transcript)	NM_006563.3
Reference allele:	*KLF1*01*

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
Common	*KLF1*01*						
In(Lu)	*KLF1*BGM01*	c.-124T>C	Promoter	p.0	PMID: 18487511		not found
In(Lu)	*KLF1*BGM02*	c.380T>A	2	p.Leu127Ter	PMID: 18487511		not found
In(Lu)	*KLF1*BGM03*	c.569delC	2	p.Pro190Leufs*47	PMID: 18487511		not found
In(Lu)	*KLF1*BGM04*	c.874A>T	2	p.Lys292Ter	PMID: 18487511		rs137852687
In(Lu)	*KLF1*BGM05*	c.895C>T	2	p.His299Tyr	PMID: 18487511		rs137852688
In(Lu)	*KLF1*BGM06*	c.954dupG	3	p.Arg319Glufs*34	PMID: 18487511		rs397514445
In(Lu)	*KLF1*BGM07*	c.983G>T	3	p.Arg328Leu	PMID: 18487511		not found
In(Lu)	*KLF1*BGM08*	c.983G>A	3	p.Arg328His	PMID: 18487511		rs140252918
In(Lu)	*KLF1*BGM09*	c.991C>G	3	p.Arg331Gly	PMID: 18487511		not found
CO:−1,−2 IN:−1,−2	*KLF1*BGM10*	c.973G>A	3	p.Glu325Lys	PMID: 29047116		rs267607201
In(Lu)	*KLF1*BGM11*	c.90G>A	1	p.Trp30Ter	PMID: 29047116		rs1427628301
Obsolete Normal BG phenotype	*KLF1*BGM12*	c.304T>C	2	p.Ser102Pro	PMID: 29047116		rs2072597
In(Lu)	*KLF1*BGM13*	c.114delC	2	p.Asp38Glufs*53	PMID: 27043150		not found
In(Lu)	*KLF1*BGM14*	c.298G>T	2	p.Glu100Ter	PMID: 27043150		not found
In(Lu)	*KLF1*BGM15*	c.304T>C c.484_484insC	2	p.Ser103Pro p.Lys162Glnfs*352	PMID: 27043150		rs2072597 rs116107190
In(Lu)	*KLF1*BGM16*	c.304T>C c.1002delGT	2, 3	p.Ser103Pro p.Thr334Glyfs*351	PMID: 27043150		rs2072597 rs764608850
In(Lu)	*KLF1*BGM17*	c.621C>G	2	p.Tyr207Ter	PMID: 27043150		not found
In(Lu)	*KLF1*BGM18*	c.948delC	3	p.Cys316Trpfs*326	PMID: 27043150		not found
In(Lu)	*KLF1*BGM19*	c.1040C>A c.1045delT	3	p.Ala347Asp p.Ser349Argfs*358	PMID: 27043150		not found

In(Lu)	*KLF1*BGM20*	c.809C>A	2	p.Ser270X	PMID: 21273267 PMID: 21821711		rs558942739
In(Lu)	*KLF1*BGM21*	c.977T>G	3	p.Leu326Arg	PMID: 21821711 PMID: 23125034		rs397514634
In(Lu)	*KLF1*BGM22*	c.994A>G	3	p.Lys332Glu	PMID: 29193102		not found
In(Lu)	*KLF1*BGM23*	c.517_519delC	2	p.Pro173ProfsX64	PMID: 28194794		rs566095433
In(Lu)	*KLF1*BGM24*	c.551_556GGACCG>A	2	p.Gly184GlufsX167	PMID: 28194794		not found
In(Lu)	*KLF1*BGM25*	c.637C>T	2	p.Glu213X	PMID: 28194794		not found
In(Lu)	*KLF1*BGM26*	c.802C>T	2	p.Arg268X	PMID: 28194794		not found
In(Lu)	*KLF1*BGM27*	c.899T>C	2	p.Leu300Pro	PMID: 28194794		rs763096758
In(Lu)	*KLF1*BGM28*	c.902insT	2	p.Arg301LeufsX52	PMID: 28194794		not found
In(Lu)	*KLF1*BGM29*	c.947G>A	3	p.Cys316Tyr	PMID: 28194794		not found
In(Lu)	*KLF1*BGM30*	c.968C>G	3	p.Ser323Trp	PMID: 23125034		not found
In(Lu)	*KLF1*BGM31*	c.991C>T	3	p.Arg331Trp	PMID: 28194794 PMID: 24443441		not found
In(Lu)	*KLF1*BGM32*	c.310_311insG	2	p.Ala104GlyfsX249	PMID: 23125034		not found
In(Lu)	*KLF1*BGM33*	c.519_520insC	2	p.Gly174ArgfsX179	PMID: 23125034		not found
In(Lu)	*KLF1*BGM34*	c.519_525dupCGGCGCC	2	p.Gly176ArgfsX179	PMID: 23125034 PMID: 23844643 (1), Abstract		rs483352838
In(Lu)	*KLF1*BGM35*	c.591C>G	2	p.Tyr197X	PMID: 23125034		not found
In(Lu)	*KLF1*BGM36*	c.663delG	2	p.Leu222SerfsX15	PMID: 23125034		not found
In(Lu)	*KLF1*BGM37*	c.862A>G	2	p.Lys288Glu	PMID: 23125034		not found
In(Lu)	*KLF1*BGM38*	c.1071C>A	3	p.His357Gln	PMID: 23125034		VCV000040066.1
In(Lu)	*KLF1*BGM39*	c.109C>T	2	p.Gln37X	PMID: 28194794		rs755193431
In(Lu)	*KLF1*BGM40*	c.196G>T	2	p.Glu66X	PMID: 28194794		not found

In(Lu)	*KLF1*BGM41*	c.204delC	2	p.Gly68GlyfsX169	PMID: 28194794		not found
In(Lu)	*KLF1*BGM42*	c.796C>T	2	p.Arg266X	PMID: 28194794		rs756046932
In(Lu)	*KLF1*BGM43*	c.1022G>A	3	p.Cys341Tyr	PMID: 28194794		rs483352839
In(Lu)	*KLF1*BGM44*	c.151delC	2	p.Leu51SerfsX3	PMID: 28194794		not found
In(Lu)	*KLF1*BGM45*	c.826C>T	2	p.Gln276X	PMID: 28194794		not found
In(Lu)	*KLF1*BGM46*	c.868T>C	2	p.Tyr290His	PMID: 28194794		not found
In(Lu)	*KLF1*BGM47*	c.914-1g>c	i2	Splicing failure	PMID: 28194794		not found
In(Lu)	*KLF1*BGM48*	c.1001C>T	3	p.Thr334Met	PMID: 28194794		rs483352841
In(Lu)	*KLF1*BGM49*	c.1048C>T	3	p.Arg350Cys	PMID: 28194794		rs1321730417
In(Lu)	*KLF1*BGM50*	c.86A>G	1	p.Lys29Arg	PMID: 28194794		not found
In(Lu)	*KLF1*BGM51*	c.262_284dup	2	p.Ala95AlafsX150	PMID: 28194794		not found
In(Lu)	*KLF1*BGM52*	c.472delG	2	p.Ala158ProfsX79	PMID: 28194794		not found
In(Lu)	*KLF1*BGM53*	c.533C>A	2	p.Ser178X	PMID: 28194794		not found
In(Lu)	*KLF1*BGM54*	c.887T>C	2	p.Leu296Pro	PMID: 28194794		rs754996390
In(Lu)	*KLF1*BGM55*	c.939G>A	3	p.Trp313X	PMID: 28194794		rs1364414353
In(Lu)	*KLF1*BGM56*	c.964C>A	3	p.Arg322Ser	PMID: 28194794		rs376711350
In(Lu)	*KLF1*BGM57*	c.1004G>C	3	p.Gly335Ala	PMID: 28194794		not found
In(Lu)	*KLF1*BGM58 (proposed)*	c.892G>C	2	p.Ala298Pro	PMID: 21821711		rs387907598
In(Lu)	*KLF1*BGM59 (proposed)*	c.1012C>T	3	p.Pro338Ser	PMID: 29047116 PMID: 25585695		rs387907599
In(Lu)	*KLF1*BGM60 (proposed)*	c.318T>G	2	p.Tyr106X	PMID: 29047116		rs956697155
In(Lu)	*KLF1*BGM61 (proposed)*	c.911C>A	2	p.Thr304Lys	PMID: 29047116		not found

In(Lu)	*KLF1*BGM62 (proposed)*	c.954G>C	3	p.Trp318Cys	PMID: 30222867		rs769526751
In(Lu)	*KLF1*BGM63 (proposed)*	c.421C>T	2	p.Arg141X	PMID: 30222867		rs1426116895
In(Lu)	*KLF1*BGM64 (proposed)*	c.199delA	2	p.Gly68AlafsTer236	PMID: 29193102		not found
In(Lu)	*KLF1*BGM65 (proposed)*	c.604G>A	2	p.Gly202Arg	PMID: 29193102		rs756658473
In(Lu)	*KLF*BGM66 (proposed)*	c.858C>A	2	p.Cys286Ter	(2), Abstract		rs778506653
In(Lu)	*KLF*BGM67 (proposed)*	c.1001C>G	3	p.Thr334Arg	(3), Abstract		rs483352841
In(Lu)	*KLF*BGM68 (proposed)*	c.175insC	2	p.Pro59Profs*66	(3), Abstract		not found
In(Lu)	*KLF*BGM69 (proposed)*	c.304C,874G	2	p.Lys292Ter	(3), Abstract		not found
In(Lu)	*KLF*BGM70 (proposed)*	c.1003A	3	p.Gly335Arg	(3), Abstract		rs750987930
In(Lu)	*KLF*BGM71 (proposed)*	c.56_74del	1	p.Pro19Leufs*29	(3), Abstract		not found
In(Lu)	*KLF*BGM72 (proposed)*	c.304C,1017G	2, 3	p.Phe339Leu	(3), Abstract		not found
In(Lu)	*KLF*BGM73 (proposed)*	c.304C,1028C	2, 3	p.Leu343Pro	(3), Abstract		not found

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	Track of changes		from version	to version
1	**Version**			**v1.0 30-JUN-2021**
2	Author	created:	n.a.	Margaret Keller
3	Review	reviewed:	n.a.	unreviewed
4	General		n.a.	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	**End Version**			**v1.0 30-JUN-2021**