

## Names for *KLF1* Alleles

General description: EKLF (erythroid Krüppel-like factor) does not represent a blood group system, but is the product of *KLF1*. Mutations in this gene are responsible for In(Lu), a phenotype inherited in a dominant fashion, and for reduced expression of antigens of the Lutheran system and for P1, In<sup>b</sup>, and AnWj antigens.

Gene name: *KLF1*  
Number of exons: 3  
Initiation codon: Within exon 1  
Stop codon: Within exon 3  
Entrez Gene ID: 10661  
LRG sequence: NG\_013087.1 (genomic)  
NM\_006563.3 (transcript)  
Reference allele: *KLF1\*01* (shaded)

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
Common	<i>KLF1*01</i>			
In(Lu)	<i>KLF1*BGM01</i>	c.-124T>C	Promoter	p.0 [1]
In(Lu)	<i>KLF1*BGM02</i>	c.380T>A	2	p.Leu127Ter [1]
In(Lu)	<i>KLF1*BGM03</i>	c.569delC	2	p.Pro190Leufs*47 [1]
In(Lu)	<i>KLF1*BGM04</i>	c.874A>T	2	p.Lys292Ter [1]
In(Lu)	<i>KLF1*BGM05</i>	c.895C>T	2	p.His299Tyr [1]
In(Lu)	<i>KLF1*BGM06</i>	c.954dupG	3	p.Arg319Glufs*34 [1]
In(Lu)	<i>KLF1*BGM07</i>	c.983G>T	3	p.Arg328Leu [1]
In(Lu)	<i>KLF1*BGM08</i>	c.983G>A	3	p.Arg328His [1]
In(Lu)	<i>KLF1*BGM09</i>	c.991C>G	3	p.Arg331Gly [1]
CO:-1,-2 IN:-1,-2	<i>KLF1*BGM10</i>	c.973G>A	3	p.Glu325Lys [2]
In(Lu)	<i>KLF1*BGM11</i>	c.90G>A	1	p.Trp30Ter [3]
<b>*Obsolete* Normal BG phenotype</b>	<b><i>KLF1*BGM12</i></b>	<b>c.304T&gt;C</b>	<b>2</b>	<b>p.Ser102Pro [3]</b>
In(Lu)	<i>KLF1*BGM13</i>	c.114delC	2	p.Asp38Glufs*53 [4]
In(Lu)	<i>KLF1*BGM14</i>	c.298G>T	2	p.Glu100Ter [4]
In(Lu)	<i>KLF1*BGM15</i>	c.304T>C, c.384insC	2	p.Ser103Pro, p.Lys162Glnfs*352 [4]
In(Lu)	<i>KLF1*BGM16</i>	c.304T>C, c.1002del2	2, 3	p.Ser103Pro, p.Thr334Glyfs*351 [4]
In(Lu)	<i>KLF1*BGM17</i>	c.621C>G	2	p.Tyr207Ter [4]
In(Lu)	<i>KLF1*BGM18</i>	c.948delC	3	p.Cys316Trpfs*326 [4]
In(Lu)	<i>KLF1*BGM19</i>	c.1040C>A, c.1045delT	3	p.Ala347Asp, p.Ser349Argfs*358 [4]
In(Lu)	<i>KLF1*BGM20</i>	c.809C>A	2	p.Ser270X [5,6]
In(Lu)	<i>KLF1*BGM21</i>	c.977T>G	3	p.Leu326Arg [6,7]
In(Lu)	<i>KLF1*BGM22</i>	c.994A>G	3	p.Lys332Glu [6]
In(Lu)	<i>KLF1*BGM23</i>	c.517_519delC	2	p.Pro173ProfsX64 [8]
In(Lu)	<i>KLF1*BGM24</i>	c.551_556GGA CCG>A	2	p.Gly184GlufsX167 [8]
In(Lu)	<i>KLF1*BGM25</i>	c.637C>T	2	p.Glu213X [8]

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In(Lu)	<i>KLF1</i> *BGM26	c.802C>T	2	p.Arg268X [8]
In(Lu)	<i>KLF1</i> *BGM27	c.899T>C	2	p.Leu300Pro [8]
In(Lu)	<i>KLF1</i> *BGM28	c.902insT	2	p.Arg301LeufsX52 [8]
In(Lu)	<i>KLF1</i> *BGM29	c.947G>A	3	p.Cys316Tyr [8]
In(Lu)	<i>KLF1</i> *BGM30	c.968C>G	3	p.Ser323Trp [8]
In(Lu)	<i>KLF1</i> *BGM31	c.991C>T	3	p.Arg331Trp [8,9]
In(Lu)	<i>KLF1</i> *BGM32	c.310_311insG	2	p.Ala104GlyfsX249 [7]
In(Lu)	<i>KLF1</i> *BGM33	c.519_520insC	2	p.Gly174ArgfsX179 [7]
In(Lu)	<i>KLF1</i> *BGM34	c.519_525dupC GGCGCC	2	p.Gly176ArgfsX179 [7,10,11]
In(Lu)	<i>KLF1</i> *BGM35	c.591C>G	2	p.Tyr197X [7]
In(Lu)	<i>KLF1</i> *BGM36	c.663delG	2	p.Leu222SerfsX15 [7]
In(Lu)	<i>KLF1</i> *BGM37	c.862A>G	2	p.Lys288Glu [7]
In(Lu)	<i>KLF1</i> *BGM38	c.1071C>A	3	p.His357Gln [7]
In(Lu)	<i>KLF1</i> *BGM39	c.109C>T	2	p.Gln37X [8]
In(Lu)	<i>KLF1</i> *BGM40	c.196G>T	2	p.Glu66X [8]
In(Lu)	<i>KLF1</i> *BGM41	c.204delC	2	p.Gly68GlyfsX169 [8]
In(Lu)	<i>KLF1</i> *BGM42	c.796C>T	2	p.Arg266X [8]
In(Lu)	<i>KLF1</i> *BGM43	c.1022G>A	3	p.Cys341Tyr [8]
In(Lu)	<i>KLF1</i> *BGM44	c.151delC	2	p.Leu51SerfsX3 [8]
In(Lu)	<i>KLF1</i> *BGM45	c.826C>T	2	p.Gln276X [8]
In(Lu)	<i>KLF1</i> *BGM46	c.868T>C	2	p.Tyr290His [8]
In(Lu)	<i>KLF1</i> *BGM47	c.914-1g>c	Intron 2	Splicing failure [8]
In(Lu)	<i>KLF1</i> *BGM48	c.1001C>T	3	p.Thr334Met [8]
In(Lu)	<i>KLF1</i> *BGM49	c.1048C>T	3	p.Arg350Cys [8]
In(Lu)	<i>KLF1</i> *BGM50	c.86A>G	1	p.Lys29Arg [8]
In(Lu)	<i>KLF1</i> *BGM51	c.262_284dup	2	p.Ala95AlafsX150 [8]
In(Lu)	<i>KLF1</i> *BGM52	c.472delG	2	p.Ala158ProfsX79 [8]
In(Lu)	<i>KLF1</i> *BGM53	c.533C>A	2	p.Ser178X [8]
In(Lu)	<i>KLF1</i> *BGM54	c.887T>C	2	p.Leu296Pro [8]

In(Lu)	<i>KLF1</i> *BGM55	c.939G>A	3	p.Trp313X [8]
In(Lu)	<i>KLF1</i> *BGM56	c.946C>A	3	p.Arg322Ser [8]
In(Lu)	<i>KLF1</i> *BGM57	c.1004G>C	3	p.Gly335Ala [8]

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- [4] Garcia-Sanchez et al. Transfusion 2016;56:1413-18.
- [5] Satta et al. Haematologica 2011;96:767-70.
- [6] Perseu et al. Blood 2011;118:4454-58.
- [7] Helias et al. Hum Mut 2013;34:221-8.
- [8] Kawai et al. Transfusion 2017;57:1072-7.
- [9] Viprakasit et al. Blood 2014;123:1586-95.
- [10] Yang et al. Transfusion 2012;52 (Suppl.):161A-162A (abstract).
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