

## Names for VEL (ISBT 034) Blood Group Alleles

General description: The Vel blood group system consists of one antigen that is dependent on the expression of SMIM1, a 78 amino acid single pass membrane protein, likely type 2. The protein is encoded by *SMIM1*, or *VEL* if analysis is to predict a blood group antigen. In addition to mutations in the coding region, Vel antigen expression is considerably influenced by the rs1175550 SNP genotype. This SNP is located in intron 2, upstream of the coding region.

Gene name: *SMIM1*  
 Number of exons: 4  
 Initiation codon: Within exon 3  
 Stop codon: Within exon 4  
 Entrez Gene ID: 388588  
 LRG sequence: NG\_033869.1 (genomic)  
 NM\_001163724.2 (transcript)  
 Reference allele: *VEL\*01*

Reference allele <i>VEL*01</i> encodes Vel				
Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
Vel+				
Vel-	<i>VEL*-01</i>	c.64_80delAGCCTAGGGGCTGTGTC	3	p.Ser22Glnfs*?
Vel <sub>weak</sub>	<i>VEL*01W.01</i>	c.152T>A	4	p.Met51Lys
Vel <sub>weak</sub> / Vel-	<i>VEL*01W.02</i>	c.152T>G	4	p.Met51Arg