Names for VEL (ISBT 034) Blood Group Alleles

Intro

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* .
Gene name:	*SMIM1*
Number of exons:	4
Initiation codon:	exon 3
Stop codon:	exon 4
Entrez Gene ID:	388588
LRG:	LRG_827
LRG sequence:	NG_033869.1 (genomic) NM_001163724.3 (transcript)
Reference allele:	*VEL*01* (shaded)
Reference allele *VEL*01* encodes:	Vel
Antithetical antigens:	n.a.

Additional information

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
VEL:1 (Vel+)	*VEL*01*						
Null phenotypes							
Vel–	*VEL*01N.01*	c.64_80delAGCCTAGGGGCTGTGTC	3	p.Ser22Glnfs*?	(1) PMID: 23505126; (2) PMID: 23563606; (3) PMID: 23563608	KC751412.1	rs566629828
Weak phenotypes							
Vel_{weak}	*VEL*01W.01*	c.152T>A	4	p.Met51Lys	(3) PMID: 23563608; (4) PMID: 25647324		rs1182690110
Vel_{weak}/ Vel–	*VEL*01W.02*	c.152T>G	4	p.Met51Arg	(3) PMID: 23563608; (4) PMID: 25647324		n.a.
Vel_{weak}	*VEL*01W.03*	c.161T>C	4	p.Leu54Pro	(6) PMID: 31218697		n.a.
Vel_{weak}	*VEL*01W.04*	c.-75-335A>G	intron 2	none	(3) PMID: 23563608; (4) PMID: 25647324; (5) PMID: 28084402		rs1175550
Vel_{weak}	*VEL*01W.05*	c.122G>A	4	p.Arg41Lys	(7) Abstract		n.a.

References

1. PMID: 23505126 Ballif BA, Helias V, Peyrard T, Menanteau C, Saison C, Lucien N, Bourgouin S, Le Gall M, Cartron JP, Arnaud L. Disruption of SMIM1 causes the Vel- blood type. EMBO Mol Med. 2013 May;5(5):751-61. doi: 10.1002/emmm.201302466. PMID: 23505126; PMCID: PMC3662317.

2. PMID: 23563606 Storry JR, Jöud M, Christophersen MK, Thuresson B, Åkerström B, Sojka BN, Nilsson B, Olsson ML. Homozygosity for a null allele of SMIM1 defines the Vel-negative blood group phenotype. Nat Genet. 2013 May;45(5):537-41. doi: 10.1038/ng.2600. PMID: 23563606.

3. PMID: 23563608 Cvejic A, Haer-Wigman L, Stephens JC, Kostadima M, Smethurst PA, Frontini M, van den Akker E, Bertone P, Bielczyk-Maczyńska E, Farrow S, Fehrmann RS, Gray A, de Haas M, Haver VG, Jordan G, Karjalainen J, Kerstens HH, Kiddle G, Lloyd-Jones H, Needs M, Poole J, Soussan AA, Rendon A, Rieneck K, Sambrook JG, Schepers H, Silljé HHW, Sipos B, Swinkels D, Tamuri AU, Verweij N, Watkins NA, Westra HJ, Stemple D, Franke L, Soranzo N, Stunnenberg HG, Goldman N, van der Harst P, van der Schoot CE, Ouwehand WH, Albers CA. SMIM1 underlies the Vel blood group and influences red blood cell traits. Nat Genet. 2013 May;45(5):542-545. doi: 10.1038/ng.2603. PMID: 23563608; PMCID: PMC4179282.

4. PMID: 25647324 Haer-Wigman L, Stegmann TC, Solati S, Ait Soussan A, Beckers E, van der Harst P, van Hulst-Sundermeijer M, Ligthart P, van Rhenen D, Schepers H, de Haas M, van der Schoot CE. Impact of genetic variation in the SMIM1 gene on Vel expression levels. Transfusion. 2015 Jun;55(6 Pt 2):1457-66. doi: 10.1111/trf.13014. PMID: 25647324.

5. PMID: 28084402 Christophersen MK, Jöud M, Ajore R, Vege S, Ljungdahl KW, Westhoff CM, Olsson ML, Storry JR, Nilsson B. SMIM1 variants rs1175550 and rs143702418 independently modulate Vel blood group antigen expression. Sci Rep. 2017 Jan 13;7:40451. doi: 10.1038/srep40451. PMID: 28084402; PMCID: PMC5233989.

6. PMID: 31218697 van der Rijst MVE, Voorn L, Veldhuisen B, Jongerius JM, van den Akker E, van der Schoot CE. Identification of a novel single-nucleotide mutation in SMIM1 gene that results in low Vel antigen expression. Transfusion. 2019 Oct;59(10):E8-E10. doi: 10.1111/trf.15411. PMID: 31218697; PMCID: PMC7079045.

7. Abstract van der Rijst M, Veldhuisen B, van der Schoot E, van den Akker E. Identification of two novel mutations in SMIM1 resulting in low Vel expression. Vox Sanguinis. 2017; 112 (Suppl. 1):53.

Track of changes		**from v1.0 160620**	**to v2.0 30-OCT-2020**
	created: reviewed:	Jill Storry	Jill Storry/NN/NN
General		Last word version published on ISBT website	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
General			rs numbers recorded where available
Intro	LRG ID line added:		LRG_827
Allele Table	Allele renamed	*VEL*−01*	*VEL*01N.01*
Allele Table	Antigen/allele added:		*VEL*01W.03* *VEL*01W.04* *VEL*01W.05*
References		References updated	References found for all alleles and collated
End of changes		**from v1.0 160620**	**to v2.0 30-OCT-2020**