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:	
Additional information:	

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number	Comment
VEL:1 (Vel+)	***VEL*01***							
Vel_{strong}	***VEL*01.01***	c.-75-335A>G	i2	none	PMID: 23563608 PMID: 25647324 PMID: 28084402		rs1175550	
Null phenotypes								
Vel–	***VEL*01N.01***	c.64_80delAGCCTAGGGGCTGTGTC	3	p.Ser22Glnfs*?	PMID: 23563608 PMID: 23505126 PMID: 23563606	KC751412.1	rs566629828	
Weak phenotypes								
Vel_{weak}	***VEL*01W.01***	c.152T>A	4	p.Met51Lys	PMID: 23563608 PMID: 25647324		rs1182690110	
Vel_{weak}/ Vel–	***VEL*01W.02***	c.152T>G	4	p.Met51Arg	PMID: 23563608 PMID: 25647324		rs1182690110	This change is a tri-allelic SNP and has the same number as *VEL*01W.01*
Vel_{weak}	***VEL*01W.03***	c.161T>C	4	p.Leu54Pro	PMID: 31218697		n.a.	
Vel_{weak}	***VEL*01W.05***	c.122G>A	4	p.Arg41Lys	(1), Abstract		n.a.	

References

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.

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.

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.

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.

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.

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.

Abstract (1) 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	to
1	**Version**		**v2.0 30-OCT-2020**	**v.2.1 30-NOV-2021**
2	Author	created:	Jill Storry	Jill Storry, November 2021
3	Review	reviewed:	n.a.	Barbera Veldhuisen, November 2021
4	Allele Table	Allele renamed	*VEL*01.−01*	*VEL*01W.04* renamed *VEL*01.01.* References remain the same but this change enhances Vel antigen expression. The consensus allele gives the weaker phenotype.
5	References	renumbered	References ordering numbers	References ordering numbers have been removed
6	References	renumbered	(7), Abstract	(7), Abstract renumbered to (1), Abstract
7	**End Version**		**v2.0 30-OCT-2020**	**v.2.1 30-NOV-2021**

	Track of changes		from	to
1	**Version**		**v1.0 160620**	**v2.0 30-OCT-2020**
2	Author	created:	Jill Storry	Jill Storry
3	Review	reviewed:	n.a.	n.a.
4	General		Last word version published on ISBT website	First Excel map version. Spread-sheets "Intro", "Allele Table", "References" and "Versioning" created.
5	General			rs numbers recorded where available
6	Intro	LRG ID line added:		LRG_827
7	Allele Table	Allele renamed	*VEL*−01*	*VEL*01.−01*
8	Allele Table	Antigen/allele added:		*VEL*01W.03*
9				*VEL*01W.04*
10				*VEL*01W.05*
11	References		References updated	References found for all alleles and collated
12	**End Version**		**v1.0 160620**	**v2.0 30-OCT-2020**