

Supplementary Note

Recruitment of study subjects

Samples of blood were obtained from healthy blood donors, patients (cases) with antibodies against Vel (anti-Vel) and first-degree relatives of these patients. All 96 samples were assigned Unique Study Numbers (USN) and the key characteristics are summarized in **Supplementary Fig. 1**. When required biological samples were obtained with informed consent and the studies were approved according to national Research Ethics regulations in Denmark, The Netherlands and UK.

Genome Wide Association Study Sentinel SNP rs1175550 Genotype	Concordance phenotype- genotype: Yes=0, No=1; nsSNP=2	Δ/Δ , deletion on both alleles, WT/ Δ deletion on single allele; WT/M50K and WT/M50R, non synonymous (ns)SNP	Vel status: negative=0; weak- Test 2=1; weak-Test 3=2	Pregnancy=1; Surgery=2; Unknown=3	Blood donor=1, Case=2, Sib of case=3	Country of sample origin	Unique Study Number (USN)
AA	0	Δ/Δ	0		1	UK	1
AA	0	Δ/Δ	0		1	UK	2
AA	1	WT/ Δ	0		1	UK	3
AA	0	Δ/Δ	0		1	UK	4
AA	0	Δ/Δ	0		1	UK	5
AA	0	Δ/Δ	0		1	UK	6
AA	0	Δ/Δ	0		1	UK	7
AA	0	Δ/Δ	0		1	UK	8
AA	0	Δ/Δ	0		1	UK	9
AA	0	Δ/Δ	0		1	UK	10
AA	0	Δ/Δ	0		1	UK	11
AA	0	Δ/Δ	0		1	UK	12
AA	0	Δ/Δ	0		1	UK	13
AG	1, 2	WT/M51R	0		1	UK	14
AA	0	Δ/Δ	0		1	UK	15
AA	1	WT/ Δ	0		1	UK	16
AA	0	Δ/Δ	0		1	UK	17
AA	0	Δ/Δ	0		1	UK	18
AA	0	Δ/Δ	0		1	UK	19
AA	0	Δ/Δ	0		1	UK	20
AA	0	Δ/Δ	0		1	UK	21
AA	0	Δ/Δ	0		1	UK	22
AA	0	Δ/Δ	0		1	UK	23
AA	0	Δ/Δ	0		1	UK	24
AA	0	Δ/Δ	0		1	UK	25

Unique Study Number (USN)	Country of sample origin	Blood donor=1, Case=2, Sib of case=3	Pregnancy=1; Surgery=2; Unknown=3	Vel status: negative=0; weak-Test 2=1; weak-Test 3=2	Δ/Δ , deletion on both alleles, WT/ Δ deletion on single allele; WT/M50K and WT/M50R, non synonymous (ns)SNP	Concordance phenotype-genotype: Yes=0, No=1; nsSNP=2	Genome Wide Association Study Sentinel SNP rs1175550 Genotype
26	UK	1		0	Δ/Δ	0	AA
27	UK	1		0	Δ/Δ	0	AA
28	UK	1		0	Δ/Δ	0	AA
29	UK	1		0	Δ/Δ	0	AA
30	UK	1		0	Δ/Δ	0	AA
31	UK	1		0	Δ/Δ	0	AA
32	UK	1		0	Δ/Δ	0	AA
33	UK	1		0	Δ/Δ	0	AA
34	UK	1		0	WT/ Δ	1	AA
35	UK	1		0	Δ/Δ	0	AA
36	UK	1		0	Δ/Δ	0	AA
37	UK	1		0	WT/ Δ	1	AA
38	UK	1		0	Δ/Δ	0	AA
39	UK	1		0	Δ/Δ	0	AA
40	UK	1		0	Δ/Δ	0	AA
41	UK	1		0	Δ/Δ	0	AA
42	UK	1		0	Δ/Δ	0	AA
43	UK	1		0	Δ/Δ	0	AA
44	UK	1		1	WT/ Δ	0	AA
45	DN	1		0	Δ/Δ	0	AA
46	DN	1		0	Δ/Δ	0	AA
47	DN	1		0	Δ/Δ	0	AA
48	DN	1		0	WT/ Δ	1	AA
49	DN	1		0	Δ/Δ	0	AA
50	DN	1		0	Δ/Δ	0	AA

Unique Study Number (USN)	Country of sample origin	Blood donor=1, Case=2, Sib of case=3	Pregnancy=1; Surgery=2; Unknown=3	Vel status: negative=0; weak-Test 2=1; weak-Test 3=2	Δ/Δ , deletion on both alleles, WT/ Δ deletion on single allele; WT/M50K and WT/M50R, non synonymous (ns)SNP	Concordance phenotype-genotype: Yes=0, No=1; nsSNP=2	Genome Wide Association Study Sentinel SNP rs1175550 Genotype
51	NL	1		0	Δ/Δ	0	AA
52	NL	1		0	Δ/Δ	0	AA
53	NL	2	1	0	Δ/Δ	0	AA
54	NL	2	2	0	Δ/Δ	0	AA
55	NL	3		0	Δ/Δ	0	AA
56	NL	1		0	Δ/Δ	0	AA
57	NL	2	1	0	Δ/Δ	0	AA
58	NL	2	2	0	Δ/Δ	0	AA
59	NL	2	2	0	Δ/Δ	0	AA
60	NL	1		0	Δ/Δ	0	AA
61	NL	2	2	0	Δ/Δ	0	AA
62	NL	2	3	0	Δ/Δ	0	AA
63	NL	2	3	0	Δ/Δ	0	AA
64	NL	2	1	0	Δ/Δ	0	AA
65	NL	2	3	0	Δ/Δ	0	AA
66	NL	3		0	Δ/Δ	0	AA
67	NL	3		0	Δ/Δ	0	AA
68	NL	3		0	Δ/Δ	0	AA
69	NL	2	3	0	Δ/Δ	0	AA
70	NL	2	3	0	Δ/Δ	0	AA
71	NL	3		0	Δ/Δ	0	AA
72	NL	2	3	0	Δ/Δ	0	AA
73	NL	2	3	0	Δ/Δ	0	AA
74	NL	2	3	0	Δ/Δ	0	AA
75	NL	2	3	0	Δ/Δ	0	AA

Unique Study Number (USN)	Country of sample origin	Blood donor=1, Case=2, Sib of case=3	Pregnancy=1; Surgery=2; Unknown=3	Vel status: negative=0; weak-Test 2=1; weak-Test 3=2	Δ/Δ, deletion on both alleles, WT/Δ deletion on single allele; WT/M50K and WT/M50R, non synonymous (ns)SNP	Concordance phenotype-genotype: Yes=0, No=1; nsSNP=2	Genome Wide Association Study Sentinel SNP rs1175550 Genotype
76	NL	1		2	<i>WT/M51K</i>	0,2	AA
77	NL	1		1	WT/Δ	0	AA
78	NL	1		1	WT/Δ	0	AA
79	NL	1		1	WT/Δ	0	AA
80	NL	1		1	WT/Δ	0	AA
81	NL	1		1	WT/Δ	0	AA
82	NL	1		1	WT/Δ	0	AA
83	NL	1		1	WT/Δ	0	AA
84	NL	1		1	WT/Δ	0	AA
85	NL	1		1	WT/Δ	0	AA
86	NL	1		1	WT/Δ	0	AA
87	NL	1		1	WT/Δ	0	AA
88	NL	1		1	WT/Δ	0	AA
89	NL	1		1	WT/Δ	0	AA
90	NL	1		1	WT/Δ	0	AA
91	NL	1		1	WT/Δ	0	AA
92	NL	1		1	WT/Δ	0	AA
93	NL	1		1	WT/Δ	0	AA
94	NL	1		1	WT/Δ	0	AA
95	NL	3		1	WT/Δ	0	AA
96	NL	3		1	WT/Δ	0	AA

Supplementary Table 1. Characteristics of the Vel phenotype and SMIM1 genotype of the 96 study samples. Samples are from blood donors (n=73; see **Supplementary Fig. 1**), Vel-negative clinical cases with anti-Vel (n=16) and relatives of Vel-negative clinical cases with anti-Vel (n=7) (see **Supplementary Fig. 3**). DN, Denmark; NL, The Netherlands; UK, United Kingdom. WT, wild-type indicating that one of the common SMIM1 haplotypes was observed; A and G, the major and minor allele of GWAS SNP rs1175550; M51R and M51K are the amino mutations at residue 50 of the SMIM1 protein for USN14 and USN76, respectively; M, methionine; R, arginine; K, lysine, both samples are italics. USN1-5 were exome sequenced and are in bold.