Genotypes of patients with factor V deficiency

Mutation (Name)	Location	Domain	Type	genotype	Origin	Activity U/dL	Antigen U/dL	Expression	comments	Reference
IVS2+1 G>A	Intron 2		Splicing	Het		30				1
Leu62Ser	Exon 3	A1	Missense	Het		43				1
Gly67Arg	Exon 3	A1	Missense	Het		40				2
Asp68His*	Exon 3	A1	Missense	Hom		5	6			3
Asp68His*	Exon 3	A1	Missense	Het	China					4
Tyr91Asn	Exon 3	A1	Missense	Het		51		Yes		2,5
nt434 del G	Exon 4	A1	Frameshift	Het		45			Also FVII deficiency	6
Glu119stop	Exon 4	A1	Nonsense	Het		47				7
nt 585 del A Undefined	Exon 4	A1	Frameshift	Comp het	China	1	2			8
Ala188Val	Exon 5	A1	Missense	Hom		15				2
Phe190Ser*	Exon 5	A1	Missense	Hom	Japan	6	<2, 19			9
nt 678 ins 78 bp	Exon 5	A1	Insertion	Het		41				1
Gly208stop	Exon 5	A1	Nonsense	Het		37				1
IVS5-2 A>C	Intron 5		Splicing	Het		55				2
Ala221Val (New Brunswick) Undefined	Exon 6	A1	Missense	Comp het		29	39	Yes		10,11
Trp227Arg Arg1698Trp*	Exon 6 Exon 16	A1 A3	Missense Missense	Comp het		3				2
Gly231Glu nt 1118 ins 1278 bp	Exon 6 Intron 7	A1 A2	Missense Splicing	Comp het		6	6			1
Ser234Leu Arg413Cys*	Exon 6 Exon 9	A1 A2	Missense Missense	Comp het	China	6	33	Yes Yes		12
Ser234Trp Gene deletion	Exon 6	A1	Missense Deletion	Comp het		9			Chromosome 1q deletion	13
Gln250Arg	Exon 6	A1	Missense	Het		42				2

Gly276Glu*	Exon 6	A1	Missense	Hom		7				2
Lys310stop (Amersfoort)	Exon 7	A1	Nonsense	Het	Netherlands	34	51			14
nt 1041 del 8 bp (Seoul I) Tyr1702Cys* (Seoul 2)	Exon 7 Exon 15	A2 A3	Frameshift Missense	Comp het	South Korea	1	3			14
Phe325Leu nt5519 ins 7 bp	Exon 7 Exon 17	A2 A3	Missense Frameshift	Comp het	China	4	4	Yes		15
Ala327Val	Exon 7	A2	Missense	Het		50				2
Arg348Ser Leu1642Arg	Exon 8 Exon 15	A2 A3	Missense Missense	Comp het		33				2
Gly392Cys* nt 4798 del G	Exon 8 Exon 14	A2 A3	Missense Splicing	Comp het	China	<1	1			16
Gly392Cys*	Exon 8	A2	Missense	Hom	Taiwan	1.7	2.5	Yes		17
IVS8+6 T>C	Intron 8		Splicing	Hom	Iran	5	6			18
IVS8- 2A>G	Intron 8		Splicing	Hom	China					19
Arg413Cys*	Exon 9	A2	Missense	Het						2
Ile417Thr (Ogden) nt4699 ins 4 bp*	Exon 9 Exon 13	A2 B	Missense Frameshift	Comp het		<5	<5			20
Gln448stop*	Exon 10	A2	Nonsense	Het		67			In trans with FV Leiden	2
Gln448stop*	Exon 10	A2	Nonsense	Het		30				2
Cys472Gly nt 5038 ins A	Exon 10 Exon 15	A2 A3	Missense Frameshift	Comp het	UK	<1	2	Yes		21
nt 1516 del TAC nt 2862 del T*	Exon 10 Exon 13	A2 B	Del Tyr478 Frameshift	Comp het		2		Yes		22
Gly493Arg	Exon 10	A2	Missense	Het		27, 37	38			23
nt 1599 del G	Exon 10	A2	Frameshift	Hom	South Asia	<1	4			24
Arg506stop	Exon 10	A2	Nonsense	Het		33				25

nt 1611 G>T	Exon 10	A2	Splicing	Hom		<1	8			26
Gln517stop	Exon 11	A2	Nonsense	Het						2
Tyr530Ser*	Exon 11	A2	Missense		China					27
Tyr530Ser* IVS16-1 G>T	Exon 11 Intron 16	A2	Missense Splicing	Comp het	China			Yes Yes		28,29
Cys585Arg (Nijkerk)	Exon 12	A2	Missense	Het	Netherlands	52	64			14
nt 1853 ins C Y1702C*	Exon 12 Exon 15	A2 A3	Frameshift Missense	Comp het	Japan	<3	<1	Yes Yes		30
Lys650Glu	Exon 13	A2	Missense	Het	Japan	50	36			9
nt 2148 del AG Gly 2079Val*	Exon 13 Exon 23	B C2	Frameshift Missense	Comp het	China	0.1	1.5			19,31
Arg712stop*	Exon 13	B	Nonsense	Het					In trans with FV Leiden	32
Arg712stop* Val1813Met*	Exon 13 Exon 17	B A3	Nonsense Missense	Comp het	UK	<1	5	Yes		21
Arg712stop* nt 3510 del AA	Exon 13 Exon 13	B B	Nonsense Frameshift	Comp het	UK	1				33
Gln773stop (Casablanca)	Exon 13	B	Nonsense	Hom	Morocco	1	3			34
nt 2539 del A nt 3170 del 5 bp	Exon 13 Exon 13	B B	Frameshift Frameshift	Comp het						2
nt 2572 del AA or AG	Exon 13	B	Frameshift	Hom	India		2.4			35
Trp896stop	Exon 13	B	Nonsense	Het		36	22			1
nt 2743 del AC	Exon 13	B	Frameshift	Hom		<1	<1			36
nt 2862 del T* nt 5403 ins G (Debrecen)	Exon 13 Exon 16	B A3	Frameshift Frameshift	Comp het		<1	1			37
nt 2862 del T*	Exon 13	B	Frameshift	Het		50				2
nt 2862 del T*	Exon 13	B	Frameshift	Comp het		10				2

Thr1870Met	Exon 18	A3	Missense							
Arg952Cys Gln1109stop	Exon 13 Exon 13	B B	Missense Nonsense	Comp het	China					4
Arg1002stop*	Exon 13	B	Nonsense	Hom	Puerto Rica	<1	<1			24
Arg1002stop*	Exon 13	B	Nonsense	Hom		1				2
Arg1002stop* nt 6032 ins 5bp	Exon 13 Exon 21	B C1	Nonsense Frameshift	Comp het	South India	<1	<1			21
Arg1002stop* IVS21+1 G>A	Exon 13 Intron 21	B	Nonsense Splicing	Com het	Italy	<1	<1	- Yes		18
nt3296 del A	Exon 13	B	Frameshift	Hom	India		2.9			35
nt3471 del 4 bp	Exon 13	B	Frameshift	Hom	African American	<1				24
Arg1133stop*	Exon 13	B	Nonsense	Hom	Italy	<1	<1		In cis with FV Leiden	38
Arg1133stop*	Exon 13	B	Nonsense	Hom		1				2
Arg1133stop* nt 6026 del 6 bp*	Exon 13 Exon 21	B C1	Nonsense Frameshift	Comp het		Korea	3	4		39
nt 3616 ins TC	Exon 13	B	Frameshift	Het		42	54		In trans with FV Leiden	40,41,42
nt 3799 del C	Exon 13	B	Frameshift	Hom	India		<1			35
nt 3924 del 4 bp	Exon 13	B	Frameshift	Hom		2	<1			43
nt 4204 del C (Utrecht)	Exon 13	B	Frameshift	Hom	Turkey	<1	3			14
Gln1425stop	Exon 13	B	Nonsense	Hom		1				2
nt 4699 ins 4 bp* (Stanford)	Exon 13	B	Frameshift	Het		56			In trans with FV Leiden	44,45
nt 4699 ins 4 bp* (Stanford)	Exon 13	B	Frameshift	Het		44				44
nt 4746 del TTA	Exon 13	B	Frameshift	Comp het		10				2

Met1939Lys	Exon 21	C1								
Arg1606stop*	Exon 14	A3	Nonsense	Het		37				2
Arg1606stop* Tyr1702Cys*	Exon 14 Exon 15	A3 A3	Nonsense Missense	Comp het	Italy	<1	<1			21
Glu1608Lys*	Exon 14	A3	Missense	Het		56	51		In trans with FV Leiden	42,46
Glu1608Lys*	Exon 14	A3	Missense	Het		38	50			46
Pro1618Arg	Exon 14	A3	Missense	Hom	Japan	<3	<2, 50			9
Arg1631Pro	Exon 15	A3	Missense	Het		41				1
Arg1698Trp* Arg2019stop	Exon 16 Exon 22	A3 C1	Missense Nonsense	Comp het		6				2
Tyr1702Cys*	Exon 15	A3	Missense	Het	Italy	50	50		In trans with FV Leiden	47
Tyr1702Cys*	Exon 15	A3	Missense	Hom	Italy	1				47,48
Tyr1702Cys* Undefined	Exon 15	A3	Missense	Comp het	Italy	<1				48
Tyr1702Cys*	Exon 15	A3	Missense	Hom	Slovenia	<1	<1			21
Phe1745Tyr	Exon 16	A3	Missense	Het			30			2
His1775Arg* Leu1821Ser*	Exon 16 Exon 17	A3 A3	Missense Missense	Comp het		3	5			1
His1775Arg* Ser2058stop	Exon 16 Exon 23	A3 C2	Missense Nonsense	Comp het		<1				1
nt 5419 G>A	Exon 16	A3	Splicing	Het		72	73		In trans with FV Leiden	49
nt 5419 G>A	Exon 16	A3	Splicing	Het		30				2
Pro1788Ser	Exon 17	A3	Missense	Het		47				2
nt 5593 del T	Exon 17	A3	Frameshift	Het		50	48		In cis with FV Leiden	50
Val1813Met*	Exon 17	A3	Missense	Hom	Japan	<1	4	Yes		51
Val1813Met*	Exon 17	A3	Missense	Het		35				2
Val1813Met* nt 6157 del 5bp	Exon 17 Exon 22	A3 C1	Missense Frameshift	Comp het	Japan	<1	9			51

Leu1821Ser*	Exon 17	A3	Missense	Het			35			1
Gly1852Val	Exon 18	A3	Missense	Hom						52
Trp1854stop*	Exon 18	A3	Nonsense	Hom	Turkey	2	1			21
Trp1854stop*	Exon 18	A3	Nonsense	Het		31				2
Thr1870Met*	Exon 18	A3	Missense	Hom		26				2
IVS18-12 T>A	Intron 18		Splicing	Hom		<1				53
Gln1894stop	Exon 19	C1	Nonsense	Het		62	46		In trans with FV Leiden	42
IVS19+3 A>T	Intron 19		Splicing	Hom	Iran	<1	<1	Yes		54
nt 6026 del 6 bp*	Exon 21	C1	Deletion	Hom	Japan	<1	<2			9
Pro2006Ala	Exon 22	C1	Missense	Hom	China					4
Gly2032Asp	Exon 22	C1	Missense	Het	Italy	65	53	Yes	In cis with FV Leiden	55
nt 6212 del G	Exon 23	C1	Frameshift	Het		56	60		In trans with FV Leiden	42
Pro2070Leu	Exon 23	C2	Missense	Hom	Norway				The first patient described (Mary)	56
Arg2074Cys	Exon 23	C2	Missense	Hom	Italy	4	2	Yes		57,58
Arg2074His*	Exon 23	C2	Missense	Hom	Tunis	2	3			59
Arg2074His* IVS24+1 del 4bp	Exon 23 Intron 24	C2	Missense Splicing	Comp het	Austria	3		Yes		18
Gly2079Val*	Exon 23	C2	Missense	Hom	China	2	2			3
Asp2098Tyr	Exon 24	C2	Missense	Hom		4		Yes		2,5
Gly2112Asp	Exon 24	C2	Missense	Het		59	52		In trans with FV Leiden	42
nt 6491 del 21 bp and nt 6523 del G	Exon 24	C2	Frameshift	Het		45	40		In trans with FV Leiden	50

Pro2167Leu	Exon 25	C2	Missense	Het						2
Arg2174Cys	Exon 25	C2	Missense	Hom	China					4
Arg2174Leu	Exon 25	C2	Missense	Hom		1	5	Yes		51

Nucleotide numbers are based on the Genebank file M16967 using the A of the ATG initiator methionine as +1
*A mutation that was identified in more than 1 family

Mutations causing Factor V deficiency according to their types

Missense			Nonsense	Splice	Deletion/Insertion	
L62S	G493R	G2032N	E119X	IVS2+1 G>A	nt 434 delG	nt 4204 del C
G67R	Y530S*	P2070L	G208X	IVS5-2 A>C	nt 585 del A	nt 4699 ins 4 bp*
D68H*	C585R	R2074C	K310X	IVS8+6 T>C	nt 683-760 dup 78	nt 4746 del TTA
Y91N	K650E	R2074H*	Q448X*	IVS8-2 A>G	nt 1041 del 8 bp	nt 4798 del G
A188V	R952C	G2079V*	R506X	nt 1611 G>T	nt 1118 + 1278 bp	nt 5038 ins A
F190S*	E1608K*	D2098Y	Q517X	nt 5419 G>A*	nt 1516 del TAG	nt 5403 ins G
A221V	P1618R	G2112N	R712X*	IVS16-1 G>T	nt 1599 del G	nt 5519 ins 7 bp
W227R	R1631P	P2167L	Q773X	IVS18-12 T>A	nt 1853 ins C	nt 5593 del T
G231E	L1642R	R2174C	W896X	IVS19+3 A>T	nt 2148 del AG	nt 6023 del 6 bp
S234L	R1698W*	R2174L	R1002X*	IVS21+1 G>A	nt 2539 del A	nt 6032 ins 5 bp
S234W	Y1702C*		Q1109X	IVS24+1 del 4 bp	nt 2572 del 2 bp	nt 6026 del 6 bp*
Q250R	F1745Y		R1133X*		nt 2743 del AC	nt 6157 del 5 bp
G276E*	H1775R*		Q1425X		nt 2862 del T*	nt 6212 del G
F325L	P1788S		R1606X*		nt 3170 del ACACA	nt 6491 del 21 bp
A327V	V1813M*		W1854X*		nt 3296 del A	nt 6523 del G
R348S	L1821S*		Q1894X		nt 3471 del 4 bp	gene deletion
G392C*	G1852V		R2019X		nt 3510 del AA	
R413C*	T1870M*		S2058X		nt 3616 ins TC	
I417T	M1939K				nt 3799 del C	
C472G	P2006A				nt 3924 del 4 bp	

* Mutation was identified in more than one family

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