Genotypes of patients with factor VII deficiency

Mutation (Name)	Location	Domain	Type	Genotype	Origin	Activity U/dL**	Antigen U/dL	Study of dysfunctional protein	Comments***	Reference
-96 C>T IVS4+1 G>A*	Promoter Intron 4	Promoter	Regulatory Splicing	Comp het	Japan	<1	6	Expressed	Sp1 binding site	1
-94 C>G	Promoter	Promoter	Regulatory	Hom	French-Canadian	<1	<1	Expressed	Sp1 binding site	2
-79 C>T and nt 9702 del 9	Promoter	Promoter	Regulatory	Hom	USA					3
-61 T>G*	Promoter	Promoter	Regulatory	Hom	French-Canadian	<1	<1	Expressed	HNF4 binding site	4
-61 T>G* -55 C>T*	Promoter Promoter	Promoter Promoter	Regulatory Regulatory	Comp het Comp het	France	2				5
-59 T>G Ala294Val*	Promoter Exon 8	Promoter Catalytic	Regulatory Missense	Comp het	Finland	8	24		M1M2	6
-55 C>G*	Promoter	Promoter	Regulatory	Het	Germany	41				3
-55 C>T*	Promoter	Promoter	Regulatory	Het	Germany	42				3
-55 C>T* Ala294Val+ nt 11128 del C*	Promoter Exon 8	Promoter Catalytic	Regulatory Missense+ Frameshift	Comp het	Poland	2	2	Expressed	HNF4 binding site	7
-44 T>C*	Promoter	promoter	Regulatory	Het	Germany	58				3
-39A>G*	Promoter	Promoter	Regulatory	Het	Germany	33				3
-32 A>C*	Promoter	Promoter	Regulatory	Hom	Finland	2	1	Expressed		6
-32 A>C*	Promoter	Promoter	Regulatory	Hom	Sweden	5				3
-30 A>C*	Promoter	Promoter	Regulatory	Hom	Pakistan	16	20			8
-30 A>C*	Promoter	Promoter	Regulatory	Hom	Germany	2				3
Met-60Val Undefined	Exon 1a	Initiation codon	Missense	Comp het	France	2			M1M2	5
Met-60Ile nt 8973 del G	Exon 1a Exon 6	Initiation codon Activation	Missense Frameshift	Comp het	Switzerland	1	1			8
Gln-57stop Ala294Val+ nt 11128 del C*	Exon 1a Exon 8	Prepro Catalytic	Missense Missense+ Frameshift	Comp het	Romania	8-12				9

nt 16 del C	Exon 1a	Prepro	Frameshift	Hom	India	<1				10
nt 27 del CT *	Exon 1a	Prepro	Frameshift	Hom	China	<1	<1			11
nt 27 del CT * IVS6+1 G>T*	Exon 1a Intron 6	Prepro	Frameshift Splicing	Comp het	Taiwan	<1	1			12
Leu-48Pro (Morioka)	Exon 1a	Prepro	Missense	Hom	Japan	11	11			13
Leu-42Pro Val252Met*	Exon 1a Exon 8	Prepro Catalytic	Missense Missense	Comp het	Germany	11				9
nt 64 G>A (reported also as Val-17Ile)	Exon 1a	Splicing	Missense	Hom	Turkey	8	4			9
IVS1a+5 G>A*	Intron 1a		Splicing	Hom	Iran	6	3			14
IVS1a+5 G>A* Gly179Arg*	Intron 1a Exon 7	- Catalytic	Splicing Missense	Comp het	France	2	62			5
IVS1a+5 G>A* His348Gln*	Intron 1a Exon 8	- Catalytic	Splicing Missense	Comp het	China					15
IVS1b-11 G>A nt 3933 G>C*	Intron 1b Intron 2		Splicing Splicing	Comp het	France	<1	17			5
Ala-10Asp	Prepro	Prepro	Missense	Het?		12				3
Val-7Ile IVS7+1 G>A	Exon 2 Intron 7	Prepro	Missense Splicing	Comp het	Venezuela	1				3
Arg-1Cys	Exon 2	Prepro	Missense	Hom	India	21				10
Phe4Leu	Exon 2	Gla	Missense	Hom	Turkey	1	2			9
Leu13Gln* Ala294Val*	Exon 2 Exon 8	Gla Catalytic	Missense Missense	Comp het	Latvia	2	7			8
Leu13Gln* and Ala294Val* Gly78Asp*	Exon 2 Exon 8 Exon 4	Gla Catalytic EGF1	Missense Missense Missense	Comp het	Germany	<1				3
Leu13Gln* Ala294Val+ nt 11128 del C*	Exon 2 Exon 8	Gla Catalytic	Missense Frameshift	Comp het	France	2			M2M2	5
nt 3865 ins G Ala294Val+ nt 11128 del C*	Exon 2	Gla Exon 8	Frameshift Catalytic	Comp het	Germany	<1				3
Glu16Lys IVS3+5 G>A	Exon 2 Intron 3	Gla	Missense Missense	Comp het	France	<1	25			5
Glu19Gln	Exon 2	Gla	Missense	Het	Algeria	28	34		M2M2	8
Cys22Arg*	Exon 2	Gla	Missense	Het	Israeli Arab	17	36	Expressed		16
Cys22Arg* Ala191Pro	Exon 2 Exon 6	Gla Activation	Missense Missense	Comp het	India	<1	46			17

Cys22Arg* Arg353Pro	Exon 2 Exon 8	Gla Catalytic	Missense Missense	Comp het		2	10			18
Ser23Pro	Exon 2	Gla	Missense	Het	Iran	<1	77			14
nt 3892 del 3bp	Exon 2	Gla	Del Phe 24	Hom	Israeli Arab	<1	30	Expressed		16,19
Glu25Lys His348Gln*	Exon 2 Exon 8	Gla Catalytic	Missense Missense	Comp het	Japan	1	21			1
Arg28Gly* Arg277Cys*	Exon 2 Exon 8	Gla Catalytic	Missense Missense	Comp het	Switzerland	2				5
Arg28Gly*	Exon 2	Gla	Missense	Het	Germany	46				3
Glu29Lys Gene deletion	Exon 2	Gla	Missense	Comp het	Caucasian	<1				20
nt 3933 G>C*	Intron 2		Splicing	Hom	Switzerland	1	19			8
IVS2+1 G>A	Intron 2		Splicing	Hom	Iran	<1	<1			14
IVS2+1 G>C*	Intron 2		Splicing	Hom	Israeli Arab	1				19
IVS2+1 G>C*	Intron 2		Splicing	Het	Germany	32				3
IVS2+1 G>C* Arg247Cys*	Intron 2 Exon 8	- Catalytic	Splicing Missense	Comp het	Germany	10				9
IVS2+5 G>T Val252Met*	Intron 2 Exon 8	- Catalytic	Splicing Missense	Comp het	Germany	4				9
IVS2-2 A>G	Intron 2		Splicing	Hom	Tunis					21
IVS3-1 G>A Tyr68Cys*	Intron 3 Exon 4	- EGF1	Splicing Missense	Comp het	Germany	1				9
Gln49Stop Gln100Arg*	Exon 4 Exon 5	EGF1 EGF2	Nonsense Missense	Comp het	France	<1				5
Ser52Stop	Exon 4	EGF1	Nonsense	Hom	Turkey	<1				22
Cys55Stop Gly283Ser*	Exon 4 Exon 8	EGF1 Catalytic	Nonsense Missense	Comp het		<10				23
Asn57Asp* (Hamilton)	Exon 4	EGF1	Missense	Het		35	62	Expressed		24
Asn57Ile	Exon 4	EGF1	Missense	Hom	France	<1	2			5
Ser60Pro IVS7+7 A>G	Exon 4 Intron 7	EGF1	Missense Splicing	Comp het	Germany	2-9				9
Cys61Phe Gln100Arg*	Exon 4 Exon 5	EGF1 EGF2	Missense Missense	Comp het	Switzerland	1	7			8
Cys61Stop Cys329Gly*	Exon 4 Exon 8	EGF1 Catalytic	Nonsense Missense	Comp het	China	2	49	.		25
Leu65Pro Gly375Glu*	Exon 4 Exon 8	EGF1 Catalytic	Missense Missense	Comp het	Sweden	1	9			8
Tyr68Cys*	Exon 4	EGF1	Missense	Het	UK	41	44			8

Tyr68Cys* Ala294Val+ nt 11128 del C*	Exon 4 Exon 8	EGF1 Catalytic	Missense Missense	Comp het	Russia	8				3
Cys72stop*	Exon 4	EGF 1	Missense	Hom	Algeria	<1	<1			26
Cys72stop* Cys310Phe	Exon 4 Exon 8	EGF 1 Catalytic	Missense Missense	Comp het	Algeria	3	34			26
Gly78Asp* Cys194Tyr	Exon 4 Exon 7	EGF1 Catalytic	Missense Missense	Comp het	Germany	1				3
Arg79Gln*	Exon 4	EGF1	Missense	Hom	Japan	h 100 r 11 b 150 s 25	100	Expressed		27,28,29,30
Arg79Gln* Arg152Gln* (Charlotte)	Exon 4 Exon6	EGF1 Activation	Missense Missense	Double hom	African American	<1	100	Protein purified studies. Expressed		31
Arg79Gln* Thr324Met*	Exon 4 Exon 8	EGF1 Catalytic	Missense Missense	Com het	India	h 63, 78 r 4, 10				32
Arg79Trp Gly97Val	Exon 4 Exon 5	EGF1 EGF2	Missense Missense	Comp het	UK	10	52		M1M2	8
nt 7780 del 7bp nt 7773 ins 251bp	Intron 4 Intron 4		Complex rearrange-ment	Double hom	Algeria	<1	<1			33
IVS4+1 G>A*	Intron 4		Splicing	Hom	Germany	5-7				9,34
IVS4+1 G>A*	Intron 4		Splicing	Hom	Turkey	4				35
IVS4+1 G>A*	Intron 4		Splicing	Het	Germany	63				3
IVS4+1 G>A* Cys135Arg*	Intron 4 Exon 6	- Activation	Splicing Missense	Comp het	France	2	7			5
IVS4+1 G>A* Thr359Met*	Intron 4 Exon 8	- Catalytic	Splicing Missense	Comp het	Italy	<1	<1	Expressed		36
IVS4+1 G>A* Gly375Glu*	Intron 4 Exon 8	- Catalytic	Splicing Missense	Comp het		3				3
IVS4+1 G>A* Val252Met*	Intron 4 Exon 8	- Catalytic	Splicing Missense	Comp het	UK	3				8
IVS4+1 G>A* Gln221Stop	Intron 4 Exon 8	- Catalytic	Splicing Nonsense	Comp het	Japan	<1.5	<5			37
Cys91Ser	Exon 5	EGF2	Missense	Hom	UK	1	4			8
Glu94Lys*	Exon 5	EGF2	Missense	Hom	Germany	13				3,9

Gly96Ser*	Exon 5	EGF2	Missense	Hom	Germany	<4				3
Gly96Ser* nt 10743 del G	Exon 5 Exon 8	EGF2 Catalytic	Missense Frameshift	Comp het	France	2	15			5
Gly97Cys*	Exon 5	EGF2	Missense	Hom	Italy	<1	2	Expressed		38
Gly97Cys*	Exon 5	EGF2	Missense	Het	Germany Sweden	34				3
Gly97Cys* Gln100Arg*	Exon 5 Exon 5	EGF2 EGF2	Missense Missense	Comp het	France	<1	8		M1M2	5
Gly97Cys* IVS7+5 G>A* (Lazio)	Exon 5 Intron 7	EGF2	Missense Splicing	Comp het	Italy	2	10		M2M2	39
Gly97Ser* Ala294Val+ nt 11128 del C*	Exon 5 Exon 8	EGF2 Catalytic	Missense Missense+ Frameshift	Comp het	Germany	14-40				9
Gly97Ser*	Exon 5	EGF2	Missense	Het	Italy	22	26		M2M2	39
Gln100Arg*	Exon 5	EGF2	Missense	Hom	Norway	<2	18	Expressed	Additional 11 unrelated Norwegian families and others were described	14,38,40,41
Gln100Arg*	Exon 5	EGF2	Missense	Het	Germany Sweden	15	21		M1M2	3,8,9
Gln100Arg* Cys135Arg*	Exon 5 Exon 6	EGF2 Activation	Missense Missense	Comp het	UK	2	6			8
Gln100Arg* Thr272Met	Exon 5 Exon 8	EGF2 Catalytic	Missense Missense	Comp het	Norway	7	43		M2M2	8
Gln100Arg* Ala294Val+ nt 11128 del C*	Exon 5 Exon 8	EGF2 Catalytic	Missense Missense	Comp het	Slovakia	3				3
Gln100Arg* Gly331Ser*	Exon 5 Exon 8	EGF2 Catalytic	Missense Missense	Comp het	France	<1	64			5
Gln100Arg* nt 10983 del T*	Exon 5 Exon 8	EGF2 Catalytic	Missense Frameshift	Comp het	UK	1	6			8
Cys102Tyr Undefined	Exon 5	EGF2	Missense	Comp het	France	2	19			5
Ser103Gly	Exon 5	EGF2	Missense	Hom		<2	13		Inhibitor	42
Arg110Cys*	Exon 5	EGF2	Missense	Hom	Japan	25	28			43
Arg110Cys* Asp123Tyr	Exon 5 Exon 5	EGF2 EGF2	Missense Missense	Comp het	Italy	<1	<1			44

Gly117Arg*	Exon 5	EGF2	Missense	Hom	India	<1				10
Gly117Arg* Arg152stop*	Exon 5 Exon 6	FGF2 Activation	Missense Nonsense	Comp het	India	<1	1.1		M1M1	45
Gly117Arg* Leu263Arg*	Exon 5 Exon 8	FGF2 Catalytic	Missense Missense	Comp het	India	<1	2			17
IVS5-12 T>A	Intron 5		Splicing	Hom	Asia	“Low”			M2M2	8
IVS5-2 A>G*	Intron 5		Splicing	Het	Malaysia	72				8
IVS5-2 A>G* IVS6+1 G>T*	Intron 5 Intron 6		Splicing Splicing	Comp het	India	2				46
IVS5-1 G>A*	Intron 5		Splicing	Hom	China	5				47
IVS5-1 G>A* Cys389Gly	Intron 5 Exon 8		Splicing Catalytic	Comp het	China	4	39			48
Pro134Thr*	Exon 6	Activation	Missense	Hom	Germany					3
Pro134Thr* (Malta I) Ala244Val* (Malta II)	Exon 6 Exon 8	Activation Catalytic	Missense Missense	Comp het	Malta	12	14			49
Cys135Arg*	Exon 6	Activation	Missense	Hom	Germany	1-4	2			5,8,9
Cys135Arg* nt 10586 del 17 bp*	Exon 6 Exon 8	Activation Catalytic	Missense Frameshift	Comp het		<1	<1			14
Cys135Arg* Val281Phe*	Exon 6 Exon 8	Activation Catalytic	Missense Missense	Comp het		7	7			50
Cys135Arg* Arg304Gln*	Exon 6 Exon 8	Activation Catalytic	Missense Missense	Comp het	France	<5	55			5
Cys135Arg* Thr359Met*	Exon 6 Exon 8	Activation Catalytic	Missense Missense	Comp het	Germany	5				3
Lys137Glu (Undefined)	Exon 6	Activation	Missense	Comp het	Japan	<1	18			30
Ile138Thr Leu263Arg*	Exon 6 Exon 8	Activation Catalytic	Missense Missense	Comp het	India	4	6			17
Ile140Ser	Exon 6	Activation peptide	Missense	Het	Brazil	h 62 r 54	65		M1M2	51
Arg152Stop* Ala294Val+ nt 11128del C*	Exon 6 Exon 8	Activation Catalytic	Nonsense Missense+ Frameshift	Comp het	Germany	1	1			9
Arg152stop*	Exon 6	Activation	Nonsense	Het		35-52				3
Arg152Gln*	Exon 6	Activation	Missense	Hom	Iran	<1	78		Additional German heterozygote	9,14

Arg152Gln*	Exon 6	Activation	Missense	Hom	India	<1				10
Arg152Gln*	Exon 6	Activation	Missense	Hom	India	<1	2			17
Arg152Gln*	Exon 6	Activation	Missense	Het	Germany	32				3
Arg152Gln* Ala294Val+ nt 11128 del C*	Exon 6 Exon 8	Activation Catalytic	Missense Missense+ Frameshift	Comp het	France	<1			M1M2	5
Arg152Leu Arg304Trp* nt 10968 del C	Exon 6 Exon 8 Exon 8	Activation Catalytic Catalytic	Missense Missense Frameshift	Comp het	China					52
Gly156Asp Ala294Val+ nt 11128 del C*	Exon 6 Exon 8	Activation Catalytic	Missense, Missense+ Frameshift	Comp het	Hungary	15				9
IVS6+1 G>T* Gly283Ser*	Intron 6 Exon 8	- Catalytic	Splicing Missense	Comp het	Venezuela	3				3
IVS6+1 G>T* Undefined	Intron 6		Splicing	Comp het		<1	30		Another heterozygote is from Malaysia	8,14
Cys178Tyr	Exon 7	Catalytic	Missense	Het	Italy	37	30		M1M2	53
Gly179Arg* Ala244Val*	Exon 7 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	France	3	10		M2M2	5
Gly180Arg*	Exon 7	Catalytic	Missense	Hom	Bedouin	<1				54
Gly180Arg* Arg304Gln*	Exon 7 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	France	3	23		M1M2	5
Thr181Asn* Undefined	Exon 7	Catalytic	Missense	Comp het	Vietnam	1	1			8
Thr181Asn* Met306Val	Exon 7 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	China					55
Ala191Val	Exon 7	Catalytic	Missense	Hom	Algeria	5	5		M2M2	56
Ala191Glu*	Exon 7	Catalytic	Missense	Hom	India	<1				10
Ala191Glu* Trp364Cys*	Exon 7 Exon 8	Catalytic Catalytic	Missense Missense	Double hom	India	<1				10
Ala191Thr*	Exon 7	Catalytic	Missense	Hom	North Africa	<5	25			5
Ala191Thr*	Exon 7	Catalytic	Missense	Hom	Germany	13				3
Ala191Thr* Arg224Gln*	Exon 7 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Algeria	6	7			56
Cys194Tyr* Undefined	Exon 7	Catalytic	Missense	Comp het	Germany	<1				9

Cys194Tyr* Ala294Val+ nt 11128 del C*	Exon 7	Catalytic	Missense	Comp het	Germany	1				3
Cys194Arg Ala294Val+ nt 11128 del C*	Exon 7	Catalytic	Missense	Comp het	Russia	3				3
Leu204Pro	Exon 7	Catalytic	Missense	Het	Norway	7.5	23	Expressed	M1M2	57
Ala206Thr*	Exon 7	Catalytic	Missense	Het	Germany	24				3
Ala206Thr* IVS7+7 A>G*	Exon 7 Intron 7	Catalytic	Missense Splicing	Comp het		30				50
Ala206Thr* Pro303Arg	Exon 7 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Germany	15	60			9
IVS7+2 T>G Arg224Gln*	Intron 7 Exon 8	- Catalytic	Splicing Missense	Double hom	Algeria	<1	<1			58
IVS7+3 del GGGT*	Intron 7		Splicing	het	Italy	61	58		3 more heterozygotes are from Spain and Germany	8,9,59
IVS7+5 G>A* (Lazio)	Intron 7		Splicing	Hom	Italy	1	1		Common in Italy; another heterozygote is from UK	8,39,59
IVS7+7 A>G*	Intron 7		Splicing	Het	Italy	43	42			14,39,59
IVS7+7 A>G* Ala294Val*	Intron 7 Exon 8	- Catalytic	Splicing Missense	Comp het		23	52			50
IVS7+7 A>G* Met298Ile*	Intron 7 Exon 8	 Catalytic	Splicing Missense	Comp het		41				50
nt 10543 del 15bp Cys310Phe*	Exon 8 Exon 8	Catalytic Catalytic	Frameshift Missense	Comp het	France	<1				5
nt 10554 del 15bp Ala294Val*	Exon 8 Exon 8	Catalytic Catalytic	5 AA deletion Missense	Comp het		4				9
nt 10567 ins 15bp	Exon 8	Catalytic	5 AA insertion	Hom	Oman	<1	10	Expressed		60
10586 del 17 bp* Ala294Val*	Exon 8 Exon 8	Catalytic Catalytic	Frameshift Missense	Comp het	Italy	7	25	Plasma studied	M1M2	61
Asp212Asn*	Exon 8	Catalytic	Missense	Het	Germany	52				50
Arg223Trp*	Exon 8	Catalytic	Missense	Het	Italy	44	49		M1M2	62

Arg223Trp*	Exon 8	Catalytic	Missense	Het	Germany	30				50
Gln227stop	Exon 8	Catalytic	Nonsense	Hom	India	<1			Additional 2 families	10
Thr239Pro	Exon 8	Catalytic	Missense	Hom	Algeria	25	95			56
Asp242His* Val252Met*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Germany	4	1		M2M2	8
Asp242His* Thr359Met*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Germany	2-4				9
Asp242Asn*	Exon 8	Catalytic	Missense	Hom	Pakistan	1	9		M2M2	8
Asp242Asn* His348Arg*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	India	<1			M1M	63
Ala244Val*	Exon 8	Catalytic	Missense	Hom	Moroccan Jew	5	9	Expressed	M2M2 Common in Moroccan and Iranian Jews. Rare in other Jewish ethnic groups and Arabs	9,16,64,65
Ala244Val* Undefined	Exon 8	Catalytic	Missense	Comp het	France	5	7		M2M2	5
Ala244Val* Arg304Gln*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	North Africa	5	44		M1M2	5,16,64
Ala244Thr* Undefined	Exon 8	Catalytic	Missense	Comp het	Spain	3	13		Another Moroccan Jew heterozygote	8,16
Arg247His* (Mie)	Exon 8	Catalytic	Missense	Hom	Japan	26	26	Expressed		66
Arg247His*	Exon 8	Catalytic	Missense	Het	Germany	35				3
Arg247Cys*	Exon 8	Catalytic	Missense	Hom					M1M2	8
Arg247Cys* Cys310Phe*	Exon8 Exon8	Catalytic Catalytic	Missense Missense	Comp het		<1	54			14
Val252Met* Ala294Val*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Germany	9	31			9
Val252Met*	Exon 8	Catalytic	Missense	Het	Germany	63				3
nt 10698 del C nt 10785 del C	Exon 8 Exon 8	Catalytic Catalytic	Frameshift Frameshift	Comp het		<2	1			67
Leu261Phe	Exon 8	Catalytic	Missense	Hom		1	1			3

Leu263Arg* Undefined	Exon 8	Catalytic	Missense	Comp het	India	<1	1			17
Glu265Lys*	Exon 8	Catalytic	Missense	Het	Italy	42	46		M1M2.	62
Glu265Lys*	Exon 8	Catalytic	Missense	Het	Germany	35				3,9
Arg277Cys*	Exon 8	Catalytic	Missense	Hom	Iran	<1	1	Expressed	M2M2	14,68
Arg277Cys*	Exon 8	Catalytic	Missense	Hom	Germany	6	12			3
Arg277His	Exon 8	Catalytic	Missense	Het	Yemenite Jewish	25	44			19
Val281Phe* Ala294Val	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het		1-5				50
Val281Phe* Ala294Val+ nt 11128 del C*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense+ frameshift	Comp het	Germany	<1	10			9
Ser282Arg Undefined	Exon 8	Catalytic	Missense	Comp het	Iran	<1	6		.	14
Gly283Ser*	Exon 8	Catalytic	Missense	Hom	Italy	3	100	Plasma studied Expressed		69
Gly283Ser*	Exon 8	Catalytic	Missense	Hom	Germany	7				3
Trp284Arg Undefined	Exon 8	Catalytic	Missense	Comp het	India	3		65		17
Trp284stop*	Exon 8	Catalytic	Missense	Het	Germany	30				3
Arg290Cys*	Exon 8	Catalytic	Missense	Het	Germany	29-33				3
Ala294Val*	Exon 8	Catalytic	Missense	Hom	Poland	11	47	Expressed	M2M2 Common in Europe	9,70,71
Ala294Val* Gly331Ser*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Germany	6	100		Additional patient from Turkey	72
Ala294Val* Gly375Glu*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Sweden	4				3
Ala294Val* Ala294Val+ nt 11128 del C*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense + Frameshift	Comp het	France	4	48		M2M2	5
Ala294Val+ nt 11128 del C*	Exon 8	Catalytic	Missense + Frameshift	Double hom	Poland	<2	1	Expressed Expressed	M2M2 Common in Europe	9,16,70
Ala294Val+ nt 11128 del C*	Exon 8	Catalytic	Missense + Frameshift	Double hom	Poland	3	17		M1M1	70
Met298Val*	Exon 8	Catalytic	Missense	Het	Italy	50	87		M1M2	62

Met298Ile*	Exon 8	Catalytic	Missense	Hom	Italy	6	85		Common	5,8,9,73
Met298Ile*	Exon 8	Catalytic	Missense	Hom	Algeria	10	136		M1M1	56
Met298Ile* Cys310Phe*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	North Africa	1	9			5
Met298Ile* Gly331Ser*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Spain	3				74
Met298Ile* Trp364Stop	Exon 8 Exon 8	Catalytic Catalytic	Missense Nonsense	Comp het	France	5				5
Leu300Pro Cys310Phe*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Yemenite Jewish	<1	52		Expressed	16,19
Pro303Thr*	Exon 8	Catalytic	Missense	Hom	Iran	<1	135	Expressed	Additional homozygote from Taiwan	12,14,75
Pro303Thr*	Exon 8	Catalytic	Missense	Het	Germany	30-48				3
Arg304Gln* (Padua)	Exon 8	Catalytic	Missense	Hom	Italy	h 30 r<1	100	Purified	Common in other populations too	5,14,16,21,30, 62,72,73,76, 79,80
Arg304Gln*	Exon 8	Catalytic	Missense	Het	Italy	r 40 b 100	70			73,81
Arg304Gln*	Exon 8	Catalytic	Missense	Het	Brazil	r 40	26		M1M2	51
Arg304Gln*	Exon 8	Catalytic	Missense	Hom	African American	r 5-8 h 68-87				82
Arg304Gln* Gly 365Cys	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	USA	6				3
Arg304Gln* Arg304Trp*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	China					83
Arg304Trp*	Exon 8	Catalytic	Missense	Hom	Japan	h 16 r<5 b 60	100	Expressed		5,84
Cys310Phe*	Exon 8	Catalytic	Missense	Hom	Iran	<1	104	Expressed	Common worldwide	5,8,9,14,16,17, 51,73,81
Cys310Phe*	Exon 8	Catalytic	Missense	Het	Tunis	14	38		M2M2	21
Cys310Phe* Gly331Ser*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Italy	4	95			72
Cys310Phe* Trp356Stop	Exon 8 Exon 8	Catalytic Catalytic	Missense Nonsense	Comp het	Italy	7	41			73
Arg315Trp Arg304Gln*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	African American	26	67			85
Thr324Met*	Exon 8	Catalytic	Missense	Hom	India	4	70			17

Met327Ile	Exon 8	Catalytic	Missense	Hom	North Africa	<1	47		M1M1	5
Met327Thr*	Exon 8	Catalytic	Missense	Het	Germany	24				3
Met327Val	Exon 8	Catalytic	Missense	Het	Germany	31				3
nt 10896 del 18 bp	Exon 8	Catalytic	In frame del	Hom	Saudi Arabia	<1	21			45
Phe328Ser* (Central)	Exon 8	Catalytic	Missense	Hom	Hispanic	<1	38	Expressed		86
Phe328Ser*	Exon 8	Catalytic	Missense	Hom	Venezuela	1				3
Phe328Ser* Asp343Asn	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	France	<1	65			5
Cys329Gly*	Exon 8	Catalytic	Missense	Hom	China	3	55	Expressed		25,87
Cys329Gly* Unidentified	Exon 8	Catalytic	Missense	Comp het	Taiwan	1	30		Another Indonesian patient	8,12
Cys329Arg*	Exon 8	Catalytic	Missense			12	50			88
Gly331Cys	Exon 8	Catalytic	Missense	Hom		1				3
Gly331Asp*	Exon 8	Catalytic	Missense	Het		h 85 r 23 b 46	77		Another Brazilian heterozygote	51,89
Gly331Ser*	Exon 8	Catalytic	Missense	Hom	Italy	<2	85-100	Expressed	Common in Europe	69,72
Gly331Ser*	Exon 8	Catalytic	Missense	Hom	Japan	2	84	Expressed	M1M1	90
Gly331Ser*	Exon 8	Catalytic	Missense	Hom	Germany	7				3
Gly331Ser*	Exon 8	Catalytic	Missense	Het	UK	30	64		M1M2	8,72
Asp338Glu	Exon 8	Catalytic	Missense	Hom	India	<1	95			17
Ser339Cys	Exon 8	Catalytic	Missense	Het	Japan	34	40	Expressed		91
Ser339Phe*	Exon 8	Catalytic	Missense	Hom	Tunis	1.5		Expressed		21
Ser339Phe*	Exon 8	Catalytic	Missense	Het	Tunis	40	56		M1M2	21
Gly342Arg	Exon 8	Catalytic	Missense	Het	Italy	35	64		M1M2	73
Gly342Glu*	Exon 8	Catalytic	Missense	Het	Italy	42	85			62
Asp343His	Exon 8	Catalytic	Missense		Germany	18	74			9
His348Arg*	Exon 8	Catalytic	Missense	Hom	India	<1	6			17
His348Gln*	Exon 8	Catalytic	Missense	Hom	Japan	5	<10	Expressed	Additional Asian patients were described	5,12,92
Arg353Gly	Exon 8	Catalytic	Missense	Het	Germany	46				3
Gly354Cys	Exon 8	Catalytic	Missense	Hom	Japan	5	7	Expressed		93
Trp356stop*	Exon 8	Catalytic	Nonsense	Het	Germany	32-45				3
Thr359Met*	Exon 8	Catalytic	Missense	Hom	Japan	<2	2			5,14,94,95

(Toyama)										
Thr359Met*	Exon 8	Catalytic	Missense	Hom	Russia					3
Thr359Met* R402stop*	Exon 8 Exon 8	Catalytic catalytic	Missense Nonsense	Comp het	Japan	<3	<1			96
Ser363Ile	Exon 8	Catalytic	Missense	Hom	Iran	<1	80	Expressed		14,97
Trp364Cys*	Exon 8	Catalytic	Missense	Hom	Iran	<1	132	Expressed		14,97
Trp364Phe	Exon 8	Catalytic	Missense	Hom	Iran	6	77			5
Ala369Thr*	Exon 8	Catalytic	Missense	Het	Germany Russia	45				3
Gly375Glu*	Exon 8	Catalytic	Missense	Hom	Sweden	2	45			3
Gly375Glu*	Exon 8	Catalytic	Missense	Het	Sweden Costa Rica	24-27				3
Gly375Glu* Undefined	Exon 8	Catalytic	Missense	Hom	Sweden	1				3
Arg379Gly	Exon 8	Catalytic	Missense	Hom	North Africa	<1	6			5
Gln382stop	Exon 8	Catalytic	Nonsense	Hom	India	<1				10
Glu385Lys	Exon 8	Catalytic	Missense	Het		35			M1M2	98
R402stop*	Exon 8	Catalytic	Nonsense	Het	Japan	24		Expressed		99
Gene deletion				Het		32				100
Complexed rearrangement Arg304Gln*	Exon 8	Catalytic	Missense	Comp het		3				100

Nucleotide numbers are based on the full sequence published by O'hara et al 1987 using the A of the ATG initiator methionine as +1. Numbering of the amino acids is based on Genebank file NM_000131. Methionine is numbered as -60 and the mature protein starts at Ala +1.

*A mutation that was identified in more than one family.

** Source of tissue factor used for measurement of FVII activity: h-human, r-rabbit, b-bovine, s-simian

*** M1- Arg at position 353, M2- Gln at position 353 of the common Arg353Gln polymorphism

Mutations causing Factor VII deficiency according to their types

Promoter	Missense					Nonsense	Splice	Deletion/Insertion	Big Deletion/ Rearrangement
nt-96 C>T	M-60I	C91S	T181N	R277C*	D338E	*E26X*	nt 64 G>A	nt 16 del C	3
nt-94 C>G	M-60V	E94K*	A191V	R277H	S339C	*K32X*	IVS1a +5 G>A*	nt 27 del CT*	
nt-79 C>T	L-48P	G96S*	A191E*	V281F*	S339F	Q-57X	*IVS1-8 del 14*	*nt 156 del G*	
nt-65 G>C	L-42P	G97C*	A191T*	S282R	G342R	Q49X	IVS1b -11 G>A	nt 3865 ins G	
nt -62 C>T	*V-15A*	G97V	A191P	G283S*	G342E*	S52X	IVS2+1 G>A	nt 3892 del 3 bp	
nt-61T>G*	A-10D	G97S	*A191F*	W284R	D343N	C55S	IVS2+1 G>C*	nt 7780 del 7bp	
nt-60 T>G	V-7I	Q100R*	C194Y*	*G285S*	D343H	C61X	*IVS2+1 del G*	nt 7773 ins 251 bp	
nt-59 T>G	R-1C	C102Y	C194R	R290C	*S344N*	C72X*	IVS2+5 G>T	nt 8973 del G	
nt -55 C>T*	F4L	S103G	L204P	A294V*	H348Q*	R152X*	*IVS2-3 C>G*	nt 9702 del 9 bp	
nt-55 C>G	L13Q*	R110C*	A206T*	M298V*	H348R*	Q221X	IVS2-2 A>G	nt 10543 del 15 bp	
nt-44 T>C*	*L13E*	G117R*	D212N	M298I*	R353P	Q227X	*IVS3 +1 G>A*	nt 10554 del 15 bp	
nt-39 A>G*	E16K	*S111F*	*D217N*	L300P*	R353G	W284X	IVS3 +5 G>A	nt 10567 ins 15 bp	
nt-32 A>C*	E19Q	*L121P*	R223W*	P303R	G354C	W356X*	IVS3-1 G>A	nt 10586 del 17*	
nt-30 A>C*	C22R*	*D123N*	R224Q*	P303T*	T359M*	W364X	IVS4+1 G>A*	nt 10698 del C	
	C22F	D123Y	*T238I*	R304Q*	S363I	Q382X	nt 3933 G>C*	nt 10743 del G	
	S23P	*S126F*	T239P	R304W*	W364C*	R402X*	IVS5-12 T>A	nt 10785 del C	
	E25K	*P129S*	*H241Q*	M306V	W364F		IVS5-2 A>G*	nt 10896 del 18 bp	
	R28G*	*P134L*	D242H*	C310F*	G365C		IVS5-1G>A*	nt 10968 del C	
	R28P	P134T*	D242N*	R315W	*G365A*		IVS6+1 G>T*	nt 10983 del T	
	E29K	C135R*	A244V*	T324M*	A369T*		*IVS6+1 G>A*	nt 11128 del C*	
	N57D	K137E	R247C*	*M327T*	G375E*		*IVS6+3 A>G*		
	N57I	I138T	R247H*	M327I	*Y377F*		IVS7+1 G>A	*Glu14 del A*	
	S60P	I140S	R247C	M327V*	R379G		IVS7+2 T>G	*Glu14 ins G*	
	C61F	R152Q*	V252M*	F328S*	E385K		IVS7+3 del GGGT	*Arg271 ins GG*	
	L65P	R152L	L261F	C329G*	C389G		IVS7+5 G>A*	*Pro303 del CC*	
	Y68C*	G156D	L263R*	C329R*			IVS7+7 A>G*	*Thr106 del C*	
	G78D*	C178Y	E265K*	G331D*			*IVS7+8 C>G*	*Leu170 del TTG*	
	R79W	G179R*	*R266Q*	G331S*				*Val 188 del C?*	
	R79Q*	G180R*	T272M	G331C					

*Mutations that were identified in more than one family

Mutations in Italic letters indicate publications with no information on the patients (101,102). They are therefore not included in the Table that list genotypes of patients with factor VII deficiency.

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