Genotypes of patients with factor X deficiency

Mutation	Location	Domain	Type	Genotype	Origin	Activity (PT based assay) U /dL	Antigen U/dL	Studies of Dysfunctional protein	Reference
Met-40Val (Nice I) Pro304Ser (Nice II)	Exon 1 Exon 8	Prepro Catalytic	Initiation Met Missense	Comp het		4	7		1
nt 8 del GC Phe31Ser*	Exon 1 Exon 2	Prepro Gla	Frameshift	Comp het	China	<1	5		2
Val-33Phe* Undefined	Exon 1	Prepro	Missense	Comp het	India	<1	6		3
Val-33Phe* Gln316stop	Exon 1 Exon 8	Prepro Catalytic	Missense Nonsense	Comp het	India	<1			3
Leu-32Pro	Exon 1	Prepro	Missense	Hom	Germany	23	15		4
Ser-30Arg (Shanghai)	Exon 1	Prepro	Missense	Hom	China	2.6	8.2	Expressed	5
Ala-26Asp*	Exon 1	Prepro	Missense	Hom	India	<1	<1		3
Gly-20Arg* (Santo Domingo)	Exon 1	Prepro	Missense	Hom	Santo Domingo	<2	<5	Expressed	4,6,7,8
Gly-20Arg*	Exon 1	Prepro	Missense	Hom	Iran	<1	4		9
Gly-20Arg*	Exon 1	Prepro	Missense	Hom	India	<1	0		3
Gly-20Arg* (Santo Domingo) Gly204Arg*	Exon 1 Exon 7	Prepro Catalytic	Missense Missense	Comp het	Venezuela	<1[#]			4

IVS1+1 G>A Arg347His	Intron 1 Exon 8	- Catalytic	Splicing Missense	Comp het	China	2.5	59	Expressed	10
IVS1+3 A>T	Intron 1		Splicing	Hom	Iran	<1	7		11
IVS1-1 G>C	Intron 1		Splicing	Hom	Asia	0	<1		8
Thr-2Met Cys111Tyr	Exon 2 Exon 5	Prepro EGF 2	Missense Missense	Comp het	France	12	26		8
Arg-1Thr*	Exon 2	Prepro	Missense	Hom		<1-7	43-66		11
Ser3Cys	Exon 2	Gla	Missense	Hom		10	42		12
Glu7Gly* (St Louis II)	Exon 2	Gla	Missense	Hom		1-3	100	Expressed	13
Glu7Lys	Exon 2	Gla	Missense	Het	Germany	47[#]			4
Glu14Lys* (Vorarlberg)	Exon 2	Gla	Missense	Hom		5-10	20	Purified	4,14
Glu14Gly* (Ketchikan)	Exon 2	Gla	Missense	Hom		5	20		15
Glu14Gly* (Ketchikan) Ser379Lys	Exon 2 Exon 8	Gla Catalytic	Missense Missense	Comp het	Poland	2[#]			4
Glu16Lys Val298Met*	Exon 2 Exon 8	Gla Catalytic	Missense Missense	Comp het	Caucasian	<1	66		16
Glu19Ala	Exon 2	Gla	Missense	Hom	Italy	<1	17	Expressed	17
Glu25Lys (Frankfurt I)	Exon 2	Gla	Missense	Het		56	55	Expressed	18
Glu29Lys	Exon 2	Gla	Missense	Hom	Iran	<1	19		9
Phe31Ser*	Exon 2	Gla	Missense	Hom	Algeria	<1	4-16	5 families founder effect	19
Phe31Ser*	Exon 2	Gla	Missense	Comp het	Algeria	<1	2		19

Tyr130stop	Exon 6	EGF 2	Nonsense						
Phe31Ser* nt 514 del T and nt 516 T>C	Exon 2 Exon 6	Gla EGF 2	Missense Frameshift	Comp het	Nepal	<1			20
Glu32Gln (Tokyo)	Exon 2	Gla	Missense	Hom	Japan	3	61		21
IVS2-3 T>G	Intron 2		Splicing	Hom	Iran	2	3		11
Glu51Lys* (Riyadh)	Exon 4	EGF 1	Missense	Hom	Saudi Arabia	4	Normal	2 families Normal PTT	22
Asn57Thr (Wenatchee II) Arg139Cys (Wenatchee I)	Exon 4 Exon 6	EGF 1 EGF 2	Missense Missense	Comp het		10-20	30-35		23
Gly59Arg	Exon 4	EGF 1	Missense	Hom	India	<1	20		3
Asp63His	Exon 4	EGF 1	Missense	Hom		<1			9
Gly78Asp	Exon 4	EGF 1	Missense	Hom	Iran	<1	8		11
Cys81Tyr*	Exon 4	EGF 1	Missense	Hom	Iran	<1			11
IVS4-8 del CTT	Intron 4		Splicing	Hom	Japan	2.5	<5		24
Gly94Arg*	Exon 5	EGF 2	Missense	Hom	Poland	<2	<2		4
Gly94Arg*	Exon 5	EGF 2	Missense	Hom	Iran	<1	3		9
Gly94Arg* Asp95Glu	Exon 5 Exon 5	EGF 2 EGF 2	Missense Missense	Comp het	Iran-Turkey	<1-2	3-4		11
Gly94Arg* Arg142Met	Exon 5 Exon 6	EGF 2 Catalytic	Missense Missense	Comp het	Germany	<3[#]			4
Gly94Arg* Tyr344Cys	Exon 5 Exon 8	EGF 2 Catalytic	Missense Missense	Comp het	Poland	<3[#]			4
Glu102Lys	Exon 5	EGF2	Missense	Hom	Germany	36			4

Cys109Tyr	Exon 5	EGF 2	Missense	Hom		<1			8
Gly114Arg* (Kanazawa)	Exon 5	EGF 2	Missense	Hom	Venezuela	19	25		4
Gly114Arg* (Kanazawa)	Exon 5	EGF 2	Missense	Het	Japan	45	50		25
Gly133Arg	Exon 6	EGF 2	Missense	Hom	India	<			20
Arg139Ser (Kurayoshi)	Exon 6	EGF 2	Missense	Hom	Japan	27	72		26
nt556 del C and Lys408Asn*	Exon 6 and exon 8	Catalytic Catalytic	Frameshift Missense	Double het	Italy	9	72		27
Gly152Arg and Ala234Ser*	Exon 6 and exon 7	Catalytic	Missense	Double hom	Venezuela	6	120		4
nt 608 del AT	Exon 6	Catalyic	Frameshift	Hom		<1			4,28
Val196Met (Hofu)	Exon 6	Catalytic	Missense	Het	Japan	51	100		29
Gly204Arg*	Exon 6	Catalytic	Missense	Hom	Iran	6	7		11
Gly204Arg*	Exon 6	Catalytic	Missense	Hom	Iran	<1	<1		9
Gly204Arg* (Debrecen)	Exon 6	Catalytic	Missense	Hom		1	1		30
Gly222Asp*	Exon 7	Catalytic	Missense	Hom	Iran	<1	10		4,11
Gly223Arg	Exon 7	Catalytic	Missense	Hom	India	<1			20
nt814 del C Arg326Cys (San Antonio)	Exon 7	Catalytic	Frameshift Missense	Comp het		14	36	Normal PTT	31
Ala234Ser*	Exon 7	Catalytic	Missense	Het	Germany	52[#]			4

Lys245Met	Exon 7	Catalytic	Missense	Hom	India	<1	<1		3
Val246Met Del exon 6	Exon 7	Catalytic	Missense	Com het		<1			32
Val248Ala*	Exon 7	Catalytic	Missense	Hom	India	<1	46		3
nt 865 G>C	Exon 7	Catalytic	Splicing	Het	Spain	60	53		8
IVS7-1 G>A	Introns 7		Splicing	Hom	Germany	<1			4
Arg251Trp (Padua)	Exon 8	Catalytic	Missense	Hom		26	100	Normal PTT	33,34
nt 882 ins C Gly366Ser*	Exon 8 Exon 8	Catalytic Catalytic	Frameshift Missense	Comp het	China	<1	45		35
Glu264Lys	Exon 8	Catalytic	Missense	Het	France	68	74		8
nt 929 del TCA	Exon 8	Catalytic	Frameshift	Het	Slovakia				4
Phe281Leu	Exon 8	Catalytic	Missense	Hom	Poland	<1	<2		4
Asp282Asn* Arg287Trp	Exon 8 Exon 8	Catalytic	Missense Missense	Comp het	U.K	6			8
Asp282Asn* (Stockton)	Exon 8	Catalytic	Missense	Het		43	101	Plasma studied	36
Val298Met* (Stuart)	Exon 8	Catalytic	Missense	Hom		<1	<1		37
Val298Met* (Stuart)	Exon 8	Catalytic	Missense	Het	Germany	45[#]			4
Val298Met* nt1151 del17bp	Exon 8 Exon 8		Missense Frameshift	Comp het	UK	<1	2		8
Arg306Cys (Nagoya I) Undefined	Exon 8	Catalytic	Missense	Comp het		3	<10		38
Glu310Lys	Exon 8	Catalytic	Missense	Comp het	Germany	8	10		8

Undefined									
Met314Arg Undefined	Exon 8	Catalytic	Missense	Comp het	India	<1	<1		3
Thr318Met* (Roma)	Exon 8	Catalytic	Missense	Hom	Italy	<1	100	Plasma studied	8,39
Thr318Met*	Exon 8	Catalytic	Missense	Hom		3	80		40
Thr318Met*	Exon 8	Catalytic	Missense	Hom	India	24			20
Thr318Met* Gly323Ser*	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	UK	<1	64		8
Gly323Ser*	Exon 8	Catalytic	Missense	Hom	India	<1			20
Glu329Gly	Exon 8	Catalytic	Missense	Het	Slovakia	32[#]			4
Ser334Pro* (Marseille)	Exon 8	Catalytic	Missense	Hom	France	21-26	100	Purified	41,42,43
Ser334Pro* Gly380Arg* (Padua 3)	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het	Italy	8	40		44
Val342Ala	Exon 8	Catalytic	Missense	Het		73		Expressed	45
Pro343Ser* (Friuli)	Exon 8	Catalytic	Missense	Hom	Italy	4-9	100		46,47,48
Arg347Cys Undefined	Exon 8	Catalytic	Missense	Comp het	Germany	<1[#]			4
Cys350Phe* (Padua 4)	Exon 8	Catalytic	Missense	Hom	Italy	5	5		33,49
Cys350Phe*	Exon 8	Catalytic	Missense	Hom	Germany	<1			4,50
Ser353Tyr	Exon 8	Catalytic	Missense	Het	Germany	47[#]			4
Ser354Arg	Exon 8	Catalytic	Missense	Hom	India	<1			20
Phe356Cys	Exon 8	Catalytic	Missense	Comp het		3	25		51

Glu7Gly*	Exon 2	Gla	Missense						
Phe363Ile	Exon 8	Catalytic	Missense	Het	Germany	44[#]			4
Cys364Arg*	Exon 8	Catalytic	Missense	Hom	Iran	<1			52
Cys364Arg*	Exon 8	Catalytic	Missense	Het		48	31		8
Gly366Ser* (Nagoya II)	Exon 8	Catalytic	Missense	Het		34	80		1,8
Gly366Ser*	Exon 8	Catalytic	Missense	Hom	Sri Lanka	1	67	Expressed	53
Gly366Ser*	Exon 8	Catalytic	Missense	Hom	India	<1			20
Gly366Ser*	Exon 8	Catalytic	Missense	Hom	India	<1	90		3
Gly380Arg*	Exon 8	Catalytic	Missense	Hom	Costa Rica	<1	<1		4,28
Gly381Asp*	Exon 8	Catalytic	Missense	Hom	Oman	<1	Normal	Expressed	54
Gly381Asp*	Exon 8	Catalytic	Missense	Hom		<1	18		9
Pro382Leu	Exon 8	Catalytic	Missense	Hom		<1	10		51
His383Gln Trp421Arg	Exon 8 Exon 8	Catalytic Catalytic	Missense Missense	Comp het		6			40
Ala404Thr (Nottingham)	Exon 8	Catalytic	Missense	Hom		1-3	110		55
Arg405Gly (Taunton)	Exon 8	Catalytic	Missense	Het		54	98		55
Lys408Asn* (San Giovanni Rotondo)	Exon 8	Catalytic	Missense	Hom		16	98		27
Ile411Phe* (Leicester)	Exon 8	Catalytic	Missense	Hom	Gujarati Indian	<1	8		3,56
nt 1429 del 5 bp and del 8 bp in 3'	Exon 8 Non coding	Catalytic	Frame shift	Double Hom	African American	<1	<1		57

Del exons 7-8 Undefined			Big deletion	Comp het		4	<10		58
Gene deletion ? Del exons 7-8			Big deletion	Comp het		1	18		59

Nucleotide numbers are based on the Genebank file NM_000504 using the A (nucleotide 26) of the ATG initiator methionine as +1.
*A mutation that was identified in more than 1 family.
[#] Value was obtained by personal communication with the authors

Mutations causing Factor X deficiency according to their types

Missense					Nonsense	Splice	Deletion/Insertion
M-40V	E32Q	V196M	T318M	G380R	Y130X	IVS1+1 G>A	nt 8 del GC
V-33F	E51K	G204R	G323S	G381D	Q316X	IVS1+3 A>T	nt 514 del T and nt 516 T>C
L-32P	N57T	G222D	R326C	P382L		IVS1-1 G>C	nt 556 del C
S-30R	G59R	G223R	E329G	H383Q		IVS2-3 T>G	nt 608 del AT
A-26D	D63H	A234S	S334P	A404T		IVS4-8 del CTT	nt 814 del C
G-20R	G78D	K245M	V342A	R406G		nt 865 G>C	nt 882 ins C
T-2M	C81Y	V246M	P343S	K408N		IVS7-1 G>A	nt 929 del TCA
R-1T	G94R	V248A	Y344C	I411F			nt 1151del 17 BP
S3C	D95E	R251W	R347C	W421R			nt 1429 del 5 bp and del 8 bp in 3'
E7K	E102K	E264K	R347H				
E7G	C109Y	F281L	C350F				
E14K	C111Y	D282N	S353Y				
E14G	G114R	R287W	S354R				
E16K	G133R	V298M	F356C				
E19A	R139S	P304S	F363I				
E25K	R139C	R306C	C364R				
E29K	R142M	E310K	G366S				
F31S	G152R	M314R	S379K				

* Mutation was identified in more than 1one family

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