Table I. Detrimental Mutations

Pedigree[a]	Position and Mutation[b] (codon)	Predicted amino acid change[b]	Total PS antigen % of normal	Free PS antigen[c] % of normal	PS activity % of normal	Cosegregation[d]	Comments	References
PS-34-275	5'UT exon 1 cDNA 143 C→G		53	19	14	2(2)	Double variant for -27 CTC→CAC mutation	124
PS-46-27	-34, TC (del G)	-34, C→S	61	4[PEG]		2(3)	Frameshift, stop at codon -24	Rezende, Simmonds Pers. Comm.
PS-46-03	-34, TC (del G)	-34. C→S	52[5]	2[5, PEG]		2(5)	Frameshift, stop at codon -24	Rezende, Simmonds Pers. Comm.
PS-34-275	-27, CTC→CAC	-27, L→H	53	19	14	2(2)	Double variant for 5'UT exon 1 cDNA 143 C→G	124
PS-31-006	-25, CT (ins T)	-25, L→L	28[1]			5(8)	Frameshift, stop at codon -4	125
PS-31-142	-25, CT (ins T)	-25, L→L	35[1]			3(6)		126
PS-46-02	-24, CTG→GAG	-24, V→E	62	4[PEG]		8(17)		Rezende, Simmonds Pers. Comm.
PS-46-16	-24, GTG→GAG	-24, V→E	61[5]	1[5, PEG]		2(3)		Rezende, Simmonds Pers. Comm.
PS-1-B	intron a, A→G, exon 1, +4		28	<12.5[PEG]	5	2(2)	Abnormal mRNA splicing, exon skipping	127
PS-33-088	-12, CAA→TAA	-12, Q→Stop			58	2(3)		Borgel, Gandrille Pers. Comm.
PS-33-022	-12, C(del AACAGGC)	-12, Q→L	55	12[PEG]		3(3)	Frameshift, stop at codon 43	128
PS-33-001	-2, CGT→CTT	-2, R→L	87	112[PEG]	57	1(2)	Qualitative deficiency	129
PS-33-002	-2, CGT→CTT	-2, R→L	130	60[PEG]	50	1(2)	Qualitative deficiency	129
PS-33-003	-1, CGT→CAT[4]	-1, R→H	126	101[PEG]	64	2(2)	Qualitative deficiency	129
PS-44-740	9, AAA→GAA	9, K→E	108	70	24		Qualitative deficiency	130
PS-81	9, AAA→GAA	9, K→E	83	75	60	2(4)	Qualitative deficiency	131
PS-33-087	19, GAA→TAA	19, E→Stop	45	35	32			Borgel, Gandrille Pers. Comm.
PS-34-276	19, GAA→TAA	19, E→Stop	41	19	16	5(6)		124
PS-34-232	21 C(del TG)	21, L→L	49	25[PEG]	19	8/13	Frameshift, stop at codon 23	118
PS-33-004	22, TGC→TGA	22, C→Stop	57	22[PEG]	33	3(4)		129
PS-33-005	26, GAA→GCA	26, E→A	54	45[PEG]	27	2(2)		129
PS-33-006	26, GAA→GCA	26, E→A	42	25[PEG]	38			129
PS-39-290	26, GAA→GCA	26, E→A	52	57[PEG]	46	2(5)	PS deficient son did not carry the mutation	130
PS-33-086	26, GAA→GCA	26, E→A	52	44	25	2(2)		Borgel, Gandrille Pers. Comm.

PS-33-089	26, GAA→GCA	26, E→A	45	35	32			Borgel, Gandrille Pers. Comm.
PS-33-090	26, GAA→GCA	26, E→A	57	49	44	2(2)		Borgel, Gandrille Pers. Comm.
PS-33-091	27, GCC→GAC	27, A→D	62	44				Borgel, Gandrille Pers. Comm.
PS-33-007	31, TTT→TGT	31, F→C	63	72[PEG]	45			129
PS-33-008	37, ACG→ATG[4]	37, T→M	60	42[PEG]	37			129
PS-43-22	intron b, A→G, exon3 -2		47	25[PEG]		2(3)	Cosegregation with type I and III	132
PS-1-unnamed	43, AA (del A)	43, K→N	0.2			2(2)	Homozygote for type I deficiency; Frameshift, stop at codon 45	133
PS-33-057	44, TA (del CTTA)	44, Y→Stop	47	<10[PEG]		3(3)	Frameshift, stop at codon 44	134
PS-33-023	44, TA (del CTTA)	44, Y→Stop	35	37[PEG]		6(7)	Frameshift	128
PS-34-21	45-46 TTAGTT→ TTGTTT	45-46 L, V→L, F	41	9.5[PEG]		17(33)	In splice donor of intron c (positions -1 and -2)	118
PS-81-07[3]	46, GTT→CTT	46, V→L	71	49	36	2(4)	Mutation also affecting the donor splice site of intron c (position -1) Mutated mRNA +	97
PS-81-09	46, GTT→CTT	46, V→L	50	38	44	1(1)	Designated as PS Osaka 2	131
PS-46-18	49, CGC→TGC[4]	49, R→C	70[5]	12[5, PEG]		9(76)		Rezende, Simmonds Pers. Comm.
PS-44-762	54, GGG→GAG	54, G→E	61	59	42			130
PS-81-08	54, GGG→AGG	54, G→R	74	50	34	1(1)	Variant designated as PS Fukuoka	Tsuda, et al. Pers. Comm.
PS-81-05	62, TCA→TGA	62, S→Stop	41	15	<5	1(4)	Mutation inducing exon 4 skipping Inframe deletion of 87 nt encoding the thrombin-sensitive loop	135
PS-33-058	67, C(del C)	67, P→L	45	21[PEG]	30	1(2)	Frameshift, Stop at codon 69	134
PS-33-059	70, AGA→AGC	70, R→S	61	40[PEG]		2(4)		134
PS-44-01	Intron d, G→A[4], exon 4 + 1		60	14		4(7)	Results in both skipping of exon IV and utilization of cryptic splice site 48 bp 3' to donor splice site Mutated mRNA +	121
PS-44-742	Intron d, G→A[4], exon 4 + 1		31	28[PEG]			In splice donor site	130
PS-33-010	82, CC (del T)	82, P→P	40	12[PEG]	14	5(11)	Frameshift, stop at codon 91	129, 136
PS-34-32	96, CGA→TGA	96, G→Stop	59	14	22	3(5)		124

PS-33-011	103, ACT→AAT	103, T→N	85	80[PEG]	46		Qualitative deficiency Mutation on a residue involved in interaction with aPC (68)	129
PS-34-003	108, TGG→TGA	108, W→Stop	35	14[PEG]		1(2)		118
PS-45-12	108, TGG→TGT	108, W→C	46	8[6, PEG]		2(4)		Lind Pers. Comm.
PS-33-101	113, TGT→TTT	113, C→F	75	48	55		Subject bearing also an ivs g, G→A, exon 8-20	Borgel, Gandrille Pers. Comm.
PS-33-012	intron e, G→A, exon 5 + 5		95	20[PEG]	31			129
PS-33-108	intron e, G→A, exon 5 + 5		70	50	27	2(3)	Compound heterozygous subject bearing also the 460 Ser→Pro mutation	137
			100	97	30		Results of the propositus son who bore only this mutation. Qualitative deficiency Abnormal mRNA transcript with exon 5 skipping leading to the presence of a circulating truncated PS lacking EGF-1	
PS-33-060	116, GAC→GGC	116, D→G	53	60[PEG]	36	3(4)		134
PS-33-061	120, TGC→GGC	120, C→G	63	53[PEG]	40	2(2)		134
PS-44-1322	122, GA (del T)	122, D→D	50[5]	35[5]		12(22)	Frameshift, stop at codon 126	130
PS-44-385	145, TGT→TAT	145, C→Y		50				130
PS-66-1	146-147 Ins A	147, N→K	-/48[5]	0[1]/Traces[5]		2(4)	Frameshift, stop at 155 Compound heterozygous subject for R 410→Stop mutation	95, 138, 139
PS-81-02	155, AAG→GAG	155, K→E		101	37			140
PS-81-03	155, AAG→GAG	155, K→E	70	70	31	7(9)		141, 142
PS-33-024	186, (ins T)	186, E→stop	36	10[PEG]			Frameshift	128
PS-33-097	187, TGC→AGC	187, C→S	69	31	30			Borgel, Gandrille Pers. Comm.
PS-33-062	193, TAT→TAA	193, Y→Stop	45	43[PEG]				134
PS-33-093	197, TCA→TAA	197, S→Stop						Borgel, Gandrille Pers. Comm.
PS-44-293	200, TGT→TCT	200, C→S	60	36			Asymptomatic family member	130
PS-39-58	Intron g, A→T, exon 8-2	Del I 203-D 204	80	73	28	3(7)	Splice site mutation activating a cryptic splice site Qualitative deficiency	Bernardi Pers. Comm.
PS-33-013	204, GAT→GGT	204, D→G		45[PEG]				129

PS-33-092	206, TGC→CGC	206, C→R	64	19	21			Borgel, Gandrille Pers. Comm.
PS-33-014	208, GAG→AAG	208, E→K	46	57[PEG]				129
PS-39-17	217, AAT→AGT	217, N→S	71	45	38	8(26)	Predicted to lead to loss of calcium binding affinity Mutated mRNA+	121, 143
PS-46-04	217, AAT→AGT	217, N→S	67[5]	8[5, PEG]		2(5)	Two additional base changes in the same family: 192 AGA→AAA intron b, G→A, exon 2 +5	Rezende, Simmonds Pers. Comm.
PS-44-07	220, G (del G)	220, G→E		18		3(4)	Frameshift, stop at codon 250 Mutated mRNA -	121
PS-33-015	224, TGC→TGG	224, C→W	55	35[PEG]	29	7(9)		129
PS-33-016	224, TGC→CGC	224, C→R	46	15[PEG]	16			129
PS-34-218	238, CAG→TAG	238, Q→Stop	58	14	35	5(5)	Cosegregates with type I and III	118
PS-34-234	238, CAG→TAG	238, Q→Stop	50	21	27	5(10)		118
PS-34-CPI	238, CAG→TAG	238, Q→Stop	37	7		3(5)		118
PS-34-C4	238, CAG→TAG	238, Q→Stop	46	11		3(3)		118
PS-33-024	259, CTT→CCT	259, L→P	55	50[PEG]		3(3)		128
PS-31-061	261, T (del T)	261, L→W	66			8(15)	Frameshift, stop at codon 272	126
PS-351-564	265, TTT (Ins T)	266, A→C					Frameshift, stop at codon 287	144
PS-351-474	265, TTT (Ins T)	266, A→C					Frameshift, stop at codon 287	144
PS-351-732	265, TTT (Ins T)	266, A→C	48	26		6(13)	Frameshift, stop at codon 287	144
PS-31-081	267, GG (del G)	267, G→G	30[1]				Frameshift, stop at codon 272	126
PS-44-261	283, TCA→CCA	283, S→P		23		4(4)	Asymptomatic family member	130
PS-33-098	292, GAT→AAT	292, D→N			<5			Borgel, Gandrille Pers. Comm.
PS-34-244	293, TCA→TGA	293, S→Stop	49	16	12	10(12)		118
PS-34-249	293, TCA→TGA	293, S→Stop	40	20	16	3(3)		118
PS-81-10	295, GGC→AGC	295, G→S	50	29	24	2(3)	Designated as PS Osaka 1	131
PS-46-19	295, GGC→GTC	295, G→V	69[5]	8[5, PEG]		8(17)		Rezende, Simmonds Pers. Comm.
PS-46-01	295, GGC→GTC	295, G→V	60[5]	3[5, PEG]		44(122)		117, 145
PS-54-9	310, CTG→CCG	310, L→P	26[1]	2[1, PEG]		2(2)		146
PS-33-104	316, GGA→AGA	316, G→R	62	33[PEG]	32		Subject with a 35, Pro→Leu mutation	Borgel, Gandrille Pers Comm
PS-33-063	323, AAG→GAG	323, K→E	54	44[PEG]	51	2(5)		134
PS-33-017	335, GAT→AAT	335, D→N	50	28	20	1(3)		129

PS-31-086	340, GGT→GTT	340, G→V	19[1] 37/71			3(4)	PS levels in son/father of the propositus who were not on oral anticoagulant therapy	126
PS-221-276	340, GGT→GAT	340, G→D	37	7[PEG]	8		Apparent homozygote	130
PS-33-064	342, TGG→TAG[4]	342, W→stop	64		40			134
PS-45-6	342, TGG→CGG	342, W→R	44	1[6, PEG]		2(8)		Lind Pers Comm
PS-44-06	Intron j, G→A, exon 10 + 1			13		3(3)	In donor splice site Mutated mRNA +	121
PS-44-665	Intron j, A→C, exon 10 +3		54[5]	47[5]	43[5]	5(13)	In donor splice site	130
PS-31-023	Intron j, G→A[4], exon 10 +5		28[1]				In donor splice site	125
PS-31-069	Intron j, G→A[4], exon 10 +5		47			3(4)	In donor splice site	125
PS-43-2	Intron j, G→A[4], exon 10 +5		31[1]	3[1, PEG]		4(6)	In donor splice site	146
PS-43-14	Intron j, G→A[4], exon 10 +5		30[1]	3[1, PEG]		1(1)	In donor splice site Mutated mRNA + Use of a cryptic splice site leading to the deletion of 32 nt in exon 10	147
PS-43-3	Intron j, G→A[4], exon 10 +5		81	4[PEG]		2(2)	In donor splice site	147
PS-34-26	Intron j, G→A[4], exon 10 +5				39	8(17)	In donor splice site	118
PS-46-10	Intron j, G→A[4], exon 10 +5		56	1[PEG]		4(6)		Rezende, Simmonds Pers. Comm.
PS-34-1	Intron j, G→T, exon 11 -1		55	5[PEG]		5(8)	In invariant AG of acceptor splice site of intron j	118
PS-45-7	349, GAA→AAA	349, E→K	36	5[6, PEG]		2(2)		Lind Pers. Comm.
PS-45-13	349, GAA→AAA	349, E→K	38	1[6, PEG]		6(11)		Lind Pers. Comm.
PS-45-16	349, GAA→AAA	349, E→K	65	5[6, PEG]		2(2)		Lind Pers. Comm.
PS-45-3	349, GAA→AAA	349, E→K	62	4[6, PEG]		4(6)		Lind Pers. Comm.
PS-45-19	349, GAA→AAA	349, E→K	70	8[6, PEG]		1(1)		Lind Pers. Comm.
PS-45-22	349, GAA→AAA	349, E→K	53	4[6, PEG]		3(5)		Lind Pers. Comm.
PS-44-833	368, AAA→TAA	368, K→Stop	25[1]	5[1, PEG]		3(3)		130

PS-33-094	372, del CTTTTTT, Ins AA	372, L→K	42	17		2(2)		Borgel, Gandrille Pers Comm.
PS-33-100	372, del CTTTTTT, Ins AA	372, L→K	51	22	36	1(3)		Borgel, Gandrille Pers. Comm.
PS-34-33	383, AA (del A)	383, K→K	43	22	37	2(11)	Frameshift, stop at codon 405 Cosegregates with type I and type III in the pedigree	124
PS-33-065	Intron k, exon 11 del +1 to +4		52	10[PEG]	10			134
PS-44-unnamed	Intron k, A→G, exon 12, -9		29[1]	4[1]		5(7)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-44-unnamed	Intron k, A→G, exon 12, -9		27[1]	3[1]		2(6)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-44-unnamed	Intron k, A→G, exon 12, -9		27[1]	7[1]		4(4)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-44-unnamed	Intron k, A→G, exon 12, -9		51	21		3(3)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-44-unnamed	Intron k, A→G, exon 12, -9		29[1]	4[1]		5(7)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-44-unnamed	Intron k, A→G, exon 12, -9		23[1]	9[1]		4(11)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-44-unnamed	Intron k, A→G, exon 12, -9		61	27		2(3)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-44-unnamed	Intron k, A→G, exon 12, -9		35[1]	4[1]		1(1)	Abnormal mRNA splicing Insertion of 8 bp of intron k	148
PS-33-066	405, CTA→CCA	405, L→P	80[5]	24[5, PEG]	34[5]	3(6)		134
PS-32-02	405, CTA→CCA	405, L→P	50	35[PEG]		7(12)		149
PS-44-835	408, TGT→AGT	408, C→S	64	20[PEG]		4(4)	Mutated mRNA +	130
PS-33-067	410, CGA→TGA[4]	410, R→Stop	60	45[PEG]	36	2(2)	Polymorphism in 3'-UT part See table II	134
PS-33-068	410, CGA→TGA[4]	410, R→Stop	11[1]	9[1, PEG]				134
PS-33-069	410, CGA→TGA[4]	410, R→Stop	52	59[PEG]	35	2(2)	Polymorphism in 3'-UT part See table II	134
PS-33-070	410, CGA→TGA[4]	410, R→Stop	57	40[PEG]	36		Polymorphism in 3'-UT part See table II	134
PS-33-071	410, CGA→TGA[4]	410, R→Stop	62	30[PEG]	15	3(3)	Polymorphism in 3'-UT part See table II	134
PS-43-10	410, CGA→TGA[4]	410, R→Stop	23[1]	4[1, PEG]		3(5)	Mutated mRNA -	146
PS-45-10	410, CGA→TGA[4]	410, R→Stop	56	5[6, PEG]	48	3(6)	Founder effect with PS-45-11 and 45-1	150, 151
PS-45-11	410, CGA→TGA[4]	410, R→Stop	35	3[6, PEG]	18	3(3)	Founder effect with PS-45-10 and 45-1	150, 151

PS-45-1	410, CGA→TGA[4]	410, R→Stop	70	1[6, PEG]	44	3(9)	Founder effect with PS-45-10 and 45-11	150, 151
PS-34-265	410, CGA→TGA[4]	410, R→Stop	77	30	12	5(12)	Cosegregates with type I and III in the pedigree	124
PS-66-1	410, CGA→TGA[4]	410, R→Stop	-/70[5]	0/Traces[5, PEG]		3(4)	Compound heterozygous subject for 146-147 Ins A	95, 138, 139
PS-46-05	431, AA (ins A)	431, N→K	68	6[PEG]		1(1)	Frameshift, stop at codon 342	Rezende, Simmonds Pers. Comm.
PS-46-06	431, AA (ins A)	431, N→K	50	6[PEG]		2(5)	Frameshift, stop at codon 342	Rezende, Simmonds Pers. Comm.
PS-33-072	434, TGC→CGC	434, C→R	64	46[PEG]	33			134
PS-34-23	441, GGC→TGC	441, G→C	35	35[PEG]	51	11(22)		118, 120
PS-34-2	444, TAT→TGT	444, Y→C	25[1]	6[1, PEG]		7(10)		118, 120
PS-44-09A	448, GGA→TGA	448, G→Stop	37			1(2)	Mutated mRNA -	121
PS-45-8	448, 449 del (A)	449, I→L	51	3[6, PEG]	24	10(13)	Frameshift, stop at codon 454	150
PS-33-073	460, TCC→GCC	460, S→A	65	39[PEG]	55			134
PS-34-24	465, TGG→TGA	465, W→Stop	67	17		2(10)	Cosegregates with type I and III in the pedigree	124
PS-351-551	465, TGG→TGA	465, W→Stop						144
PS-31-010	467, GTA→GGA	467, V→G	32[1]			4(6)		126
PS-31-026	467, GTA→GGA	467, V→G	23[1]					126
PS-33-074	469, GTG→ATG	469, V→M	73	60[PEG]	57	1(3)		134
PS-33-075	474, CGT→TGT[4]	474, R→C	77	37[PEG]	42	4(5)		134
PS-81-01	474, CGT→TGT[4]	474, R→C	45	7	11	6(9)	Designated as PS Nagoya Mutated mRNA +	152
PS-33-076	484, GCC→CCC	484, A→P	50	36[PEG]		1(4)	De novo mutation	134, 153
PS-45-18	485, TTG→TCG	485, L→S	58	3[6, PEG]		2(3)		Lind Pers. Comm.
PS-33-077	491, ACA→GCA	491, T→A	53	52		2(4)		134
PS-43-8	511, TTA→TCA	511, L→S	26[1]	2[1, PEG]		5(6)	Mutated mRNA +	146
PS-34-214	520, CGG→GGG	520, R→G	36	16	14	4(4)	Type I in homozygous propositus Type III in heterozygous relatives	118,120
PS-81-04	522, CAG→TAG	522, Q→Stop	55	<30	<10	3(5)		154
PS-33-078	522, CAG→TAG	522, Q→Stop	31[1]	15[1, PEG]	31[1]	5(10)		134
PS-33-079	522, CAG→TAG	522, Q→Stop	62	35[PEG]	41	2(5)		134
PS-33-080	522, CAG→TAG	522, Q→Stop	41	30[PEG]				134
PS-33-081	527, TGT→TAT	527, C→Y	55	20[PEG]		2(4)		134
PS-33-082	534, CTG→CGG	534, L→R	36	<10[PEG]		1(2)		134
PS-33-083	534, CTG→CGG	534, L→R	39	35[PEG]		2(2)		134

PS-33-095	534, CTG→CGG	534, L→R	51	25	28	2(2)		Borgel, Gandrille Pers. Comm.
PS-34-274	536, TT (del T)	536, F→L	49	20	8	3(5)	Frameshift, stop at codon 551 Cosegregates with type I and type III in the pedigree	124
PS-32-01	539, ins TC	539, N→S	33[5]	19[5, PEG]		5(15)		139
PS-43-7	543, CTG→CAG	543, L→Q	86	30[PEG]		2(2)	Mutated mRNA +	146
PS-33-103	547, ACA→GCA	547, T→A	66	46		2(4)		Borgel, Gandrille Pers. Comm.
PS-44-04	547, (del AC)	547, T→T	44	13		2(4)	Exhibits allelic exclusion Frameshift, stop at codon 549	121, 155
PS-31-070	565, GT (ins T)	565, V→V	19[1]/35[5]			3(6)	Frameshift, stop at codon 596	126
PS-44-05	570, ATG→ACG	570, M→T	24[1]/74[5]	<10[1]/54[5]		2(2)	May affect C4bBP binding Mutated mRNA +	121, 155
PS-43-6	575, GCC→CCC	575, A→P	30[1]	2[1]		3(4)	Mutated mRNA+	146
PS-31-022	578, C (ins C)	578, P→L	19[1]/51[5]			4(6)	Frameshift, stop at codon 596	126
PS-33-096	Intron n, (del CTAATATT), exon 15 -28 to -21		61	44	62			Borgel, Gandrille Pers. Comm.
PS-81-06[2]	Intron n, G→T, exon 15 -1		50	17	28	2(4)	Activation of a cryptic splice site. Deletion of the first 13 nt of exon 15 Frameshift, stop at codon 587 Mutated mRNA +	97
PS-33-099	588, GCC (ins C)	589, T→H	53	28[PEG]		2(3)	Frameshift, stop at codon 596	Borgel, Gandrille Pers. Comm.
PS-44-37	595, TAT→TAA	595, Y→Stop			23[5]			130
PS-351-734	598, TGC→TTC	598, C→F						144
PS-34-27	599, ATG→ACG	599, M→T		7[1]	24	4(6)		124
PS-1-A	603, ATT→AGT	603, I→S	71	5[PEG]		3(3)		127
PS-46-09	623, CAC→CCC	623, H→P	43[5]	4[5, PEG]		4(5)		Rezende, Simmonds Pers. Comm
PS-33-107	624, TCA→TTA	624, S→L	50	33	19	5(10)		Borgel, Gandrille Pers. Comm. 152
PS-33-026	625, TGT→CGT	625, C→R	43	18[PEG]		5(6)		128
PS-33-027	625, TGT→CGT	625, C→R	48	25[PEG]		3(3)		128
PS-33-028	625, TGT→CGT	625, C→R	42	16[PEG]		2(2)		128
PS-33-029	625, TGT→CGT	625, C→R	44	38[PEG]		3(3)		128

PS-33-084	625, TGT→CGT	625, C→R	30	25[PEG]				134
PS-44-743	625, TGT→CGT	625, C→R	32	13		2(2)	Mutated mRNA +	130
PS-34-APC13	626, CCA→CTA	626, P→L	59	30	12	3(3)		120, 124
PS-45-4	632 AC(delAAAG)	633, K→I	66	10[6, PEG]		2(2)	Frameshift, stop at new codon 642	Lind Pers. Comm.
PS-43-1	633, (del AA)	633, K→Q	26[1]	7[1, PEG]		4(8)	Frameshift, new stop at codon 656 Mutated mRNA +	146
PS-43-4	633, (del AA)	633, K→Q	8[1]	2[1, PEG]		1(2)	Frameshift, new stop at codon 656 Mutated mRNA +	146
PS-43-12	633, (del AA)	633, K→Q	26[1]	6[1, PEG]			Frameshift, new stop at codon 656	Mannhalter Pers. Comm.
PS-31-014	636, TAA→TAT	636, stop→Y	24[1]			3(5)	New stop at codon 649	125
PS-31-039	636, TAA→TAT	636, stop→Y	22[1]			2(4)	New stop at codon 649	125

Table I Notes

[a] [b] All data are from heterozygous, symptomatic propositi, unless otherwise indicated in the Comments column
Amino acids are numbered as in Schmidel et al (60)

[c] [d] PEG=Free protein S levels evaluated after PEG precipitation of C4b-BP bound protein S. Free PS antigen is expressed as % of the free PS antigen in normal plasma, otherwise indicated
Cosegregation: x=number of PS deficient heterozygotes and/or homozygotes bearing the mutation; y=total number of tested family members (normal plus PS deficient heterozygotes and/or homozygotes)

[1]Plasma antigen levels measured during oral anticoagulant treatment
[2]Results from the mother of a propositus compound heterozygous for this mutation and a 46 V→L mutation (see PS-81-07)
[3]Results from the father of a propositus compound heterozygous for this mutation and an intron n, G→T, exon 15 -1 mutation (see PS-81-06)
[4]Mutation on CpG
[5]Protein S levels of related subject bearing the mutation but with no oral anticoagulant therapy
[6]Normal range = 18-47%

Mutated mRNA +/- corresponds to the presence/absence of mutated mRNA.
OAC = oral anticoagulant therapy
Pers. Comm. = personal communication