Table II: Sequence variations that are probably polymorphisms (detected in patients or in normal subjects)

Pedigree	Codon with the sequence variation	Predicted mutation	Total PS antigen % of normal	Free PS antigen % of normal	PS Activity % of normal	Comments	Ref
PS-34-normal	5'-UT C→T, exon 1, -296				91	Normal subject	124
PS-34-232	5'-UT T→C, exon 1, -286		49	25	19	No cosegregation with PS deficiency Compound heterozygous See 21, C (del TG)	118
PS-34-201	-30, CTG→CTC	L→L	92	31			124
PS-44-1306	Intron a, A→G, exon 1 +7		<25	<25[PEG]			130
PS-39-288	Intron a, del ATT, exon 2 -67		69	62	37	PS deficient mother did not carry the mutation	130
PS-33-018	35, CCG→CTG	P→L	78	67[PEG]	120		129
PS-33-019	35, CCG→CTG	P→L	84	62[PEG]			129
PS-33-020	35, CCG→CTG	P→L	75	95[PEG]	84		129
PS-39-278	35, CCG→CCA	P→P	46	43[PEG]	60		130
PS-31-006	intron b, G→A, exon 2 +5		Patient with insertion in codon -25, see table I			No co-segregation with PS deficiency	124
PS-46-04	intron b, G→A, exon 2 +5		67	8[PEG]		Co-segregates with PS deficiency	Rezende, Simmonds Pers. Comm.
PS-33-009	49, CGC→CAC[4]	R→H	66	60[PEG]		No co-segregation with PS deficiency	129
PS-34-17	57, ACT→AGT	T→S	44	41		No co-segregation with PS deficiency	120, 124
PS-33-010	192, AGA→AAA	R→K	Patient with deletion in codon 82, see Table I				129, 136
PS-33-092	192, AGA→AAA	R→K	80	74[PEG]	124	Normal subject	Borgel, Gandrille Pers. Comm.
PS-33-093	192, AGA→AAA	R→K				Normal subject	Borgel, Gandrille Pers. Comm.
PS-33-094	192, AGA→AAA	R→K				Normal subject	Borgel, Gandrille Pers. Comm.
PS-34-11	192, AGA→AAA	R→K	93	57		No co-segregation with PS deficiency	124
PS-46-04	192, AGA→AAA	R→K	67	8[PEG]		No co-segregation with PS deficiency	Rezende, Simmonds Pers. Comm.
PS-44-919	intron g, G→A, exon 8 -20		54	61	65	Asymptomatic family member	130
PS-33-001	303, ATC→ATT	I→I	Patient with -2, R→L mutation, see table I				129
PS-33-002	303, ATC→ATT	I→I	Patient with -2, R→L mutation, see table I				129
PS-33-021	344, ATG→GTG	M→V	90	60[PEG]		Homozygous patient	129
	intron k, C→T, exon 11 +54		Frequent polymorphism, see text				71
PS-33-096	418, GGA→GGG	G→G				Normal subject	Borgel, Gandrille

							Pers. Comm.
	460, TCC→CCC	S→P	See text				
PS-33-095	477, ACG→ATG	T→M				Normal subject	Borgel, Gandrille Pers. Comm.
PS-34-5	518, ATA→ATG	I→M	49	26		No co-segregation with PS deficiency	120, 124
PS-34-225	518, ATA→ATG	I→M	102	52	47	No co-segregation with PS deficiency	118, 120
	626, CCA/CCG	P/P	Frequent polymorphism, see text				
PS-33-067	Exon 15, T→G 18 nt after the Stop codon		Patient with 410 R→Stop mutation, see Table I				134
PS-33-069	Exon 15, T→G 18 nt after the Stop codon		Patient with 410 R→Stop mutation, see Table I				134
PS-33-070	Exon 15, T→G 18 nt after the Stop codon		Patient with 410 R→Stop mutation, see Table I				134
PS-33-071	Exon 15, T→G 18 nt after the Stop codon		Patient with 410 R→Stop mutation, see Table I				134
	Exon 15, T→G 520 nt after the Stop codon		Frequent polymorphism, see text				71