Table S3- Novel sequence variations in LTBP2

No.	cDNA position[£]	Reference SNP No.	Exon/ Intron	Effect on protein
	POAG			
1	c.1295C>T	rs137854861	E6	p.Pro432Leu
2	c.1484G>A	rs137854858	E7	p.Arg495Gln
3	c.3571G>A	rs137854862	E24	p.Glu1191Lys
4	c.4250A>G	rs137854863	E29	p.Gln1417Arg
5	c.4912G>A	rs137854860	E34	p.Val1638Met
	PEX syndrome			
1	c.1295C>T	rs137854861	E6	p.Pro432Leu
6*	c.5376delC	rs137854895	E36	p.Tyr1792fsX55
	Other novel variations probably not disease associated			
7	c.1553G>T	rs137854857	E7	p.Ser518Ile
8	c.4356G>A	rs137854865	E29	p.Pro1452Pro
9	c.4668G>C	rs137854866	E32	p.Pro1556Pro
10	c.1999A>C	rs137854859	E11	p.Ile667Leu
11	c.3527-14T>C	rs137854886	I23	Intronic
12	c.4699A>G	rs137854864	E32	p.Met1567Val

£A of the initiation codon was designated +1; * Not novel, previously observed in PCG affected sons