Table S2- *LTBP2* sequence variations observed among 90 POAG and PEX Syndrome patients

cDNA location[*,‡]	Gene location	Effect on protein	Disease associated/ Disease[†]	No. of Patients		Total no. of alleles	Reference SNP No.
				Homo	Het		
c.1-232G>A	5' UTR			0	3	3	rs61980926
c.956C>A	E4	Pro319Gln		1	21	23	rs2304707
c.1287G>A	E6	Leu429Leu		1	16	18	rs61738025
c.1295C>T	E6	Pro432Leu	+/POAG, PEXG	0	2	2	rs137854861
c.1484G>A	E7	Arg495Gln	+/POAG	0	1	1	rs137854858
c.1553G>T	E7	Ser518Ile	-/POAG, PEX[¶]	0	4	4	rs137854857
c.1864+22C>A	I9			25	11	61	rs3742794
c.1987+21G>A	I10			7	34	48	rs862025
c.1999A>C	E11	Ile667Leu	?/PEX	0	1	1	rs137854859
c.2406T>C	E14	Thr802Thr		10	40	60	rs699374
c.2429-31A>G	I14			13	1	27	rs862030
c.2502T>C	E15	Thr834Thr		40	32	112	rs862031
c.2966C>G	E19	Pro989Arg		0	4	4	rs76172717
c.3262G>A	E21	Gly1088Ser		0	1	1	rs61505039
c.3278-16C>T	I21			0	3	3	rs117613718
c.3527-21C>T	I23			0	1	1	rs78703020
c.3527-14T>C	I23		-/PEX	0	1	1	rs137854886
c.3571G>A	E24	Glu1191Lys	+/POAG	0	1	1	rs137854862
c.3908-57G>A	I26			20	22	62	rs8020768
c.3908-55A>G	I26			2	23	27	rs60130392
c.3908-53A>G	I26			16	23	55	rs45537036
c.3908-51A>G	I26			0	4	4	rs61980865
c.4034-48C>T	I27			3	33	39	rs2286411
c.4250A>G	E29	Gln1417Arg	+/POAG	1	0	2	rs137854863
c.4356G>A	E29	Pro1452Pro	-/POAG	0	1	1	rs137854865
c.4369del(+25to+35)	I29			1	7	9	rs36016624
c.4668G>C	E32	Pro1556Pro	-/POAG	0	1	1	rs137854866
c.4699A>G	E32	Met1567Val	?/PEXG	0	1	1	rs137854864
c.4912G>A	E34	Val1638Met	+/POAG	0	1	1	rs137854860
c.5376delC[§]	E36	Tyr1792fsX55	+/PCG, PEX				rs137854895

* A of the initiation codon was designated +1

‡ Reference sequences used: NM_000428.2 and NP_000419.1

† Novel sequence varaitons that were (+) or were not (-) disease associated and disease of individual in whom variation was observed

¶ Also observed in two control individuals

§ Mutation previously identified as PCG causing in homozygous proband and now seen in his PEX affected mother in heterozygous state

E, exon; I, intron; Homo, homozygous; Het, heterozygous