

## Appendix 1

Allelic and genotype association testing results of rs1048661, rs3825942 and rs2165241 in LOXL1 for Greek cases with PEX and controls are shown. The single asterisk indicates the p values, OR values and CI% for G versus T of rs1048661, G versus A of rs3825942 and T versus C of rs2165241, respectively.

SNP ID	Genotype Frequency (%)				Allele Frequency (%)				
	Genotype	Cases PEX (n=48)	Controls (n=52)	p-value	Allele	Cases PEX (n=96)	Controls (n=104)	p-value	OR (CI%)
<b>rs1048661 (R141L)</b>	GG	34 (70.8)	34 (65.4)	0.339	G	82 (85.4)	83 (79.8)	0.297*	1.48 (0.706-3.112)*
	GT	14 (29.2)	15 (28.8)		T	14 (14.6)	21 (20.2)		
	TT	0 (0.0)	3 (5.8)						
<b>rs3825942 (G153D)</b>	GG	36 (75)	27 (51.9)	0.004	G	83 (86.5)	67 (64.4)	3.24x10 <sup>-4</sup> *	3.52 (1.735-7.166)*
	GA	11 (22.9)	13 (25)		A	13 (13.5)	37 (35.6)		
	AA	1 (2.1)	12 (23.1)						
<b>rs2165241</b>	TT	29 (60.4)	18 (34.6)	0.005	T	74 (77.1)	55 (52.9)	3.53x10 <sup>-4</sup> *	2.99 (1.625-5.527)*
	CT	16 (33.3)	19 (36.5)		C	22 (22.9)	49 (47.1)		
	CC	3 (6.3)	15 (28.8)						