Chr.	Position	MAF	Ref	Mut (*)	Function	Gene	CADD/MSC
2	61144011	.	G	A	Essential splicing	REL	26.7/2.31
2	68385590	2.09e-4	T	C	Missense	PNO1	22.9/2.31
2	70406718	1.80e-3	C	A*	Missense	C2orf42	15.8/2.31
2	71896835	3.69e-3	G	A*	Missense	DYSF	26.2/0.01
2	130872821	4.57e-4	A	T	Missense	POTEF	23/2.31
2	238978106	.	G	A	Missense	SCLY	13.16/2.31
3	108324270	2.15e-3	A	G*	Missense	DZIP3	23/4.76
3	108788596	1.11e-4	G	T	Missense	MORC1	5.84/2.31
5	131705723	1.25e-3	T	A*	Missense	SLC22A5	18.61/5.88
5	131726578	1.25e-3	A	G*	Missense	SLC22A5	11.09/5.88
5	131995889	6.75e-4	G	A*	Missense	IL13	11.01/8.13
6	21595777	1.37e-3	A	G*	Missense	SOX4	23/5.24
6	36076169	9.94e-4	A	G*	Missense	MAPK14	21.5/17.46
6	161152819	9.59e-3	C	T*	Missense	PLG	26.8/0
8	127568857	9.82e-5	T	A*	Missense	FAM84B	21.9/2.31
8	132966167	1.44e-3	T	G*	Missense	EFR3A	24.3/2.31
9	131583067	3.33e-4	G	A	Missense	ENDOG	34/2.31
10	81372080	2.58e-3	C	T*	Missense	SFTPA1	22.5/2.31
12	94603406	8.08e-3	G	C*	Missense	PLXNC1	3.14/2.31
13	52508989	2.07e-3	G	A*	Missense	ATP7B	24.1/1.46
16	14234467	8.14e-5	G	T*	Missense	MKL2	26.6/2.31
19	16008417	1.88e-4	G	A	Missense	CYP4F2	10.72/2.31

Supplemental Table 2: Homozygous variants found in the whole exome sequencing of the patient after filtering the data. Chr: chromosome; MAF: Minor allelic frequency; Ref: reference allele; Mut: Mutation; CADD: combined annotation-dependent depletion; MSC: mutation significance cutoff. * refers to variants for which homozygosity is reported in the general population (gnomAD, GME databases)