Supplementary Table 1. List of 27 rare, functional and homozygous variants from Patient 1. Damaging SIFT and Polyphen-2 values are displayed in bold.

Gene	Amino acid change	Genomic coordinate (hg19)	Nucleotide change	Read number (non-reference read / total coverage depth)	SIFT	Polyphen-2
PRCC	p.H202L	chr1:156756488	A>T	37/37	0.69	0.842
ATP1A4	p.M865I	chr1:160147313	G>A	45/45	**0.00**	0.862
MPV17	p.R50W	chr2:27535899	G>A	37/37	**0.00**	**1.000**
CYP1B1	p.R368H	chr2:38298394	C>T	33/33	**0.00**	**1.000**
DGKD	p.E652K	chr2:234358693	G>A	51/51	0.50	0.187
BBX	p.S4G	chr3:107429317	A>G	27/27	0.17	0.014
VIPR2	p.G317S	chr7:158826926	C>T	38/38	0.08	**1.000**
RAD54B	p.R415H	chr8:95411776	C>T	12/12	0.13	**1.000**
EIF3A	p.R803K	chr10:120810098	C>T	192/192	0.49	**0.956**
PRDX3	p.M138L	chr10:120931979	T>G	117/117	0.26	0.367
SLC22A18	p.R377Q	chr11:2946282	G>A	89/90	0.08	**1.000**
OR52K2	p.I31L	chr11:4470660	A>C	138/138	0.45	**0.985**
OR52K2	p.F285L	chr11:4471424	C>G	83/83	0.30	0.048
MMP26	p.D89G	chr11:5011044	A>G	112/112	0.45	0.002
OR10A4	p.L286F	chr11:6898736	G>C	73/73	0.42	**1.000**
NLRP10	p.F533C	chr11:7981561	A>C	29/29	**0.00**	**0.998**
TTC17	p.K202E	chr11:43413443	A>G	13/13	0.54	0.491
GAS6	p.M172K	chr13:114530112	C>T	164/165	**0.00**	0.863
GRB7	p.R255Q	chr17:37900423	G>A	66/66	0.06	**1.000**
KRT34	p.T9I	chr17:39538599	G>A	80/80	**0.01**	0.067
FBN3	p.R861Q	chr19:8191204	C>T	48/48	0.29	0.118
CD97	p.N33D	chr19:14499537	A>G	94/94	0.20	0.122

COLGALT1	p.V582I	chr19:17692128	G>A	165/165	0.30	0.373
ZNF14	p.D54G	chr19:19824930	T>C	22/22	0.23	**0.973**
IFNL1	p.R175C	chr19:39789076	C>T	64/64	**0.00**	**1.000**
SPTBN4	p.E312K	chr19:41008071	G>A	48/48	0.30	0.870
CYP2A7	p.A166T	chr19:41386147	C>T	93/93	0.12	0.102