Supplementary Table 2. List of 23 rare, functional and homozygous variants from Patient 2. Damaging SIFT and Polyphen-2 values are displayed in bold. na = not applicable.

Gene	Amino acid change	Genomic coordinate (hg19)	Nucleotide change	Read number (non-reference read / total coverage depth)	SIFT	Polyphen-2
OR5H1	p.P79S	chr3:99334466	C>T	277/277	**0.00**	0.662
SERAC1	1 bp downstream of exon 12	chr6:158458746	C>G	445/445	na	na
COL28A1	p.V155M	chr7:7537722	C>T	355/355	**0.01**	**1.000**
FAM75A1	p.A134P	chr9:39877413	G>C	117/117	0.2	**0.993**
DGKK	p.K286N	chrX:50180225	T>G	709/709	**0.00**	**0.999**
SHROOM4	p.A93T	chrX:50398041	C>T	92/92	0.39	0.002
SAGE1	p.N643S	chrX:134820303	A>G	358/358	0.12	0.712
VGLL1	p.T21M	chrX:135445907	C>T	136/136	0.28	**1.000**
ANKRD13B	p.Q599R	chr17:24964641	A>G	51/51	1	0.000
LOC401052	p.P21L	chr3:10025103	G>A	279/279	0.1	0.938
TNRC18	p.A2084T	chr7:5331303	C>T	159159	0.92	0.002
MUC12	p.S428Y	chr7:100421847	C>A	238/238	**0.04**	0.845
MUC12	p.P1271L	chr7:100424376	C>T	138/138	0.33	0.02
GIT1	p.T66M	chr17:24934171	G>A	233/233	**0.01**	**1.000**
FGD1	p.R910Q	chrX:54489424	C>T	169/169	1	0.003
TESK2	p.R18H	chr1:45695992	C>T	501/501	**0.01**	0.317
DNAH9	p.A1791V	chr17:11548465	C>T	451/451	0.1	0.279
KCND1	p.S575A	chrX:48705007	A>C	181/181	0.31	0.004
UBXN8	p.X209W	chr8:30740388	A>G	944/944	na	na
FBLN2	p.R866X	chr3:13645431	C>T	289/289	na	na
NRK	p.P789S	chrX:105046393	C>T	462/462	0.58	0.000

CDHR3	p.A798S	chr7:105460113	G>T	200/200	0.17	0.017
FLG	p.T938R	chr1:150551173	G>C	622/622	0.27	0.000