

Supplementary Table 3. List of 41 rare and functional variants from Patient 3. 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
ABCA4	L1970F	chr1:94473287	G>A	58/107	<b>0.05</b>	<b>1.000</b>
ACTN3	1 bp downstream of exon 13	chr11:66327548	G>T	21/40	na	na
ARID1A	G889D	chr1:27089710	G>A	19/64	0.33	<b>0.998</b>
ASPM	R2039G	chr1:197072266	T>C	18/42	0.36	0.931
ATR	R2533X	chr3:142176504	G>A	90/168	na	na
CACNB2	1 bp upstream of exon 2	chr10:18439811	G>T	18/86	na	na
CEP63	I477T	chr3:134280319	T>C	100/206	0.51	0.972
CFTR	S1426Y	chr7:117306996	C>A	18/38	<b>0.00</b>	<b>0.997</b>
COG4	E639Q	chr16:70516637	C>G	94/182	0.18	0.892
CTC1	I1155V	chr17:8131872	T>C	63/131	0.65	0.004
DDB1	S97N	chr11:61097467	C>T	62/136	0.52	0.000
EYS	V2040D	chr6:65016935	A>T	131/275	0.62	<b>1.000</b>
FLG	G1015S	chr1:152284319	C>T	22/108	0.78	0.000
GP6	R528C	chr19:55525731	G>A	48/96	<b>0.00</b>	0.003
GPR179	E861D	chr17:36486869	C>A	27/40	<b>0.03</b>	<b>0.996</b>
GPR98	Q235H	chr5:89923060	A>T	32/56	0.16	0.001
HLA-C	L315P	chr6:31237814	A>G	7/35	<b>0.00</b>	0.956
HPS3	L339S	chr3:148863186	T>C	79/190	<b>0.01</b>	<b>0.973</b>
KIR3DL1	V440I	chr19:55341713	G>A	40/184	<b>0.00</b>	<b>0.996</b>

KIR3DL1	F30L	chr19:55329789	C>G	60/188	<b>0.02</b>	0.016
KIR3DL1	N335I	chr19:55340819	A>T	115/519	<b>0.02</b>	0.003
MALT1	2 bp upstream of exon 3	chr18:56363596	A>T	13/35	na	na
MLL	R503W	chr11:118343381	C>T	24/43	<b>0.01</b>	<b>0.985</b>
MMP20	Y180X	chr11:102480745	A>T	32/76	na	na
MUC5B	D682G	chr11:1253980	A>G	13/43	0.00	0.000
NOTCH2	H1882Y	chr1:120462072	G>A	20/39	<b>0.00</b>	<b>1.000</b>
NOTCH2	S1741L	chr1:120464424	G>A	90/159	0.12	<b>0.981</b>
NOTCH3	E813K	chr19:15295235	C>T	29/58	<b>0.00</b>	<b>1.000</b>
NTRK1	R55S	chr1:156834186	C>A	95/179	0.67	0.000
OPN1LW	L153M	chrX:153418460	C>A	29/29	0.32	0.066
OPN1MW	Y116S	chrX:153490611	A>C	14/50	0.29	0.002
PHF8	R809C	chrX:54011473	G>A	26/50	<b>0.02</b>	0.929
PRICKLE2	S525G	chr3:64132593	T>C	25/64	0.17	0.000
PRSS1	N29I	chr7:142458451	A>T	78/284	<b>0.04</b>	0.000
PRSS1	K170E	chr7:142460335	A>G	107/236	0.57	0.000
PRSS1	S181N	chr7:142460369	G>A	70/225	0.77	0.000
PRX	R1391W	chr19:40900088	G>A	26/59	<b>0.01</b>	0.000
RAD50	A985V	chr5:131945006	C>T	19/42	0.29	0.000
STIL	V56L	chr1:47768005	C>A	69/167	0.79	0.002
TAZ	F128S	chrX:153642450	T>C	35/72	0.73	0.001
TLL1	G740V	chr4:166996060	G>T	61/109	0.10	<b>1.000</b>