

Supplementary Figure 3

Reference SNP (rs) number	Position (GRCh38/hg38)	Patient	Father	Mother
rs1553637932 (the mutation in the proband)	chr2:240783109	G/C	G/G	G/G
rs2288748	chr2:240784010	A/A	A/A	A/G
rs891991042	chr2:240784437	C/T	C/T	C/T
rs768776074	chr2:240784440	C/T	C/T	C/T
No rs number	chr2:240784443	C/T	C/T	C/T
No rs number	chr2:240784621	C/G	C/G	C/G
No rs number	chr2:240784679	G/C	G/C	G/C