Supplementary Table

Title: Exome filtering scheme and the list of survived and potential pathogenic variants found in Family one and Family two.

	Family One	**Family Two**
Filtering Scheme		
Total number of variants	38019	166318 + 171659
Homozygous variants	15731	94128 + 100282
Coding/splicing	9991	3596 + 3516
Family One: 1000 genomes MAF <0.001	621	na
Family Two: concordance	na	26
Within the autozygome	22	23
Family One: Human Saudi Genome Project database: frequency <0.01 **Family Two: gnomAD frequency <0.001**	1. *PLCH1*:NM_001130960.1:c.2109T>C, p.(=) (synonymous SNV) 2. *PLCH1*:NM_001130960.1: c.2065C>T: p.(Arg689*) (stopgain) 3. *SMC4*:NM_005496:c.2673C>A:p.I891I (synonymous SNV) 4. *C20orf141*:NM_080739: c.265G>A:p.A89T (nonsynonymous SNV) 5. *PCNA*: NM_002592:c.388-18A>C	1. *PLCH1*:NM_001130960.1: c.3236CA>C: p.(Cys1079ValfsTer16) (frameshift)
Predicted to be pathogenic	1. *PLCH1*:NM_001130960.1: c.2065C>T: p.(Arg689*) (stopgain)	1. *PLCH1*:NM_001130960.1: c.3236CA>C: p.(Cys1079ValfsTer16) (frameshift)