

**Supplementary Table**

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

	<b>Family One</b>	<b>Family Two</b>
<b>Filtering Scheme</b>		
<b>Total number of variants</b>	38019	166318 + 171659
<b>Homozygous variants</b>	15731	94128 + 100282
<b>Coding/splicing</b>	9991	3596 + 3516
<b>Family One: 1000 genomes MAF &lt;0.001</b>	621	na
<b>Family Two: concordance</b>	na	26
<b>Within the autozygome</b>	22	23
<b>Family One: Human Saudi Genome Project database: frequency &lt;0.01</b>  <b>Family Two: gnomAD frequency &lt;0.001</b>	<ol style="list-style-type: none"> <li>1. <i>PLCH1</i>:NM_001130960.1:c.2109T&gt;C, p.(=) (synonymous SNV)</li> <li>2. <i>PLCH1</i>:NM_001130960.1:c.2065C&gt;T: p.(Arg689*) (stopgain)</li> <li>3. <i>SMC4</i>:NM_005496:c.2673C&gt;A:p.I891I (synonymous SNV)</li> <li>4. <i>C20orf141</i>:NM_080739:c.265G&gt;A:p.A89T (nonsynonymous SNV)</li> <li>5. <i>PCNA</i>: NM_002592:c.388-18A&gt;C</li> </ol>	<ol style="list-style-type: none"> <li>1. <i>PLCH1</i>:NM_001130960.1:c.3236CA&gt;C: p.(Cys1079ValfsTer16) (frameshift)</li> </ol>
<b>Predicted to be pathogenic</b>	<ol style="list-style-type: none"> <li>1. <i>PLCH1</i>:NM_001130960.1:c.2065C&gt;T: p.(Arg689*) (stopgain)</li> </ol>	<ol style="list-style-type: none"> <li>1. <i>PLCH1</i>:NM_001130960.1:c.3236CA&gt;C: p.(Cys1079ValfsTer16) (frameshift)</li> </ol>