



Fig. S2. Sanger sequence traces for cases 1 and 2. Sequence trace data for case 3 was not available. Case 1 was compound heterozygous for 2 mutations: a maternally inherited missense mutation, c.178G>A p.(Glu60Lys) in exon 2 and a paternally inherited in-frame deletion c.218_220del p.(Cys73del) in exon 3. Case 2 was homozygous for *SLURP1* c.178G>A p.(Glu60Lys) in exon 2.