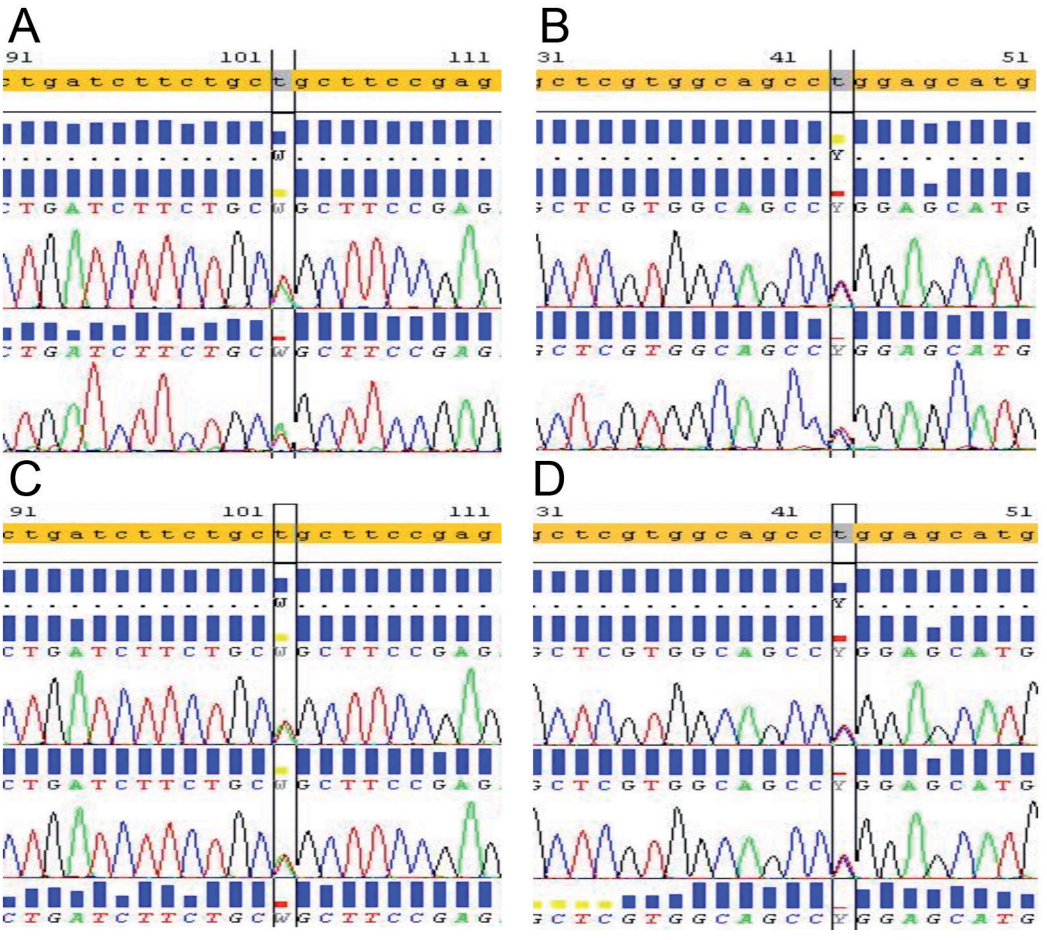

Fig. S1. Compound heterozygous mutations in *SLURP1* in one patient affected by PPK-GN. A) The c.280T>A mutation in exon 3 results in a cysteine to serine change in position 94. B) The c.43T>C mutation in exon 1 changes a tryptophan in position 15 to an arginine. C) One parent carries the c.280T>A mutation. D) The other parent carries the c.43T>C mutation. Reference sequence is NM_020427.2.