



**Suppl. figure (1):** p.T54A mutation: (a.) amino acids sequence of 17 $\beta$ HSD-3 protein, the underlying sequence is the cofactor binding domain and the dashed box is the cofactor binding motif. (b.) RFLP analysis on 3% agarose gel of exon 2 of HSD17B3 gene digested with BsrI restriction enzyme. Lanes 1 & 2 showing the wild type pattern, Lanes 3, 4, 6, 7, 9-13 showing the heterozygous pattern, and Lane 8 showing the homozygous mutant pattern of the patient.