



**Supplementary Figure S3.** The heterozygous *EDNRB* $\Delta$ 3 isoform mutation in the patient. (A) Family members of the proband are shown. The proband is shown in the black square (arrow). (B) The heterozygous c.140G>A mutation is found in the patient and the mother. (C) The structure of *EDNRB* isoforms and the location of the p.Gly47Asp (c.140G>A) mutation are shown.