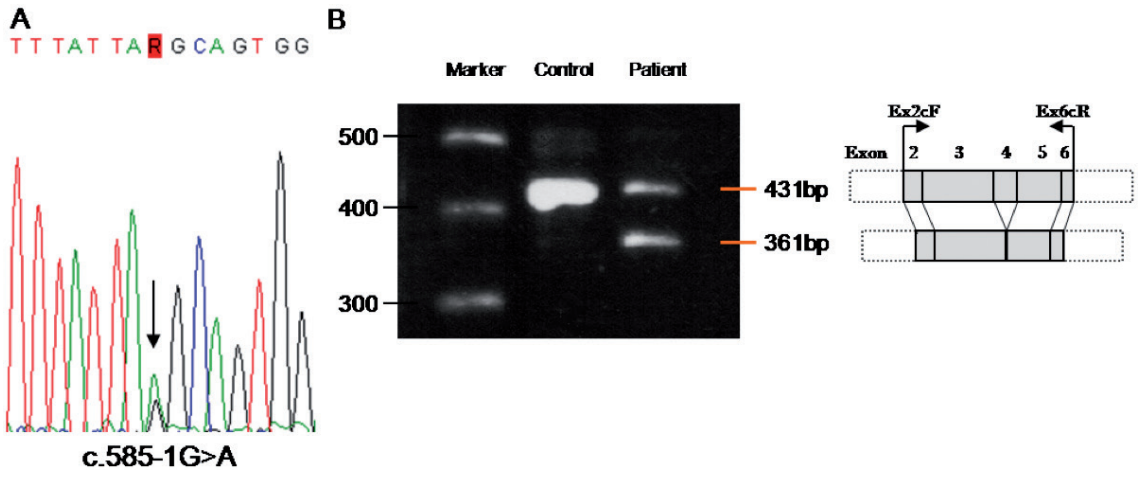

Fig. S2. Results of analysis for the *PTCH1* mutation identified in the patient. DNA sequencing analysis showing the splice site mutation c.585-1G>A (denoted with arrow) in heterozygosis (A). RT-PCR analysis (B). A cDNA fragment (431 bp) was amplified with the forward primer Ex2cF:5'-TAAAAGCAGCGAACCTCGAG-3' and the reverse Ex6cR:5'-AAGTTTGTCCACCGCAAAGGA-3' encompassing part of exon 2 (49 bp), exons 3 (190 bp), 4 (70 bp), 5 (92 bp) and part of exon 6 (30 bp). An abnormal band of smaller size (361 bp) was identified revealing that the mutated transcript was resulted from the skipping of exon 4.