Supplemental Table 1. *In silico* pathogenicity scores for *TRPV4* splice variants affected by our patient's mutation. Ensembl transcript (ENST) and protein (ENSP) identification numbers are shown. Scores for SIFT,[12] PolyPhen2,[13] and CADD[14] scores were calculated using the Ensembl Variant Effect Predictor website (https://useast.ensembl.org/Tools/VEP). SIFT and PolyPhen2 scores are presented with interpretations. CADD PHRED scores represent raw CADD scores normalized to all potential single nucleotide variations genome-wide.[15] A score greater than 20 indicates that a variant is in the top 1% genome-wide in terms of estimated pathogenicity.

Transcript	Protein	Protein position	SIFT	PolyPhen2	CADD PHRED
ENST00000261740.7	ENSP00000261740	435	Deleterious (0.03)	Benign (0.141)	41
ENST00000418703.6	ENSP00000406191	435	Deleterious (0.03)	Benign (0.141)	41
ENST00000538125.5	ENSP00000437449	435	Deleterious (0.03)	Benign (0.141)	41
ENST00000541794.5	ENSP00000442167	388	Deleterious (0.03)	Possibly damaging (0.664)	41
ENST00000536838.1	ENSP00000444336	401	Deleterious (0.02)	Possibly damaging (0.767)	41