**Supplemental Table**

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,5 PolyPhen2,6 and CADD7 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.8 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 |