



**Additional Figure A1.** Trio quantitative PCR analyses confirmed the *de novo* deletion of the (a) *UBA2* and (b) *SCN1B* genes in the proband. The patient's sample showed a delayed amplification curve relative to his parents and control samples, indicating a single copy of each gene. The amplification curve of each sample is according to the colour chart at the left, and target genes are indicated with arrows. The ribonuclease P-RNA component H1 gene (*RPPHI*) was used as a diploid reference assay, and two different sets of pooled samples were included for comparison (sets A and B, 10 healthy controls each). Assays were performed in quadruplicate.