**Fig. S2.** Mutation analysis in Family 1. Multiplex Ligation-Dependent Probe Amplification (a) identifies the mutation del23-29 in the proband. The patient sample represented by the blue peaks and control sample represented by red peaks demonstrate the deletion of exons 28, 23, 24, 27, 25 and 26 (arrows), indicating heterozygosity for the del23-29 mutation. The results of the multiplex ligation-dependent probe amplification tests were confirmed in members of the Family 1 by PCR analysis of the mutation del23-29; ctrl; control sample heterozygous for the del23-29 mutation. (b). In addition, nucleotide sequencing revealed the presence of a missense mutation R1339C in individual II-2 (c).