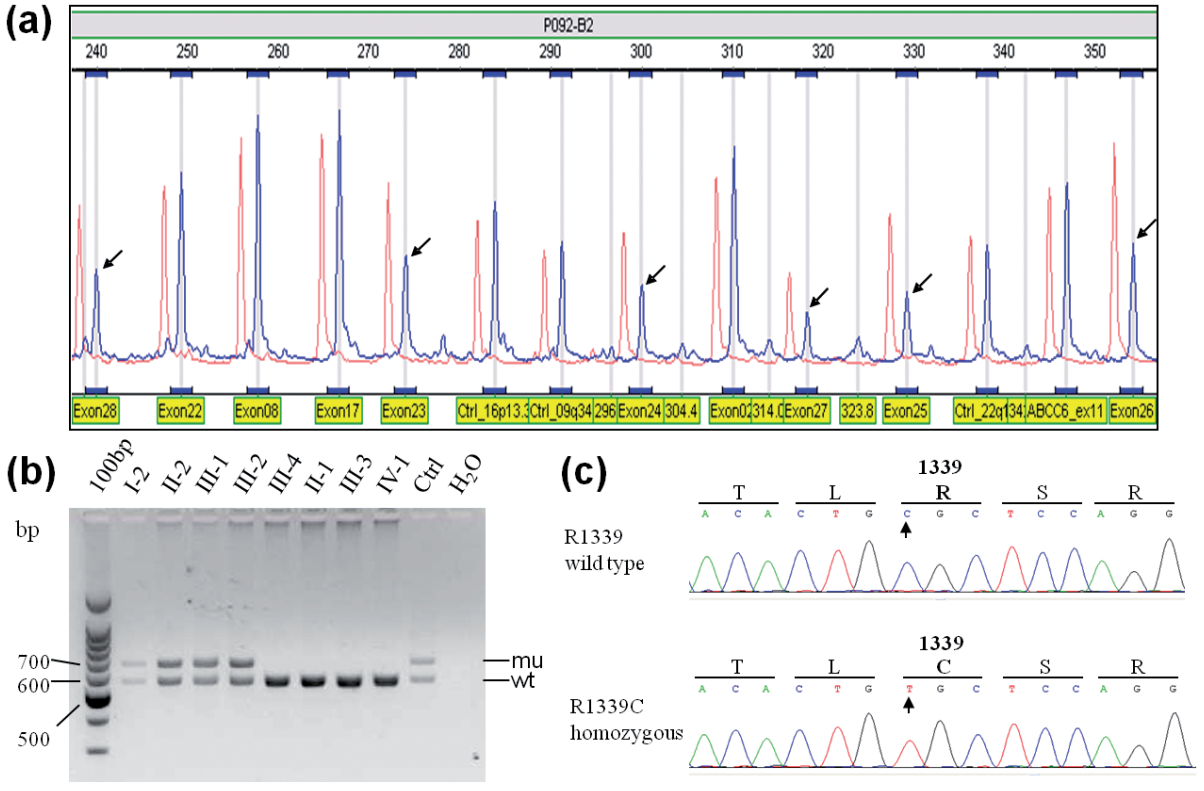

Fig. S2. Mutation analysis in Family 1. Multiplex Ligation-Dependent Probe Amplification (a) identifies the mutation del23-29 in the proband. The patient sample represented by the blue peaks and control sample represented by red peaks demonstrate the deletion of exons 28, 23, 24, 27, 25 and 26 (*arrows*), 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 heterogous for the del23-29 mutation. (b). In addition, nucleotide sequencing revealed the presence of a missense mutation R1339C in individual II-2 (c).