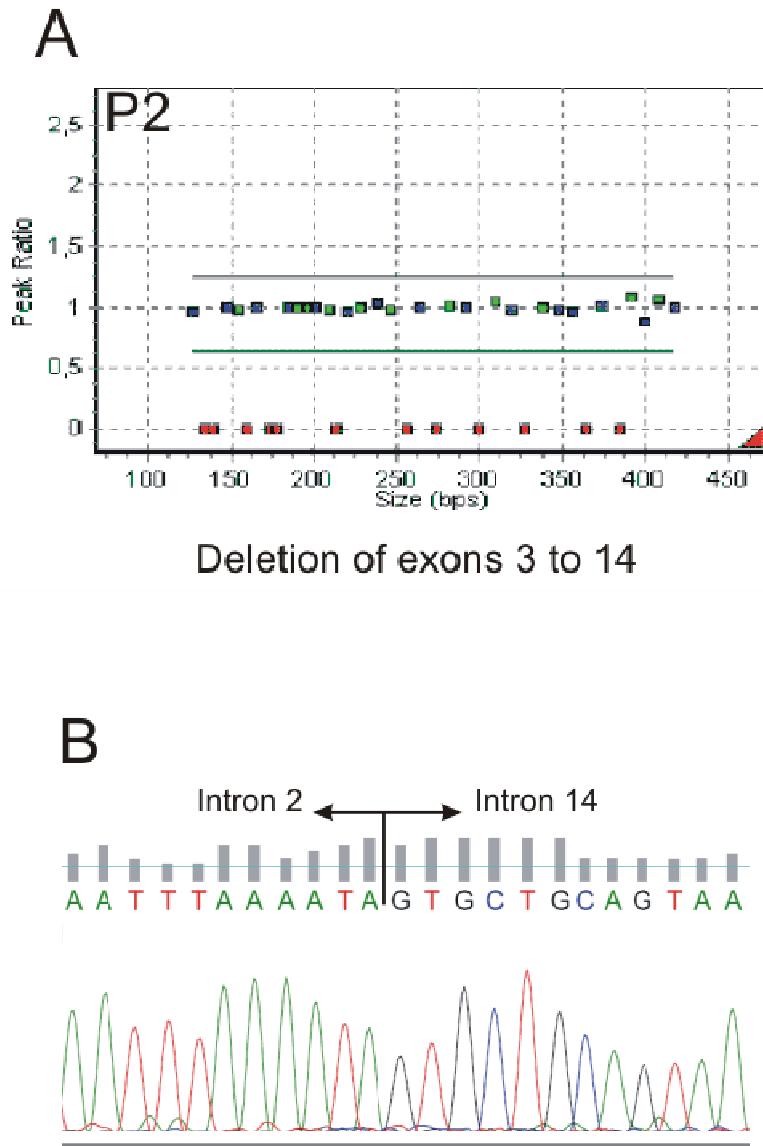


Supplementary data

Figure S5 - Multi-exonic *MTM1* deletion identified in patient 2



Legend: (A) Deletion of exons 3 to 14 of *MTM1* in patient 2 (P2) confirmed by MLPA technique. Red squares correspond to the deleted regions (peak ratios are equal to 0). (B) Electropherogram of the junction fragment (introns 2 and 14), initially obtained by long-range PCR.