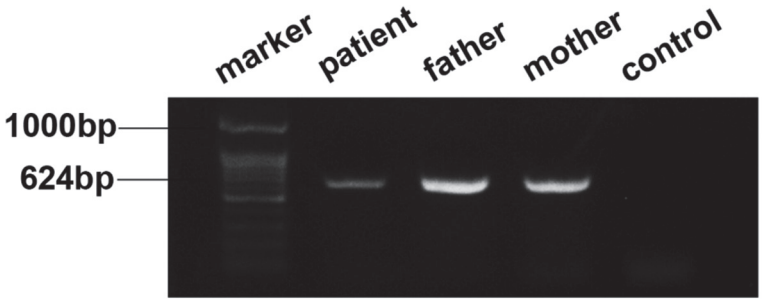

Fig. S1. Electrophoresis of the amplicons. One percent agarose gel electrophoresis of the amplicons of PCR using the mutation-specific primers (designed to amplify the sequence spanning 2 deletion breakpoints) revealed a 628-bp band in the patient and his parents, but not in a normal control, suggesting the mutation was only present in the patient and his parents. To detect the breakpoints of the deletion, the following steps were taken: (*i*) Multiple primers were designed to amplify sequences downstream of exon 10 to determine the 3'-breakpoint. (*ii*) Several mutation-specific primers were designed to amplify regions spanning the suspected deletion site (forward primers at intron 5 and reverse primers at non-deleted 3'-region successfully amplified by (*i*)). Multiple PCRs were performed using the mutation-specific primers from (*ii*), and the only primer pair that successfully amplified the ECM1 deletion region in the patient is: Forward 5'-TCATCCATTCCATGCCGAGAG-3' at *ECM1* intron 5; Reverse 5'-GCTCTAAAGCATCTCCGGGAA-3' at ncRNA-a1 exon 1, product 628 bp).