



Fig. S2. The deletion containing part of *ECM1* and ncRNA-a1 in the patient. (a) Direct sequencing of the 628-bp amplicon showed the deletion breakpoint between the intron 5 of *ECM1* gene and ncRNA-a1, indicated by the red line. (b) Schematic illustration of the genomic DNA structure of *ECM1* gene and ncRNA-a1 and of *ECM1* protein, along with coding regions affected by the 5141-bp deletion. Deleted coding regions (*ECM1* exons 6–10 and the first half of ncRNA-a1 exon 1) are marked in grey boxes; and untranslated regions in black boxes.