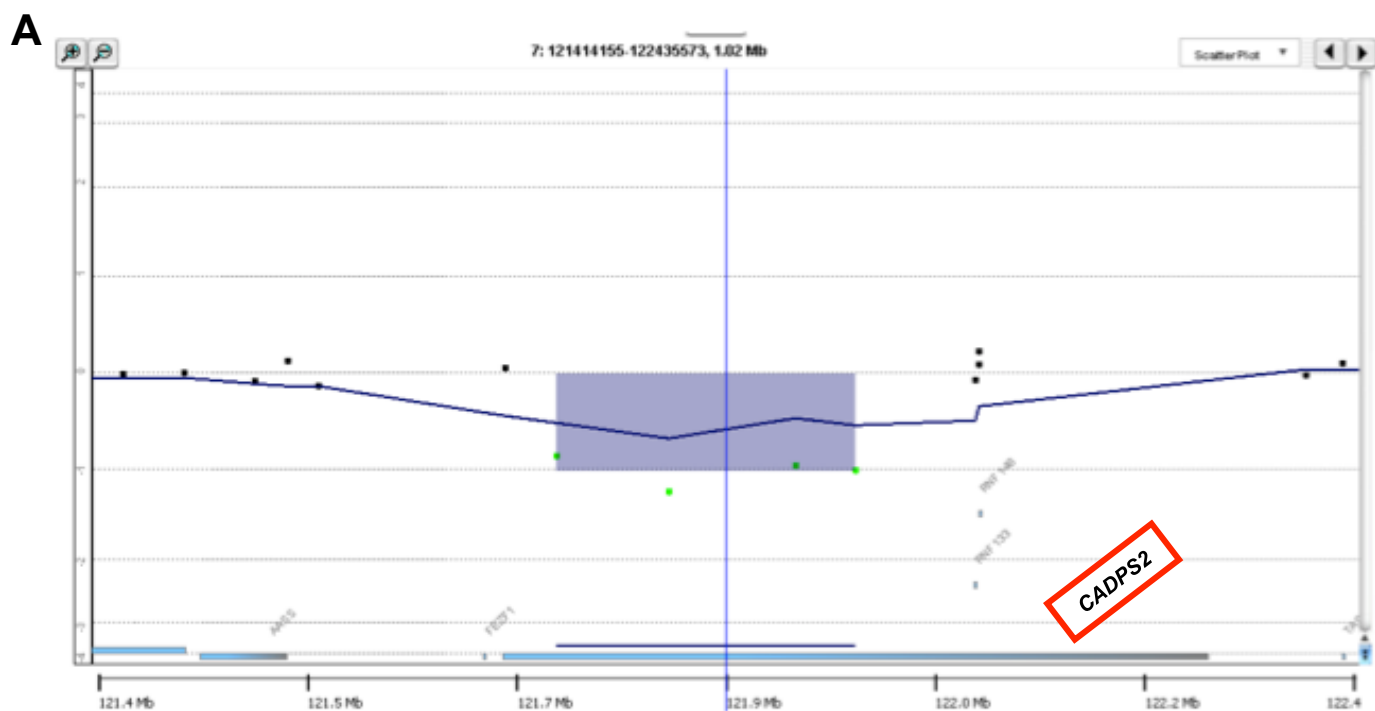
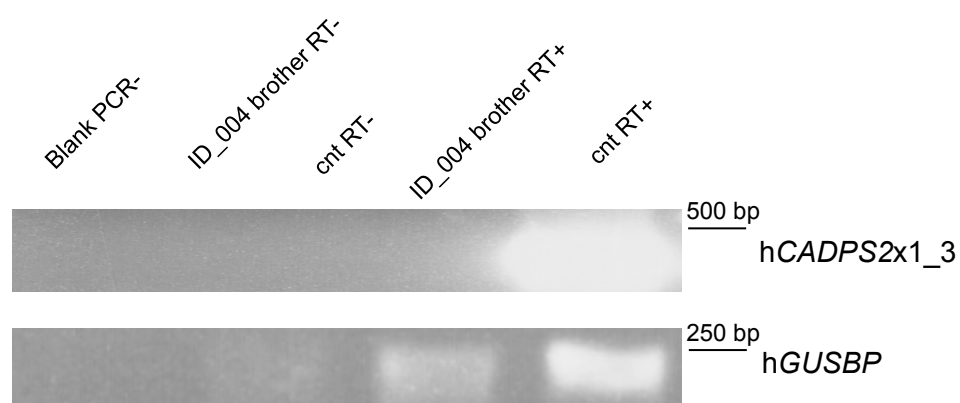




B



C

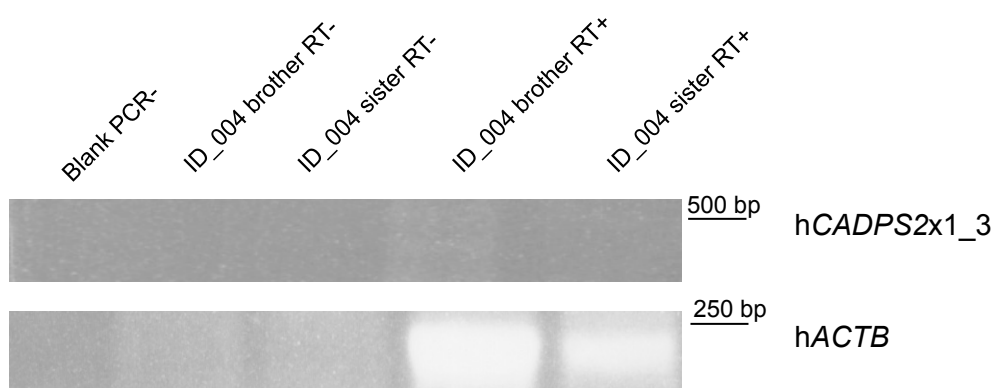


Figure S1: (A) Array-CGH profile centered on chromosome 7q31.32 deletion. This view is derived from Genomic Workbench software (Agilent Technologies), which visualizes normal and deleted oligonucleotides as black and green filled circles, respectively, and genes as light blue bars. *CADPS2* gene is highlighted by a red box. Numbers indicate genomic positions (hg18); (B,C) RT_PCR for *CADPS2* expression in ID_004 patients carrying the deletion vs a control: (B) upper lane, *CADPS2* expression (primer mapping in exon 1 and exon 3) from patient ID_004.01 and from a control cDNA, lower lane, *GUSBP* expression (control gene); (C) upper lane, *CADPS2* expression in the two ID_004 sibs, lower lane, *ACTB* expression (control gene). Rt+= reverse transcription in presence of the RT enzyme, RT-= reverse transcription in absence of the RT enzyme.