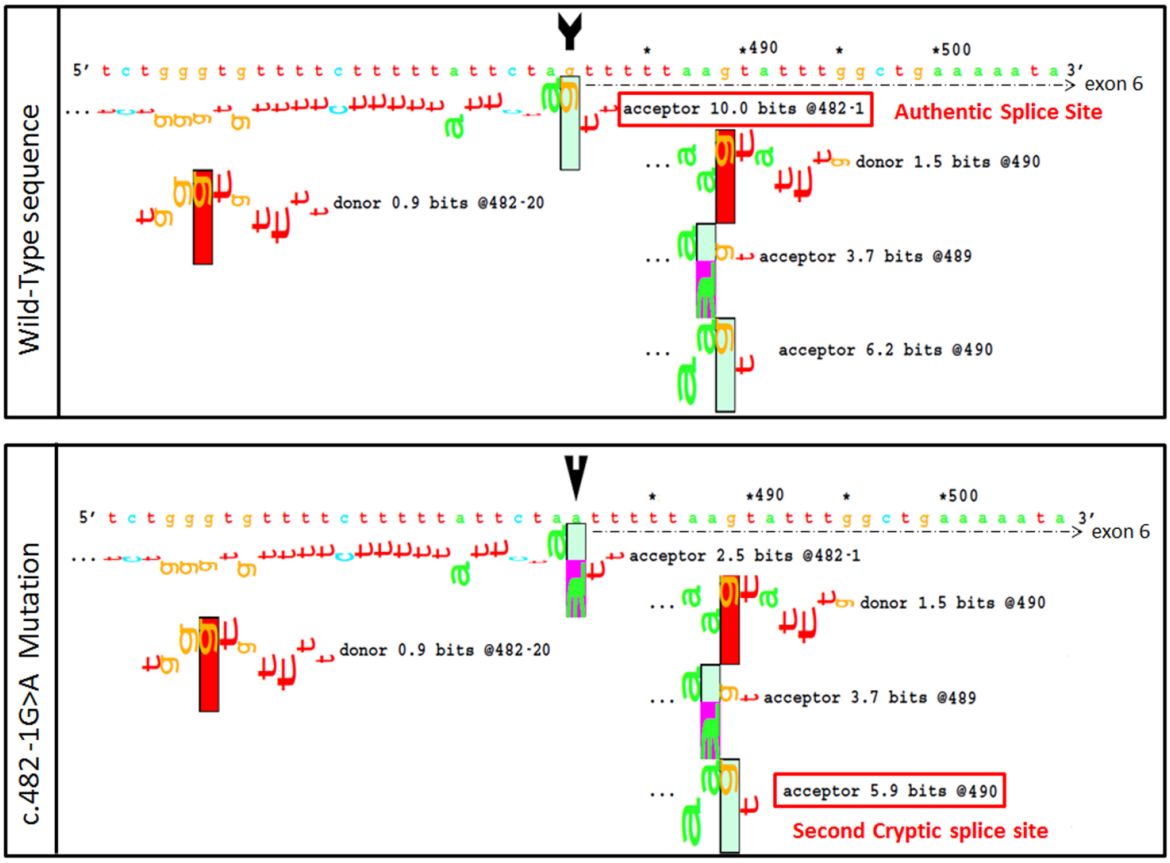
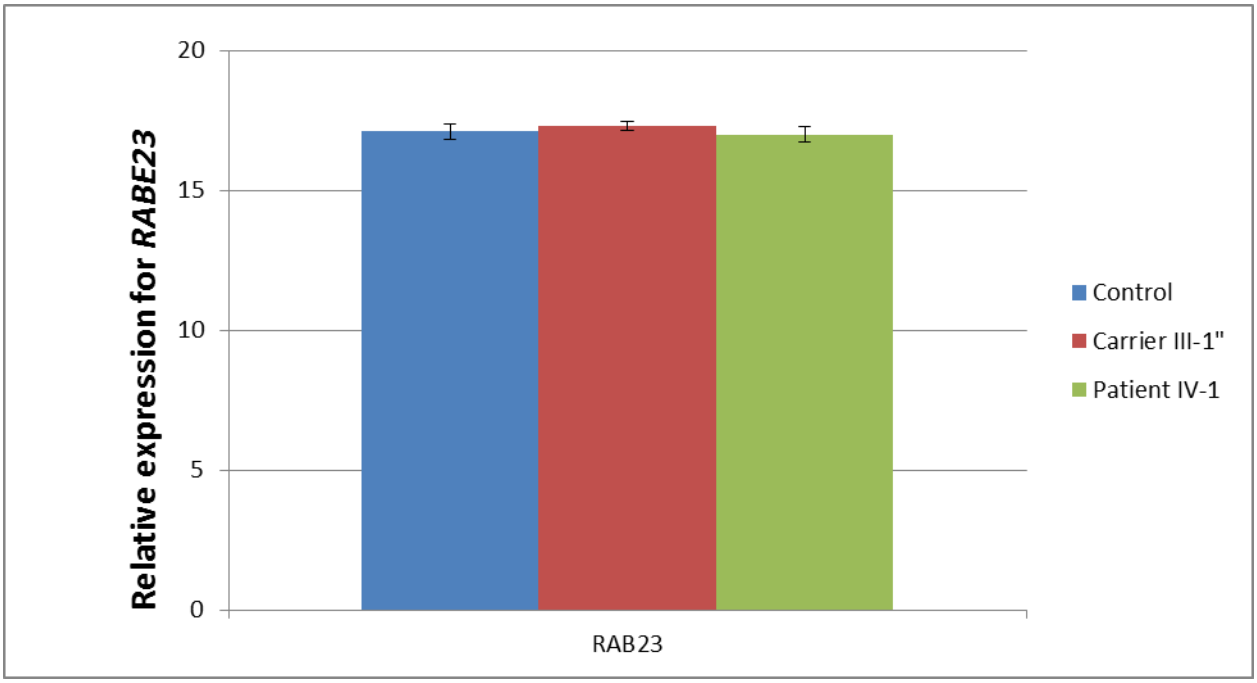


Suppl. Figure 1. Computational analyses of exon 5 of *RAB23* gene in the presence of c.482-1G>A. Walker diagram of the c.482-1G>A substitution in exon 5 of *RAB23* gene. Splice sites are shown by walkers, in which the height of each letter is the contributions of that base to the total conservation of the site in bits [16 Schneider, 1997a]. Upper and lower bound of the vertical rectangles are at +2 bits and -3 bits respectively, and letters that are upside down denote negative contributions. Horizontal rectangles represent the conserved and cryptic splice sites and their relative position in the sequence. This mutation abolishes the canonical 5' splice site with Ri decreased from 10 to 2.5 bits activating a second cryptic splice site at position c.490 with a highest Ri of 5.9 compare to other potential acceptor splice sites. This mutation is predicted to result in eight nucleotides deletion causing a frameshift and leading to premature termination codon 3 amino acids downstream within exon 5.



Supp. Figure 2. Quantitative expression of *RAB23* transcripts. Relative expression of *RAB23* mRNA were measured using qRT-PCR in patient (IV-2), carrier (III-1) and control samples normalized according to an internal control *GAPDH*. X axis depict the $2e^{-\Delta\Delta CT}$ values ; Y axis represent control, carrier and patient samples respectively.