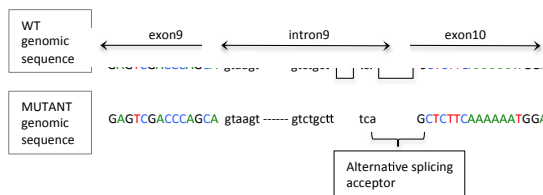


## Supplementary Figure 1. *RINT1* c.1334-5delA, c.1334-1\_1335delGTT minigene assay

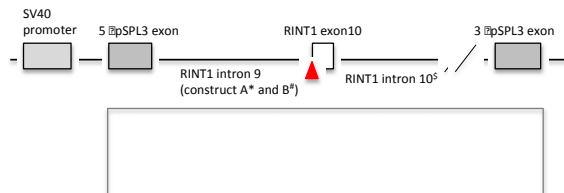
A) The genomic DNA region of interest that contains a putative splicing mutation found in the patient with breast cancer. Exonic and intronic sequences are in capital and small letters, respectively. Black squares denote the splice-acceptor site deletions. B) Schematic representation of minigene assay construct. The dark and light gray boxes represent SV40 promoter and 5' and 3' exons in pSPL3 vector, respectively. The white box represents *RINT1* exon10. Double parallel diagonal bars indicate the border of *RINT1* intron10 and pSPL3 intron. C) Agarose gel electrophoretic profile of RT-PCR products. Migration patterns of RT-PCR products from HEK2993 cells and COS-7 cells with or without cycloheximide treatment are shown in the picture. The expected wild-type product size is 399bp, and there is no visible difference in size between wild-type and mutant RT-PCR products. D) Chromatograms derived using construct A. Left chromatograms show the intron-exon junction in wild-type and mutant construct A. Right chromatograms show the splicing outcomes of wild-type and mutant. The dashed lines indicate the sites of deletions. The same profiles were observed using construct B (data not shown).

### RINT1 c.[1334-5delA;1334-1\_1335delGTT] Minigene Assay

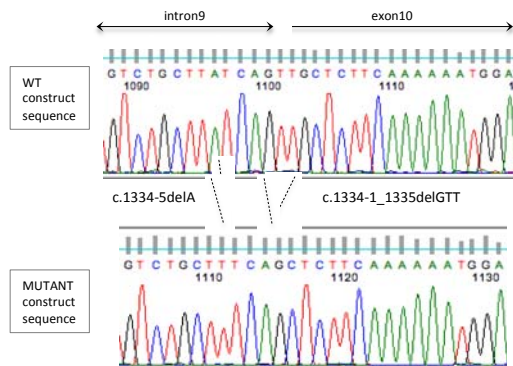
#### A) WT and MUTANT genomic DNA sequence



#### B) Minigene Assay constructs



#### D) Chromatograms derived using construct A (similar profiles were observed using construct B)



#### C) RT-PCR result

