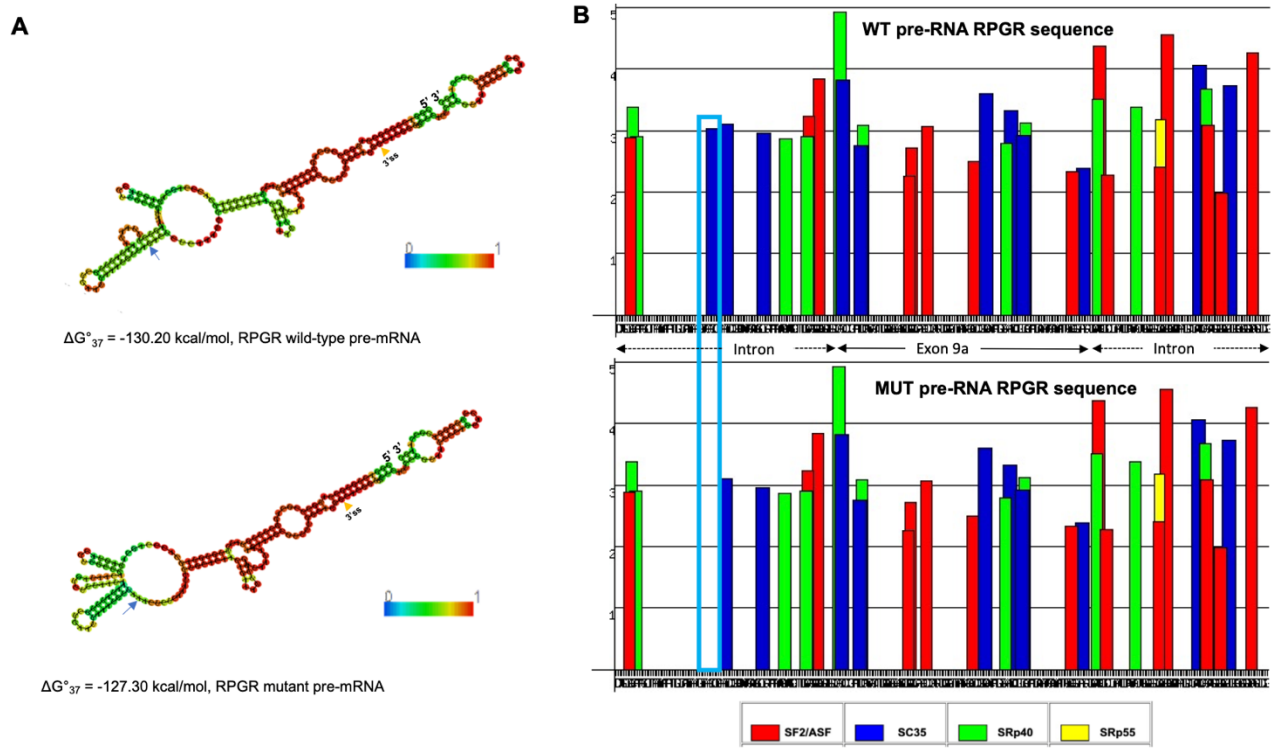

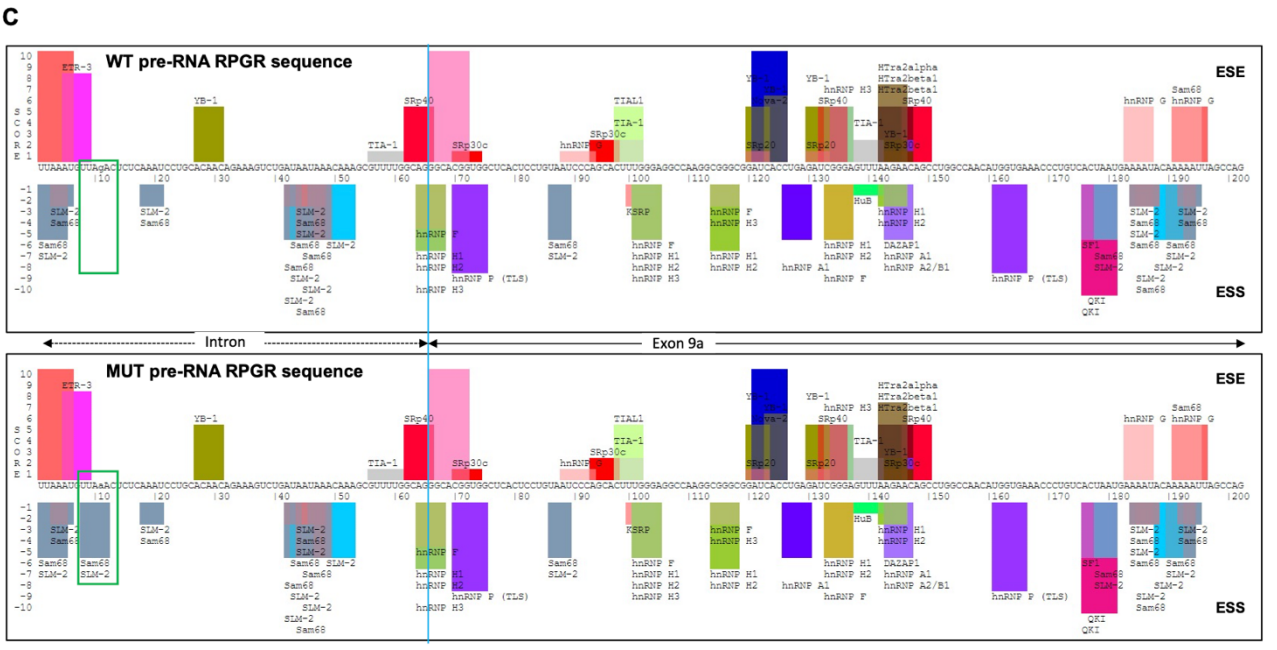

Supplemental Figure S1. Computational analysis of splicing sequences in *RPGR* exon 9a. (a) Folding of a portion of the wild-type and mutant *RPGR* pre-mRNA sequence (from position 31551 to position 31751 of NCBI Refseq: NG_009553.1, covering the first 45 nucleotides of exon 9a and the preceding 155 nucleotides in intron 9) and $\Delta G°_{37}$ free energy was predicted by using RNAfold algorithms. Blue arrows indicate the nucleotide position G and A in wild-type or mutant pre-mRNA sequences of *RPGR,* respectively. The 3'splice site (3'ss) of exon 9a is indicated by a yellow arrow. Colour code bar (0 to 1) indicates base-pairing probability. **(b)** Exonic Splice Enhancers (ESEs) in *RPGR* pre-mRNA sequence (from position 31605 to position 31935 of NCBI Refseq: NG_009553.1, covering the whole exon 9a, the preceding 102 nucleotides in intron 9, and the 92 nucleotides downstream of exon 9a) were predicted using ESEfinder. The program searches the binding sites of four different RNA-binding proteins, all with a splicing enhancer function when binding to exons: in red, protein SF2/ASF, in blue, protein SC35, in green, protein SRp40, in yellow, protein SRp55. The light blue rectangular box indicates that mutation c.1059+363G>A abolishes a binding site for SC35 protein in the

mutant *RPGR* pre-mRNA, 54 to 46 nucleotides upstream the 3'ss. **(c)** Exonic Splice Enhancers (ESEs) and Exonic Splice Silencers (ESSs) in *RPGR* pre-mRNA sequence (from position 31642 to position 31843 of NCBI Refseq: NG_009553.1, covering the whole exon 9a and the preceding 65 nucleotides in intron 9) were predicted using SpliceAid. The program searches for the binding sites of 71 different splicing factors. **Top panel:** wild-type RPGR pre-mRNA sequence. **Bottom panel:** *RPGR* pre-mRNA sequence with mutation c.1059+363G>A. The mutated nucleotide is indicated in lowercase in the sequence in both panels. In the upper part of each panel RNA-binding proteins predicted to bind the pre-mRNA and having a splicing enhancing function are depicted (ESE). In the lower part of each panel RNA-binding proteins predicted to bind the pre-mRNA and having a splicing silencer function are depicted (ESS). All ESEs predicted in exon 9a are overlapping with predicted ESS thus a clear ESE is not defined. The green rectangular boxes indicate that mutation c.1059+363G>A introduces a new binding site for Sam68 and Sam68-like SLM-2 proteins in the mutant *RPGR* pre-mRNA, 58 to 52 nucleotides upstream the 3'ss.