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Supplemental Data

Systematic Computational Identification of Variants That Activate Exonic and Intronic Cryptic Splice Sites

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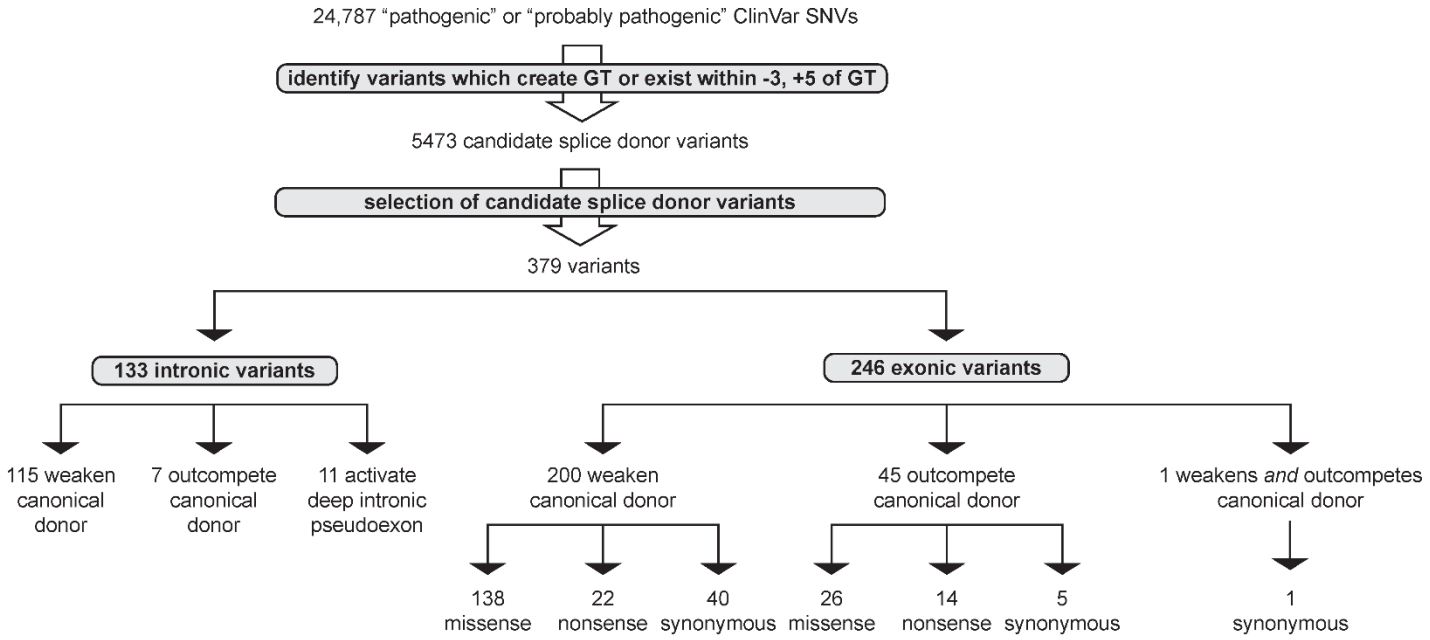
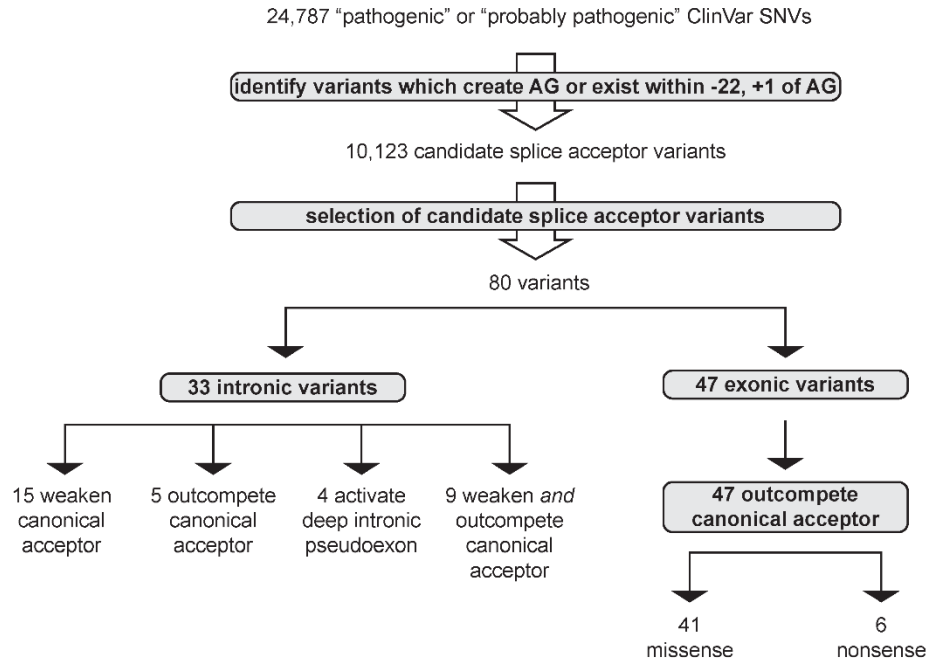
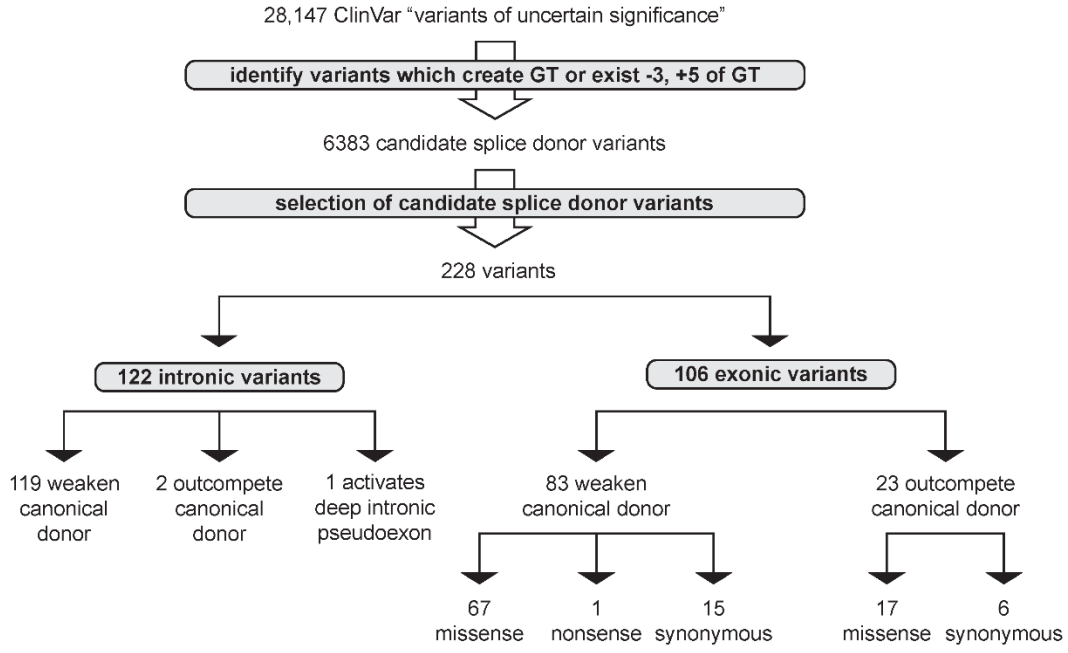
Figure S1. Flowcharts of selected high confidence splice variants from ClinVar "pathogenic" or "probably pathogenic" variants.

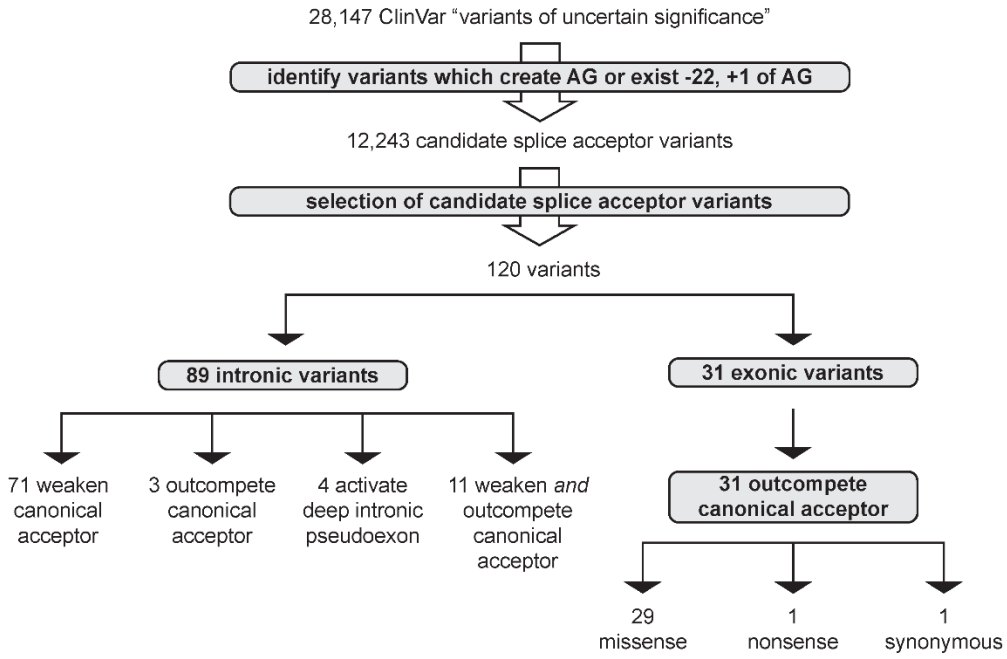




Predicted high confidence splice variants broken down by impact on splicing and predicted impact on protein. Flowcharts are split by predicted impact on splice donors or splice acceptors.

Figure S2. Flowcharts of selected high confidence splice variants from ClinVar "variants of uncertain clinical significance."





Predicted high confidence splice variants broken down by impact on splicing and predicted impact on protein. Flowcharts are split by predicted impact on splice donors or splice acceptors.

Table S1. Table of selected high confidence splice variants from CFTR2 database. Predicted high confidence splice variants from CFTR2 with predicted splicing consequence. Some variants are predicted to alter in splicing in multiple ways (i.e. weaken canonical splice site *and* create a novel splice site) and therefore have multiple predictions.

Table S2. Table of selected high confidence splice variants from ClinVar "pathogenic" or "probably pathogenic" variants. Predicted high confidence splice variants with hg19 coordinates, RefSeq transcript accession number, ClinVar cDNA and protein names, and predicted splicing consequence. Some variants are predicted to alter in splicing in multiple ways (i.e. weaken canonical splice site *and* create a novel splice site) and therefore have multiple predictions.

Table S3. Table of selected high confidence splice variants from ClinVar "variants of uncertain clinical significance." Predicted high confidence splice variants with hg19 coordinates, RefSeq transcript accession number, ClinVar cDNA and protein names, and predicted splicing consequence. Some variants are predicted to alter in splicing in multiple ways (i.e. weaken canonical splice site *and* create a novel splice site) and therefore have multiple predictions.