

Figure S5.

SJST030310

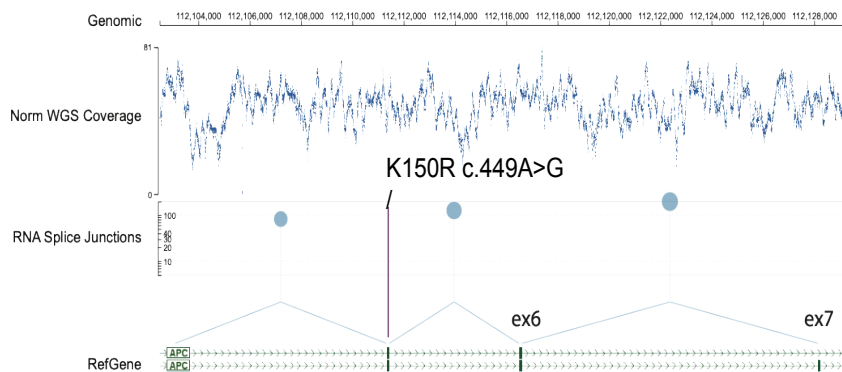


Figure S5. Molecular phenotypes refine functional classification of variants. Examination of splicing for APC variant identified in the germline of patient SJST030310. GenomePaint view of the APC locus at chr5:112104000-112128000[hg19] in SJST030310. The upper scatterplot shows whole genome sequencing coverage with each point representing a single 100 base-pair window. Blue links show splice junctions relative to the APC gene model in green. A purple bar indicates the position of a predicted de novo splice site in APC exon 5 (NM_000038). However, we saw no abnormal splicing patterns in the vicinity of this prediction.