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.