



Supplemental Figure 1. Recommended step-by-step pipeline for variant-calling and variant-splicing linkage detection. Reads from raw fastq files were processed to trim adapters and low quality 3' ends (names of the tools shown in blue). These reads were then aligned by STAR using the 2-pass alignment option. This option allows for accurate mapping of splice junctions which is important for accurate identification of variant-splicing linkage. After alignment, PCR duplicates were marked using Picard Tools. Duplicated reads amplify the frequency of random errors which can lead to false positive calls. Finally, scAllele calls variants and identifies variant-splicing linkage.



