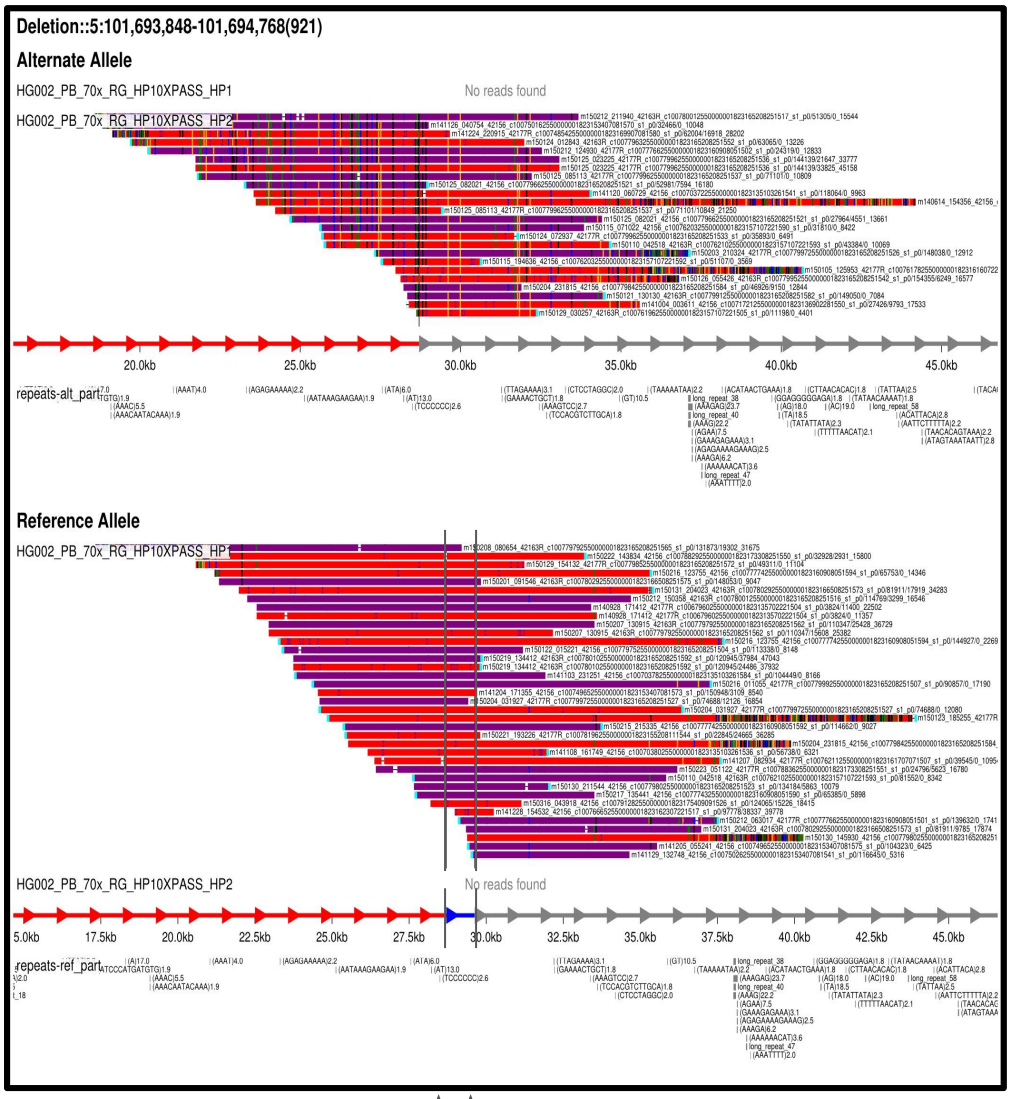


Reads that support an **alternate** allele

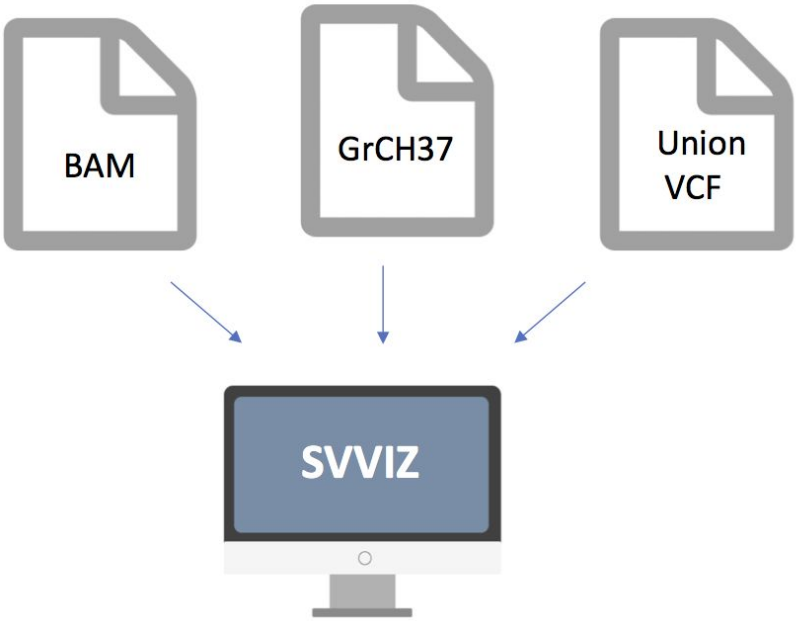
Reads that support a **reference** allele

Annotations to note repeats

structural variant breakpoints

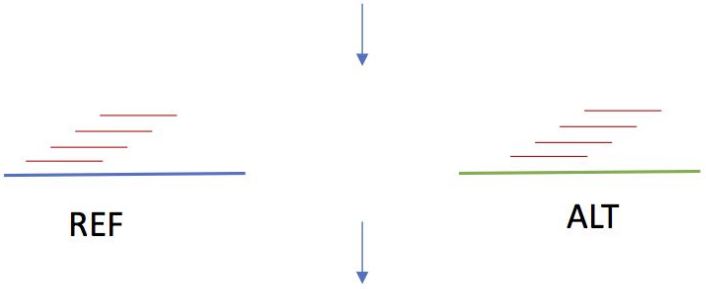
1

SVVIZ takes in:
-<u>read data</u> in BAM format from short read and long read sequencing technologies
- <u>reference genome</u>
- <u>putative SVs</u> listed in a VCF



2

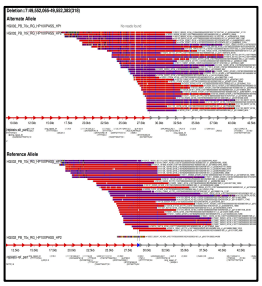
Reads from the BAM file are mapped to **reference allele** (from GrCH37)



Reads from the BAM file are mapped to **alternate allele** (from union VCF). Reads that do not map to the ref or alt allele are placed in an **ambiguous** bin

3

SVVIZ generates read aligned images (right), dotplots, and summary statistics.



Putative SV
Deletions (top)
Insertions (bottom)
DEL/INS:size:uniqeID

PacBio Tracks
-Haplotype separated reads
-HP2 shows evidence of a ~920bp deletion

10X Tracks
-Haplotype separated reads
-HP2 shows evidence of a ~920bp deletion

Illl250bp Track

Additional Annotations
-Segmental duplications
-Repeats