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Input =
demultiplexed
bam file
containing
pacbio long
reads

Use bam2fastq
to split bam file
by barcode tags
into **fastq files**



Use Canu - de novo assembly of reads into contigs, without reference

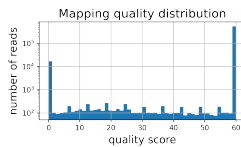
Use mummer to
call SNPs

View mutations in IGV and Ribbon

Use our custom
GUI to
accept/reject
mutations

Analyze
validated mutations
from biological
point of view

Plot mapping quality



Map reads to reference genome using minimap2 (bam file format)

Sort and index mapped bam files

Use sniffles and SVIM to identify structural variants (**vcf files**)

Use SURVIVOR to compare SVs from different SV callers and merge vcf files

Sequence

Filtered

Assembled

Mutations

| | | | | | | |
|----------|----------|-------------|--------|------------|---|----------------------|
| Accepted | Reject | AC_000094.3 | 11391 | v16.p16.1 | C | CCCCCCCCCCCCCCCCCCCC |
| Accepted | Reject | AC_000094.3 | 22089 | v16.p16.2 | A | AAAAAAAAAAAAAAAAAAAA |
| Accept | Rejected | AC_000094.3 | 31094 | v16.p16.10 | E | CCCCCCCCCCCCCCCCCCCC |
| Accept | Reject | AC_000094.3 | 40749 | v16.p16.13 | A | AAAAAAAAAAAAAAAAAAAA |
| Accepted | Reject | AC_000094.3 | 50520 | v16.p16.27 | N | NNNNNNNNNNNNNNNNNN |
| Accept | Rejected | AC_000094.3 | 58732 | v16.p16.32 | N | NNNNNNNNNNNNNNNNNN |
| Accepted | Reject | AC_000094.3 | 68741 | v16.p16.37 | T | TTTTTTTTTTTTTTTTTTTT |
| Accept | Reject | AC_000094.3 | 77143 | v16.18 | C | CCCCCCCCCCCCCCCCCCCC |
| Accept | Reject | AC_000094.3 | 139310 | v16.p16.24 | T | TTTTTTTTTTTTTTTTTTTT |

