Software note.

pauvre is useful to check the sequencing stats (pauvre stats)

https://github.com/conchoecia/pauvre

clair3 is efficient in calling SNPs from Nanopore data  
NanoSNP in inferior to clair3 but superior to other SNP callers for long reads when the coverage is low.

<https://github.com/wdecoster/nanopack>

many useful tools

SMRT Toolsis PacBio’s official tools working with their reads

SequelTools is a program that provides a collection of tools for working with multiple SMRTcells of BAM format PacBio Sequel raw sequece data

* Structural variant calling: the long and the short of it
* Comprehensive evaluation and characterization of short read general-purpose structural variant calling software
* NextSV3: automated structrual variation detection from long-read sequencing using state-of-the-art tools.
* Dysgu: efficient structural variant calling using short or long reads
* Jasmine and Iris: population-scale structural variant comparison and analysis

1. Population genetics
   1. genodive version 3.0
   2. ANGSD 0.94 stable
2. Demography, Site frequency spectrum
   1. <https://dadi.readthedocs.io/en/latest/>
   2. <http://cmpg.unibe.ch/software/fastsimcoal26/>
   3. https://speciationgenomics.github.io/easysfs/
3. Long read assemble.
   1. BlasR [76], MUMmmer [77], or Minimap2
   2. specialized methods to align long reads such as BLASR [76], Minimap2 [35], and NGMLR
4. cuteSV, LRcaller, Sniffles, SVJedi, and VaPoR
5. SV caller or workflow, Short-read alignment approach
   1. Manta (v1.1.0), DELLY (v0.7.7), LUMPY (v0.2.13) and GRIDSS (v1.3.4)
   2. DELLY [41]: the larger events remain hard
   3. TARDIS
   4. sv-callers: a highly portable parallel workflow for structural variant detection in whole-genome sequence data
6. SV caller or workflow, LOOOOOOONG-read mapping-based approach
   1. SVIM: structural variant identification using mapped long reads (newer)
   2. Sniffles operates on a per read base, also capable of reporting very low-frequency SVs in the sample.
   3. PBHoney: For PacBio, relying on BLASR alignments
   4. PacBio structural variant calling and analysis tools (PBSV) : For PacBio, SVs within the range of 20+ bp
   5. SMRT-SV: For PacBio, includes de novo assembly and a specialized genotyping module
   6. NanoSV: For Oxford Nanopore, preferentially uses as input an alignment from LAST. NanoSV reports only breakpoints (BND) which again makes the interpretation of the SVs type difficult.
   7. Overall, long-read mapping-based methods for SV calling often show a better performance than short-read ones
7. SV caller or workflow, De novo assembly-based approach: heterozygous SVs are often missed
   1. SGVar [32] is a more recent string graph-based (see Table 2 for definition) de novo assembly pipeline based on the SGA assembler [75] that also uses short-read sequencing data
   2. Assemblytics [34] is a web application that relies on MUMmer and identifies insertions and deletions
   3. paftools.js [35] uses Minimap2 alignments
   4. SMARTie-SV was recently introduced to detect insertions, deletions, and inversions, using BlasR
8. SV caller or workflow, Other approach
   1. Hic\_breakfinder (1+ Mbp), can potentially identify all types of SVs
   2. Strand-Seq, Strandseq-InvertR [68] (min ~ 1 kbp)
9. misc
   1. BEDOPS v2.4.41: [https://bedops.readthedocs.io/en/latest/index.html#](https://bedops.readthedocs.io/en/latest/index.html)
   2. GenomeScope: Estimate genome heterozygosity, repeat content, and size from sequencing reads using a kmer-based statistical approach.
   3. GenomeScope 2.0 and Smudgeplot for reference-free profiling of polyploid genomes