

CNVnator with JBrowse

Abyzov Alexej

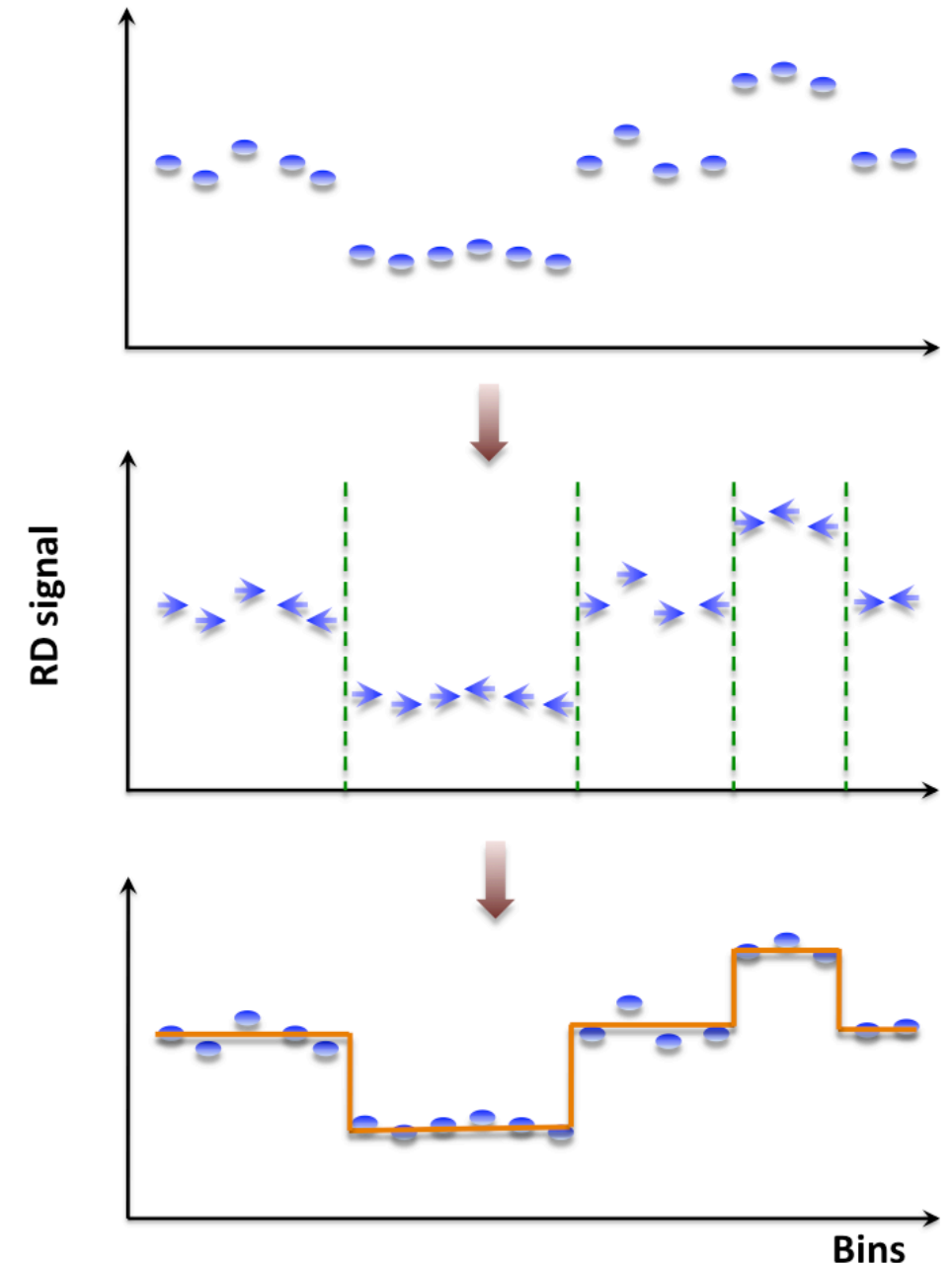
Mayo Clinic

March 6, 2020

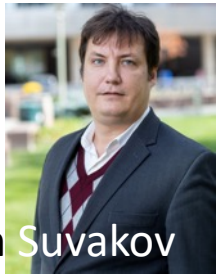
CNVnator and CNVpytor

CNVnator: mean-shift approach

- Derived from image-processing applications
- For each bin, attraction (mean-shift) vector points the direction of bins with most similar RD signal
- Not model-based, with minimum assumptions and parameters
 - Local segmentation
 - No prior assumptions about number, sizes, frequency and density of CNV regions
- Achieves discontinuity-preserving smoothing
- Highest sensitivity to find CNVs detected by aCGH and fosmid end sequencing



CNVpytor: a Python extension of CNVnator



Milovan Suvakov



- Easy to install

```
> pip install cnvpytor  
> cnvpytor -download
```

- Can be used as python module with all functionalities

```
import cnvpytor
```



- Can be used in Jupyter notebook
- Installable in Colab Google

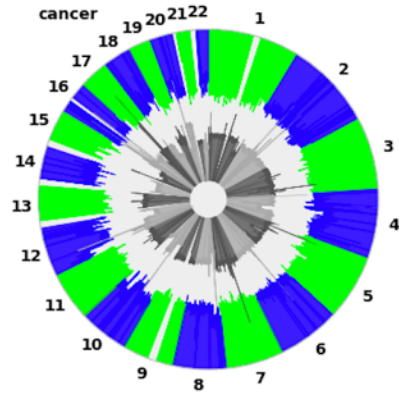
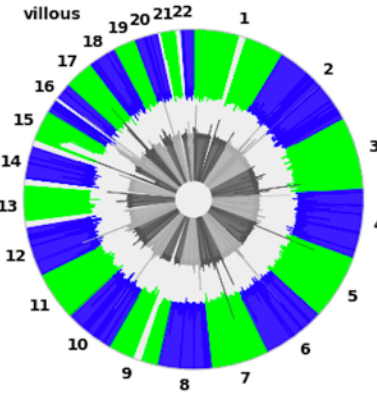
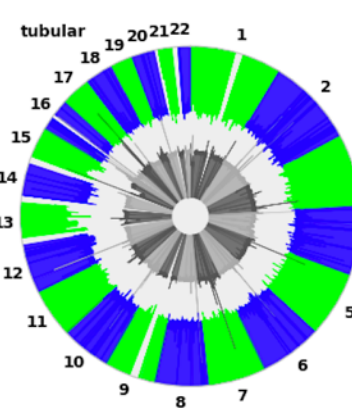
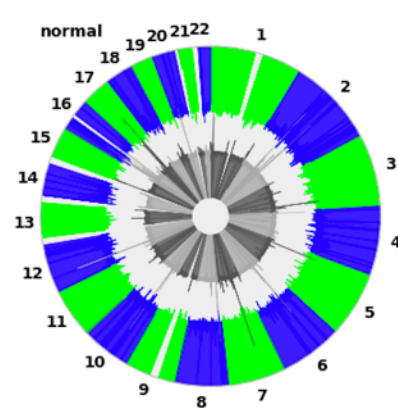


- GitHub:
<https://github.com/abyzovlab/CNVpytor>

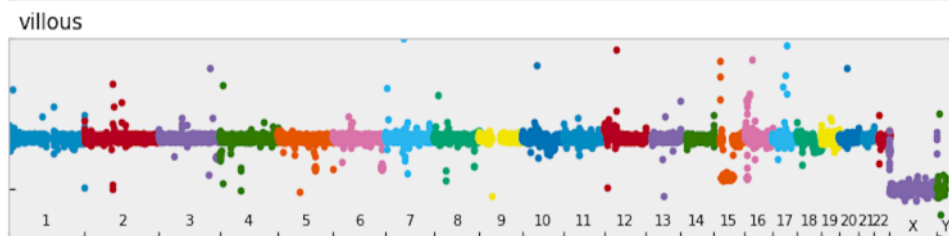
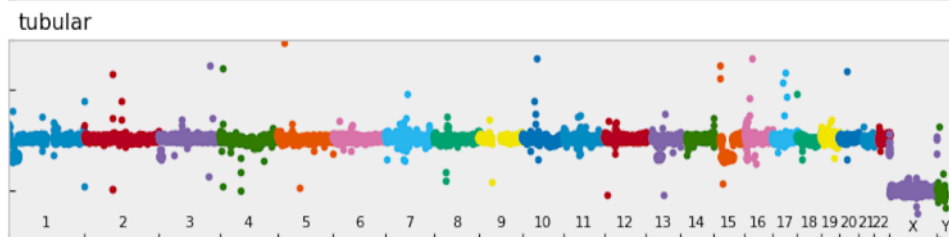
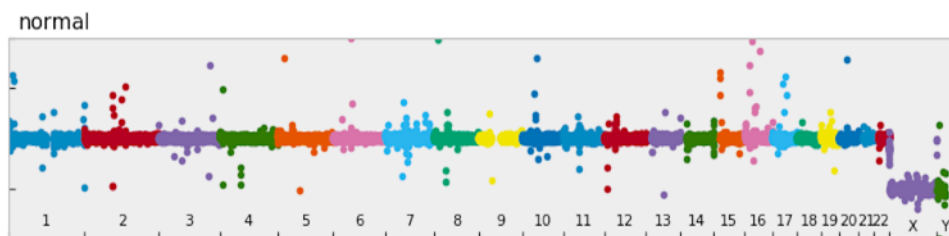
- HDF5 format to store data:
 - RD & BAF information *.pytor* file takes less than 50Mb
- Interactive plotting command line interface with tab completion
- Different plot styles and image formats

Plotting

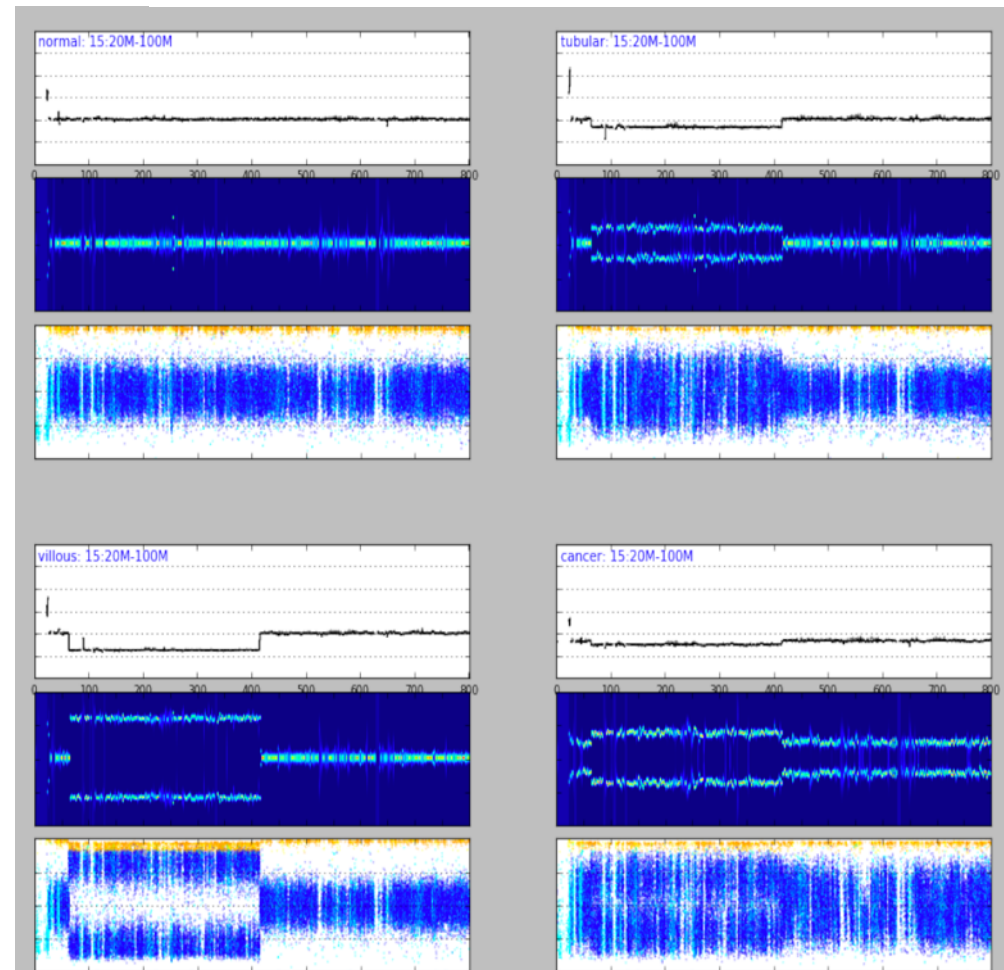
Circular plots



Manhattan plot



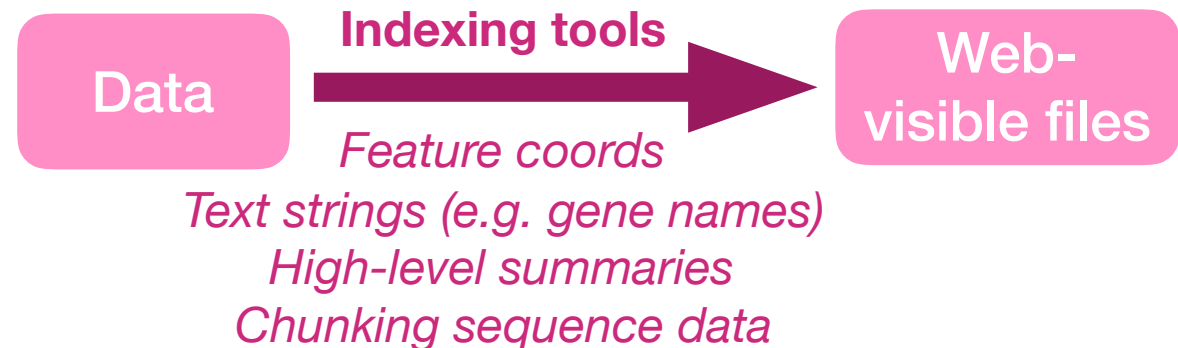
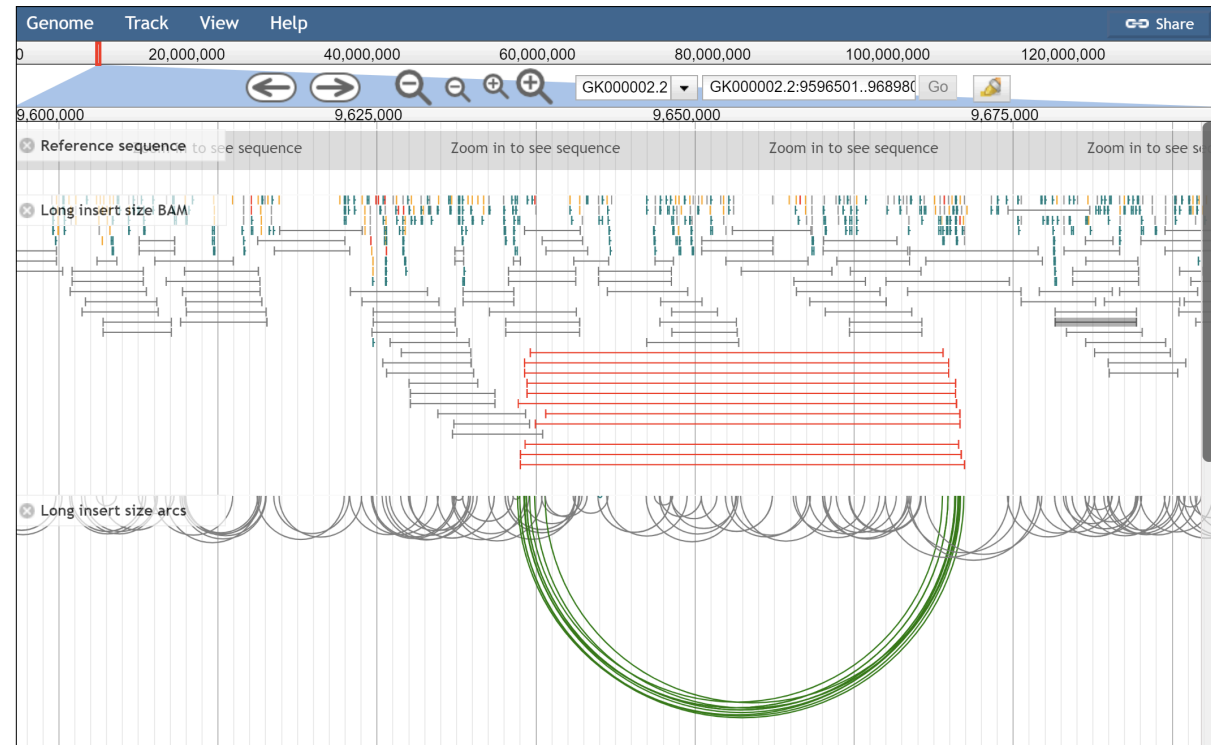
Multiple signals in genomic regions



JBrowse

JBrowse is...

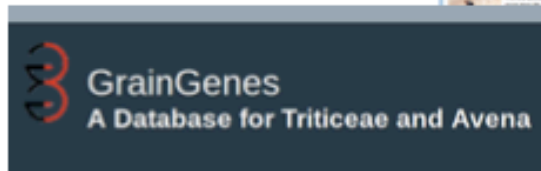
- A general-purpose genome browser
- Written entirely in JavaScript
- Web-publishable as a static site (low cost, high security)
- Fluent in most common bioinformatics formats



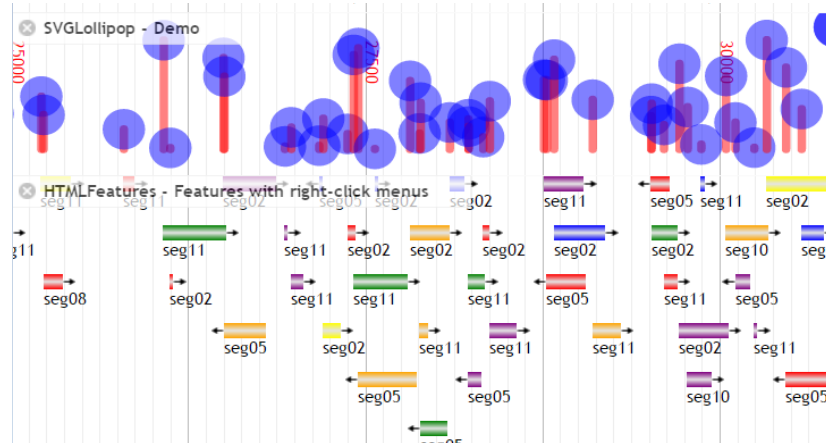
Apps & sites using JBrowse

- Sequencing analysis pipelines: GenSas, DNASubway, InterMine, SeqWare...
- Collaborative online genome annotation: WebApollo, Afra
- Model orgs (AGR), non-models (i5k, Phytozome), cancer (Cosmic), many “small” genome projects

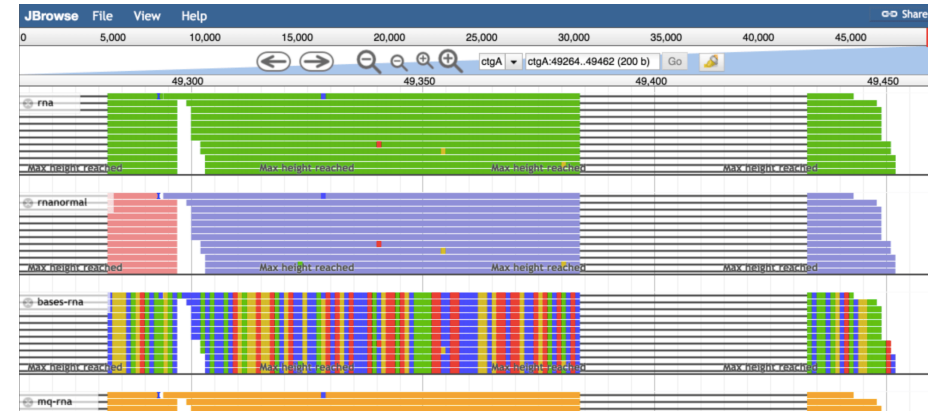
ALLIANCE
of GENOME RESOURCES



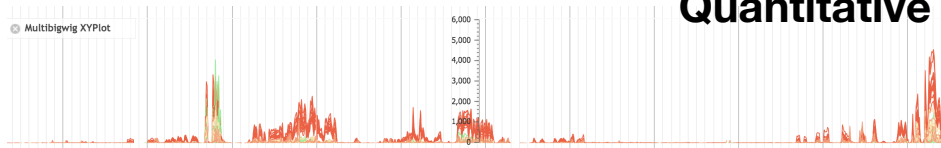
JBrowse plugin registry



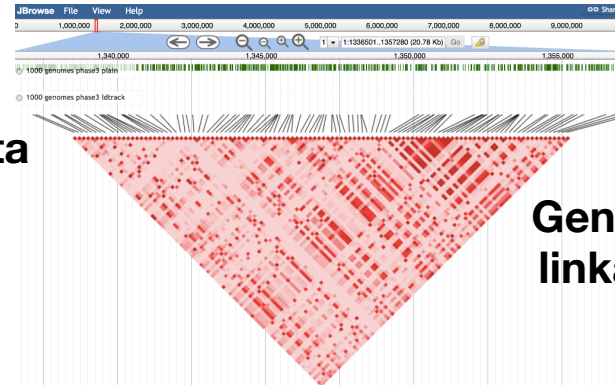
Population genomics



Variants



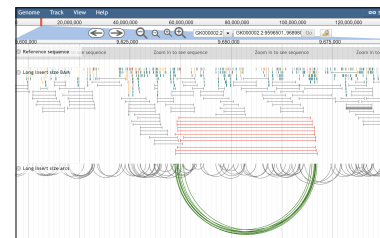
Quantitative data



Genetic linkage

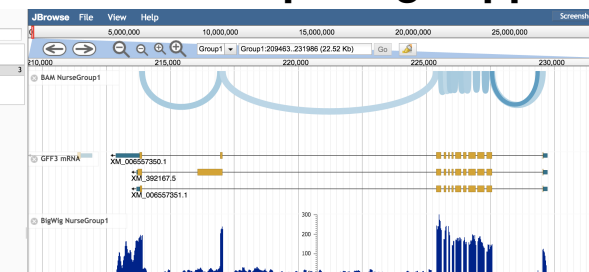
Dynamix: AI that prioritizes deep track lists
Apollo: Google Docs for genome curators
Plugins for Galaxy, CoGe...

**~50 plugins as of
January 2020**



Paired-end visualization

Splicing support



Integrating CNVpytor and JBrowse



Arijit Panda

How to integrate

- Install CNVpytor with pyBigWig dependency
- Install JBrowse with MultiBigwig and MultiScaleBigwig plugins

- Export file generation

```
CNVpytor -root [root files] -export jbrowse [Directory Name(optional)]
```

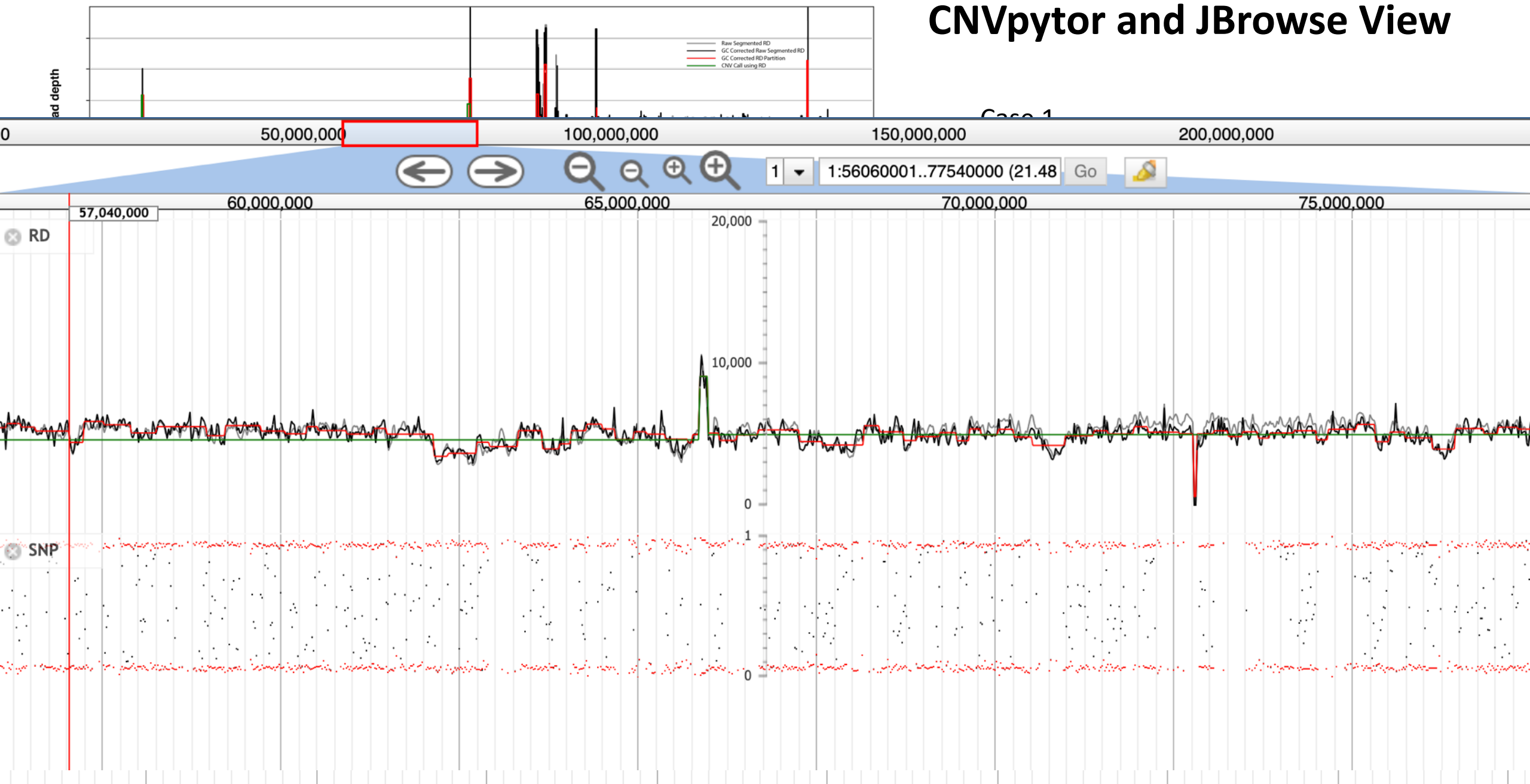
- Visualization

```
http://localhost/jbrowse/?data=\[Directory Name\]
```

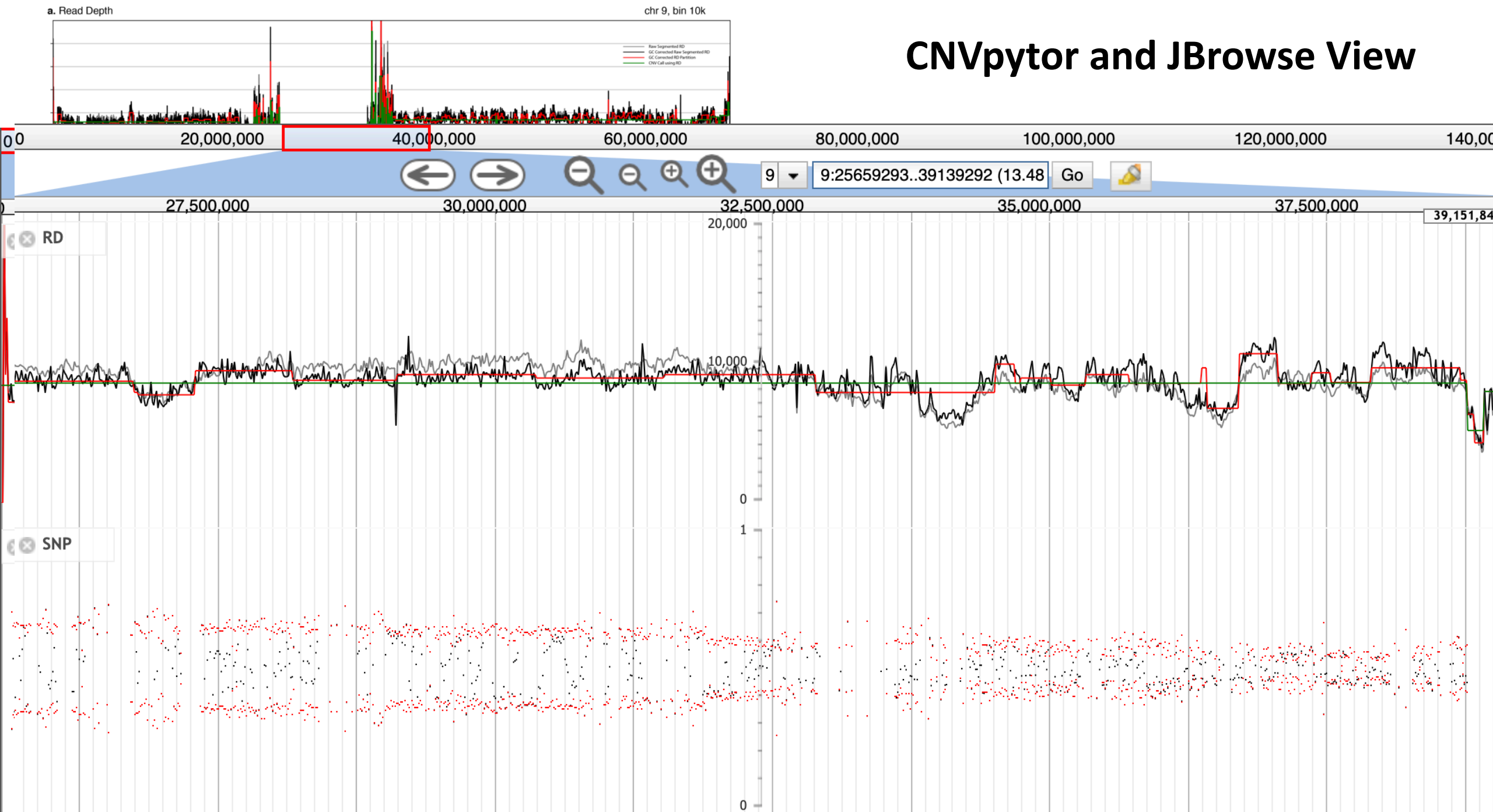
Note:

If export directory is not the JBrowse Installed directory, then the user need to copy to JBrowse directory

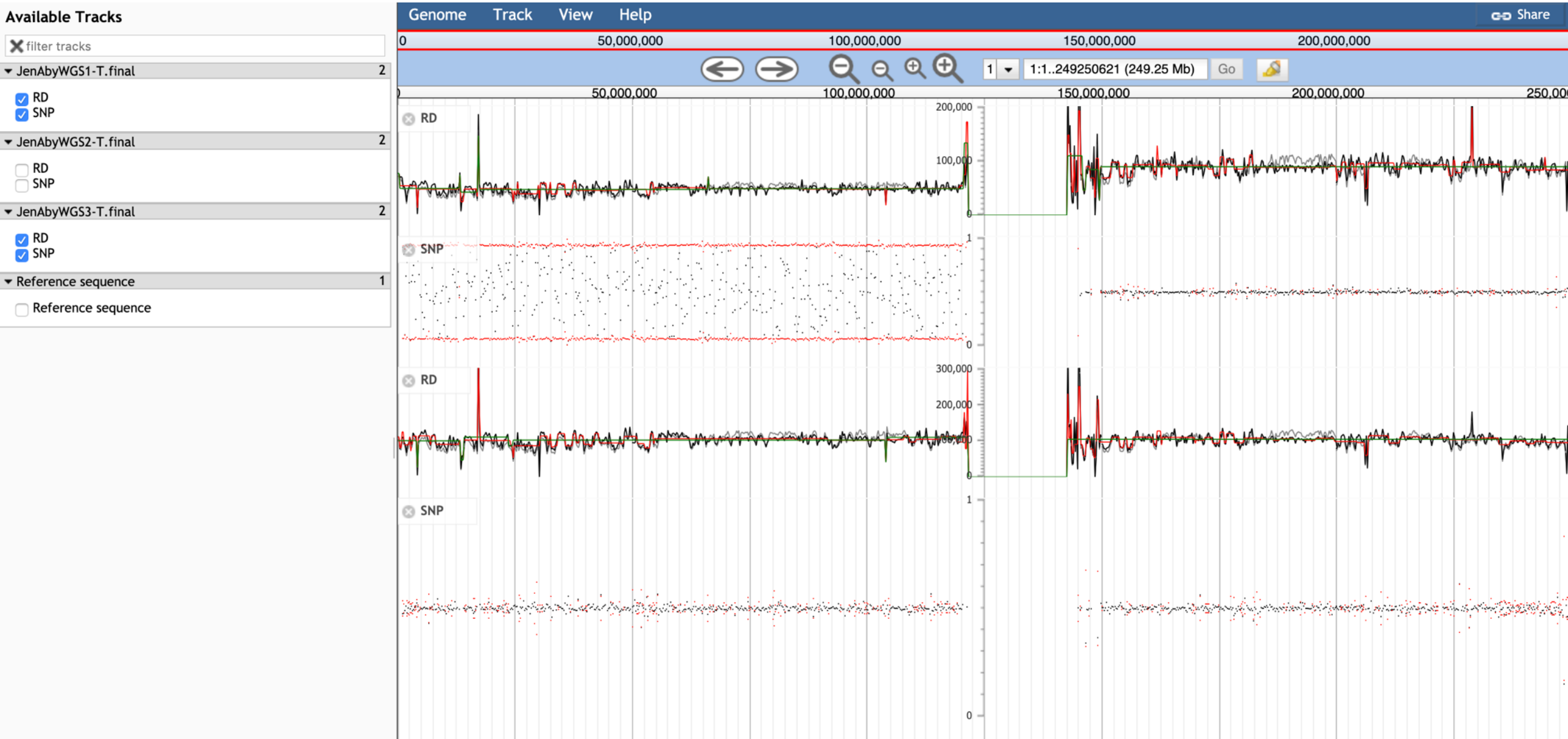
a. CNVpytor View: Read depth data for chr1



CNVpytor and JBrowse View



CNVpytor and JBrowse View with Multiple cases



Future developments

- Porting some of CNVpytor's segmentation and calling functionality into JBrowse
- Improvements and maintenance