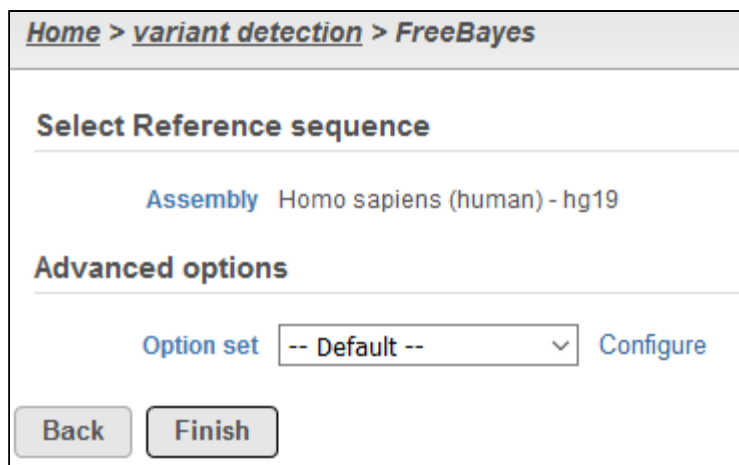


FreeBayes

A very popular variant detection approach that performs well in many situations, FreeBayes (version 1.0.1) employs a Bayesian statistical framework to determine the most likely combination of genotypes in the sample(s) at each position in a reference genome for any number of individuals from a population. It is haplotype-based, calling variants based on the literal sequences of reads aligned to a particular target and not their precise alignment. This method can identify both single nucleotide variants and insertions/deletion events. Information on the model underlying the variant detection are detailed by Garrison et al.¹

FreeBayes dialog

Selecting FreeBayes from the context sensitive menu will bring up the Freebayes task dialog (Figure 1), which contains two sections: *Select Reference sequence* and *Advanced options*.



Home > variant detection > FreeBayes

Select Reference sequence

Assembly Homo sapiens (human) - hg19

Advanced options

Option set -- Default -- Configure

Back Finish

Figure 1. Components of the FreeBayes task dialog

Select Reference sequence will specify the reference assembly to utilize for variant detection. If the alignment was generated in Partek® Flow®, the *Assembly* will be displayed as text in the section, and you do not have the option to change the reference. In the event that alignment was performed outside of Partek Flow, you will need to select the appropriate *Assembly* utilized for alignment in the drop-down list. Assemblies previously added to library files (see [Library File Management](#)) will be available for selection or *New assembly...* can be utilized to import the reference sequence to library files from within the task.

Advanced options provides a means to tune parameters in the variant detection for optimal performance. Upon invoking the task dialog, *Option set* is set to *Default*, and these parameters are provided by the FreeBayes developers. Clicking *Configure* button will open a window to tune advanced options (Figure 2). Freebayes has advanced options for *Population model*, *Allele scope*, *Indel realignment*, *Input filters*, *Mappability priors*, *Genotype likelihoods*, *Algorithmic features*, and *Report options*. Moving the mouse cursor over the info button  will provide details for each parameter. Please refer to the [FreeBayes documentation](#) for further information on tuning these parameters.

Advanced options

▼ Population model

Expected mutation rate *i*

0.001

Ploidy *i*

2

Samples result from pooled sequencing *i*

☐

Output all alleles *i*

☐

▼ Allele scope

Ignore SNP alleles *i*

☐

Ignore insertion and deletion alleles *i*

☐

Ignore MNPs *i*

☐

Ignore complex events *i*

☐

Use best alleles *i*

4

Haplotype length *i*

3

Min repeat size *i*

5

Min repeat entropy *i*

0

No partial observations *i*

☐

▼ Indel realignment

Don't left align indels *i*

☐

▼ Input filters

Use duplicate reads *i*

☐

Min mapping quality *i*

1

Min base quality *i*

0

Min supporting allele qsum *i*

0

Min supporting mapping qsum *i*

0

Apply

Save as new

Cancel

Figure 2. Configuring advanced FreeBayes options

References

1. Garrison E, Marth G. Haplotype-based variant detection from short-read sequencing. July 2012. <https://arxiv.org/abs/1207.3907>.

Additional Assistance

If you need additional assistance, please visit [our support page](#) to submit a help ticket or find phone numbers for regional support.



Your Rating: Results: 39 rates