# LoFreq

LoFreq (version 2.1.3a) is a very sensitive and fast variant caller that can be employed to robustly call low-frequency variants. Uilizing sources of sequencing error in the detection model, LoFreq can identify variants below the sequencing error rate. The significance of each variant is calculated to allow for control of false positives. This method can identify both single nucleotide variants and insertions/deletion events, although the current implementation does not produce discrete genotype calls. Information on the model underlying the variant detection is detailed by Wilm et al.[1]

## LoFreq dialog

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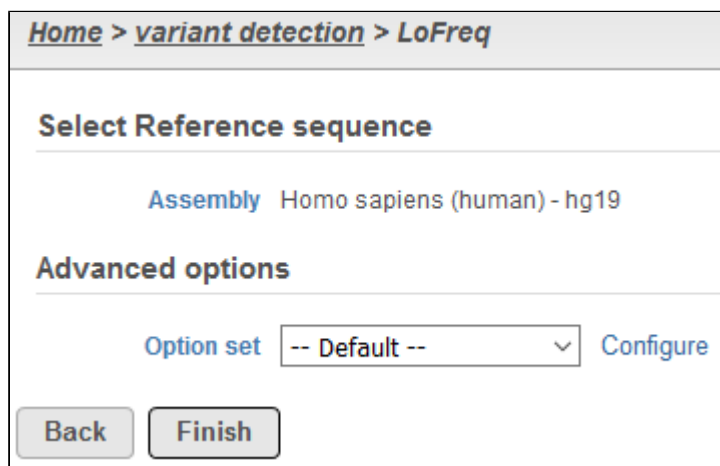


*Figure 1. Components of the LoFreq task dialog*

*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 LoFreq developers. Clicking *Configure* will open a window to tune advanced options (Figure 2). LoFreq has advanced options for *Region control*, *Base-call quality*, *Base-alignment (BAQ)* and *indel-alignment (IDAQ) qualities*, *Mapping quality*, *Indels*, *Source quality*, *P-values*, and *Other*. Moving the mouse cursor over the info button 🛈 will provide details for each parameter. Please refer to the LoFreq documentation for further suggestions on tuning these parameters.

*Figure 2. Configuring advanced LoFreq options*

# References

1. Wilm A, Aw PPK, Bertrand D, et al. LoFreq: a sequence-quality aware, ultra-sensitive variant caller for uncovering cell-population heterogeneity from high-throughput sequencing datasets. *Nucleic Acids Res*. 2012;40(22):11189-11201.

# Additional Assistance

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

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