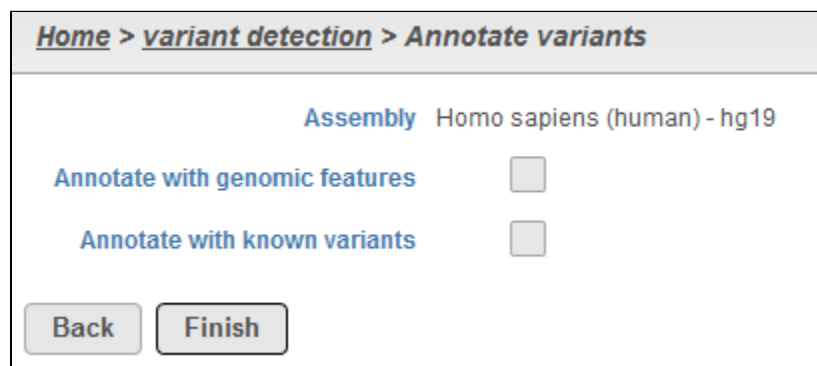


Annotate Variants

The *Annotate variants* task in Partek Flow provides a means to add information with regards to genomic features, such as transcript models, and existing variant databases to the variants contained in the projects. This information can be useful for filtering, interpreting, and prioritizing variants for downstream investigation. The *Annotate variants* task can be invoked from any *Variants* or *Annotated variants* data node, and the task will be added to and supplement any existing annotation in the underlying vcf files. Annotation information will also be visible in the [View variants Variant report](#) and the [Summarize Cohort Mutations Cohort mutation summary report](#).

Annotate variants dialog

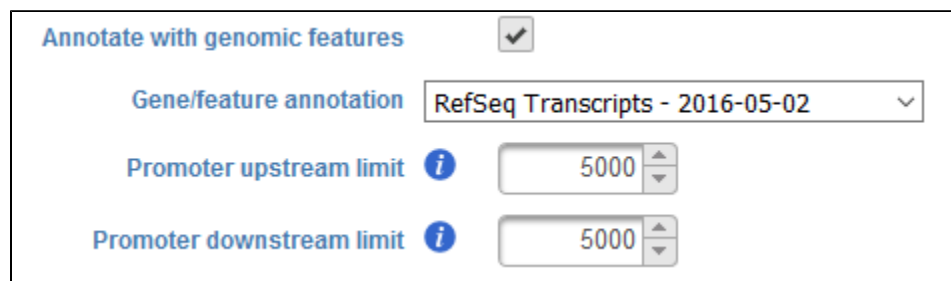
The task dialog for **Annotate variants** contains three sections: *Assembly*, *Annotate with genomic features*, and *Annotate with known variants* (Figure 1). If variant detection was performed in Partek Flow, the *Assembly* will be displayed as text in the section, and you do not have the option to change the reference. In the event that variant detection was performed outside of Partek Flow, you will need to select the appropriate Assembly utilized for variant detection 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 from within the task.



The screenshot shows the 'Annotate variants' dialog box. At the top, there is a breadcrumb trail: 'Home > variant detection > Annotate variants'. Below this, the 'Assembly' section displays 'Homo sapiens (human) - hg19'. There are two checkboxes: 'Annotate with genomic features' and 'Annotate with known variants', both of which are currently unchecked. At the bottom left, there are two buttons: 'Back' and 'Finish'.

Figure 1. Components of the annotate variants dialog

Selecting *Annotate with genomic features* provides the means to add gene/feature information to the variants (Figure 2). This typically takes the form of overlaying a transcript model (such as RefSeq). Annotation models previously added to library files (see [Library File Management](#)) will be available for selection or *Add annotation model* in the drop-down list can be utilized to import an annotation model to library files within the task. *Promoter upstream limit* and *Promoter downstream limit* provides a means to set the number of bases flanking the transcription start site, and this region will be considered the promoter of a feature.



The screenshot shows the configuration for 'Annotate with genomic features'. The checkbox is checked. Below it, the 'Gene/feature annotation' dropdown menu is set to 'RefSeq Transcripts - 2016-05-02'. There are two input fields for promoter limits, each with an information icon (i) to its left. The 'Promoter upstream limit' is set to 5000, and the 'Promoter downstream limit' is also set to 5000. Both input fields have up and down arrow buttons for adjustment.

Figure 2. Configuration of Annotate with genomic features

Selecting *Annotate with known variants* will provide the ability to specify a *Variant annotation* database (Figure 3). Known variant databases in vcf format, such as dbSNP¹ and 1000 Genomes² for human variants, can be used in the task. Additional databases not provided for automated download in Partek[®] Flow[®], such as the Catalogue of Somatic Mutations in Cancer (COSMIC)³, can be obtained and employed by the user. Variant databases previously added to library files (see [Library File Management](#)) will be available for selection or *Add variant database* in the menu can be utilized to import the variant database to library files from within the task.

Annotate with known variants
☒

Variant annotation
1000 Genomes

Figure 3. Configuration of Annotate with known variants

References

1. Sherry ST. dbSNP: the NCBI database of genetic variation. *Nucleic Acids Research*. 2001;29(1):308-311. doi:10.1093/nar/29.1.308
2. Auton A, Abecasis GR, Altshuler DM, et al. A global reference for human genetic variation. *Nature*. 2015;526(7571):68-74. doi:10.1038/nature15393.
3. Forbes SA, Bhamra G, Bamford S, et al. The Catalogue of Somatic Mutations in Cancer (COSMIC). In: Haines JL, Korf BR, Morton CC, Seidman CE, Seidman JG, Smith DR, eds. *Current Protocols in Human Genetics*. Hoboken, NJ, USA: John Wiley & Sons, Inc.; 2008. <http://doi.wiley.com/10.1002/0471142905.hg1011s57>.

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