

# Filter Variants

Variant detection can identify large numbers of variants, dependent both on the size of the regions being interrogated and the parameters utilized during detection. As such, filtering variants is often a necessary task to aid in identifying variants that may be relevant for downstream investigation. Partek Flow provides tools for the filtering variant data both in regards to quality metrics generated during detection and annotation information. The task can be invoked from any *Variants* or *Annotated variants* data node.

## Filter variants dialog

The *Filter variants* task dialog can contain two to five sections, dependent on the variant caller used for detection and the level of annotation (Figure 1). All instances of the task will include the following: *Include region overlapping variants* and a section for *Quality*.

The screenshot shows the 'Filter variants' dialog box with the following sections and controls:

- Include region overlapping variants:** A checkbox that is currently unchecked.
- Variant Novelty:** Radio buttons for 'Known variants' (selected), 'Known only', 'Novel only', and 'All'.
- Variant Type:** A grid of checkboxes for various mutation types and feature sections.
  - Mutation type:** Synonymous, Missense, Nonsense.
  - Feature section:** Splice-5, Intron, Splice-3, Promoter, Non-coding RNA, Intergenic, 5-prime UTR, 3-prime UTR.
- Quality:** Two input fields: 'Minimum read depth' (set to 2.0) and 'Minimum variant quality' (set to 30.0).
- Buttons:** 'Back' and 'Finish' buttons at the bottom left.

Figure 1. Components of the Filter variants dialog

Selecting *Include region overlapping variants* will bring up a dialog to include variants located within genomic regions of interest (Figure 2), which could be regions such as transcript models or amplicons. 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 to library files from within the task. The *Gene/feature* section will allow for the use of any annotation model specified in the library files (see [Library File Management](#)) in the drop-down menu or can be imported from within the task by selecting *Add annotation model*. If an annotation that contains gene-level information is selected, this filter will include both intronic and exonic regions.

This dialog box is titled 'Include region overlapping variants' and has a checked checkbox. It contains the following fields:

- Assembly:** A text field displaying 'Homo sapiens (human) - hg19'.
- Gene/feature annotation:** A dropdown menu currently showing 'RefSeq Transcripts - 2016-05-02'.

Figure 2. Filtering with Include region overlapping variants

If the filter is invoked from an *Annotated variants* data node, the *Variant Novelty* section can be utilized to filter known variants as identified in a variant database used for annotation (Figure 3). Selecting *Known only*, *Novel only*, or *All* will include only these types of variants in the resulting filtered variants.

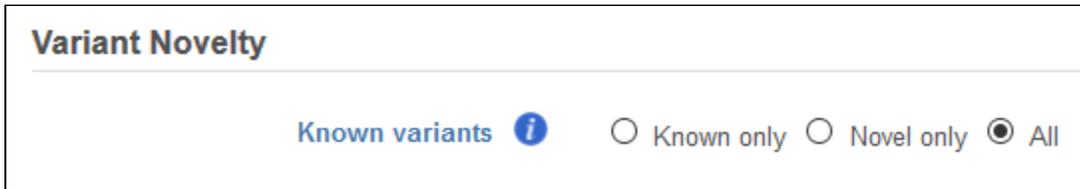


Figure 3. Filtering with Variant Novelty for data annotated with a variant database

Variants annotated with a transcript model will include a filter for *Variant Type* (Figure 4). For variants in coding regions, *Mutation type* allows for the inclusion of *Synonymous*, *Missense*, and/or *Nonsense* variants when selecting the appropriate type. For variants located outside of coding regions, *Feature section* allows for the inclusion of 5-prime splice site (*Splice-5*), 3-prime splice site (*Splice-3*), *Non-coding RNA*, *5-prime UTR*, *3-prime UTR*, *Intron*, *Promoter*, and/or *Intergenic* variants by selecting the appropriate type.



Figure 4. Filter with Variant Type for data annotated with a transcript model

The *Allele Frequency* section is available for variants detected using *LoFreq* and provides a means to identify low frequency variants within samples by excluding variant alleles that are present above the frequency specified for *Maximum estimated allele frequency* (Figure 5). This may be useful for determining variants identified in samples with heterogeneous DNA populations, such as cell-free DNA.



Figure 5. Filtering by Allele frequency derived from LoFreq variant detection

The *Quality* section of the filter task will involve metrics generated from the variant detection, and these will be dependent upon the method utilized for variant detection. Moving the mouse cursor over the info button  will provide details for each parameter. Decisions on quality filtering parameters should be based upon sequencing assay design as well as goal or the study, either identification of all potential variants or identification of high confidence variants. At the very least, the use of *Minimum read depth* should be considered for filtering to ensure sufficient read evidence was available to call a variant. In instances where paired variant detection was performed in *SAMtools*, *Minimum genotype log ratio* may be employed to ensure sufficient evidence of genotype differences in case and control sample pairs. Please refer to the *Samtools*, *FreeBayes*, and *LoFreq* documentation for further details on any of these parameters.

### Quality

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Maximum strand bias	<i>i</i>	<input type="checkbox"/>	60.0
Minimum read depth	<i>i</i>	<input type="checkbox"/>	1.0
Minimum high-quality alternate calls	<i>i</i>	<input type="checkbox"/>	0.0
Minimum mean-square mapping quality	<i>i</i>	<input type="checkbox"/>	39.0
Minimum variant quality	<i>i</i>	<input type="checkbox"/>	30.0

Figure 6. Example of filtering variants based on Samtools quality metrics

### 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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Your Rating: ☆☆☆☆☆ Results: ★★★★★ 39 rates