Annotate Variants (SnpEff)

An important aspect of variant analysis is the ability to prioritize specific variants for further investigation. As variant detection can often identify a large number of variants, it may be difficult to determine which variants may impact phenotypes. SnpEff (version 4.1k) provides a means to annotate and predict the effects of variants on genes, allowing for prioritization of variants within the project. In addition, the SnpEff databases utilized for prediction support a large number of genome assemblies. Information regarding the implementation of the predictions is detailed by Cingolani et al.[1] The predicted effect of the variant is categorized by impact:

- HIGH - frame shifts, addition/deletion of stop codons, etc;
- MODERATE – codon change/deletion/insertion, etc;
- LOW – synonymous changes, etc;
- MODIFIER – changes outside coding regions, etc.

Further details about output metrics can be found in the SnpEff documentation. The *Annotate variants (SnpEff)* task can be invoked from any *Variants* or *Annotated variants* data node, and the task will supplement any existing annotation in the 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 (SnpEff) dialog

The task dialog for **Annotate variants (SnpEff)** contains two sections: *Select SnpEff database* and *Advanced options* (Figure 1). *Select SnpEff database* will specify the reference assembly to utilize for variant detection. If the 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. *Select SnpEff database* will allow selection of databases utilized for prediction, and Partek Flow provides automated download of a limited number of these databases. Databases previously added to library files (see Library File Management) will be available for selection or *Add SnpEff variant database* in the menu can be utilized to import the reference sequence to library files from within the task. Additional information of SnpEff databases can be found in the SnpEff documentation.

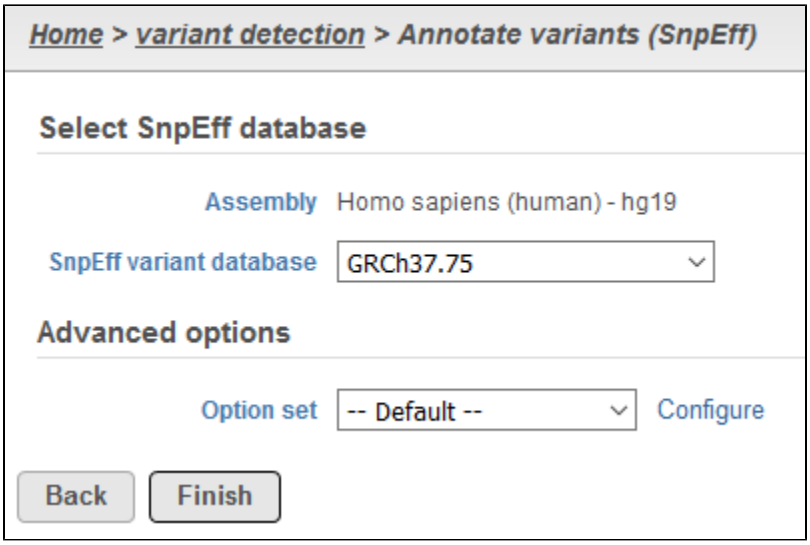

Figure 1. Components of the SnpEff dialog

Advanced options provides a means to tune parameters for annotation generated from the SnpEff database. Upon invoking the task dialog, *Option set* is set to *Default*, and these parameters are prescribed by the developers of SnpEff. Clicking *Configure* will open a window to tune advanced options (Figure 2). SnpEff has *Advanced options* for *Results filter options*, *Annotation options*, and *Database options*. Moving the mouse cursor over the info button will provide details for each parameter.

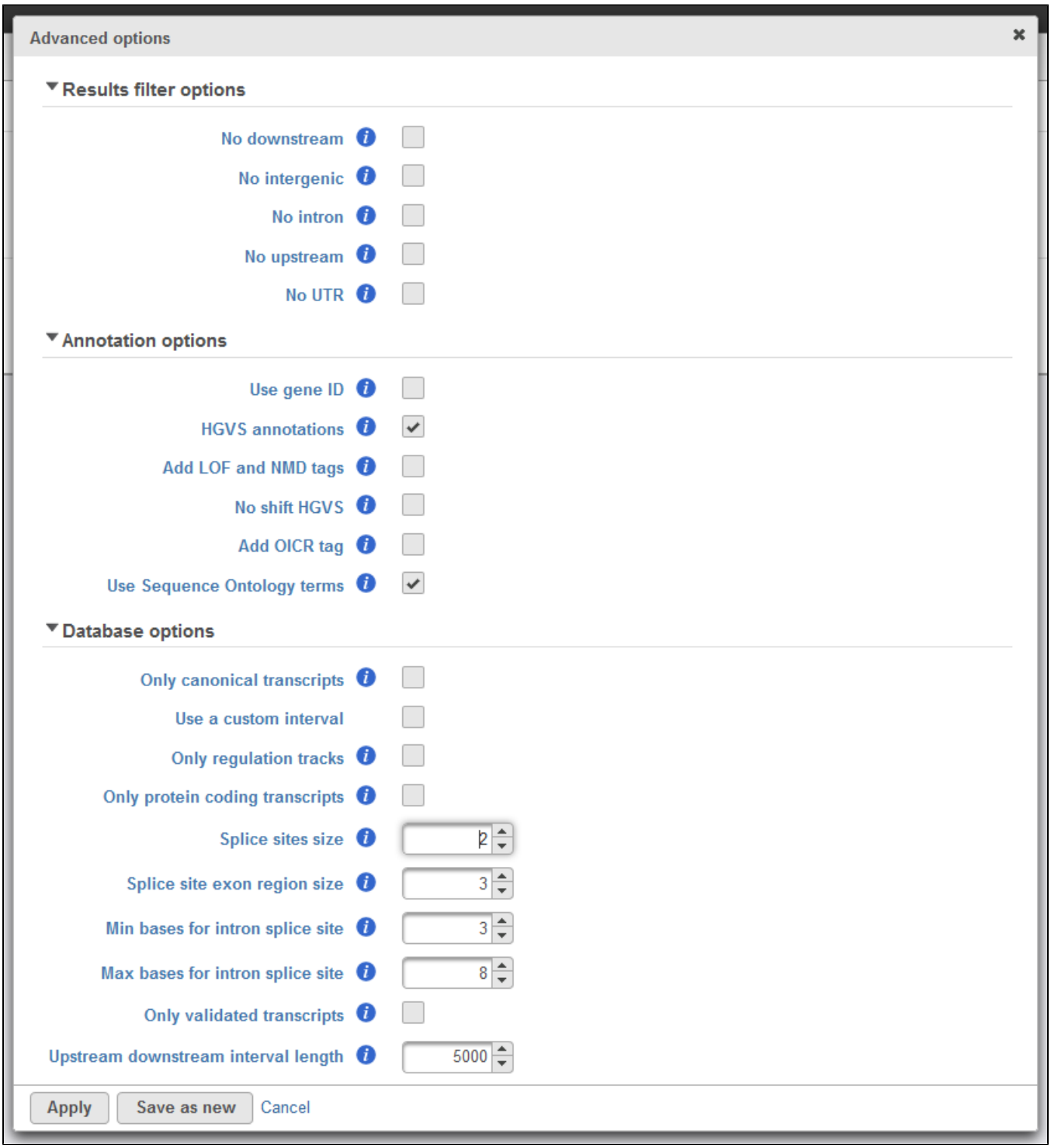

Figure 2. Configuration of SnpEff advanced options

References

1. Cingolani P, Platts A, Wang LL, et al. A program for annotating and predicting the effects of single nucleotide polymorphisms, SnpEff: SNPs in the genome of Drosophila melanogaster strain w1118; iso-2; iso-3. *Fly (Austin)*. 2012;6(2):80-92.

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: 38 rates