Annotate Variants (VEP)

An important aspect of variant analysis is the ability to prioritize variants for downstream analysis. As variant detection can often identify a large number of variants, it may be difficult to determine which variants may impact phenotypes. As implemented in Partek® Flow®, the Ensembl Variant Effect Predictor (VEP, version 84)[1] provides a means to add detailed annotation to variants in the project such as discrete aspects of transcript models and variant databases not available in the Annotate Variants task. For variants identified in human data, information from popular tools that predict the impact of variants that cause amino acid changes, SIFT[2–4] and PROVEAN[5] (available for the hg19 genome assembly), will be included. VEP databases can be obtained for multiple species, and content will be dependent on available transcript and variant information for that organism. The *Annotate variants (VEP)* t ask 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 (VEP) dialog

The task dialog for **Annotate variants (VEP)** contains two sections: *Select Variant Effect Predictor database* and *Advanced options* (Figure 1). *Select Variant Effect Predictor 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. Upon initial task usage, click the *Create variant effect predictor database* button to import a database. The VEP database for hg19 is available for automated download in Partek® Flow®, and information regarding obtaining additional databases for other species and genome assemblies can be found in the VEP documentation.

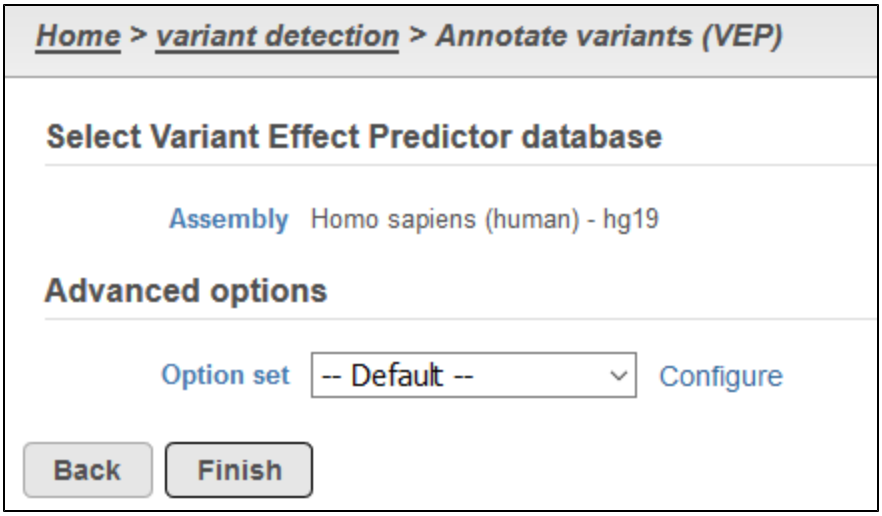

Figure 1. Components of the VEP dialog

Advanced options provides a means to specify aspects of the annotation generated from the VEP annotation task. Upon invoking the task dialog, *Option set* is set to *Default*. Clicking *Configure* will open a window to specify additional components of annotation (Figure 2). VEP has *Advanced options* for *Identifiers*, *Output options*, and *Co-located variants*. Moving the mouse cursor over the info button will provide details for each parameter.

Figure 2. Configuration of VEP advanced options

In the report, there variant impact information, it is a subjective classification of the severity of the variant consequence:

Low: a variant that is assumed to be mostly harmless or unlikely to change protein behavior

Moderate: a non-disruptive variant that might change protein effectiveness

Modifier: usually non-coding variants or variants affecting non-coding genes, where predictions are difficult or there is no evidence of impact

High: a variant is assumed to have high disruptive impact in the protein, probably causing protein truncation, loss of function or triggering nonsense mediated decay.

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