

Veridical – User Documentation

This program attempts to validate putative splicing variants by checking for concordance between splicing predictions and corresponding RNA-Seq data. This program utilizes all non-variant containing files as controls and can be provided with an additional set of normal samples to be used as controls as well.

This program creates a log file and a file filtered by p-value (defaulting to 0.05).

Options:

- h [--help] Produce help message and exit.
- t [--mutations] arg REQUIRED: provide full path to the mutation (variant) file.
- t [--exomeAn] arg REQUIRED: provide full path to the exome annotation file.

NB: variants should be derived and analyzed with respect to a conservative exome (i.e. one in which exon-intron boundaries require a moderately high level of biological evidence as opposed to those with purely EST-based transcripts or those reliant upon *in silico* methods. This will serve to limit the number of "boutique exons" and decrease the number of biologically-irrelevant validation events. For example, RefSeq would be appropriate, while Ensembl would not.

- t [--tumour] arg REQUIRED: provide full path to the list of tumour RNA-Seq BAM files.
- n [--normals] arg Provide full path to the list of normal RNA-Seq BAM files.
- b [--base] arg Provide the base full path for all output files. Defaults to ./VeridicalOut if not specified.
- d [--direct] Specify that only direct-evidence-based variants should be output.
- p [--filterPVal] arg Override the default p-value cutoff for filtered data. Variants will appear in the filtered data set iff its p-value is strictly less than this parameter.
- P [--nopvals] Specify that no p-values should be output. Useful for downstream programs that directly parse output tables.
- T [--transformOutput] Specify that all output data should be transformed via the Yeo-Johnson transformation. Overrides the default wherein only the p-values output use the transformed data.
- v [--verbose] arg Specify the verbosity of the program's output. An argument is optional and this option may be specified multiple times. If the argument is a single digit it will be used, otherwise the length of any given argument will be used as the verbosity level (i.e. "-vvv", "-v ab", and "-v 3" => verbosity level 3)