BiForce Toolbox Quick Start Guide

This quick start guide runs through the analyses possible with BiForceToolbox using the sample data provided with the software download.

For details of the full options please see the user manual.

BiForceToolbox is available at http://bioinfo.utu.fi/biforcetoolbox

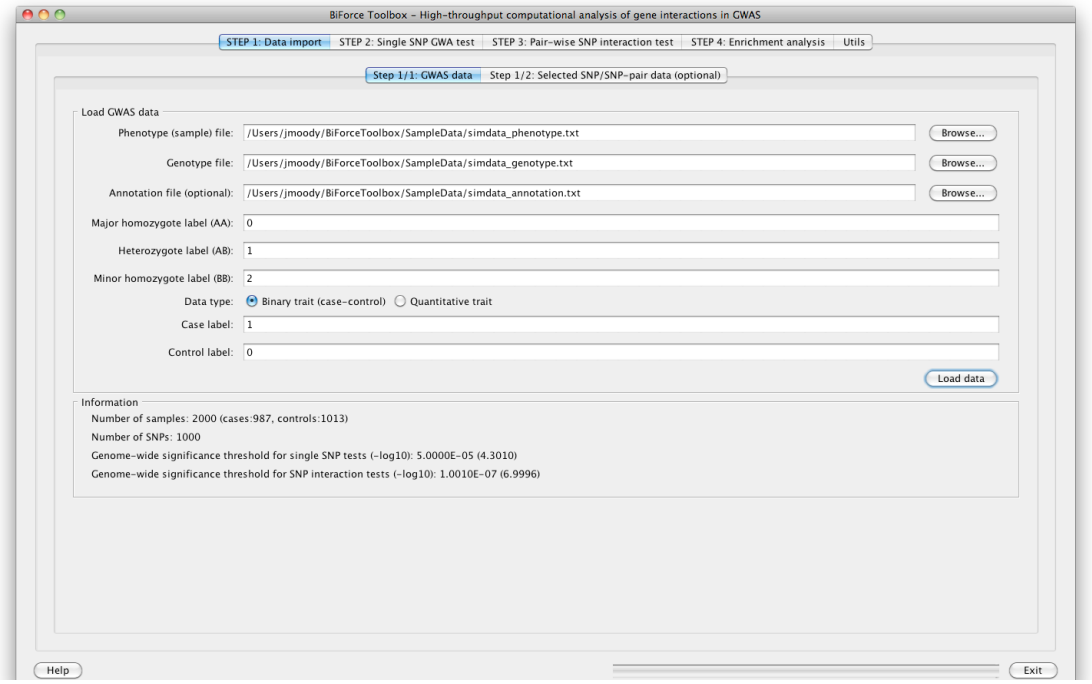
Running BiForceToolbox

To run the software navigate to the directory extracted from the program download and simply double click on the BiForceToolbox.jar file or under mac OSX/Linux run:

java -jar BiForceToolbox.jar

Loading Data

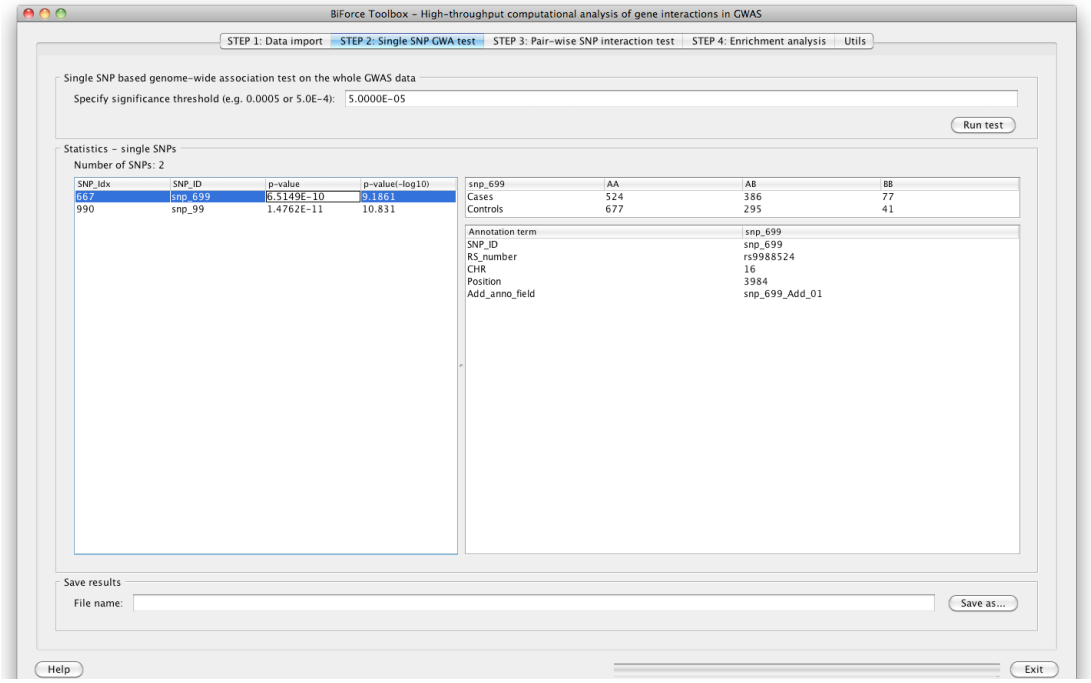
To load the sample data in the tab "STEP1 Data Import" use the buttons provided to specify the simdata_phenotype.txt, simdata_genotype.txt and simdata_annotation.txt files. These files use the default coding of 0/1/2 for major homozygote, heterozygote, minor homozygote and 1/0 for case and control phenotype. So these parameters do not need to be changed.



Pressing "Load Data" will read these files into the program and display some summary statistics in the information panel.

Running a single SNP association test

Click the tab “STEP2 Single SNP GWA test” and specify your significance threshold in the box provided. The default is the Bonferroni corrected P=0.05 threshold.

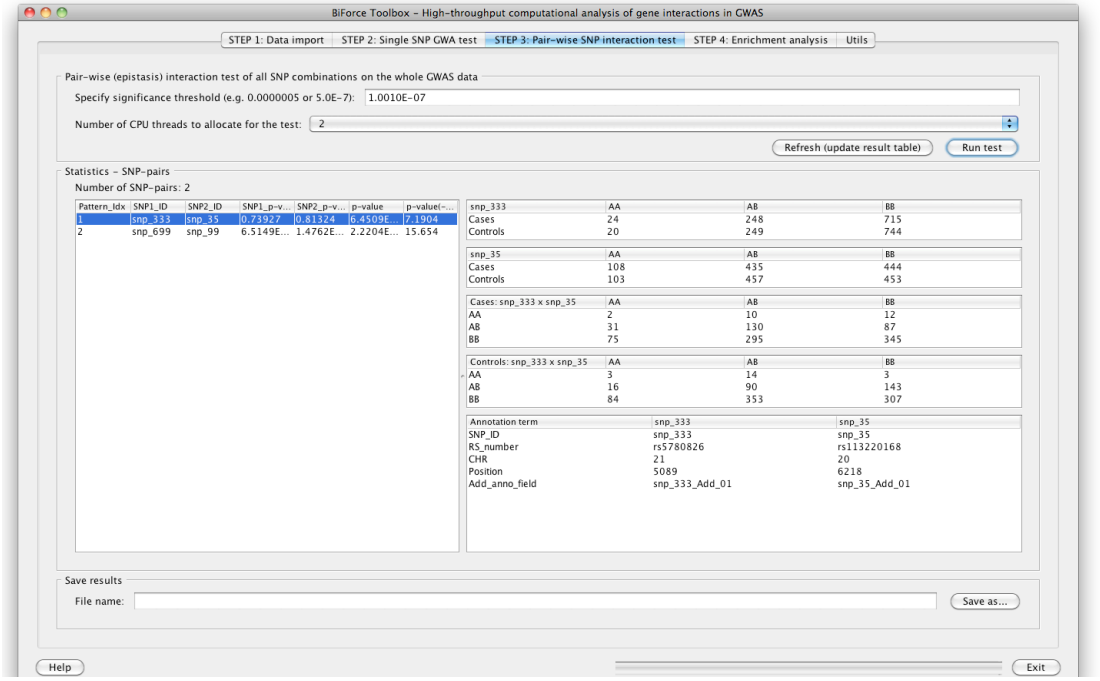


Clicking “Run Test” will perform this test and report results in the table which pass this threshold. Clicking on the results will display the annotations and contingency tables for the selected SNP.

The results table may be saved using the “save as” button below.

Running a Pairwise interaction test

Click the tab “STEP3 Pair-wise SNP interaction test”, again the default significance threshold for keeping results is the Bonferroni corrected threshold for the number of tests being performed. And select the number of CPUs to use for the tests, the default being the maximum available.

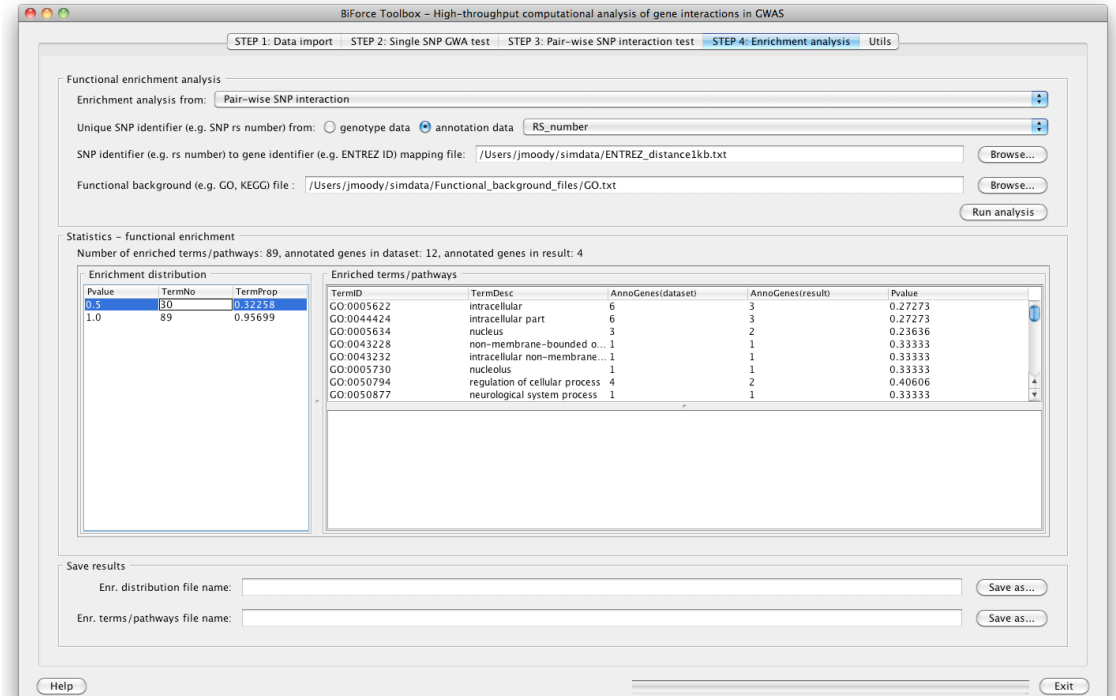


Clicking “Run test” will perform the scan displaying the results in the table, and selecting a result will display its annotations and contingency tables to the right.

The results table may be saved using the “save as” button below.

Performing Enrichment analysis

Select the tab “STEP4 Enrichment Analysis”, the top panel allows selection of which test results to perform enrichment in, as well as specifying the mapping files. Mapping files are provided on the BiForceToolbox website to map SNPs to genes and genes to either GO terms or KEGG terms.



To perform GO term enrichment in the pairwise SNP results select ''Pair-wise SNP interaction” in the first dropdown box, and select SNP identifiers in annotation data column RS_number.

Download from the BiForceToolbox website the SNP to genes mapping file ENTREZ_distance1kb.txt and the functional background file GO.txt. And select these files in the next two fields.

Clicking “Run Analysis” will perform the enrichment analysis, a hypergeometric test for each functional term. Results can be sorted by P value by clicking on the column header, selecting a term will display below the genes and SNPs annotated to that term.

The results table may be saved using the “save as” button below.