

NucleOlap

Program Documentation

1. System Requirements

- a. Minimum 1024MB RAM
- b. Java Runtime Environment (Version 6, update 14 or later)

2. Installation

- a. NucleOlap does not require installation, but instead runs as a standalone executable application. Simply unzip the contents of the NucleOlap.zip file and place the NucleOlap folder wherever you like.

3. Launching the Program

- a. Within the NucleOlap folder, there are a few items: Launcher.jar, NucleOlap.jar, a folder called "lib," and this documentation file.
- b. Double clicking either NucleOlap.jar or Launcher.jar will launch the NucleOlap graphical user interface. However, you should use Launcher.jar because it allocates 512MB of physical memory so that your computer properly handles the large SNP files.
- c. In short: Double-click Launcher.jar.
- d. Note: Neither the Launcher.jar nor the NucleOlap.jar files can be moved from this folder or the program will not run. However, shortcuts can be created.

4. The input files

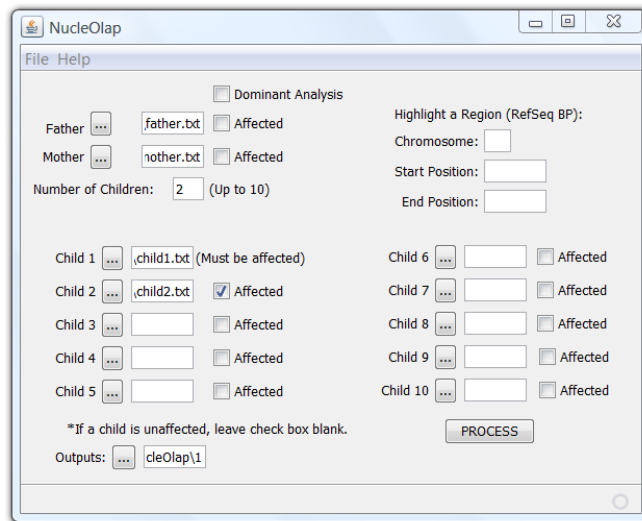
- a. NucleOlap requires at least four input files of SNP microarray data. It expects to have data for all 22 autosomes.
- b. The data file must have a row of headers and four columns for: refSNP id, chromosome, position, genotype. Ex.:

rsid	chromosome	position	genotype
rs3094315	1	742429	AA
rs1256203	1	758311	GG
rs3934834	1	995669	CC
rs9442372	1	1008567	AG
rs3737728	1	1011278	AG
rs1126058	1	1011521	GG
rs6687776	1	1020428	CC
rs9651273	1	1021403	GG
rs4970405	1	1038818	AA
rs1272625	1	1039813	AA

- c. Important: The genotype column must contain two letters for each cell. This only affects data from the X chromosome. Male files must contain two letters for the input into this program. If only one letter is entered, the program will fail.
- d. NullCalls: For null calls, either "NN" or "--" can be entered into the cell, and the data will be ignored (SNP will not be identified as informative).
- e. The file can be either tab or space separated.

5. Using the Program

a. Below is a picture of the NucleOlap GUI:



- b. A SNP file must be inputted for the father, the mother, and at least two children. The program will not analyze only one child (the analysis would not show anything).
- c. By default, the program performs an analysis assuming that you are looking at a recessive trait.
- d. To run a dominant analysis, you must check the dominant analysis box as well as which parent is affected.
- e. Output selection: if you leave the output box blank, the results file will be printed in the location of the Launcher.jar file.
- f. Highlight a region: an optional feature is the ability to highlight any chromosomal region (in pink). If left blank, nothing will happen. For consistency with the chromosomal lengths used in the program, use the location from build 36.1 of the human chromosome. Future updates will include new builds. Note: a minimum of 1,500,000 bp is

recommended for visualizing a region or the mark may be too small.

6. The Output

- a. Two files are created from this program: a region.summary.txt file, and an image.
- b. The region summary file contains a list of candidate regions for causing the recessive trait based on the SNP files and affected/non-affected statuses.
- c. The image draws these regions to allow for visualization.
- d. Saving the image: File->Save, opens up a save dialogue box. To save the file, you must give it a name with a .jpg or .png extension. Note: without typing the .jpg or .png, the image will not be saved.