

This software is provided “as is” without any warranty of any kind. In no event shall the author be held responsible for any damage resulting from the use of this software. The program package, including source codes, executables, and this documentation, is provided free of charge.

1 Description

The program can be run in batch mode, with all settings being given at the command line, or interactively via a menu. A pedigree file is used as the input file. The program produces the EFBAT statistic, S –the number of informative transmissions of the allele of interest, as well as the expectation and variance of S , and the exact p-value. When the program is run interactively, this information is output to the screen. The user has the option of creating a log file containing this output as well. When run from the command line, EFBAT will always create a log file to contain the output. In addition to the log file, EFBAT automatically generates a file, “informative.txt” which lists the families considered informative. The program accepts complex pedigrees, but these are parsed into separate nuclear families. The program is also limited to analyzing 25 markers at a time.

2 The pedigree file

A typical pedigree file for the X chromosome appears as:

```
m1 m2
100 1 0 0 1 1 1 1 3 3
100 2 0 0 2 1 1 2 1 3
100 3 1 2 1 2 2 2 3 3
100 4 1 2 2 1 1 1 1 3
101 1 0 0 1 1 1 1 1 1
101 2 0 0 2 1 1 2 1 3
101 3 1 2 2 2 0 0 1 1
```

The first line in this file is a list of the marker names. On each subsequent line, the first six columns are: family ID, individual ID, father’s ID (this will be 0 if the father is a founder of the pedigree - i.e. his parents are unknown), mother’s ID (again, 0 if a founder), sex (1=male, 2=female), and affection status (1=unaffected, 2=affected). Following this are two columns (representing the two alleles) for each marker. For X-chromosome data, all males are recorded as homozygous. 0 represents an unknown or missing marker value.

Notes:

1. All IDs must be numeric.
2. The last line of input must be the last line in the pedigree file. No blank lines should follow.

3 Menu and Settings

The EFBAT menu (Figure 1) enables the user to adjust the settings for analysis of the data set. For a description of how the settings are managed in batch mode, see the next section. A description of the menu options follows (the default options are in square brackets):

Menu Option

- 1) Log to File. [off]
- 2) X-linked trait. [off]
- 3) Choose null hypothesis. [no linkage]
- 4) Choose inheritance model. [add]
- 5) Choose marker to test. [*first marker*]
- 6) Choose allele. [*smallest allele number*]
- 7) Analyze.
- 8) Quit.

Explanation

When this is on, the output is written to a log file as well as to the screen.

When this is set to off, the program precesses the data as autosomal otherwise it assumes the marker is on the X-chromosome.

Data can be analyzed under either of the null hypotheses “no linkage and no association”, or “linkage, but no association”. The user can select the additive, dominant, or recessive inheritance model.

The user has the option to test all markers or to choose a single marker.

Choose a specific allele or test each allele for the selected marker.

Perform the analysis using the current settings.

Exit the program.

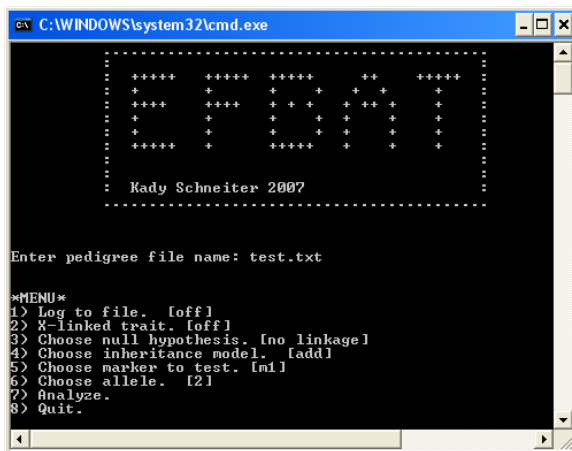


Figure 1: The EFBAT menu.

4 Running in batch mode

To run in batch mode, the user must specify the following options:

1. **pedfilename**: The name of the pedigree file, with extension ‘.ped’ or ‘.txt’.
2. **logfile**: The name of the output file.
3. **xlinked**: 1 for X-linked data, 0 for autosomal data.
4. **H_0**: 0 for the null hypothesis “no linkage, no association”, 1 for the null hypothesis “linkage, no association”
5. **model**: 1 for the additive model, 2 for the dominant model, 3 for the recessive model.
6. **marker**: set to m to select the m th marker, set to 0 to analyze all markers.

The command is as follows:

```
./EFBAT pedfilename logfile xlinked H_0 model marker
```

For example, the command

```
./EFBAT test.ped out.txt 1 0 1 3
```

will analyze the 3rd marker in the pedigree file ‘test.ped’ assuming X-linked data under the null hypothesis of ‘no linkage and no association’ and the additive inheritance model.

Note: In the interactive mode, the user has the option of specifying which allele to test. In batch mode, all alleles are analyzed for each selected marker. If a marker has more than two alleles, each is tested against all others as a single class.