POLYUNPHASED

Version 1.0

Complement to the UNPHASED user guide

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This extension of Unphased was made to perform polytomous analyses by treating jointly two binary outcomes, T1 and T2, or using one polytomous outcome. The creation of this tool was motivated by a complex genetic disease scenario hypothesized in Bureau et al (2014).

Typically, T2 can be the disease and T1 can be a related endophenotype. The reference phenotype in this joint analysis is defined by T1=T2=0, i.e. not affected by the disease nor the endophenotype. The effect of the tested SNP on the three non-reference phenotypes can be estimated with or without conditioning on another SNP located at another locus.

Input files

The outcome names must be contained in a data file. No phenotype file should be provided. Below is an example of data file with two affection status entries (A) in the case of two binary outcomes. In the case of one polytomous outcome, only one affection status entry (A) is needed.

```
A T1
A T2
M SNP1
M SNP2
 …
```

The columns of the ped file following the 5th column have to correspond to the data file description, in the same order.

Options

-joint <Pheno1:Pheno2>
or
-polytomous <Polytomous_outcome_name>

Here's an example of command line for this analysis:
polyunphased -pedfile pedfile.txt -datafile datafile.txt -joint T1:T2

Output

In the case of two binary outcomes, level 4 corresponds to T1=T2=0, which is the reference. So the output includes allele effect estimations for each one of the levels 1, 2 and 3, which are defined by:

Level 1 : T1=2 and T2=1
Level 2 : T1=1 and T2=2
Level 3 : T1=2 and T2=2

Additionnally, frequencies of transmitted and untransmitted alleles (from both parents) are given for each level of the polytomous outcome.