

# Generating Marker Summary Reports Using the *GeneticsBase*

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## 1 Introduction

This document demonstrates how to use the *GeneticsBase* (version 0.6.0) package to generate marker summary tables. It is written as a step-by-step tutorial. For additional details on each of the R functions utilized, please see the individual help pages

## 2 Example

### 2.1 Prepare phenotype data

The first step is to prepare the phenotype data. It may be in the form of a SAS dataset, SAS export file, comma-delimited text file (CSV), tab-delimited text file (TSV), or Microsoft Excel spreadsheet file (XLS). It should have one row per observation and one column per variable, and must contain a subject identifier variable that can be used to match observations with the corresponding genotype data.

### 2.2 Prepare genotype data

You also need to store the genetic call data in a file that can be read into R. *GeneticsBase* package accepts genotype data in a variety of formats:

- standard pedigree (ped) format.

```
a2m apoe
 50103  5010004  5090005  5090004      2      2      1
    2      3      4
 50103  5010005  5090005  5090004      2      2      1
    1      3      4
 50105  5010049  5090021  5090022      2      2      1
    1      4      4
 50105  5010070  5090020  5090019      1      2      1
    1      3      4
```

- hapmap format : The hapmap .ped format is a variant of the standard pedigree format. A portion of the first two lines of the hapmap file for chromosome 1 are shown below:



## 2.4 Reviewing the data

For the purpose of this example, we are going to use CAMP data set, which can be loaded manually as shown in the previous sub-section, or via

```
> library(GeneticsBase)
> data(CAMP)
```

```
Reading 8 markers and 2011 subjects from `CAMP.ped` ...
generating 'geneSet' object...
```

```
Successfully read the pedigree file `CAMP.ped`.
```

```
Number of Markers: 8
Number of Subjects: 2011
Number of Families: 651
```

```
Reading 12 vars from `CAMPZ.phe` ... Done.
```

```
Number of Phenotype Variables: 12
Number of Observations      : 2011
```

Now you can see a brief summary of the data that was loaded by simply entering the name of the object on a line by itself:

```
> CAMP
```

```
geneSet object
-----
```

```
Number of Markers:      8
Number of Observations: 2011
```

```
Sample variables: family, pid, father, mother, sex, affected, zposfevp,
```

```
zposfvcp, zlog
```

```
Genetic data:
```

```
      1.1900 1.1667 1.978 2.1391 2.1988 2.109      649.1837 650.1736 650.1908
m709 1/1    1/1    1/1    1/1    1/1    1/1    ... 1/1    1/1    1/1
m654 1/1    1/1    1/1    1/1    2/1    1/1    ... 2/1    <NA>  1/1
m47  1/1    1/2    1/2    1/2    2/2    1/2    ... 2/2    1/1    1/2
p46  2/2    2/2    2/2    2/2    1/2    2/2    ... 1/2    2/2    2/2
p79  2/2    2/1    2/1    2/1    1/1    2/1    ... 1/1    <NA>  2/1
p252 2/2    1/2    1/2    1/2    1/2    <NA>  ... 1/2    2/2    1/2
p491 1/1    1/1    1/1    1/1    1/1    1/1    ... 1/1    <NA>  1/1
p523 1/1    1/2    1/2    1/2    1/2    1/2    ... 1/2    1/1    1/2
      650.1675 651.568 651.1725
m709 1/1      1/1      1/1
m654 1/1      2/2      2/1
m47  1/2      2/2      2/2
p46  1/2      1/1      1/2
p79  2/1      1/1      1/1
```

```
p252 1/2      2/2      1/2
p491 1/1      1/1      1/1
p523 1/2      1/1      1/2
```

Warning messages:

```
1: geneSet Object has 121 observations. Only first and last 6 displayed
in: .local(object, ...)
```

The phenotype data can be extracted from the CAMP data object using the `sampleInfo` command:

```
> pdata <- sampleInfo(CAMP)
> summary(pdata)
```

| family        | pid            | father         | mother         |
|---------------|----------------|----------------|----------------|
| Min. : 1.0    | Min. : 1.0     | Min. : 0.0     | Min. : 0.0     |
| 1st Qu.:165.0 | 1st Qu.: 503.5 | 1st Qu.: 0.0   | 1st Qu.: 0.0   |
| Median :327.0 | Median :1006.0 | Median : 0.0   | Median : 0.0   |
| Mean :326.9   | Mean :1006.0   | Mean : 340.9   | Mean : 367.7   |
| 3rd Qu.:489.0 | 3rd Qu.:1508.5 | 3rd Qu.: 521.0 | 3rd Qu.: 637.5 |
| Max. :651.0   | Max. :2011.0   | Max. :2009.0   | Max. :2010.0   |

| sex           | affected       | zposfevp            | zposfvcp            |
|---------------|----------------|---------------------|---------------------|
| Min. :1.000   | Min. :0.0000   | Min. :-3.234e+00    | Min. :-2.880e+00    |
| 1st Qu.:1.000 | 1st Qu.:0.0000 | 1st Qu.: -6.790e-01 | 1st Qu.: -6.250e-01 |
| Median :1.000 | Median :0.0000 | Median : 1.000e-03  | Median : -1.600e-02 |
| Mean :1.453   | Mean :0.7041   | Mean : 1.431e-05    | Mean : -3.433e-05   |
| 3rd Qu.:2.000 | 3rd Qu.:2.0000 | 3rd Qu.: 6.275e-01  | 3rd Qu.: 6.005e-01  |
| Max. :2.000   | Max. :2.0000   | Max. : 4.021e+00    | Max. : 4.041e+00    |

...

## 2.5 Generate the tables

We can generate a variety of summary tables on our genetics data.

- Allele information

```
> alleleSummary(CAMP)
```

| Gene | Marker | Position | Group | Allele | Count | Freq  | CI-Lower | CI-Upper |
|------|--------|----------|-------|--------|-------|-------|----------|----------|
| ALL  | m709   | ?        | ALL   | 1      | 2536  | 0.998 | 0.996    | 0.999    |
|      |        |          | ALL   | 2      | 6     | 0.002 | 0.001    | 0.004    |
|      |        |          | ALL   | NA     | 0     |       |          |          |
|      | m654   | ?        | ALL   | 1      | 1618  | 0.640 | 0.621    | 0.658    |
|      |        |          | ALL   | 2      | 912   | 0.360 | 0.342    | 0.379    |
|      |        |          | ALL   | NA     | 0     |       |          |          |
|      | m47    | ?        | ALL   | 1      | 935   | 0.374 | 0.355    | 0.393    |
|      |        |          | ALL   | 2      | 1565  | 0.626 | 0.607    | 0.645    |
|      |        |          | ALL   | NA     | 0     |       |          |          |

```
p46    ?      ALL    1      1000 0.400 0.380    0.419
```

```
...
```

- Genotype information

```
> genotypeSummary(CAMP)
```

| Gene | Marker | Position | Group | Genotype | Count | Freq  | CI-Lower | CI-Upper | Expected |
|------|--------|----------|-------|----------|-------|-------|----------|----------|----------|
| ?    | m709   | ?        | ALL   | 1/1      | 1265  | 0.995 | 0.991    | 0.998    | 1265.007 |
|      |        |          |       | 1/2      | 6     | 0.005 | 0.002    | 0.009    | 5.986    |
|      |        |          |       | 2/2      | 0     | 0.000 |          | 0.007    |          |
|      |        |          |       | NA       | 0     |       |          |          |          |
| ?    | m654   | ?        | ALL   | 1/1      | 537   | 0.425 | 0.398    | 0.452    | 517.376  |
|      |        |          |       | 1/2      | 544   | 0.430 | 0.403    | 0.458    | 583.247  |
|      |        |          |       | 2/2      | 184   | 0.145 | 0.126    | 0.165    | 164.376  |
|      |        |          |       | NA       | 0     |       |          |          |          |
| ?    | m47    | ?        | ALL   | 1/1      | 175   | 0.140 | 0.121    | 0.159    | 174.845  |
|      |        |          |       | 1/2      | 585   | 0.468 | 0.440    | 0.496    | 585.310  |
|      |        |          |       | 2/2      | 490   | 0.392 | 0.365    | 0.419    | 489.845  |

```
...
```

- Marker information
- Linkage disequilibrium, text view

```
> ld <- LD(CAMP)
> ld
```

```
-----
Pairwise LD
-----
```

|                     | m709 | m654    | m47    | p46    | p79    | p252   | p491   | p523   |
|---------------------|------|---------|--------|--------|--------|--------|--------|--------|
| m709 D              |      | 0.002   | -0.001 | 0.001  | -0.001 | 0.000  | 0.000  | 0.000  |
| m709 D'             |      | 1.000   | 0.999  | 1.000  | 0.999  | 0.641  | 1.000  | 0.288  |
| m709 Corr.          |      | 0.066   | -0.038 | 0.061  | -0.039 | -0.018 | -0.005 | -0.007 |
| m709 R <sup>2</sup> |      | 0.004   | 0.001  | 0.004  | 0.001  | 0.000  | 0.000  | 0.000  |
| LD X <sup>2</sup>   |      | 6.000   | 2.000  | 5.000  | 2.000  | 0.000  | 0.000  | 0.000  |
| P-value             |      | 0.00941 | 0.0907 | 0.0193 | 0.086  | 0.577  | 0.703  | 0.808  |
| m709 LOD            |      | 1.464   | 0.622  | 1.188  | 0.640  | 0.067  | 0.032  | 0.013  |
| m709 n              |      | 1303    | 1303   | 1303   | 1303   | 1303   | 1303   | 1303   |
| m654 D              |      |         | -0.134 | 0.214  | -0.137 | -0.080 | -0.004 | -0.069 |

```
...
```

- Linkage disequilibrium, matrix plot

```
> plot(ld)
```

## Linkage Disequilibrium

|      | m709    | m654     | m47     | p46      | p79     | p252     | p491     | p523  |
|------|---------|----------|---------|----------|---------|----------|----------|-------|
| m709 | m709    | 0.004    | 0.001   | 0.004    | 0.001   | 0.000    | 0.000    | 0.000 |
| m654 | 0.00941 | m654     | 0.333   | 0.827    | 0.346   | 0.159    | 0.007    | 0.133 |
| m47  | 0.09066 | < 2e-16  | m47     | 0.390    | 0.972   | 0.162    | 0.008    | 0.140 |
| p46  | 0.01931 | < 2e-16  | < 2e-16 | p46      | 0.401   | 0.181    | 0.008    | 0.153 |
| p79  | 0.08600 | < 2e-16  | < 2e-16 | < 2e-16  | p79     | 0.164    | 0.008    | 0.142 |
| p252 | 0.57719 | < 2e-16  | < 2e-16 | < 2e-16  | < 2e-16 | p252     | 0.042    | 0.811 |
| p491 | 0.70263 | 8.33e-05 | 0.00351 | 1.95e-05 | 0.00268 | 2.93e-13 | p491     | 0.052 |
| p523 | 0.80818 | < 2e-16  | < 2e-16 | < 2e-16  | < 2e-16 | < 2e-16  | 3.62e-16 | p523  |

P-value

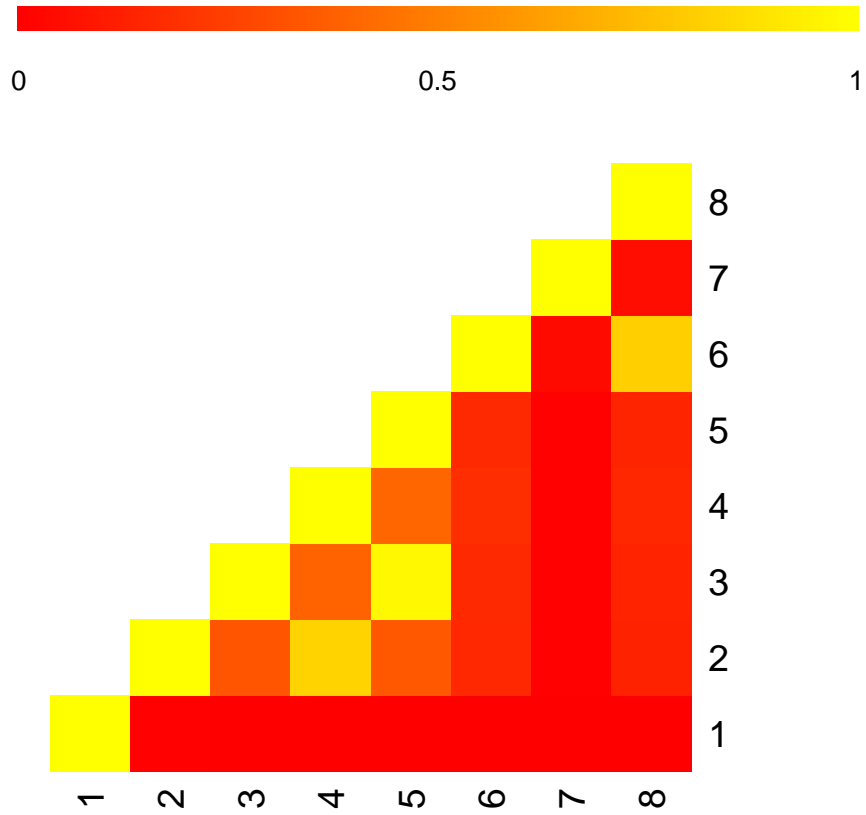
|            |               |               |               |             |            |          |
|------------|---------------|---------------|---------------|-------------|------------|----------|
| [0, 0.001] | (0.001, 0.01] | (0.01, 0.025] | (0.025, 0.05] | (0.05, 0.1] | (0.1, 0.5] | (0.5, 1] |
|------------|---------------|---------------|---------------|-------------|------------|----------|

R<sup>2</sup>

|           |             |            |            |             |           |
|-----------|-------------|------------|------------|-------------|-----------|
| [0, 0.25] | (0.25, 0.5] | (0.5, 0.8] | (0.8, 0.9] | (0.9, 0.95] | (0.95, 1] |
|-----------|-------------|------------|------------|-------------|-----------|

- Linkage disequilibrium, graphical view using LDView

```
> r2 <- t(ld@"R^2")  
> diag(r2) <- 1  
> LDView(r2, labRow = markerNames(CAMP))
```



### 3 Generating tables for inclusion in reports

To make it simple to include the summary tables in written reports, they can be written to files in a variety of formats, including plain text, html, and LaTeX.

#### 3.1 Plain text files

```
> aS <- alleleSummary(CAMP)
> txt(aS, file = "CAMP_alleleSummary.txt")
```

#### 3.2 LaTeX files

```
> aS <- alleleSummary(CAMP)
> latex(aS)
```

|    | Gene | Marker | Position | Group | Allele | Count | Freq  | CI-Lower | CI-Upper |
|----|------|--------|----------|-------|--------|-------|-------|----------|----------|
| 1  | ALL  | m709   | ?        | ALL   | 1      | 2536  | 0.998 | 0.996    | 0.999    |
| 2  |      |        |          | ALL   | 2      | 6     | 0.002 | 0.001    | 0.004    |
| 3  |      |        |          | ALL   | NA     | 0     |       |          |          |
| 4  |      |        |          |       |        |       |       |          |          |
| 5  |      | m654   | ?        | ALL   | 1      | 1618  | 0.640 | 0.621    | 0.658    |
| 6  |      |        |          | ALL   | 2      | 912   | 0.360 | 0.342    | 0.379    |
| 7  |      |        |          | ALL   | NA     | 0     |       |          |          |
| 8  |      |        |          |       |        |       |       |          |          |
| 9  |      | m47    | ?        | ALL   | 1      | 935   | 0.374 | 0.355    | 0.393    |
| 10 |      |        |          | ALL   | 2      | 1565  | 0.626 | 0.607    | 0.645    |
| 11 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 12 |      |        |          |       |        |       |       |          |          |
| 13 |      | p46    | ?        | ALL   | 1      | 1000  | 0.400 | 0.380    | 0.419    |
| 14 |      |        |          | ALL   | 2      | 1502  | 0.600 | 0.581    | 0.620    |
| 15 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 16 |      |        |          |       |        |       |       |          |          |
| 17 |      | p79    | ?        | ALL   | 1      | 1544  | 0.621 | 0.601    | 0.639    |
| 18 |      |        |          | ALL   | 2      | 944   | 0.379 | 0.361    | 0.399    |
| 19 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 20 |      |        |          |       |        |       |       |          |          |
| 21 |      | p252   | ?        | ALL   | 1      | 517   | 0.221 | 0.205    | 0.238    |
| 22 |      |        |          | ALL   | 2      | 1819  | 0.779 | 0.762    | 0.795    |
| 23 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 24 |      |        |          |       |        |       |       |          |          |
| 25 |      | p491   | ?        | ALL   | 1      | 2505  | 0.988 | 0.983    | 0.992    |
| 26 |      |        |          | ALL   | 2      | 31    | 0.012 | 0.008    | 0.017    |
| 27 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 28 |      |        |          |       |        |       |       |          |          |
| 29 |      | p523   | ?        | ALL   | 1      | 2057  | 0.810 | 0.795    | 0.825    |
| 30 |      |        |          | ALL   | 2      | 481   | 0.190 | 0.175    | 0.205    |
| 31 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 32 |      |        |          |       |        |       |       |          |          |



```

> gs <- genotypeSummary(CAMP[-2, J])
> latex(gs)

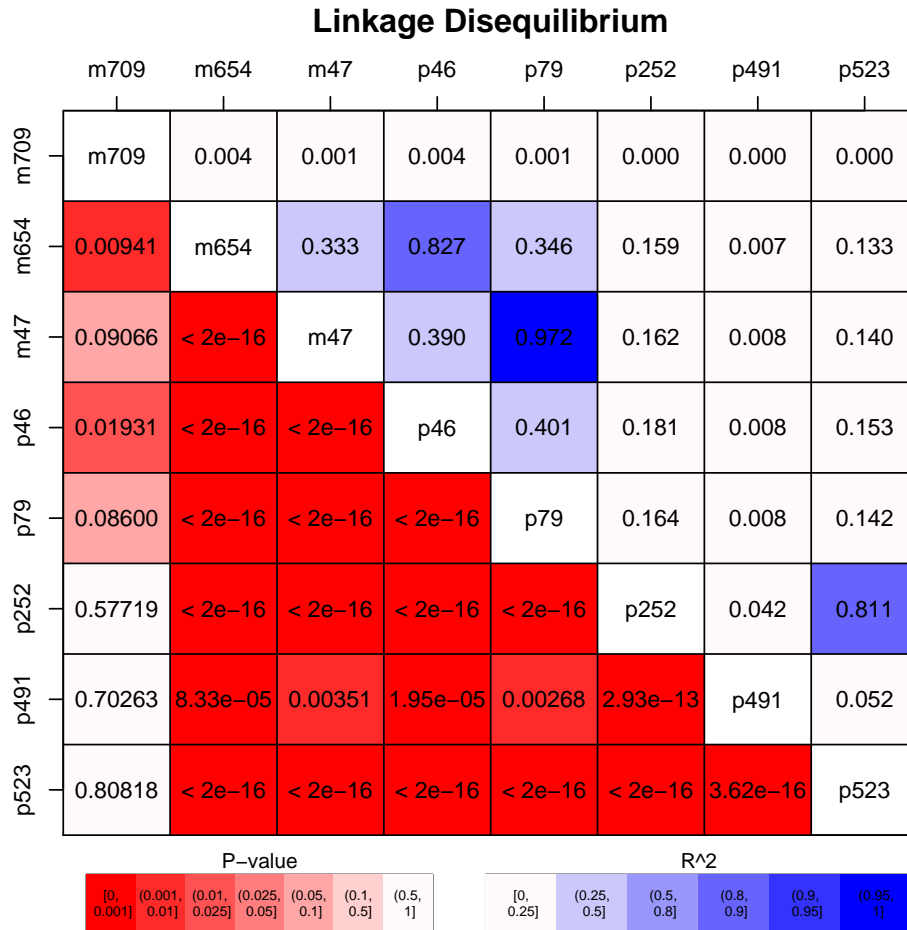
```

|    | Gene | Marker | Position | Group | Genotype | Count | Freq  | CI-Lower | CI-Upper | Expected | Obs-Exp | HWE $X^2$ | P-value |
|----|------|--------|----------|-------|----------|-------|-------|----------|----------|----------|---------|-----------|---------|
| 1  | ?    | m709   | ?        | ALL   | 1/1      | 1265  | 0.995 | 0.991    | 0.998    | 1265.007 | -0.007  | 0.007     | 1       |
| 2  |      |        |          |       | 1/2      | 6     | 0.005 | 0.002    | 0.009    | 5.986    | 0.014   |           |         |
| 3  |      |        |          |       | 2/2      | 0     | 0.000 |          |          | 0.007    | -0.007  |           |         |
| 4  |      |        |          |       | NA       | 0     |       |          |          |          |         |           |         |
| 5  |      |        |          |       |          |       |       |          |          |          |         |           |         |
| 6  | ?    | m47    | ?        | ALL   | 1/1      | 175   | 0.140 | 0.121    | 0.159    | 174.845  | 0.155   | 0.000     | 1       |
| 7  |      |        |          |       | 1/2      | 585   | 0.468 | 0.440    | 0.496    | 585.310  | -0.310  |           |         |
| 8  |      |        |          |       | 2/2      | 490   | 0.392 | 0.365    | 0.419    | 489.845  | 0.155   |           |         |
| 9  |      |        |          |       | NA       | 0     |       |          |          |          |         |           |         |
| 10 |      |        |          |       |          |       |       |          |          |          |         |           |         |
| 11 | ?    | p46    | ?        | ALL   | 1/1      | 209   | 0.167 | 0.146    | 0.188    | 199.840  | 9.160   | 1.165     | 0.288   |
| 12 |      |        |          |       | 1/2      | 582   | 0.465 | 0.437    | 0.493    | 600.320  | -18.320 |           |         |
| 13 |      |        |          |       | 2/2      | 460   | 0.368 | 0.341    | 0.395    | 450.840  | 9.160   |           |         |
| 14 |      |        |          |       | NA       | 0     |       |          |          |          |         |           |         |
| 15 |      |        |          |       |          |       |       |          |          |          |         |           |         |
| 16 | ?    | p79    | ?        | ALL   | 1/1      | 486   | 0.391 | 0.363    | 0.418    | 479.087  | 6.913   | 0.693     | 0.437   |
| 17 |      |        |          |       | 1/2      | 572   | 0.460 | 0.432    | 0.487    | 585.826  | -13.826 |           |         |
| 18 |      |        |          |       | 2/2      | 186   | 0.150 | 0.130    | 0.170    | 179.087  | 6.913   |           |         |
| 19 |      |        |          |       | NA       | 0     |       |          |          |          |         |           |         |
| 20 |      |        |          |       |          |       |       |          |          |          |         |           |         |
| 21 | ?    | p252   | ?        | ALL   | 1/1      | 70    | 0.060 | 0.047    | 0.074    | 57.211   | 12.789  | 4.715     | 0.0331  |
| 22 |      |        |          |       | 1/2      | 377   | 0.323 | 0.296    | 0.349    | 402.578  | -25.578 |           |         |
| 23 |      |        |          |       | 2/2      | 721   | 0.617 | 0.589    | 0.645    | 708.211  | 12.789  |           |         |
| 24 |      |        |          |       | NA       | 0     |       |          |          |          |         |           |         |
| 25 |      |        |          |       |          |       |       |          |          |          |         |           |         |
| 26 | ?    | p491   | ?        | ALL   | 1/1      | 1237  | 0.976 | 0.967    | 0.983    | 1237.189 | -0.189  | 0.194     | 1       |
| 27 |      |        |          |       | 1/2      | 31    | 0.024 | 0.017    | 0.033    | 30.621   | 0.379   |           |         |
| 28 |      |        |          |       | 2/2      | 0     | 0.000 |          |          | 0.189    | -0.189  |           |         |
| 29 |      |        |          |       | NA       | 0     |       |          |          |          |         |           |         |
| 30 |      |        |          |       |          |       |       |          |          |          |         |           |         |
| 31 | ?    | p523   | ?        | ALL   | 1/1      | 840   | 0.662 | 0.636    | 0.688    | 833.579  | 6.421   | 1.377     | 0.241   |
| 32 |      |        |          |       | 1/2      | 377   | 0.297 | 0.272    | 0.322    | 389.841  | -12.841 |           |         |
| 33 |      |        |          |       | 2/2      | 52    | 0.041 | 0.031    | 0.052    | 45.579   | 6.421   |           |         |
| 34 |      |        |          |       | NA       | 0     |       |          |          |          |         |           |         |
| 35 |      |        |          |       |          |       |       |          |          |          |         |           |         |

```
> ld <- LD(CAMP)
> latex(ld)
```

|                     | m709    | m654    | m47     | p46     | p79     | p252     | p491     | p523   |
|---------------------|---------|---------|---------|---------|---------|----------|----------|--------|
| m709 D              | 0.002   | -0.001  | 0.001   | 0.001   | -0.001  | 0.000    | 0.000    | 0.000  |
| m709 D'             | 1.000   | 0.999   | 1.000   | 0.999   | 0.999   | 0.641    | 1.000    | 0.288  |
| m709 Corr.          | 0.066   | -0.038  | 0.061   | -0.039  | -0.018  | -0.005   | -0.005   | -0.007 |
| m709 R <sup>2</sup> | 0.004   | 0.001   | 0.004   | 0.001   | 0.000   | 0.000    | 0.000    | 0.000  |
| LD χ <sup>2</sup>   | 6       | 2       | 5       | 2       | 0       | 0        | 0        | 0      |
| P-value             | 0.00941 | 0.09066 | 0.01931 | 0.08600 | 0.57719 | 0.70263  | 0.80818  |        |
| m709 LOD            | 1.464   | 0.622   | 1.188   | 0.640   | 0.067   | 0.032    | 0.013    |        |
| m709 n              | 1303    | 1303    | 1303    | 1303    | 1303    | 1303     | 1303     |        |
|                     |         |         |         |         |         |          |          |        |
| m654 D              |         | -0.134  | 0.214   | -0.137  | -0.080  | -0.004   | -0.069   |        |
| m654 D'             |         | 0.992   | 0.979   | 0.996   | 0.991   | 1.000    | 1.000    |        |
| m654 Corr.          |         | -0.577  | 0.909   | -0.588  | -0.399  | -0.084   | -0.365   |        |
| m654 R <sup>2</sup> |         | 0.333   | 0.827   | 0.346   | 0.159   | 0.007    | 0.133    |        |
| LD χ <sup>2</sup>   |         | 620     | 1758    | 676     | 303     | 15       | 277      |        |
| P-value             |         | < 2e-16 | < 2e-16 | < 2e-16 | < 2e-16 | 8.33e-05 | < 2e-16  |        |
| m654 LOD            |         | 134.831 | 381.769 | 146.932 | 65.982  | 3.362    | 60.366   |        |
| m654 n              |         | 1303    | 1303    | 1303    | 1303    | 1303     | 1303     |        |
|                     |         |         |         |         |         |          |          |        |
| m47 D               |         |         | -0.148  | 0.231   | -0.081  | -0.005   | -0.071   |        |
| m47 D'              |         |         | 0.996   | 0.991   | 0.973   | 0.998    | 0.990    |        |
| m47 Corr.           |         |         | -0.625  | 0.986   | -0.403  | -0.087   | -0.374   |        |
| m47 R <sup>2</sup>  |         |         | 0.390   | 0.972   | 0.162   | 0.008    | 0.140    |        |
| LD χ <sup>2</sup>   |         |         | 750     | 2240    | 267     | 8        | 245      |        |
| P-value             |         |         | < 2e-16 | < 2e-16 | < 2e-16 | 0.00351  | < 2e-16  |        |
| m47 LOD             |         |         | 162.928 | 486.549 | 58.183  | 1.851    | 53.335   |        |
| m47 n               |         |         | 1303    | 1303    | 1303    | 1303     | 1303     |        |
|                     |         |         |         |         |         |          |          |        |
| p46 D               |         |         |         | -0.150  | -0.087  | -0.005   | -0.075   |        |
| p46 D'              |         |         |         | 0.996   | 0.977   | 1.000    | 0.991    |        |
| p46 Corr.           |         |         |         | -0.633  | -0.426  | -0.090   | -0.391   |        |
| p46 R <sup>2</sup>  |         |         |         | 0.401   | 0.181   | 0.008    | 0.153    |        |
| LD χ <sup>2</sup>   |         |         |         | 783     | 319     | 18       | 283      |        |
| P-value             |         |         |         | < 2e-16 | < 2e-16 | 1.95e-05 | < 2e-16  |        |
| p46 LOD             |         |         |         | 170.105 | 69.424  | 3.960    | 61.586   |        |
| p46 n               |         |         |         | 1303    | 1303    | 1303     | 1303     |        |
|                     |         |         |         |         |         |          |          |        |
| p79 D               |         |         |         |         | -0.081  | -0.005   | -0.072   |        |
| p79 D'              |         |         |         |         | 0.974   | 0.999    | 1.000    |        |
| p79 Corr.           |         |         |         |         | -0.404  | -0.088   | -0.377   |        |
| p79 R <sup>2</sup>  |         |         |         |         | 0.164   | 0.008    | 0.142    |        |
| LD χ <sup>2</sup>   |         |         |         |         | 275     | 9        | 266      |        |
| P-value             |         |         |         |         | < 2e-16 | 0.00268  | < 2e-16  |        |
| p79 LOD             |         |         |         |         | 59.797  | 1.958    | 57.918   |        |
| p79 n               |         |         |         |         | 1303    | 1303     | 1303     |        |
|                     |         |         |         |         |         |          |          |        |
| p252 D              |         |         |         |         |         | 0.009    | 0.146    |        |
| p252 D'             |         |         |         |         |         | 1.000    | 0.994    |        |
| p252 Corr.          |         |         |         |         |         | 0.204    | 0.901    |        |
| p252 R <sup>2</sup> |         |         |         |         |         | 0.042    | 0.811    |        |
| LD χ <sup>2</sup>   |         |         |         |         |         | 53       | 1321     |        |
| P-value             |         |         |         |         |         | 2.93e-13 | < 2e-16  |        |
| p252 LOD            |         |         |         |         |         | 11.564   | 286.890  |        |
| p252 n              |         |         |         |         |         | 1303     | 1303     |        |
|                     |         |         |         |         |         |          |          |        |
| p491 D              |         |         |         |         |         |          | 0.010    |        |
| p491 D'             |         |         |         |         |         |          | 1.000    |        |
| p491 Corr.          |         |         |         |         |         |          | 0.228    |        |
| p491 R <sup>2</sup> |         |         |         |         |         |          | 0.052    |        |
| LD χ <sup>2</sup>   |         |         |         |         |         |          | 66       |        |
| P-value             |         |         |         |         |         |          | 3.62e-16 |        |
| p491 LOD            |         |         |         |         |         |          | 14.426   |        |
| p491 n              |         |         |         |         |         |          | 1303     |        |
|                     |         |         |         |         |         |          |          |        |
| p523 D              |         |         |         |         |         |          |          |        |
| p523 D'             |         |         |         |         |         |          |          |        |
| p523 Corr.          |         |         |         |         |         |          |          |        |
| p523 R <sup>2</sup> |         |         |         |         |         |          |          |        |
| LD χ <sup>2</sup>   |         |         |         |         |         |          |          |        |
| P-value             |         |         |         |         |         |          |          |        |
| p523 LOD            |         |         |         |         |         |          |          |        |
| p523 n              |         |         |         |         |         |          |          |        |

```
> plot(ld)
```



### 3.3 HTML files

### 3.4 Graphics files

As usual, plots can be generated in any format R supports.

We can also output everything all at once to a set of files, encoded as plain text (`format="print"`), html (`format="html"`), or LaTeX (`format="latex"`):

```
> PGtables(CAMP, filename = "CAMP", sep = "_", format = "html")
```

```
Creating CAMP_alleleSummary.html ...
Creating CAMP_genotypeSummary.html ...
Creating CAMP_LD.html ...
Creating CAMP_LD.pdf ...
Done.
```

which creates a set of html and a PDF files in the current directory.

Figure 1: HTML allele summary table

|    | Gene | Marker | Position | Group | Allele | Count | Freq  | CI-Lower | CI-Upper |
|----|------|--------|----------|-------|--------|-------|-------|----------|----------|
| 1  | ALL  | m709   | ?        | ALL   | 1      | 2534  | 0.998 | 0.997    | 1.000    |
| 2  |      |        |          | ALL   | 2      | 4     | 0.002 | 0.000    | 0.003    |
| 3  |      |        |          | ALL   | NA     | 0     |       |          |          |
| 4  |      |        |          |       |        |       |       |          |          |
| 5  |      | m654   | ?        | ALL   | 1      | 1630  | 0.647 | 0.629    | 0.666    |
| 6  |      |        |          | ALL   | 2      | 888   | 0.353 | 0.334    | 0.371    |
| 7  |      |        |          | ALL   | NA     | 0     |       |          |          |
| 8  |      |        |          |       |        |       |       |          |          |
| 9  |      | m47    | ?        | ALL   | 1      | 924   | 0.371 | 0.352    | 0.390    |
| 10 |      |        |          | ALL   | 2      | 1564  | 0.629 | 0.610    | 0.648    |
| 11 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 12 |      |        |          |       |        |       |       |          |          |
| 13 |      | p46    | ?        | ALL   | 1      | 990   | 0.395 | 0.376    | 0.414    |
| 14 |      |        |          | ALL   | 2      | 1516  | 0.605 | 0.586    | 0.624    |
| 15 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 16 |      |        |          |       |        |       |       |          |          |
| 17 |      | p79    | ?        | ALL   | 1      | 1556  | 0.625 | 0.607    | 0.644    |
| 18 |      |        |          | ALL   | 2      | 932   | 0.375 | 0.356    | 0.393    |
| 19 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 20 |      |        |          |       |        |       |       |          |          |
| 21 |      | p252   | ?        | ALL   | 1      | 546   | 0.231 | 0.214    | 0.247    |
| 22 |      |        |          | ALL   | 2      | 1822  | 0.769 | 0.753    | 0.786    |
| 23 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 24 |      |        |          |       |        |       |       |          |          |
| 25 |      | p491   | ?        | ALL   | 1      | 2499  | 0.989 | 0.985    | 0.993    |
| 26 |      |        |          | ALL   | 2      | 27    | 0.011 | 0.007    | 0.015    |
| 27 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 28 |      |        |          |       |        |       |       |          |          |
| 29 |      | p523   | ?        | ALL   | 1      | 2031  | 0.799 | 0.783    | 0.814    |
| 30 |      |        |          | ALL   | 2      | 511   | 0.201 | 0.186    | 0.217    |
| 31 |      |        |          | ALL   | NA     | 0     |       |          |          |
| 32 |      |        |          |       |        |       |       |          |          |

Confidence intervals width is 95%, computed using the exact quantiles for the binomial distribution.

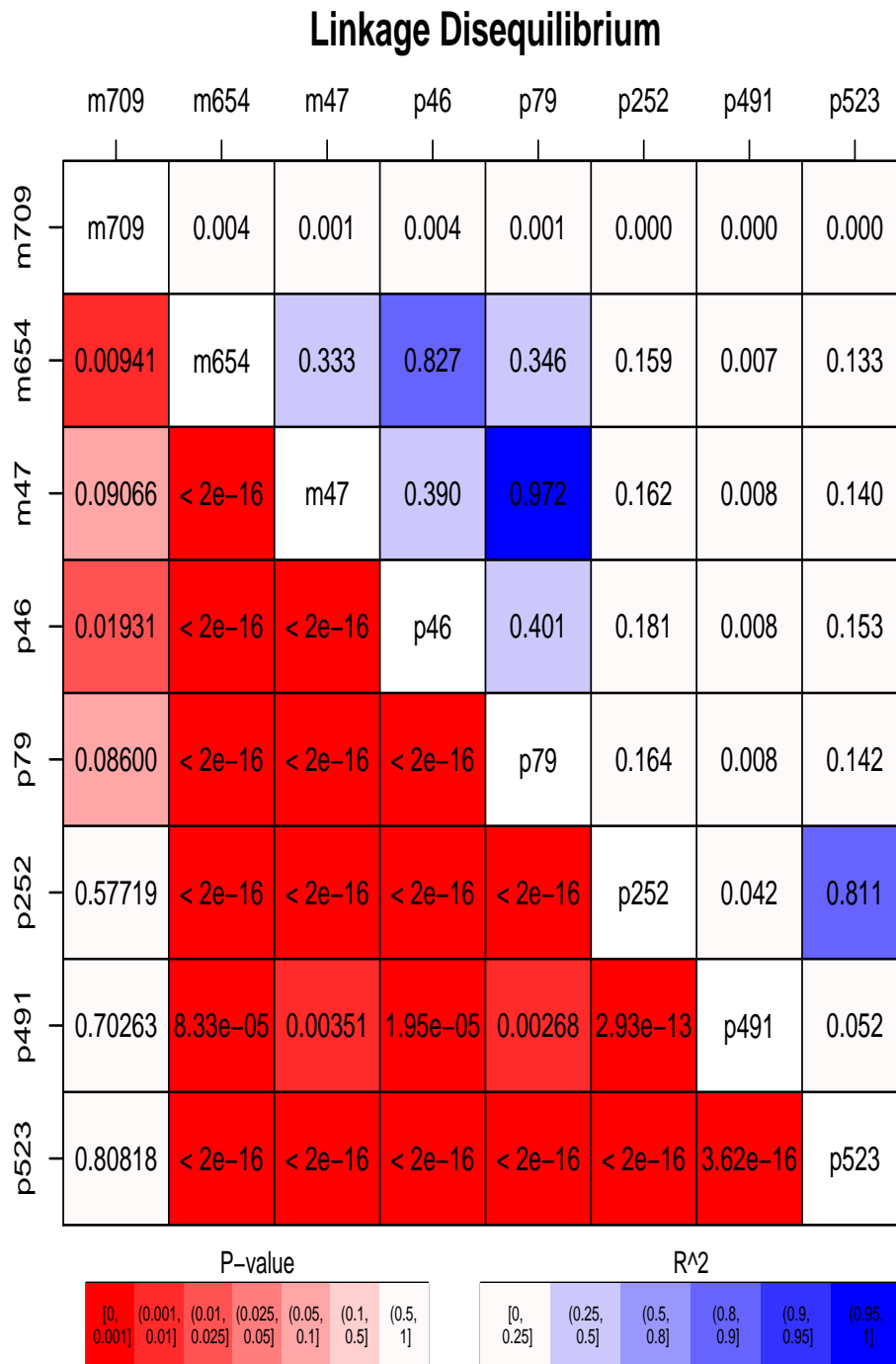
Figure 2: HTML genotype summary table

|    | Gene | Marker | Position | Group | Genotype | Count | Freq  | CI-Lower | CI-Upper | Expected | Obs-Exp | HWE X <sup>2</sup> | P-value |
|----|------|--------|----------|-------|----------|-------|-------|----------|----------|----------|---------|--------------------|---------|
| 1  | ?    | m709   | ?        | ALL   | 1/1      | 1265  | 0.997 | 0.994    | 0.999    | 1265.003 | -0.003  | 0.003              | 1       |
| 2  |      |        |          |       | 1/2      | 4     | 0.003 | 0.001    | 0.006    | 3.994    | 0.006   |                    |         |
| 3  |      |        |          |       | 2/2      | 0     | 0.000 |          |          | 0.003    | -0.003  |                    |         |
| 4  |      |        |          |       | NA       | 0     |       |          |          |          |         |                    |         |
| 5  |      |        |          |       |          |       |       |          |          |          |         |                    |         |
| 6  | ?    | m654   | ?        | ALL   | 1/1      | 536   | 0.426 | 0.399    | 0.453    | 527.581  | 8.419   | 1.080              | 0.323   |
| 7  |      |        |          |       | 1/2      | 558   | 0.443 | 0.416    | 0.471    | 574.837  | -16.837 |                    |         |
| 8  |      |        |          |       | 2/2      | 165   | 0.131 | 0.113    | 0.150    | 156.581  | 8.419   |                    |         |
| 9  |      |        |          |       | NA       | 0     |       |          |          |          |         |                    |         |
| 10 |      |        |          |       |          |       |       |          |          |          |         |                    |         |
| 11 | ?    | m47    | ?        | ALL   | 1/1      | 171   | 0.137 | 0.119    | 0.157    | 171.579  | -0.579  | 0.005              | 0.955   |
| 12 |      |        |          |       | 1/2      | 582   | 0.468 | 0.441    | 0.496    | 580.842  | 1.158   |                    |         |
| 13 |      |        |          |       | 2/2      | 491   | 0.395 | 0.367    | 0.422    | 491.579  | -0.579  |                    |         |
| 14 |      |        |          |       | NA       | 0     |       |          |          |          |         |                    |         |
| 15 |      |        |          |       |          |       |       |          |          |          |         |                    |         |
| 16 | ?    | p46    | ?        | ALL   | 1/1      | 197   | 0.157 | 0.137    | 0.178    | 195.551  | 1.449   | 0.029              | 0.906   |
| 17 |      |        |          |       | 1/2      | 596   | 0.476 | 0.448    | 0.504    | 598.899  | -2.899  |                    |         |
| 18 |      |        |          |       | 2/2      | 460   | 0.367 | 0.341    | 0.394    | 458.551  | 1.449   |                    |         |
| 19 |      |        |          |       | NA       | 0     |       |          |          |          |         |                    |         |
| 20 |      |        |          |       |          |       |       |          |          |          |         |                    |         |
| 21 | ?    | p79    | ?        | ALL   | 1/1      | 488   | 0.392 | 0.365    | 0.420    | 486.563  | 1.437   | 0.030              | 0.905   |
| 22 |      |        |          |       | 1/2      | 580   | 0.466 | 0.439    | 0.494    | 582.875  | -2.875  |                    |         |
| 23 |      |        |          |       | 2/2      | 176   | 0.141 | 0.122    | 0.161    | 174.563  | 1.437   |                    |         |
| 24 |      |        |          |       | NA       | 0     |       |          |          |          |         |                    |         |
| 25 |      |        |          |       |          |       |       |          |          |          |         |                    |         |
| 26 | ?    | p252   | ?        | ALL   | 1/1      | 68    | 0.057 | 0.045    | 0.071    | 62.947   | 5.053   | 0.685              | 0.419   |
| 27 |      |        |          |       | 1/2      | 410   | 0.346 | 0.319    | 0.373    | 420.106  | -10.106 |                    |         |
| 28 |      |        |          |       | 2/2      | 706   | 0.596 | 0.568    | 0.624    | 700.947  | 5.053   |                    |         |
| 29 |      |        |          |       | NA       | 0     |       |          |          |          |         |                    |         |
| 30 |      |        |          |       |          |       |       |          |          |          |         |                    |         |
| 31 | ?    | p491   | ?        | ALL   | 1/1      | 1236  | 0.979 | 0.970    | 0.987    | 1236.144 | -0.144  | 0.147              | 1       |
| 32 |      |        |          |       | 1/2      | 27    | 0.021 | 0.013    | 0.030    | 26.711   | 0.289   |                    |         |
| 33 |      |        |          |       | 2/2      | 0     | 0.000 |          |          | 0.144    | -0.144  |                    |         |
| 34 |      |        |          |       | NA       | 0     |       |          |          |          |         |                    |         |

Figure 3: HTML linkage disequilibrium table

|            | m709     | m654     | m47      | p46      | p79      | p252     | p491     | p523  |
|------------|----------|----------|----------|----------|----------|----------|----------|-------|
| m709 D     | 0.001    | -0.001   | 0.001    | -0.001   | 0.000    | 0.000    | 0.000    | 0.000 |
| m709 D'    | 1.000    | 0.999    | 0.999    | 0.999    | 0.047    | 1.000    | 0.151    |       |
| m709 Corr. | 0.055    | -0.031   | 0.050    | -0.031   | 0.004    | -0.004   | 0.012    |       |
| m709 R^2   | 0.003    | 0.001    | 0.002    | 0.001    | 0.000    | 0.000    | 0.000    |       |
| LD X^2     | 4        | 2        | 3        | 2        | 0        | 0        | 0        |       |
| P-value    | 0.039830 | 0.127371 | 0.069309 | 0.125046 | 0.895257 | 0.767100 | 0.702909 |       |
| m709 LOD   | 0.917    | 0.505    | 0.716    | 0.511    | 0.004    | 0.019    | 0.032    |       |
| m709 n     | 1303     | 1303     | 1303     | 1303     | 1303     | 1303     | 1303     |       |
| m654 D     |          | -0.130   | 0.212    | -0.132   | -0.082   | -0.004   | -0.070   |       |
| m654 D'    |          | 0.996    | 0.978    | 0.996    | 0.991    | 1.000    | 0.990    |       |
| m654 Corr. |          | -0.563   | 0.905    | -0.570   | -0.405   | -0.077   | -0.366   |       |
| m654 R^2   |          | 0.317    | 0.818    | 0.325    | 0.164    | 0.006    | 0.134    |       |
| LD X^2     |          | 588      | 1698     | 619      | 300      | 12       | 255      |       |
| P-value    |          | < 2e-16  | < 2e-16  | < 2e-16  | < 2e-16  | 0.000391 | < 2e-16  |       |
| m654 LOD   |          | 127.716  | 368.786  | 134.516  | 65.332   | 2.730    | 55.408   |       |
| m654 n     |          | 1303     | 1303     | 1303     | 1303     | 1303     | 1303     |       |
| m47 D      |          |          | -0.143   | 0.231    | -0.083   | -0.004   | -0.075   |       |
| m47 D'     |          |          | 0.992    | 0.991    | 0.966    | 0.998    | 1.000    |       |
| m47 Corr.  |          |          | -0.608   | 0.985    | -0.407   | -0.081   | -0.388   |       |
| m47 R^2    |          |          | 0.370    | 0.970    | 0.166    | 0.007    | 0.150    |       |
| LD X^2     |          |          | 699      | 2213     | 268      | 7        | 273      |       |
| P-value    |          |          | < 2e-16  | < 2e-16  | < 2e-16  | 0.007251 | < 2e-16  |       |
| m47 LOD    |          |          | 151.847  | 480.561  | 58.250   | 1.566    | 59.344   |       |
| m47 n      |          |          | 1303     | 1303     | 1303     | 1303     | 1303     |       |
| p46 D      |          |          |          | -0.145   | -0.090   | -0.004   | -0.078   |       |
| p46 D'     |          |          |          | 0.993    | 0.985    | 1.000    | 0.991    |       |
| p46 Corr.  |          |          |          | -0.615   | -0.439   | -0.085   | -0.400   |       |
| p46 R^2    |          |          |          | 0.379    | 0.192    | 0.007    | 0.160    |       |
| LD X^2     |          |          |          | 719      | 335      | 14       | 282      |       |
| P-value    |          |          |          | < 2e-16  | < 2e-16  | 0.000113 | < 2e-16  |       |
| p46 LOD    |          |          |          | 156.342  | 72.789   | 3.237    | 61.413   |       |
| p46 n      |          |          |          | 1303     | 1303     | 1303     | 1303     |       |
| n79 D      |          |          |          |          | -0.083   | -0.004   | -0.075   |       |

Figure 4: Linkage disequilibrium plot



## 4 Subsetting by Group

The `alleleSummary` and `genotypeSummary` functions also allow you to create tables which show the summary information separated out by a grouping variable, which must be discrete “factor” variables (such as Sex).

To accomplish this, add the argument `by=Sex` to the function call. For example:

```
> alleleSummary(CAMP, by = "sex")
```

| Gene | Marker | Position | Group | Allele | Count | Freq  | CI-Lower | CI-Upper |       |       |
|------|--------|----------|-------|--------|-------|-------|----------|----------|-------|-------|
| ALL  | m709   | ?        | 1     | 1      | 2536  | 0.998 | 0.996    | 0.999    |       |       |
|      |        |          | 1     | 2      | 6     | 0.002 | 0.001    | 0.004    |       |       |
|      |        |          | 1     | NA     | 0     |       |          |          |       |       |
|      |        |          | 2     | 1      | 2536  | 0.998 | 0.996    | 0.999    |       |       |
|      |        |          | 2     | 2      | 6     | 0.002 | 0.001    | 0.004    |       |       |
|      |        |          | 2     | NA     | 0     |       |          |          |       |       |
|      |        |          | m654  | ?      | 1     | 1     | 1618     | 0.640    | 0.621 | 0.658 |
|      |        |          |       |        | 1     | 2     | 912      | 0.360    | 0.342 | 0.379 |
|      |        |          |       |        | 1     | NA    | 0        |          |       |       |
| 2    | 1      | 1618     |       |        | 0.640 | 0.621 | 0.658    |          |       |       |
| 2    | 2      | 912      |       |        | 0.360 | 0.342 | 0.379    |          |       |       |
| 2    | NA     | 0        |       |        |       |       |          |          |       |       |
| m47  | ?      | 1        |       |        | 1     | 935   | 0.374    | 0.355    | 0.393 |       |
|      |        | 1        |       |        | 2     | 1565  | 0.626    | 0.607    | 0.645 |       |
|      |        | 1        |       |        | NA    | 0     |          |          |       |       |
|      |        | 2        | 1     | 935    | 0.374 | 0.355 | 0.393    |          |       |       |
|      |        | 2        | 2     | 1565   | 0.626 | 0.607 | 0.645    |          |       |       |
|      |        | 2        | NA    | 0      |       |       |          |          |       |       |
|      |        | p46      | ?     | 1      | 1     | 1000  | 0.400    | 0.380    | 0.419 |       |
|      |        |          |       | 1      | 2     | 1502  | 0.600    | 0.581    | 0.620 |       |
|      |        |          |       | 1      | NA    | 0     |          |          |       |       |
| 2    | 1      |          |       | 1000   | 0.400 | 0.380 | 0.419    |          |       |       |
| 2    | 2      |          |       | 1502   | 0.600 | 0.581 | 0.620    |          |       |       |
| 2    | NA     |          |       | 0      |       |       |          |          |       |       |
| p79  | ?      |          |       | 1      | 1     | 1544  | 0.621    | 0.601    | 0.639 |       |
|      |        |          |       | 1      | 2     | 944   | 0.379    | 0.361    | 0.399 |       |
|      |        |          |       | 1      | NA    | 0     |          |          |       |       |
|      |        | 2        | 1     | 1544   | 0.621 | 0.601 | 0.639    |          |       |       |
|      |        | 2        | 2     | 944    | 0.379 | 0.361 | 0.399    |          |       |       |
|      |        | 2        | NA    | 0      |       |       |          |          |       |       |
|      |        | p252     | ?     | 1      | 1     | 517   | 0.221    | 0.205    | 0.238 |       |
|      |        |          |       | 1      | 2     | 1819  | 0.779    | 0.762    | 0.795 |       |
|      |        |          |       | 1      | NA    | 0     |          |          |       |       |



|      |   |   |    |      |       |       |       |
|------|---|---|----|------|-------|-------|-------|
|      |   | 2 | 1  | 517  | 0.221 | 0.205 | 0.238 |
|      |   | 2 | 2  | 1819 | 0.779 | 0.762 | 0.795 |
|      |   | 2 | NA | 0    |       |       |       |
| p491 | ? | 1 | 1  | 2505 | 0.988 | 0.983 | 0.992 |
|      |   | 1 | 2  | 31   | 0.012 | 0.008 | 0.017 |
|      |   | 1 | NA | 0    |       |       |       |
|      |   | 2 | 1  | 2505 | 0.988 | 0.983 | 0.992 |
|      |   | 2 | 2  | 31   | 0.012 | 0.008 | 0.017 |
|      |   | 2 | NA | 0    |       |       |       |
| p523 | ? | 1 | 1  | 2057 | 0.810 | 0.795 | 0.825 |
|      |   | 1 | 2  | 481  | 0.190 | 0.175 | 0.205 |
|      |   | 1 | NA | 0    |       |       |       |
|      |   | 2 | 1  | 2057 | 0.810 | 0.795 | 0.825 |
|      |   | 2 | 2  | 481  | 0.190 | 0.175 | 0.205 |
|      |   | 2 | NA | 0    |       |       |       |

Footer:

Confidence intervals width is 95%, computed using  
the exact quantiles for the binomial  
distribution.

This will display a table within a separate block within each marker for each level of the variable `Sex`.

To control whether the summary table for entire data in addition to individual factor levels, add `includeOverall=TRUE` or `includeOverall=FALSE` (the default) as appropriate.

## A Example R script

```
> library(GeneticsBase)
> data(CAMP)
> PGtables(CAMP, filename = "test", format = "html")
> PGtables(CAMP, filename = "test", format = "latex")
```

## References

- Warnes GR. “The Genetics Package: Utilities for handling genetic data” *R News*, Volume 3, Issue 1, June 2003.
- Warnes GR. “genetics”, a package for handling marker-based genetic data within the open-source statistical package “R”. The package includes function to compute allele frequencies, use genetic markers in statistical models, estimate disequilibrium, and test for departure from Hardy-Weinberg equilibrium.  
<http://cran.us.r-project.org/src/contrib/PACKAGES.html#genetics>, 2002-