

The R package *trioClasses* for definition of the class *SNPTrioExperiment*, an extension of *SummarizedExperiment*, for use in trio based analyses of genetic data.

Samuel G. Younkin, Robert Scharpf, Holger Schwender, Ingo Ruczinski

March 7, 2013

1 Packages

```
> library("trioClasses")
```

2 Sample Data

```
> data("sample")
> se <- SummarizedExperiment(assays = SimpleList(geno = t(sm)),
  colData = col.DF, rowData = gr)
> ped <- PedClass(ped.DF)
> ste <- SNPTrioExperiment(se, pedigree = ped)

> ste
```

```
class: SNPTrioExperiment
dim: 511 968
exptData(0):
assays(1): geno
rownames(511): chr14:54383433 chr14:54383470 ...
  chr14:54444385 chr14:54444750
rowData metadata column names(0):
colnames(968): H_ME-CSc175_1-CSc175_1_a.1
  H_ME-CSc175_2-CSc175_2 ... H_ME-DS11418_2-DS11418_2
  H_ME-DS11418_3-DS11418_3
colData names(28): vcfid jfb_famid ... id ped
pedigree(4139): famid id fid mid sex dx
```

3 Methods

We can create a list of three SnpMatrices for trio analyses using `GenoTrio()`.

```
> (gtrio <- GenoTrio(ste))

$0
A SnpMatrix with 320 rows and 511 columns
```

```

Row names: H_ME-CSc175_1-CSc175_1_a.1 ... H_ME-DS11418_1-DS11418_1
Col names: chr14:54383433 ... chr14:54444750

```

```
$F
```

```

A SnpMatrix with 320 rows and 511 columns
Row names: H_ME-CSc175_2-CSc175_2 ... H_ME-DS11418_2-DS11418_2
Col names: chr14:54383433 ... chr14:54444750

```

```
$M
```

```

A SnpMatrix with 320 rows and 511 columns
Row names: H_ME-CSc175_3-CSc175_3 ... H_ME-DS11418_3-DS11418_3
Col names: chr14:54383433 ... chr14:54444750

```

To convert to Holger's format we use `ctcbind()` and `GenoTrio()`. ("`ctcbind()`") gets its name from the composition of three functions `c(t(cbind()))`.)

```

> geno <- ctcbind(gtrio)
> sum(aTDT(geno)$transMinor[1:10])

```

```
[1] 250
```

```

> wd <- 10000
> block <- GRanges(seqnames = "chr14", IRanges(start(gr[1]),
width = wd), strand = "*")

```

```
> TransCount(ste[MAF(ste) <= 0.01, ], block)
```

```
[1] 23
```

4 Classes

4.1 SnpMatrix, DataFrame, GRanges

The four key ingredients are the SNP matrix, the pedigree information as a `DataFrame`, position of the SNPs given by a `GRanges` object, and covariate data given as a `DataFrame`.

```
> sm
```

```
A SnpMatrix with 968 rows and 511 columns
Row names: H_ME-DS10776_2-DS10776_2 ... H_ME-DS11313_1-DS11313_1
Col names: chr14:54383433 ... chr14:54444750
```

```
> ped.DF
```

```
DataFrame with 4139 rows and 6 columns
```

| | famid | | id |
|------|--------------------------------|--------------------------------|--------------------------|
| | <character> | | <character> |
| 1 | 4778 | | H_ME-4778_1-4778_1.2 |
| 2 | 4778 | | H_ME-4778_2-4778_2.2 |
| 3 | 4778 | | H_ME-4778_3-4778_3.2 |
| 4 | 4783 | | H_ME-4783_1-4783_1.1 |
| 5 | 4783 | | H_ME-4783_2-4783_2.1 |
| 6 | 4783 | | H_ME-4783_3-4783_3.1 |
| 7 | 20000492 | H_ME-20000492_1-20000492_1.1 | |
| 8 | 20000492 | H_ME-20000492_2-20000492_2_a.1 | |
| 9 | 20000492 | H_ME-20000492_3-20000492_3.1 | |
| ... | ... | | ... |
| 4131 | DS12332 | | H_ME-DS12332_1-DS12332_1 |
| 4132 | DS12332 | | H_ME-DS12332_2-DS12332_2 |
| 4133 | DS12332 | | H_ME-DS12332_3-DS12332_3 |
| 4134 | DS99998 | H_ME-DS10193_5-DS10193_5.2 | |
| 4135 | DS99998 | H_ME-DS10193_6-DS10193_6 | |
| 4136 | DS99998 | H_ME-DS10193_7-DS10193_7 | |
| 4137 | DS99999 | H_ME-DS10707_4-DS10707_4.2 | |
| 4138 | DS99999 | H_ME-DS10707_5-DS10707_5 | |
| 4139 | DS99999 | H_ME-DS10707_6-DS10707_6 | |
| | famid | | mid |
| | <character> | | <character> |
| 1 | H_ME-4778_2-4778_2.2 | | H_ME-4778_3-4778_3.2 |
| 2 | NA | | NA |
| 3 | NA | | NA |
| 4 | H_ME-4783_2-4783_2.1 | | H_ME-4783_3-4783_3.1 |
| 5 | NA | | NA |
| 6 | NA | | NA |
| 7 | H_ME-20000492_2-20000492_2_a.1 | H_ME-20000492_3-20000492_3.1 | |
| 8 | NA | | NA |
| 9 | NA | | NA |
| ... | ... | | ... |
| 4131 | H_ME-DS12332_2-DS12332_2 | H_ME-DS12332_3-DS12332_3 | |
| 4132 | NA | | NA |
| 4133 | NA | | NA |

| | | |
|------|----------------------------|--------------------------|
| 4134 | H_ME-DS10193_6-DS10193_6 | H_ME-DS10193_7-DS10193_7 |
| 4135 | NA | NA |
| 4136 | NA | NA |
| 4137 | NA | NA |
| 4138 | H_ME-DS10707_4-DS10707_4.2 | H_ME-DS10707_6-DS10707_6 |
| 4139 | NA | NA |

| | sex | dx |
|------|-----------|-----------|
| | <numeric> | <numeric> |
| 1 | 1 | 1 |
| 2 | 1 | 0 |
| 3 | 2 | 0 |
| 4 | 2 | 1 |
| 5 | 1 | 0 |
| 6 | 2 | 1 |
| 7 | 2 | 1 |
| 8 | 1 | 0 |
| 9 | 2 | 0 |
| ... | ... | ... |
| 4131 | 1 | 1 |
| 4132 | 1 | 0 |
| 4133 | 2 | 0 |
| 4134 | 2 | 1 |
| 4135 | 1 | 0 |
| 4136 | 2 | 0 |
| 4137 | 1 | 0 |
| 4138 | 1 | 1 |
| 4139 | 2 | 0 |

> gr

GRanges with 511 ranges and 0 metadata columns:

| | seqnames | ranges | strand |
|----------------|----------|----------------------|--------|
| | <Rle> | <IRanges> | <Rle> |
| chr14:54383433 | chr14 | [54383433, 54383433] | * |
| chr14:54383470 | chr14 | [54383470, 54383470] | * |
| chr14:54383819 | chr14 | [54383819, 54383819] | * |
| chr14:54383945 | chr14 | [54383945, 54383945] | * |
| chr14:54384030 | chr14 | [54384030, 54384030] | * |
| chr14:54384232 | chr14 | [54384232, 54384232] | * |
| chr14:54384251 | chr14 | [54384251, 54384251] | * |
| chr14:54384291 | chr14 | [54384291, 54384291] | * |
| chr14:54384382 | chr14 | [54384382, 54384382] | * |
| ... | ... | ... | ... |
| chr14:54443495 | chr14 | [54443495, 54443495] | * |
| chr14:54443499 | chr14 | [54443499, 54443499] | * |
| chr14:54443514 | chr14 | [54443514, 54443514] | * |
| chr14:54443831 | chr14 | [54443831, 54443831] | * |
| chr14:54443880 | chr14 | [54443880, 54443880] | * |
| chr14:54444016 | chr14 | [54444016, 54444016] | * |
| chr14:54444124 | chr14 | [54444124, 54444124] | * |
| chr14:54444385 | chr14 | [54444385, 54444385] | * |
| chr14:54444750 | chr14 | [54444750, 54444750] | * |

```

---
seqlengths:
  chr14
    NA

```

4.2 SummarizedExperiment

We combine three of the key ingredients when we create the SummarizedExperiment object.

```

> se <- SummarizedExperiment(assays = SimpleList(geno = t(sm)),
  colData = col.DF, rowData = gr)

```

4.3 SNP TrioExperiment

Now, we include the pedigree information as an object of class PedClass. We keep PedClass independent of SNP TrioExperiment for flexibility.

```

> ped <- PedClass(ped.DF)
> ste <- SNP TrioExperiment(se, pedigree = ped)

```

Here is the show method.

```

> ste

class: SNP TrioExperiment
dim: 511 968
exptData(0):
assays(1): geno
rownames(511): chr14:54383433 chr14:54383470 ...
  chr14:54444385 chr14:54444750
rowData metadata column names(0):
colnames(968): H_ME-CSc175_1-CSc175_1_a.1
  H_ME-CSc175_2-CSc175_2 ... H_ME-DS11418_2-DS11418_2
  H_ME-DS11418_3-DS11418_3
colData names(28): vcfid jb_famid ... id ped
pedigree(4139): famid id fid mid sex dx

```

And now we verify that it is indeed an extension of SummarizedExperiment.

```

> getClass("SNP TrioExperiment")

```

```

Class "SNP TrioExperiment" [package "trioClasses"]

```

Slots:

| | | |
|--------|----------------------------|------------|
| Name: | pedigree | exptData |
| Class: | PedClass | SimpleList |
| Name: | rowData | colData |
| Class: | GenomicRangesORGRangesList | DataFrame |
| Name: | assays | |

```
Class: Assays
```

```
Extends: "SummarizedExperiment"
```

4.4 PedClass

Now we investigate the pedigree slot of the SNP Trio Experiment object.

```
> class(pedigree(ste))
```

```
[1] "PedClass"
attr(,"package")
[1] "trioClasses"
```

```
> getClass("PedClass")
```

```
Class "PedClass" [package "trioClasses"]
```

```
Slots:
```

```
Name:      rownames      nrows      listData
Class: characterORNULL   integer      list
```

```
Name:      elementType elementMetadata      metadata
Class:      character DataTableORNULL      list
```

```
Extends:
```

```
Class "DataFrame", directly
Class "DataTable", by class "DataFrame", distance 2
Class "SimpleList", by class "DataFrame", distance 2
Class "DataTableORNULL", by class "DataFrame", distance 3
Class "List", by class "DataFrame", distance 3
Class "Vector", by class "DataFrame", distance 4
Class "Annotated", by class "DataFrame", distance 5
```

```
> pedigree(ste)
```

```
PedClass with 4139 rows and 6 columns
```

| | famid | | id |
|------|----------|--------------------------------|----------------------|
| | <factor> | | <factor> |
| 1 | 4778 | | H_ME-4778_1-4778_1.2 |
| 2 | 4778 | | H_ME-4778_2-4778_2.2 |
| 3 | 4778 | | H_ME-4778_3-4778_3.2 |
| 4 | 4783 | | H_ME-4783_1-4783_1.1 |
| 5 | 4783 | | H_ME-4783_2-4783_2.1 |
| 6 | 4783 | | H_ME-4783_3-4783_3.1 |
| 7 | 20000492 | H_ME-20000492_1-20000492_1.1 | |
| 8 | 20000492 | H_ME-20000492_2-20000492_2_a.1 | |
| 9 | 20000492 | H_ME-20000492_3-20000492_3.1 | |
| ... | ... | | ... |
| 4131 | DS12332 | H_ME-DS12332_1-DS12332_1 | |
| 4132 | DS12332 | H_ME-DS12332_2-DS12332_2 | |

| | | | |
|------|--------------------------------|------------------------------|--------------------------|
| 4133 | DS12332 | H_ME-DS12332_3-DS12332_3 | |
| 4134 | DS99998 | H_ME-DS10193_5-DS10193_5.2 | |
| 4135 | DS99998 | H_ME-DS10193_6-DS10193_6 | |
| 4136 | DS99998 | H_ME-DS10193_7-DS10193_7 | |
| 4137 | DS99999 | H_ME-DS10707_4-DS10707_4.2 | |
| 4138 | DS99999 | H_ME-DS10707_5-DS10707_5 | |
| 4139 | DS99999 | H_ME-DS10707_6-DS10707_6 | |
| | | fid | mid |
| | | <factor> | <factor> |
| 1 | | H_ME-4778_2-4778_2.2 | H_ME-4778_3-4778_3.2 |
| 2 | | NA | NA |
| 3 | | NA | NA |
| 4 | | H_ME-4783_2-4783_2.1 | H_ME-4783_3-4783_3.1 |
| 5 | | NA | NA |
| 6 | | NA | NA |
| 7 | H_ME-20000492_2-20000492_2_a.1 | H_ME-20000492_3-20000492_3.1 | |
| 8 | | NA | NA |
| 9 | | NA | NA |
| ... | | ... | ... |
| 4131 | | H_ME-DS12332_2-DS12332_2 | H_ME-DS12332_3-DS12332_3 |
| 4132 | | NA | NA |
| 4133 | | NA | NA |
| 4134 | | H_ME-DS10193_6-DS10193_6 | H_ME-DS10193_7-DS10193_7 |
| 4135 | | NA | NA |
| 4136 | | NA | NA |
| 4137 | | NA | NA |
| 4138 | | H_ME-DS10707_4-DS10707_4.2 | H_ME-DS10707_6-DS10707_6 |
| 4139 | | NA | NA |
| | sex | dx | |
| | <factor> | <factor> | |
| 1 | 1 | 1 | |
| 2 | 1 | 0 | |
| 3 | 2 | 0 | |
| 4 | 2 | 1 | |
| 5 | 1 | 0 | |
| 6 | 2 | 1 | |
| 7 | 2 | 1 | |
| 8 | 1 | 0 | |
| 9 | 2 | 0 | |
| ... | ... | ... | |
| 4131 | 1 | 1 | |
| 4132 | 1 | 0 | |
| 4133 | 2 | 0 | |
| 4134 | 2 | 1 | |
| 4135 | 1 | 0 | |
| 4136 | 2 | 0 | |
| 4137 | 1 | 0 | |
| 4138 | 1 | 1 | |
| 4139 | 2 | 0 | |

4.5 geno accessor

```
> class(geno(ste))

[1] "SnpMatrix"
attr(,"package")
[1] "snpStats"

> getClass("SnpMatrix")

Class "SnpMatrix" [package "snpStats"]

Slots:

Name:      .Data
Class: matrix

Extends:
Class "matrix", from data part
Class "array", by class "matrix", distance 2
Class "mMatrix", by class "matrix", distance 2
Class "structure", by class "matrix", distance 3
Class "vector", by class "matrix", distance 4, with explicit coerce

Known Subclasses: "XSnpMatrix"

> geno(ste)

A SnpMatrix with 968 rows and 511 columns
Row names:  H_ME-CSc175_1-CSc175_1_a.1 ... H_ME-DS11418_3-DS11418_3
Col names:  chr14:54383433 ... chr14:54444750
```

4.6 RowData

```
> rowData(ste)

GRanges with 511 ranges and 0 metadata columns:
      seqnames      ranges strand
      <Rle>        <IRanges> <Rle>
chr14:54383433 chr14 [54383433, 54383433] *
chr14:54383470 chr14 [54383470, 54383470] *
chr14:54383819 chr14 [54383819, 54383819] *
chr14:54383945 chr14 [54383945, 54383945] *
chr14:54384030 chr14 [54384030, 54384030] *
chr14:54384232 chr14 [54384232, 54384232] *
chr14:54384251 chr14 [54384251, 54384251] *
chr14:54384291 chr14 [54384291, 54384291] *
chr14:54384382 chr14 [54384382, 54384382] *
...          ...          ...
chr14:54443495 chr14 [54443495, 54443495] *
chr14:54443499 chr14 [54443499, 54443499] *
chr14:54443514 chr14 [54443514, 54443514] *
```



```

chr14:54443831    chr14 [54443831, 54443831]    *
chr14:54443880    chr14 [54443880, 54443880]    *
chr14:54444016    chr14 [54444016, 54444016]    *
chr14:54444124    chr14 [54444124, 54444124]    *
chr14:54444385    chr14 [54444385, 54444385]    *
chr14:54444750    chr14 [54444750, 54444750]    *
---
seqlengths:
  chr14
    NA

```

4.7 ColData

5 Validity