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

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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 values 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
```

3 Methods

Now create a list of three SnpMatrices for trio analyses using GenoTrio().

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

Now we can do things easily, such as count transmissions.

```
> trans.011 <- with(gtrio, sum(F == 0 & M == 1 & 0 == 1,
     na.rm = TRUE))
Or run Holger's functions.
> geno <- ctcbind(gtrio)
> aTDT(geno[, 1:10])
$stat
 [1]
           NaN 5.0000000 0.3333333 15.0912548 10.3142857 1.0000000
 [7] 3.0000000 4.1666667 32.0512821 12.6420233
$pval
 [1]
             NaN 2.534732e-02 5.637029e-01 1.024367e-04 1.320044e-03
 [6] 3.173105e-01 8.326452e-02 4.122683e-02 1.501560e-08 3.771715e-04
$transMinor
 [1]
     0
         5
              2 100
                                  7 14 100
                       8
                          0
$transMajor
 [1]
     0 15
              1 163 27
                              3 17 64 157
                          1
```

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>	<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

```
H_ME-DS12332_2-DS12332_2
4132
         DS12332
4133
         DS12332
                        H_ME-DS12332_3-DS12332_3
4134
         DS99998
                      H_ME-DS10193_5-DS10193_5.2
                        H_ME-DS10193_6-DS10193_6
4135
         DS99998
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
                         <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
7
     H_ME-20000492_2-20000492_2_a.1 H_ME-20000492_3-20000492_3.1
8
                                   NA
9
                                   NA
                                                                 NA
. . .
4131
           H_ME-DS12332_2-DS12332_2
                                          H_ME-DS12332_3-DS12332_3
4132
                                   NA
4133
                                   NA
                                                                 NA
4134
           H_ME-DS10193_6-DS10193_6
                                          H_ME-DS10193_7-DS10193_7
4135
4136
                                   NA
                                                                 NA
4137
                                   NA
                                                                 NA
4138
         H_ME-DS10707_4-DS10707_4.2
                                          H_ME-DS10707_6-DS10707_6
4139
                       dx
           sex
     <numeric> <numeric>
1
             1
                        1
2
                        0
             1
                        0
3
             2
             2
4
                        1
5
             1
                        0
6
             2
                        1
7
             2
                        1
8
             1
                        0
9
             2
                        0
. . .
            . . .
4131
             1
                        1
4132
                        0
             1
             2
                        0
4133
4134
             2
                        1
                        0
4135
             1
             2
                        0
4136
             1
                        0
4137
4138
             1
                        1
             2
                        0
4139
> gr
```

GRanges with 511 ranges and 0 elementMetadata cols:

```
seqnames
                            ranges strand
                  <Rle>
                        <IRanges>
                                    <Rle>
                            [1, 1]
chr14:54383433
                  chr14
chr14:54383470
                  chr14
                            [2, 2]
chr14:54383819
                  chr14
                            [3, 3]
                            [4, 4]
chr14:54383945
                  chr14
                            [5, 5]
chr14:54384030
                  chr14
chr14:54384232
                  chr14
                            [6, 6]
chr14:54384251
                  chr14
                            [7, 7]
chr14:54384291
                  chr14
                            [8, 8]
                            [9, 9]
chr14:54384382
                  chr14
                   . . .
chr14:54443495
                  chr14 [503, 503]
                  chr14 [504, 504]
chr14:54443499
chr14:54443514
                  chr14 [505, 505]
chr14:54443831
                  chr14 [506, 506]
chr14:54443880
                  chr14 [507, 507]
chr14:54444016
                  chr14 [508, 508]
chr14:54444124
                  chr14 [509, 509]
chr14:54444385
                  chr14 [510, 510]
chr14:54444750
                  chr14 [511, 511]
seqlengths:
 chr14
   NA
```

4.2 SummarizedExperiment

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

4.3 SNPTrioExperiment

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

```
> ped <- PedClass(ped.DF)
> ste <- SNPTrioExperiment(se, pedigree = ped)

Here is the show method.
> ste

class: SNPTrioExperiment
dim: 511 968
exptData(0):
assays(1): geno
rownames(511): chr14:54383433 chr14:54383470 ...
chr14:54444385 chr14:54444750
```

rowData values names(0):
colnames(968): H_ME-CSc175_1-CSc175_1_a.1
 H_ME-CSc175_2-CSc175_2 ... H_ME-DS11418_2-DS11418_3
 H_ME-DS11418_3-DS11418_3
colData names(28): vcfid jb_famid ... id ped

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

> getClass("SNPTrioExperiment")

pedigree(4139): famid id fid mid sex dx

Class "SNPTrioExperiment" [package "trioClasses"]

Slots:

Name: pedigree exptData Class: PedClass SimpleList

Name: rowData colData Class: GenomicRangesORGRangesList DataFrame

Name: assays Class: SimpleList

Extends: "SummarizedExperiment"

4.4 PedClass

Now we investigate the pedigree slot of the SNPTrioExperiment 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>
         4778
                        H_ME-4778_1-4778_1.2
1
2
         4778
                        H_ME-4778_2-4778_2.2
3
                        H_ME-4778_3-4778_3.2
         4778
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
     DS99998
                    H_ME-DS10193_6-DS10193_6
4135
4136 DS99998
                    H_ME-DS10193_7-DS10193_7
     DS99999
                  H_ME-DS10707_4-DS10707_4.2
4137
4138
     DS99999
                    H_ME-DS10707_5-DS10707_5
4139
     DS99999
                    H_ME-DS10707_6-DS10707_6
                                 fid
                                                               mid
                            <factor>
                                                          <factor>
               H_ME-4778_2-4778_2.2
                                             H_ME-4778_3-4778_3.2
1
2
                                  NA
                                                                NΑ
3
                                  NA
                                                                NA
4
               H_ME-4783_2-4783_2.1
                                             H_ME-4783_3-4783_3.1
5
                                  NA
6
                                  NΑ
7
     H_ME-20000492_2-20000492_2_a.1 H_ME-20000492_3-20000492_3.1
8
                                  NΑ
                                                                NΑ
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
                                  NA
                                                                NA
4136
4137
                                  NA
                                                                NA
4138
         H_ME-DS10707_4-DS10707_4.2
                                         H_ME-DS10707_6-DS10707_6
4139
                                  NA
                                                                NA
```

sex

<factor> <factor>

dx

```
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
         2
4134
4135
         1
         2
4136
                 0
4137
         1
                  0
4138
          1
                 1
          2
4139
```

```
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 "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 elementMetadata cols:

	0			
	seqnames	ranges	strand	
	<rle></rle>	<pre><iranges></iranges></pre>	<rle></rle>	
chr14:54383433	chr14	[1, 1]	*	
chr14:54383470	chr14	[2, 2]	*	
chr14:54383819	chr14	[3, 3]	*	
chr14:54383945	chr14	[4, 4]	*	
chr14:54384030	chr14	[5, 5]	*	
chr14:54384232	chr14	[6, 6]	*	
chr14:54384251	chr14	[7, 7]	*	
chr14:54384291	chr14	[8, 8]	*	
chr14:54384382	chr14	[9, 9]	*	
chr14:54443495	chr14	[503, 503]	*	
chr14:54443499	chr14	[504, 504]	*	
chr14:54443514		[505, 505]		
chr14:54443831		[506, 506]		
chr14:54443880		[507, 507]		
chr14:54444016	chr14	[508, 508]	*	
chr14:54444124		[509, 509]		
chr14:54444385		[510, 510]		
chr14:54444750		[511, 511]		
		- , -		
seqlengths:				
chr14				
NA				

NA

4.7 ColData

5 Validity