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

2.1 Pedigree

> ped <- PedClass(ped.DF)

2.2 SNPTrioExperiment

```
> 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
colData names(28): vcfid jb_famid ... id ped
pedigree(4139): famid id fid mid sex dx
```

> ste <- SNPTrioExperiment(se, pedigree = ped)

3 Methods

3.1 Holger-style Genotype Matrix

Coercion to matrix for trio

```
> geno <- as(ste, "matrix")
```

Or apply the aTDT method to the SNPTrioExperiment directly.

> aTDT(ste)

```
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0 91

0 47

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                                                               5
                                                                       16
                                                                             0
                               0 154
                                                                             0
 [33]
        2 144
                  0 146 148
                                        5 159
                                                  2
                                                      2
                                                          0 141
                                                                        0
                                                                   0
 [49] 151
             1
                  3
                      1
                           1
                               0
                                    1
                                        1
                                             1
                                                  0
                                                     13
                                                          2
                                                               1
                                                                   1
                                                                        0
                                                                             2
        0
             2
                  2
                     46
                              45
                                    0
                                        2
                                            45
                                                          3 161
                                                                   3
                                                                        0
                                                                             0
 [65]
                           1
                                                 1
                                                      1
 [81]
       47
             1 154
                                   46
                                       46
                                                 0
                                                      0
                                                          0
                                                              45
                                                                        0
                      1
                           4
                               1
                                             1
                                                                   1
                                                                             1
 [97]
                                                               0
        0
             0
                  1
                      1
                           3
                               1
                                    0
                                        2
                                            73
                                                24
                                                     71
                                                          0
                                                                   0 120
                                                                             1
[113]
       14
             1
                 1
                      0
                          19
                               2
                                    1
                                        8
                                             3 121
                                                      1
                                                          1 127 161
                                                                        1
                                                                             5
                               0
                                        0
                                             0
                                                                             2
[129]
       25
             2
                48
                      0
                          1
                                    1
                                                 0 156
                                                          0
                                                               0
                                                                   1
                                                                        3
[145]
       49
             0
                  0
                     10
                           2
                               5
                                    1
                                        0
                                             0
                                                 1
                                                      0
                                                          1
                                                               2
                                                                  99
                                                                        2
                                                                             2
             2
                               2
[161]
                      1
                           0
                                    0
                                        0
                                            10 153
                                                          9
                                                               2
                                                                   4
                                                                             0
        0
                  1
                                                      1
                                                                        0
        2 154 155
                      2
                               2 162
                                                      2 159
                                                               2
[177]
                           1
                                        6
                                             1
                                                 1
                                                                  45
                                                                        1
                                                                           47
                                  63
                                        2
                                             1
                                                  3
[193]
        1
            65
                  0
                     62
                           0
                               0
                                                     50
                                                          0
                                                               4
                                                                   1
                                                                        0
                                                                             0
[209]
        2
                      2
                           0
                               6
                                   69
                                        4 164 168
                                                      0 155
                                                                  13 178
             1
                  1
                                                               1
                                                                             1
[225]
        0
             0
                  1
                     12
                           2
                               2 162
                                       67
                                             2
                                                 0
                                                      4
                                                         62
                                                               2
                                                                  13
                                                                       12
                                                                             9
        1 176
                     43
                                   29
                                             0 146
                                                          0
                                                               0
                                                                        0 180
[241]
                           1
                               1
                                        1
                                                      6
                                                                   1
                  1
[257]
        2
             1
                  1
                      1
                           0
                              13
                                    1
                                        0 143 175
                                                      2
                                                               1
                                                                             1
[273]
                                    2
                                       57
                                                                           30
             0
                  8
                      9
                               1
                                             1
                                                  6 157
                                                          3
                                                               1
        1
                           1
                                                                   1
                                                                        1
[289]
        0
             0 149
                     13
                           6
                               1
                                    2
                                       15
                                             2
                                                20 146
                                                          1
                                                              32
                                                                   1
                                                                        1
                                                                             0
[305] 149
             0
                 1
                      2 171
                               0
                                    2
                                        2
                                             1
                                                 1
                                                     12
                                                         21
                                                               2
                                                                   2
                                                                        2
                                                                             0
[321] 150 144
                14
                     17
                           2
                               2
                                   42
                                       16
                                            20
                                                  2
                                                      5
                                                         22
                                                               1
                                                                   1
                                                                             8
                                                      2
[337]
         1 177
                  0
                               3
                                    2
                                        0
                                                          0
                      1
                           2
                                            55
                                                 0
                                                               1
                                                                   0
                                                                             0
                                                                        1
[353]
             2
                55
                      2 179
                               0
                                    1
                                        1
                                             4
                                                42
                                                      1
                                                         53
                                                               1
                                                                  36
                                                                        2
                                                                             0
        1
                      0 121
                                        0
                                             0
                                                                  36
                                                                           37
[369]
         1
            56
                 1
                              94
                                    1
                                                 1
                                                      0
                                                          1
                                                               0
                                                                        0
[385]
        2
            25
                  2
                     30
                           0
                               7
                                    0
                                        2
                                             2
                                                 1
                                                      4 146
                                                              21
                                                                   1
                                                                        7
                                                                            1
[401]
        2
             5
                 7
                      2
                               5
                                    3
                                                10
                                                         57
                                                              29
                           1
                                        1
                                             1
                                                      1
                                                                   1
                                                                        1
                                                                             1
[417]
        2
             5
                  0
                      0
                               2
                                    0
                                        0
                                             0
                                                 0
                                                      0
                                                         30
                                                               1
                                                                   2
                                                                        0
                                                                             2
                          1
                           0
                              9
                                        2
                                             0
                                                      2
                                                              0
                                                                             2
[433]
        1
            14
                  3
                     31
                                    1
                                               11
                                                         49
                                                                   0
                                                                        1
                      2
[449]
        4
            1
                           3
                              0
                                    1
                                        2
                                             8
                                                 2
                                                      0
                                                          0
                                                              24
                                                                   2
                                                                       0
                                                                           13
                 1
[465]
        3
            55
                 0
                      2
                           0
                              10
                                    3
                                        1
                                             1
                                                 1
                                                      2
                                                          2
                                                              1
                                                                    4
                                                                       13
                                                                             1
[481]
         1
             0
                20
                      1
                           8
                               5
                                    0
                                        1
                                             1
                                                 1
                                                      1
                                                          2
                                                               5
                                                                   0
                                                                        7
                                                                             2
                12
                                                      1
[497]
             1
                      0
                           1
                              87
                                    0
                                        1
                                             1
                                                           1
                                                              65
                                                                    1 116
```

3.2 Count of Transmission of Rare Variants (TransCount())

3.2.1 In a block

```
> TransCount(ste[MAF(ste) <= 0.01, ], block)
[1] 26
3.2.2 Not In the block
> setdiff(world, block)
GRanges with 2 ranges and 0 elementMetadata cols:
      seqnames
                             ranges strand
         <Rle>
                          <IRanges> <Rle>
  [1]
         chr14 [54383433, 54395801]
  [2]
         chr14 [54405802, 54444750]
  seqlengths:
   chr14
     NA
> TransCount(ste[MAF(ste) <= 0.01, ], setdiff(world, block))
[1] 116
3.2.3 In the "world"
> world
GRanges with 1 range and 0 elementMetadata cols:
      seqnames
                             ranges strand
         <Rle>
                          <IRanges> <Rle>
  [1]
         chr14 [54383433, 54444750]
  seqlengths:
   chr14
     NA
> TransCount(ste[MAF(ste) <= 0.01, ], world)
[1] 142
```

4 Classes

4.1 SnpMatrix, DataFrame, GRanges

A SnpMatrix with 968 rows and 511 columns

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.

H_ME-DS10776_2-DS10776_2 ... H_ME-DS11313_1-DS11313_1

```
> sm
```

4133

Row 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
                            H_ME-4783_3-4783_3.1
            4783
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
         DS12332
                        H_ME-DS12332_2-DS12332_2
4132
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
                        H_ME-DS10707_5-DS10707_5
4138
         DS99999
4139
         DS99999
                        H_ME-DS10707_6-DS10707_6
                                                                mid
                         <character>
                                                        <character>
1
               H_ME-4778_2-4778_2.2
                                              H_ME-4778_3-4778_3.2
2
                                  NA
                                                                 NA
3
                                  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
                                                                 NΑ
9
                                  NA
                                                                 NA
           H_ME-DS12332_2-DS12332_2
                                          H_ME-DS12332_3-DS12332_3
4131
4132
                                  NA
                                                                 NA
```

NA

NA

4134 4135 4136 4137	H_M	E-DS10193_6	S-DS10193_6 NA NA NA	H_ME-DS10193_7-DS10193_7 NA NA NA
4138	H_ME-DS10707_4-DS10707_4.2			H_ME-DS10707_6-DS10707_6
4139			NA	NA
	sex	dx		
	<numeric></numeric>	<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 elementMetadata cols:

strand	ranges		seqnames	
<rle></rle>	<pre><iranges></iranges></pre>		<rle></rle>	
*	54383433]	[54383433,	chr14	chr14:54383433
*	54383470]	[54383470,	chr14	chr14:54383470
*	54383819]	[54383819,	chr14	chr14:54383819
*	54383945]	[54383945,	chr14	chr14:54383945
*	54384030]	[54384030,	chr14	chr14:54384030
*	54384232]	[54384232,	chr14	chr14:54384232
*	54384251]	[54384251,	chr14	chr14:54384251
*	54384291]	[54384291,	chr14	chr14:54384291
*	54384382]	[54384382,	chr14	chr14:54384382
*	54443495]	[54443495,	chr14	chr14:54443495
*	54443499]	[54443499,	chr14	chr14:54443499
*	54443514]	[54443514,	chr14	chr14:54443514
*	54443831]	[54443831,	chr14	chr14:54443831
*	54443880]	[54443880,	chr14	chr14:54443880
*	54444016]	[54444016,	chr14	chr14:54444016
*	54444124]	[54444124,	chr14	chr14:54444124
*	54444385]	[54444385,	chr14	chr14:54444385
*	54444750]	[54444750,	chr14	chr14:54444750

```
seqlengths:
chr14
```

Name:

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)</pre>
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_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("SNPTrioExperiment")
Class "SNPTrioExperiment" [package "trioClasses"]
Slots:
Name:
                          pedigree
                                                      exptData
                                                    SimpleList
Class:
                          PedClass
Name:
                           rowData
                                                       colData
Class: GenomicRangesORGRangesList
                                                     DataFrame
```

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: 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>	<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>
                                             H_ME-4778_3-4778_3.2
               H_ME-4778_2-4778_2.2
1
2
                                  NA
3
                                  NA
                                                                NA
                                             H_ME-4783_3-4783_3.1
4
               H_ME-4783_2-4783_2.1
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
4134
           H_ME-DS10193_6-DS10193_6
                                        H_ME-DS10193_7-DS10193_7
4135
                                  NA
4136
                                  NA
                                                                NA
4137
                                  NA
                                                                NA
4138
         H_ME-DS10707_4-DS10707_4.2
                                         H_ME-DS10707_6-DS10707_6
4139
          sex
                    dx
     <factor> <factor>
1
            1
2
            1
                     0
3
            2
                     0
            2
4
                     1
5
            1
                     0
            2
6
                     1
            2
7
8
            1
                     0
            2
9
                     0
4131
           1
                     1
4132
            1
                     0
4133
            2
                     0
            2
4134
                     1
4135
            1
            2
                     0
4136
            1
                     0
4137
            1
4138
                     1
            2
                     0
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:

	seqnames		ranges	strand
	<rle></rle>		<pre><iranges></iranges></pre>	<rle></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