

AshkenazimSonChr21: Annotated variants on the chromosome 21, human genome 19, Ashkenazim Trio son sample

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May 7, 2019

Introduction

This vignette describes AshkenazimSonChr21 dataset, example input for RareVariantVis package. This dataset is CompleteGenomics whole genome sequencing dataset, coming from Stanford Genome in a Bottle Consortium. This dataset was made fully available for public, without restrictions. This particular data refer to sample HG002- NA24385 - huAA53E0 (son). Original data can be found at: <https://sites.stanford.edu/abms/content/giab-reference-materials-and-data>

Preprocessing

Original whole genome sequencing sample was (HG002-son) was too big for purpose of R/Bioconductor test data, therefore only chromosome 21 variants were selected. Complete Genomics output provides 3 types of variants: homozygous reference, heterozygous and homozygous alternative. To minimize data size and make it similar to Illumina X Ten output homozygous reference were excluded. Finally, small indels were filtered out, since they introduced a lot of noise into visualization. This noise was not observed in Illumina X Ten samples that we analyzed in our laboratory.

Possible usage of data

Data aims to work well with RareVariantVis package, however it can be used also in other packages that aim for whole genome sequencing data analysis. Dataset includes two types of files: txt file with rare variants and vcf file obtained from sequencing, very similar to one from Illumina X Ten output. Examples of data usage and file structure are listed below.

```
## text file
library(AshkenazimSonChr21)
head(SonVariantsChr21)

##   Chromosome Start.position End.position Reference Variant
## 1         chr21      9411318     9411318          C         T
```

```

## 2      chr21      9411327      9411327      C      G
## 3      chr21      9411410      9411410      C      T
## 4      chr21      9411500      9411500      G      T
## 5      chr21      9411602      9411602      T      C
## 6      chr21      9411609      9411609      G      T
##      Quality.by.Depth Variant.type      SNP.id SNP.Frequency Gene.name
## 1              313.61 Substitution rs373567667      -1
## 2              720.44 Substitution rs75025155      -1
## 3             1128.86 Substitution rs78200054      -1
## 4             1241.14 Substitution rs71235073      -1
## 5              615.72 Substitution rs368646645      -1
## 6              603.02 Substitution rs76676778      -1
##      Gene.component phyloP DP      AD  GT
## 1              -0.177 38 25,13 0/1
## 2              -0.307 37 13,24 0/1
## 3               0.717 49 15,34 0/1
## 4               0.717 62 24,38 0/1
## 5               0.624 57 35,22 0/1
## 6              -0.163 56 35,21 0/1

## vcf file
library(VariantAnnotation)

## Loading required package: BiocGenerics
## Loading required package: parallel
##
## Attaching package: 'BiocGenerics'
## The following objects are masked from 'package:parallel':
##
##      clusterApply, clusterApplyLB, clusterCall, clusterEvalQ,
##      clusterExport, clusterMap, parApply, parCapply, parLapply,
##      parLapplyLB, parRapply, parSapply, parSapplyLB
## The following objects are masked from 'package:stats':
##
##      IQR, mad, sd, var, xtabs
## The following objects are masked from 'package:base':
##
##      Filter, Find, Map, Position, Reduce, anyDuplicated, append,
##      as.data.frame, basename, cbind, colnames, dirname, do.call,
##      duplicated, eval, evalq, get, grep, grepl, intersect,
##      is.unsorted, lapply, mapply, match, mget, order, paste, pmax,
##      pmax.int, pmin, pmin.int, rank, rbind, rounames, sapply,
##      setdiff, sort, table, tapply, union, unique, unsplit, which,
##      which.max, which.min
## Loading required package: GenomeInfoDb
## Loading required package: S4Vectors
## Loading required package: stats4
##
## Attaching package: 'S4Vectors'

```

```

## The following object is masked from 'package:base':
##
##   expand.grid
## Loading required package: IRanges
## Loading required package: GenomicRanges
## Loading required package: SummarizedExperiment
## Loading required package: Biobase
## Welcome to Bioconductor
##
##   Vignettes contain introductory material; view with
##   'browseVignettes()'. To cite Bioconductor, see
##   'citation("Biobase)", and for packages 'citation("pkgname)".
## Loading required package: DelayedArray
## Loading required package: matrixStats
##
## Attaching package: 'matrixStats'
## The following objects are masked from 'package:Biobase':
##
##   anyMissing, rowMedians
## Loading required package: BiocParallel
##
## Attaching package: 'DelayedArray'
## The following objects are masked from 'package:matrixStats':
##
##   colMaxs, colMins, colRanges, rowMaxs, rowMins, rowRanges
## The following objects are masked from 'package:base':
##
##   aperm, apply, rowsum
## Loading required package: Rsamtools
## Loading required package: Biostrings
## Loading required package: XVector
##
## Attaching package: 'Biostrings'
## The following object is masked from 'package:DelayedArray':
##
##   type
## The following object is masked from 'package:base':
##
##   strsplit
##
## Attaching package: 'VariantAnnotation'
## The following object is masked from 'package:base':
##
##   tabulate

fl <- system.file("extdata", "SonVariantsChr21.vcf.gz",
                  package="AshkenazimSonChr21")
vcf <- readVcf(fl, genome="hg19")
geno(vcf)

```

```

## List of length 8
## names(8): GT GQX AD DP GQ MQ PL VF

info(vcf)

## DataFrame with 94527 rows and 35 columns
##           AC          AF          AN          DP          QD
##   <IntegerList> <character> <integer> <integer> <numeric>
## 1           1          0.50           2           38          8.25
## 2           1          0.50           2           37          19.47
## 3           1          0.50           2           49          23.04
## 4           1          0.50           2           62          20.02
## 5           1          0.50           2           57          10.8
## ...           ...           ...           ...           ...           ...
## 94523        1          0.50           2          101          2.04
## 94524        1          0.50           2          113          2.12
## 94525        1          0.50           2          115          2.01
## 94526        1          0.50           2          155          0.14
## 94527        1          0.50           2          169          0.02
##   BLOCKAVG_min30p3a BaseQRankSum          DS          Dels          END
##   <logical>          <numeric> <logical> <numeric> <integer>
## 1           FALSE          -0.923    FALSE          0          NA
## 2           FALSE          -0.334    FALSE          0          NA
## 3           FALSE          -0.683    FALSE          0          NA
## 4           FALSE          1.395    FALSE          0          NA
## 5           FALSE          -1.436    FALSE          0          NA
## ...           ...           ...           ...           ...           ...
## 94523        FALSE          1.834    FALSE          0.01          NA
## 94524        FALSE          2.439    FALSE          0.06          NA
## 94525        FALSE          1.499    FALSE          0.01          NA
## 94526        FALSE          1.67     FALSE          0          NA
## 94527        FALSE          1.448    FALSE          0.01          NA
##           FS          HRun HaplotypeScore InbreedingCoeff          MQ
##   <numeric> <integer>          <numeric>          <numeric> <numeric>
## 1           0           0           1.9783           NA          51
## 2          1.443           1           0.9995           NA          52
## 3          11.788           1           0.8667           NA          50
## 4           1.005           0           0           NA          52
## 5           0           0           0           NA          53
## ...           ...           ...           ...           ...           ...
## 94523         0           1          128.0372           NA          25
## 94524         0           1          205.8792           NA          24
## 94525         0           1          250.5937           NA          22
## 94526         6.16           0          184.0491           NA          19
## 94527         2.884           3          195.0513           NA          18
##           MQ0 MQRankSum ReadPosRankSum          SB          VQSLOD
##   <integer> <numeric>          <numeric> <numeric> <numeric>
## 1           0          -0.031          -0.154          -55.94          2.0206
## 2           0           0.016           0.97          -261.36          4.3216
## 3           0          -0.597          -0.011          -414.78          2.9995

```

```

## 4          0      1.322      -1.192    -535.11      2.156
## 5          6      0.086       0.276   -178.59      2.1432
## ...      ...      ...      ...      ...      ...
## 94523      3     -3.844      -0.805    -88.65    -27.4198
## 94524      4     -1.997      -1.33    -89.77    -60.7511
## 94525      5     -3.745      -0.59   -110.6    -89.2046
## 94526     37     -1.952       3.132     -0.01   -63.3093
## 94527     56     -1.775       2.138     -0.01   -70.4434
##          culprit          set          CSQT
##          <character> <character> <CharacterList>
## 1          QD FilteredInAll          NA
## 2          MQ          variant          NA
## 3          MQ FilteredInAll          NA
## 4          MQ FilteredInAll          NA
## 5          QD FilteredInAll          NA
## ...      ...      ...      ...
## 94523 HaplotypeScore FilteredInAll          NA
## 94524 HaplotypeScore FilteredInAll          NA
## 94525 HaplotypeScore FilteredInAll          NA
## 94526          DP FilteredInAll          NA
## 94527          DP FilteredInAll          NA
##          CSQR          AA
##          <CharacterList> <character>
## 1          NA          NA
## 2          NA          NA
## 3          NA          NA
## 4          NA          NA
## 5          NA          NA
## ...      ...      ...
## 94523 ENSR00000684572|regulatory_region_variant          NA
## 94524 ENSR00000684572|regulatory_region_variant          NA
## 94525 ENSR00000684572|regulatory_region_variant          NA
## 94526 ENSR00000684572|regulatory_region_variant          NA
## 94527 ENSR00000684572|regulatory_region_variant          NA
##          GMAF          EVS          cosmic          clinvar
##          <CharacterList> <CharacterList> <CharacterList> <CharacterList>
## 1          NA          NA          NA          NA
## 2          NA          NA          NA          NA
## 3          NA          NA          NA          NA
## 4          NA          NA          NA          NA
## 5          NA          NA          NA          NA
## ...      ...      ...      ...
## 94523      NA          NA          NA          NA
## 94524      NA          NA          NA          NA
## 94525      NA          NA          NA          NA
## 94526      NA          NA          NA          NA
## 94527      NA          NA          NA          NA
##          phastCons      Variant.type      Gene.name      Gene.component      phyloP
##          <logical> <CharacterList> <CharacterList> <CharacterList> <numeric>

```

```

## 1      FALSE      Substitution      NA      NA      -0.177
## 2      FALSE      Substitution      NA      NA      -0.307
## 3      FALSE      Substitution      NA      NA      0.717
## 4      FALSE      Substitution      NA      NA      0.717
## 5      FALSE      Substitution      NA      NA      0.624
## ...      ...      ...      ...      ...      ...
## 94523    FALSE      Substitution      NA      NA      -100
## 94524    FALSE      Substitution      NA      NA      -100
## 94525    FALSE      Substitution      NA      NA      -100
## 94526    FALSE      Substitution      NA      NA      -100
## 94527    FALSE      Substitution      NA      NA      -100
##          SNP.Frequency
##          <numeric>
## 1          -1
## 2          -1
## 3          -1
## 4          -1
## 5          -1
## ...      ...
## 94523      -1
## 94524      -1
## 94525      -1
## 94526      -1
## 94527      -1

```