

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

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## 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 Quality.by.Depth
## 1 chr21 9411318 9411318 C T 313.61
```

```

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

## vcf file
library(VariantAnnotation)

## Loading required package: BiocGenerics
##
## Attaching package: 'BiocGenerics'
## 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, rownames, sapply, setdiff, sort, table,
##      tapply, union, unique, unsplit, which.max, which.min
## Loading required package: MatrixGenerics
## Loading required package: matrixStats
##
## Attaching package: 'MatrixGenerics'
## The following objects are masked from 'package:matrixStats':
##
##      colAlls, colAnyNAs, colAnys, colAugsPerRowSet, colCollapse,
##      colCounts, colCummaxs, colCummins, colCumprods, colCumsums,
##      colDiffs, colIQRDiffs, colIQRs, colLogSumExps, colMadDiffs,
##      colMads, colMaxs, colMeans2, colMedians, colMins, colOrderStats,
##      colProds, colQuantiles, colRanges, colRanks, colSdDiffs, colSds,
##      colSums2, colTabulates, colVarDiffs, colVars, colWeightedMads,
##      colWeightedMeans, colWeightedMedians, colWeightedSds,

```

```

##      colWeightedVars, rowAlls, rowAnyNAs, rowAnys, rowAugsPerColSet,
##      rowCollapse, rowCounts, rowCummaxs, rowCummins, rowCumprods,
##      rowCumsums, rowDiffs, rowIQRDiffs, rowIQRs, rowLogSumExps,
##      rowMadDiffs, rowMads, rowMaxs, rowMeans2, rowMedians, rowMins,
##      rowOrderStats, rowProds, rowQuantiles, rowRanges, rowRanks,
##      rowSdDiffs, rowSds, rowSums2, rowTabulates, rowVarDiffs, rowVars,
##      rowWeightedMads, rowWeightedMeans, rowWeightedMedians,
##      rowWeightedSds, rowWeightedVars
## Loading required package: GenomeInfoDb
## Loading required package: S4Vectors
## Loading required package: stats4
##
## Attaching package: 'S4Vectors'
## The following objects are masked from 'package:base':
##
##      I, expand.grid, unname
## 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)".
##
## Attaching package: 'Biobase'
## The following object is masked from 'package:MatrixGenerics':
##
##      rowMedians
## The following objects are masked from 'package:matrixStats':
##
##      anyMissing, rowMedians
## Loading required package: Rsamtools
## Loading required package: Biostrings
## Loading required package: XVector
##
## Attaching package: 'Biostrings'
## 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 BLOCKAVG_min30p3a
##           <IntegerList> <character> <integer> <integer> <numeric>           <logical>
## 1           1           0.50           2           38           8.25           FALSE
## 2           1           0.50           2           37           19.47          FALSE
## 3           1           0.50           2           49           23.04          FALSE
## 4           1           0.50           2           62           20.02          FALSE
## 5           1           0.50           2           57           10.80          FALSE
## ...           ...           ...           ...           ...           ...           ...
## 94523        1           0.50           2           101          2.04           FALSE
## 94524        1           0.50           2           113          2.12           FALSE
## 94525        1           0.50           2           115          2.01           FALSE
## 94526        1           0.50           2           155          0.14           FALSE
## 94527        1           0.50           2           169          0.02           FALSE
##           BaseQRankSum           DS           Dels           END           FS           HRun
##           <numeric> <logical> <numeric> <integer> <numeric> <integer>
## 1           -0.923           FALSE           0           NA           0.000           0
## 2           -0.334           FALSE           0           NA           1.443           1
## 3           -0.683           FALSE           0           NA           11.788          1
## 4           1.395           FALSE           0           NA           1.005           0
## 5           -1.436           FALSE           0           NA           0.000           0
## ...           ...           ...           ...           ...           ...           ...
## 94523        1.834           FALSE           0.01           NA           0.000           1
## 94524        2.439           FALSE           0.06           NA           0.000           1
## 94525        1.499           FALSE           0.01           NA           0.000           1
## 94526        1.670           FALSE           0.00           NA           6.160           0
## 94527        1.448           FALSE           0.01           NA           2.884           3
##           HaplotypeScore InbreedingCoeff           MQ           MQO MQRankSum
##           <numeric>           <numeric> <numeric> <integer> <numeric>
## 1           1.9783           NA           51           0           -0.031
## 2           0.9995           NA           52           0           0.016
## 3           0.8667           NA           50           0           -0.597
## 4           0.0000           NA           52           0           1.322
## 5           0.0000           NA           53           6           0.086
## ...           ...           ...           ...           ...           ...
## 94523        128.037           NA           25           3           -3.844
## 94524        205.879           NA           24           4           -1.997
## 94525        250.594           NA           22           5           -3.745
## 94526        184.049           NA           19           37          -1.952
## 94527        195.051           NA           18           56          -1.775
##           ReadPosRankSum           SB           VQSLOD           culprit           set
##           <numeric> <numeric> <numeric> <character> <character>

```

```

## 1      -0.154   -55.94    2.0206           QD FilteredInAll
## 2       0.970  -261.36   4.3216           MQ      variant
## 3      -0.011  -414.78   2.9995           MQ FilteredInAll
## 4      -1.192  -535.11   2.1560           MQ FilteredInAll
## 5       0.276  -178.59   2.1432           QD FilteredInAll
## ...      ...      ...      ...      ...      ...
## 94523   -0.805   -88.65  -27.4198 HaplotypeScore FilteredInAll
## 94524   -1.330   -89.77  -60.7511 HaplotypeScore FilteredInAll
## 94525   -0.590  -110.60  -89.2046 HaplotypeScore FilteredInAll
## 94526     3.132    -0.01  -63.3093           DP FilteredInAll
## 94527     2.138    -0.01  -70.4434           DP FilteredInAll
##           CSQT           CSQR           AA           GMAF
##           <CharacterList>           <CharacterList> <character> <CharacterList>
## 1           NA
## 2           NA
## 3           NA
## 4           NA
## 5           NA
## ...      ...      ...      ...      ...
## 94523           ENSR00000684572|regu..           NA
## 94524           ENSR00000684572|regu..           NA
## 94525           ENSR00000684572|regu..           NA
## 94526           ENSR00000684572|regu..           NA
## 94527           ENSR00000684572|regu..           NA
##           EVS           cosmic           clinvar phastCons           Variant.type
##           <CharacterList> <CharacterList> <CharacterList> <logical> <CharacterList>
## 1           FALSE           Substitution
## 2           FALSE           Substitution
## 3           FALSE           Substitution
## 4           FALSE           Substitution
## 5           FALSE           Substitution
## ...      ...      ...      ...      ...
## 94523           FALSE           Substitution
## 94524           FALSE           Substitution
## 94525           FALSE           Substitution
## 94526           FALSE           Substitution
## 94527           FALSE           Substitution
##           Gene.name Gene.component           phyloP SNP.Frequency
##           <CharacterList> <CharacterList> <numeric>           <numeric>
## 1           -0.177           -1
## 2           -0.307           -1
## 3           0.717           -1
## 4           0.717           -1
## 5           0.624           -1
## ...      ...      ...      ...
## 94523           -100           -1
## 94524           -100           -1
## 94525           -100           -1
## 94526           -100           -1

```

## 94527

-100

-1