

# 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
## 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, xtabs
##
## The following objects are masked from 'package:base':
##
##   Filter, Find, Map, Position, Reduce, anyDuplicated, append,
##   as.data.frame, as.vector, cbind, colnames, do.call,
##   duplicated, eval, evalq, get, grep, grepl, intersect,
##   is.unsorted, lapply, lengths, mapply, match, mget, order,
##   paste, pmax, pmax.int, pmin, pmin.int, rank, rbind, rownames,
##   sapply, setdiff, sort, table, tapply, union, unique, unlist,
##   unsplit
##
## Loading required package: GenomeInfoDb
## Loading required package: stats4

```

```

## Loading required package: S4Vectors
## 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: Rsamtools
## Loading required package: XVector
## Loading required package: Biostrings
##
## 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)

info(vcf)

```

```

END
er>
NA
NA
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      1.670      FALSE      0.00     NA
## 94527      FALSE      1.448      FALSE      0.01     NA
##           FS      HRun HaplotypeScore InbreedingCoeff      MQ
##           <numeric> <integer>      <numeric>      <numeric> <numeric>
## 1           0.000      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      0.0000      NA      52
## 5           0.000      0      0.0000      NA      53
## ...          ...          ...          ...          ...
## 94523      0.000      1      128.0372      NA      25
## 94524      0.000      1      205.8792      NA      24
## 94525      0.000      1      250.5937      NA      22
## 94526      6.160      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      0.970      -261.36      4.3216
## 3           0      -0.597      -0.011      -414.78      2.9995
## 4           0      1.322      -1.192      -535.11      2.1560
## 5           6      0.086      0.276      -178.59      2.1432
## ...          ...          ...          ...          ...
## 94523      3      -3.844      -0.805      -88.65      -27.4198
## 94524      4      -1.997      -1.330      -89.77      -60.7511
## 94525      5      -3.745      -0.590      -110.60      -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
## 2           MQ      variant
## 3           MQ FilteredInAll
## 4           MQ FilteredInAll
## 5           QD FilteredInAll
## ...          ...          ...          ...
## 94523 HaplotypeScore FilteredInAll
## 94524 HaplotypeScore FilteredInAll
## 94525 HaplotypeScore FilteredInAll
## 94526      DP FilteredInAll
## 94527      DP FilteredInAll
##           CSQR      AA
##           <CharacterList> <character>

```

```

## 1 NA
## 2 NA
## 3 NA
## 4 NA
## 5 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
## 2
## 3
## 4
## 5
## ...          ...          ...          ...          ...
## 94523
## 94524
## 94525
## 94526
## 94527
##          phastCons      Variant.type      Gene.name      Gene.component      phyloP
##          <logical> <CharacterList> <CharacterList> <CharacterList> <numeric>
## 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
##          SNP.Frequency
##          <numeric>
## 1          -1
## 2          -1
## 3          -1
## 4          -1
## 5          -1
## ...          ...
## 94523          -1
## 94524          -1
## 94525          -1
## 94526          -1

```

## 94527

-1