

# 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, aperm, 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, 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 object is masked from 'package:utils':
##
##      findMatches
## 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      MQ0 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