Package 'VariantToolsData'

July 17, 2025

Type Package

Version 1.33.0

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Title Data for the VariantTools tutorial

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Description Data from the sequencing of a 50/50 mixture of HapMap trio samples
NA12878 (CEU) and NA19240 (YRI), subset to the TP53 region.

Suggests VariantTools (>= 1.3.4), gmapR (>= 1.3.3), BiocStyle

Imports BiocGenerics, GenomicRanges

Depends R (>= 2.10), VariantAnnotation (>= 1.7.35)

biocViews ExperimentData, SequencingData, HapMap, SNPData

git_url https://git.bioconductor.org/packages/VariantToolsData

git_branch devel

git_last_commit 34b6a15

git_last_commit_date 2025-04-15

Repository Bioconductor 3.22

Date/Publication 2025-07-17

Contents

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geno   *Reference genotypes*

Description

Reference genotypes for NA12878 and NA19240, as called by the HapMap project, and the Broad GATK project.

Usage

```
data(geno)
```

Format

A VRanges with the genotypes for NA12878 and NA19240 from the HapMap pilot project and the Broad/GATK calling of NA12878. The genotypes, stored in the "freq." columns, are represented by the alt frequency, so 0/0.5/1 for hom-ref/het/hom-alt. The "expected.freq" column indicates the alt frequency expected in the 50/50 mixture.

Source

The HapMap Pilot and Broad GATK projects.

Examples

```
data(geno)
table(geno$expected.freq)
```

---

repeats   *Simple repeats*

Description

Repeat regions from the RepeatMasker track of the hg19 UCSC genome browser database, subset to low complexity and simple repeats.

Usage

```
data(repeats)
```

Format

A GRanges object with the repeat ranges, including variables classifying the repeats by name, class, and family, and information about the alignment of the repeat consensus to the genome.

Source

The "rmsk" table in the UCSC table browser (hg19). Click the "Describe Schema" button for details.

Examples

```
data(repeats)
tab <- table(repeats$repFamily)
tab[tab > 0]
```

---

sumDepths	*Sum Replicate Depths*

Description

Finds the unique variants across every element of a list of VRanges, with depths computed by summing the depths over the elements. If a variant is not found in a sample, the depth is assumed to zero. That is a very dangerous assumption.

Usage

```
sumDepths(x)
```

Arguments

x   A VRangesList, typically of replicates

Value

A VRanges

Author(s)

Michael Lawrence

Examples

```
data(tallies)
sumDepths(tallies)
```

---

tallies	*Tally VRanges*

Description

Nucleotide tallies computed over the TP53 region (+/- 1Mb) for the 50/50 NA12878/NA19240 mixture, separately for each replicate. Each replicate corresponds to a separate biochemical mixing.

Usage

```
data(tallies)
```

Format

A VRangesList, each VRanges of which corresponds to one of the three biochemical replicates.

Source

Computed from the alignments of the FASTQ files found in the 'inst/extdata' directory. Repeat regions (see repeats) were excluded. For example, for one replicate,

```
library(gmapR)
extdata.dir <- system.file("extdata",
                           package="VariantToolsData")
bams <- BamFileList(tools::list_files_with_exts(extdata.dir, "bam"))
data(repeats, package = "VariantToolsData")
param <- TallyVariantsParam(TP53Genome(), mask = repeats,
                           read_pos=TRUE, read_length=75L)
tallies <- split(tallyVariants(bams, param), ~ sampleNames)
```

This assumes that the BAM files have been generated for the current version of the TP53 genome:

```
param <- GsnapParam(TP53Genome(), unique_only = TRUE,
                    molecule = "DNA")
first.fastq <- dir(extdata.dir, "first.fastq",
                   full.names=TRUE)
last.fastq <- dir(extdata.dir, "last.fastq",
                  full.names=TRUE)
output <- gsnap(first.fastq, last.fastq, param)
bams <- as(output, "BamFileList")
```

References

Lawrence, M., Huntley, M. A., Stawiski, E., Owen, A., Wu, T. D., Goldstein, L. D., Cao, Y., Degenhardt, J., Young, J., Guillory, J., Heldens, S., Jackson, M., Seshagiri S., and Gentleman, R. (2015). Genomic variant calling: Flexible tools and a diagnostic data set. bioRxiv.

Examples

```
data(tallies)
VariantTools::callVariants(tallies[[1L]])
```

TP53Region   *Range around TP53*

Description

Returns a GRanges object consisting of the TP53 coordinates in hg19. All coordinates in these data are relative to that region.

Usage

```
TP53Region()
```

Value

A GRanges of the extents of the TP53 gene in hg19.

Author(s)

Michael Lawrence

Examples

```
TP53Region()
```

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