

# Package ‘VariantToolsData’

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**Type** Package

**Version** 1.2.0

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**Title** Data for the VariantTools tutorial

**Author** Michael Lawrence

**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

**NeedsCompilation** no

## R topics documented:

|                      |   |
|----------------------|---|
| geno . . . . .       | 1 |
| repeats . . . . .    | 2 |
| sumDepths . . . . .  | 3 |
| tallies . . . . .    | 3 |
| TP53Region . . . . . | 4 |

|              |          |
|--------------|----------|
| <b>Index</b> | <b>6</b> |
|--------------|----------|

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|      |                            |
|------|----------------------------|
| geno | <i>Reference genotypes</i> |
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## 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)
```

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|         |                       |
|---------|-----------------------|
| repeats | <i>Simple repeats</i> |
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**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]
```

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|           |                             |
|-----------|-----------------------------|
| sumDepths | <i>Sum Replicate Depths</i> |
|-----------|-----------------------------|

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**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)
```

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|         |                      |
|---------|----------------------|
| tallies | <i>Tally VRanges</i> |
|---------|----------------------|

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**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]])
```

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TP53Region

*Range around TP53*

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**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.

*TP53Region*

5

**Author(s)**

Michael Lawrence

**Examples**

TP53Region()

# Index

## \*Topic **datasets**

- geno, [1](#)
- repeats, [2](#)
- tallies, [3](#)

geno, [1](#)

repeats, [2](#), [4](#)

sumDepths, [3](#)

tallies, [3](#)

TP53Region, [4](#)