

COSMIC 67

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1 Introduction

The *COSMIC.67* package provides the curated mutations published with the COSMIC release version 67 (2013-10-24). Both variants found in coding and non-coding regions are included and offered as a single object of class 'CollapsedVCF' and a bgzipped and tabix-index 'VCF' file.

Additionally, the package contains the Cancer Gene Census, a list of genes causally linked to cancer.

2 Accessing and Using the Data

`library(VariantAnnotation)`

Loading required package: BiocGenerics

Loading required package: parallel

Attaching package: 'BiocGenerics'

The following objects are masked from 'package:parallel':

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*clusterApply, clusterApplyLB, clusterCall,
clusterEvalQ, clusterExport, clusterMap, parApply,
parCapply, parLapply, parLapplyLB, parRapply,
parSapply, parSapplyLB*

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, colAvgPerRowSet,
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,
rowAvgPerColSet, 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':

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```
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

library(GenomicRanges)

data(package = "COSMIC.67")
data(cosmic_67, package = "COSMIC.67")

tp53_range = GRanges("17", IRanges(7565097, 7590856))
vcf_path = system.file("vcf", "cosmic_67.vcf.gz", package = "COSMIC.67")
cosmic_tp53 = readVcf(vcf_path, genome = "GRCh37", ScanVcfParam(which = tp53_range))
cosmic_tp53

class: CollapsedVCF
dim: 5892 0
rowRanges(vcf):
```

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```
GRanges with 5 metadata columns: paramRangeID, REF, ALT, QUAL, FILTER
info(vcf):
  DataFrame with 5 columns: GENE, STRAND, CDS, AA, CNT
info(header(vcf)):
  Number Type      Description
  GENE    1      String  Gene name
  STRAND  1      String  Gene strand
  CDS     1      String  CDS annotation
  AA      1      String  Peptide annotation
  CNT     1      Integer How many samples have this mutation
geno(vcf):
  List of length 0:
```

```
data(cgc_67, package = "COSMIC.67")
head(cgc_67)
```

	SYMBOL	ENTREZID	ENSEMBL
1	ABI1	10006	ENSG00000136754
2	ABL1	25	ENSG00000097007
3	ABL2	27	ENSG00000143322
4	ACSL3	2181	ENSG00000123983
5	CASC5	57082	ENSG00000137812
6	MLLT11	10962	ENSG00000213190

For details on the collection and curation of the original data, please see the webpage of the COSMIC project: <http://cancer.sanger.ac.uk/cancergenome/projects/cosmic/>.

3 Data Provenance

3.1 COSMIC Mutations

The following steps are performed for importing and processing of the VCF data:

1. Downloading of the VCF files 'CosmicCodingMuts_v67_20131024.vcf.gz' and 'Cosmic-NonCodingVariants_v67_20131024.vcf.gz' from 'ftp://ngs.sanger.ac.uk/production/cosmic/' to 'inst/raw/
2. Importing of both files to R using 'readVcf'.
3. Sorting of the seqlevels and adding 'seqinfo' data for the toplevel chromosomes of 'GRCh37'.
4. Merging of both objects, sorting according to genomic position.
5. Converting the object to class `VariantAnnotation::VRanges`.
6. Converting the 'character' columns to 'factors'.
7. Saving the merged object to 'data/cosmic_v67_vcf.rda'.
8. Exporting the merged object as a bgzipped and tabix-indexed 'VCF' to 'inst/vcf/cosmic_v67.vcf.gz'.

3.2 Cancer Gene Census

The following steps are performed for importing and processing of the Cancer Gene Census data:

1. Downloading of the 'cancer_gene_census.tsv' file from ftp://ftp.sanger.ac.uk/pub/CGP/cosmic/data_export to 'inst/raw'.
2. Import of the files as a data frame.
3. Annotation of the 'HGNC' and 'ENSEMBLID' identifiers, using the 'ENTREZ gene ID' as query with the 'org.Hs.eg.db' object.
4. Saving the object to 'data/cgc_67.rda'.

4 Data Source

The mutation data was obtained from the Sanger Institute Catalogue Of Somatic Mutations In Cancer web site, <http://www.sanger.ac.uk/cosmic>

Bamford et al (2004):

The COSMIC (Catalogue of Somatic Mutations in Cancer) database and website.

Br J Cancer, 91,355-358.

For details on the usage and redistribution of the data, please see ftp://ftp.sanger.ac.uk/pub/CGP/cosmic/GUIDELINES_ON_THE_USE_OF_THIS_DATA.txt.

5 References

- <http://cancer.sanger.ac.uk/cancergenome/projects/cosmic/>
- http://nar.oxfordjournals.org/content/39/suppl_1/D945.long
- ftp://ftp.sanger.ac.uk/pub/CGP/cosmic/GUIDELINES_ON_THE_USE_OF_THIS_DATA.txt

6 Session Info

R version 4.1.0 (2021-05-18)

Platform: x86_64-pc-linux-gnu (64-bit)

Running under: Ubuntu 20.04.2 LTS

Matrix products: default

BLAS: /home/biocbuild/bbs-3.13-bioc/R/lib/libRblas.so

LAPACK: /home/biocbuild/bbs-3.13-bioc/R/lib/libRlapack.so

locale:

[1] LC_CTYPE=en_US.UTF-8	LC_NUMERIC=C
[3] LC_TIME=en_US.UTF-8	LC_COLLATE=C
[5] LC_MONETARY=en_US.UTF-8	LC_MESSAGES=en_US.UTF-8
[7] LC_PAPER=en_US.UTF-8	LC_NAME=C
[9] LC_ADDRESS=C	LC_TELEPHONE=C

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[11] LC_MEASUREMENT=en_US.UTF-8 LC_IDENTIFICATION=C

attached base packages:

[1] stats4 parallel stats graphics grDevices utils
[7] datasets methods base

other attached packages:

[1] VariantAnnotation_1.38.0 Rsamtools_2.8.0
[3] Biostrings_2.60.0 XVector_0.32.0
[5] SummarizedExperiment_1.22.0 Biobase_2.52.0
[7] GenomicRanges_1.44.0 GenomeInfoDb_1.28.0
[9] IRanges_2.26.0 S4Vectors_0.30.0
[11] MatrixGenerics_1.4.0 matrixStats_0.58.0
[13] BiocGenerics_0.38.0 knitr_1.33

loaded via a namespace (and not attached):

[1] httr_1.4.2 bit64_4.0.5
[3] assertthat_0.2.1 highr_0.9
[5] BiocManager_1.30.15 BiocFileCache_2.0.0
[7] blob_1.2.1 BSgenome_1.60.0
[9] GenomeInfoDbData_1.2.6 yaml_2.2.1
[11] progress_1.2.2 pillar_1.6.1
[13] RSQLite_2.2.7 lattice_0.20-44
[15] glue_1.4.2 digest_0.6.27
[17] htmltools_0.5.1.1 Matrix_1.3-3
[19] XML_3.99-0.6 pkgconfig_2.0.3
[21] biomaRt_2.48.0 zlibbioc_1.38.0
[23] purrr_0.3.4 BiocParallel_1.26.0
[25] tibble_3.1.2 KEGGREST_1.32.0
[27] generics_0.1.0 ellipsis_0.3.2
[29] cachem_1.0.5 GenomicFeatures_1.44.0
[31] magrittr_2.0.1 crayon_1.4.1
[33] memoise_2.0.0 evaluate_0.14
[35] fansi_0.4.2 tools_4.1.0
[37] prettyunits_1.1.1 hms_1.1.0
[39] BiocStyle_2.20.0 BiocIO_1.2.0
[41] lifecycle_1.0.0 stringr_1.4.0
[43] DelayedArray_0.18.0 AnnotationDbi_1.54.0
[45] compiler_4.1.0 rlang_0.4.11
[47] grid_4.1.0 RCurl_1.98-1.3
[49] rjson_0.2.20 rappdirs_0.3.3
[51] bitops_1.0-7 rmarkdown_2.8
[53] restfulr_0.0.13 DBI_1.1.1
[55] curl_4.3.1 R6_2.5.0
[57] GenomicAlignments_1.28.0 dplyr_1.0.6
[59] rtracklayer_1.52.0 fastmap_1.1.0
[61] bit_4.0.4 utf8_1.2.1
[63] filelock_1.0.2 stringi_1.6.2
[65] Rcpp_1.0.6 vctrs_0.3.8
[67] png_0.1-7 dbplyr_2.1.1
[69] tidyselect_1.1.1 xfun_0.23