

COSMIC 67

Julian Gehring, EMBL Heidelberg

May 1, 2018

Contents

1	Introduction	1
2	Accessing and Using the Data	1
3	Data Provenance	4
3.1	COSMIC Mutations	4
3.2	Cancer Gene Census	4
4	Data Source	5
5	References	5
6	Session Info	5

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'
```

COSMIC 67

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, sd, var, xtabs

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

*Filter, Find, Map, Position, Reduce, anyDuplicated,
append, as.data.frame, basename, cbind, colMeans,
colSums, colnames, dirname, 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, rowMeans, rowSums, rownames, sapply, setdiff,
sort, table, tapply, union, unique, unsplit, which,
which.max, which.min*

Loading required package: *GenomeInfoDb*

Loading required package: *S4Vectors*

Loading required package: *stats4*

Attaching package: '*S4Vectors*'

The following object is masked from 'package:base':

expand.grid

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: *DelayedArray*

Loading required package: *matrixStats*

Attaching package: '*matrixStats*'

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

anyMissing, rowMedians

COSMIC 67

Loading required package: BiocParallel

Attaching package: 'DelayedArray'

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

colMaxs, colMins, colRanges, rowMaxs, rowMins,
rowRanges

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

aperm, apply

Loading required package: Rsamtools

Loading required package: Biostrings

Loading required package: XVector

Attaching package: 'Biostrings'

The following object is masked from 'package:DelayedArray':

type

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):

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

COSMIC 67

```
AA      1      String Peptide annotation
CNT     1      Integer How many samples have this mutation
geno(vcf):
  SimpleList 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 3.5.0 (2018-04-23)
Platform: x86_64-pc-linux-gnu (64-bit)
Running under: Ubuntu 16.04.4 LTS
```

```
Matrix products: default
BLAS: /home/biocbuild/bbs-3.7-bioc/R/lib/libRblas.so
LAPACK: /home/biocbuild/bbs-3.7-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
[11] LC_MEASUREMENT=en_US.UTF-8 LC_IDENTIFICATION=C
```

```
attached base packages:
[1] stats4    parallel stats      graphics grDevices utils
[7] datasets methods base
```

COSMIC 67

other attached packages:

```
[1] VariantAnnotation_1.26.0   Rsamtools_1.32.0
[3] Biostrings_2.48.0         XVector_0.20.0
[5] SummarizedExperiment_1.10.0 DelayedArray_0.6.0
[7] BiocParallel_1.14.0       matrixStats_0.53.1
[9] Biobase_2.40.0            GenomicRanges_1.32.0
[11] GenomeInfoDb_1.16.0       IRanges_2.14.0
[13] S4Vectors_0.18.0         BiocGenerics_0.26.0
[15] knitr_1.20
```

loaded via a namespace (and not attached):

```
[1] Rcpp_0.12.16             highr_0.6
[3] compiler_3.5.0           prettyunits_1.0.2
[5] progress_1.1.2           GenomicFeatures_1.32.0
[7] bitops_1.0-6             tools_3.5.0
[9] zlibbioc_1.26.0          biomaRt_2.36.0
[11] digest_0.6.15           bit_1.1-12
[13] BSgenome_1.48.0         memoise_1.1.0
[15] evaluate_0.10.1         RSQLite_2.1.0
[17] lattice_0.20-35         Matrix_1.2-14
[19] DBI_0.8                  yaml_2.1.18
[21] GenomeInfoDbData_1.1.0   rtracklayer_1.40.0
[23] httr_1.3.1              stringr_1.3.0
[25] rprojroot_1.3-2         bit64_0.9-7
[27] grid_3.5.0              R6_2.2.2
[29] AnnotationDbi_1.42.0     XML_3.98-1.11
[31] rmarkdown_1.9           blob_1.1.1
[33] magrittr_1.5            GenomicAlignments_1.16.0
[35] backports_1.1.2         htmltools_0.3.6
[37] assertthat_0.2.0        BiocStyle_2.8.0
[39] stringi_1.1.7           RCurl_1.95-4.10
```