## Package 'ICAMS'

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Title In-depth Characterization and Analysis of Mutational Signatures ('ICAMS')

Author Steve Rozen, Nanhai Jiang, Arnoud Boot, Mo Liu

Type Package

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```
Maintainer Steve Rozen <steverozen@gmail.com>
Description Analysis and visualization of experimentally elucidated mutational
      signatures -- the kind of analysis and visualization in Boot et al.,
      "In-depth characterization of the cisplatin mutational signature in
      human cell lines and in esophageal and liver tumors", Genome Research 2018,
      <doi:10.1101/gr.230219.117>. 'ICAMS' stands for In-depth Characterization
      and Analysis of Mutational Signatures. 'ICAMS' has functions to read in
      variant call files (VCFs) and to collate the corresponding catalogs of
      mutational spectra and to analyze and plot catalogs of mutational spectra
      and signatures. Handles both ``counts-based" and ``density-based" catalogs
      of mutational spectra or signatures.
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      utils
```

2 R topics documented:

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Suggests testthat,
BSgenome.Hsapiens.1000genomes.hs37d5,
BSgenome.Hsapiens.UCSC.hg38,
BSgenome.Mmusculus.UCSC.mm10

# R topics documented:

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all.abundance K-mer abundances.

#### **Description**

An R list with one element each for BSgenome.Hsapiens.1000genomes.hs37d5, BSgenome.Hsapiens.UCSC.hg38 and BSgenome.Mmusculus.UCSC.mm10. Each element is in turn a sub-list keyed by exome, transcript, and genome. Each element of the sub list is keyed by the number of rows in the catalog class (as a string, e.g. "78", not 78). The keys are: 78 (DBS78Catalog), 96 (SBS96Catalog), 136 (DBS136Catalog), 144 (DBS144Catalog), 192 (SBS192Catalog), and 1536 (SBS1536Catalog). So, for example to get the exome abundances for SBS96 catalogs for BSgenome.Hsapiens.UCSC.hg38 exomes one would reference all.abundance[["BSgenome.Hsapiens.UCSC.hg38"]][["exome"]]["96"] or all.abundance\$BSgenome.Hsapiens.UCSC.hg38\$exome\$"96". The value of the abundance is an integer vector with the K-mers as names and each value being the count of that K-mer.

## Usage

all.abundance

#### **Format**

See Description.

## **Examples**

```
all.abundance$BSgenome.Hsapiens.UCSC.hg38$transcript$`144`
# AA AC AG AT CA CC ...
# 90769160 57156295 85738416 87552737 83479655 63267896 ...
# There are 90769160 AAs on the sense strands of transcripts in # this genome.
```

AnnotateDBSVCF

Add sequence context and transcript information to an in-memory DBS VCF.

## Description

Add sequence context and transcript information to an in-memory DBS VCF.

## Usage

```
AnnotateDBSVCF(DBS.vcf, ref.genome, trans.ranges = NULL)
```

## **Arguments**

trans.ranges

DBS.vcf An in-memory DBS VCF as a data.frame.
ref.genome A ref.genome argument as described in ICAMS.

a data. table which contains transcript range and strand information. Please

refer to TranscriptRanges for more details. If is.null(trans.ranges) do

not add transcript range information.

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#### Value

An in-memory DBS VCF as a data.table. This has been annotated with the sequence context (column name seq.21bases) and with transcript information in the form of a gene symbol (e.g. "TP53") and transcript strand. This information is in the columns trans.start.pos, trans.end.pos, trans.strand and trans.gene.symbol in the output. These columns are not added if is.null(trans.ranges).

#### **Examples**

AnnotateIDVCF

Add sequence context to an in-memory ID (insertion/deletion) VCF, and confirm that they match the given reference genome.

## Description

Add sequence context to an in-memory ID (insertion/deletion) VCF, and confirm that they match the given reference genome.

## Usage

```
AnnotateIDVCF(ID.vcf, ref.genome, flag.mismatches = 0)
```

## **Arguments**

ID.vcf

An in-memory ID (insertion/deletion) VCF as a data.frame. This function expects that there is a "context base" to the left, for example REF = ACG, ALT = A (deletion of CG) or REF = A, ALT = ACC (insertion of CC).

ref.genome

A ref. genome argument as described in ICAMS.

flag.mismatches

If > 0, then if there are mismatches to references, generate messages showing the mismatched rows and continue. Otherwise stop if there are mismatched rows.

## Value

A data frame with 2 new columns added to the input data frame:

- 1. seq.context The sequence embedding the variant.
- 2. seq.context.width The width of seq.context to the left

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#### **Examples**

AnnotateSBSVCF

Add sequence context and transcript information to an in-memory SBS VCF.

#### **Description**

Add sequence context and transcript information to an in-memory SBS VCF.

#### Usage

```
AnnotateSBSVCF(SBS.vcf, ref.genome, trans.ranges = NULL)
```

#### Arguments

trans.ranges

SBS.vcf An in-memory SBS VCF as a data.frame.

ref.genome A ref.genome argument as described in ICAMS.

a data.table which contains transcript range and strand information. Please

 $refer\ to\ {\tt TranscriptRanges}\ for\ more\ details.\ If\ {\tt is.null(trans.ranges)}\ do$ 

not add transcript range information.

#### Value

An in-memory SBS VCF as a data.table. This has been annotated with the sequence context (column name seq.21bases) and with transcript information in the form of a gene symbol (e.g. "TP53") and transcript strand. This information is in the columns trans.start.pos, trans.end.pos, trans.strand and trans.gene.symbol in the output. These columns are not added if is.null(trans.ranges).

6 as.catalog

as.catalog	Create a catalog f	from a matrix	data frame	or vector
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#### **Description**

Create a catalog from a matrix, data.frame, or vector.

#### Usage

```
as.catalog(object, ref.genome = NULL, region = "unknown",
  catalog.type = "counts", abundance = NULL, infer.rownames = FALSE)
```

## Arguments

object A numeric matrix, numeric data.frame, or vector. If a vector, converted

to a 1-column matrix with rownames taken from the element names of the vector and with column name "Unknown". If argument infer.rownames is FALSE than this argument must have rownames to denote the mutation types.

See CatalogRowOrder for more details.

ref.genome A ref.genome argument as described in ICAMS.

region A character string designating a region, one of genome, transcript, exome,

unknown; see ICAMS.

catalog.type One of "counts", "density", "counts.signature", "density.signature".

abundance If NULL, then inferred if ref.genome is one of the reference genomes known

to ICAMS and region is not unknown. See ICAMS. The argument abundance should contain the counts of different source sequences for mutations in the

same format as the numeric vectors in all. abundance.

infer.rownames If TRUE, and object has no rownames, then assume the rows of object are

in the correct order and add the rownames implied by the number of rows in object (e.g. rownames for SBS 192 if there are 192 rows). If TRUE, **be sure the** 

order of rows is correct.

#### Value

A catalog as described in ICAMS.

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Canonicalize1Del	Given a deletion and its sequence context, categorize it.

## **Description**

This function is primarily for internal use, but we export it to document the underlying logic.

#### Usage

```
Canonicalize1Del(context, del.seq, pos, trace = 0)
```

## **Arguments**

context	The deleted sequence plus ample surrounding sequence on each side (at least as long as del.seq).
del.seq	The deleted sequence in context.
pos	The position of del.sequence in context.
trace	If $> 0$ , then generate messages tracing how the computation is carried out.

## **Details**

See https://github.com/steverozen/ICAMS/raw/master/data-raw/PCAWG7\_indel\_classification\_ 2017\_12\_08.xlsx for additional information on deletion mutation classification.

This function first handles deletions in homopolymers, then handles deletions in simple repeats with longer repeat units, (e.g. CACACAC, see FindMaxRepeatDel), and if the deletion is not in a simple repeat, looks for microhomology (see FindDelMH).

See the code for unexported function CanonicalizeID and the functions it calls for handling of insertions.

## Value

A string that is the canonical representation of the given deletion type. Return NA and raise a warning if there is an un-normalized representation of the deletion of a repeat unit. See FindDelMH for details. (This seems to be very rare.)

```
Canonicalize1Del("xyAAAqr", del.seq = "A", pos = 3) # "DEL:T:1:2"
Canonicalize1Del("xyAAAqr", del.seq = "A", pos = 4) # "DEL:T:1:2"
Canonicalize1Del("xyAqr", del.seq = "A", pos = 3) # "DEL:T:1:0"
```

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CatalogRowOrder

Standard order of row names in a catalog.

## **Description**

This data is designed for those who need to create their own catalogs from formats not supported by this package. The rownames denote the mutation types. For example, for SBS96 catalogs, the rowname AGAT represents a mutation from AGA > ATA.

This data is designed for those who need to create their own catalogs from formats not supported by this package. The rownames denote the mutation types. For example, for SBS96 catalogs, the rowname AGAT represents a mutation from AGA > ATA.

## Usage

```
catalog.row.order
catalog.row.order.sp
```

#### **Format**

A list of character vectors indicating the standard orders of row names in catalogs.

#### Note

In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

In ID83 (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5,

## **Examples**

```
catalog.row.order$SBS96
# "ACAA" "ACCA" "ACGA" "ACTA" "CCAA" "CCCA" "CCGA" "CCTA" ...
# There are altogether 96 row names to denote the mutation types
# in SBS96 catalog.

catalog.row.order.sp$ID83
# "DEL:C:1:0" "DEL:C:1:1" "DEL:C:1:2" "DEL:C:1:3" ...
# There are altogether 83 row names to denote the mutation types
# in ID83 catalog.
```

CollapseCatalog

"Collapse" a catalog.

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## **Description**

1. Take a mutational spectrum or signature catalog that is based on a fined-grained set of features (for example, single-nucleotide substitutions in the context of the preceding and following 2 bases).

2. Collapse it to a catalog based on a coarser-grained set of features (for example, single-nucleotide substitutions in the context of the immediately preceding and following bases).

Collapse192CatalogTo96 Collapse an SBS 192 catalog to an SBS 96 catalog.

Collapse1536CatalogTo96 Collapse an SBS 1536 catalog to an SBS 96 catalog.

Collapse144CatalogTo78 Collapse a DBS 144 catalog to a DBS 78 catalog.

## Usage

```
Collapse192CatalogTo96(catalog)
Collapse1536CatalogTo96(catalog)
Collapse144CatalogTo78(catalog)
```

## **Arguments**

catalog

A catalog as defined in ICAMS.

## Value

A catalog as defined in ICAMS.

#### **Examples**

FindDelMH

Return the length of microhomology at a deletion.

## **Description**

Return the length of microhomology at a deletion.

## Usage

```
FindDelMH(context, deleted.seq, pos, trace = 0, warn.cryptic = TRUE)
```

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#### **Arguments**

context The deleted sequence plus ample surrounding sequence on each side (at least as

long as del. sequence).

deleted.seq The deleted sequence in context.

pos The position of del. sequence in context.

trace If > 0, then generate various messages showing how the computation is carried

out.

warn.cryptic if TRUE generating a warning if there is a cryptic repeat (see the example).

#### **Details**

This function is primarily for internal use, but we export it to document the underlying logic.

Example:

GGCTAGTT aligned to GGCTAGAACTAGTT with a deletion represented as:

```
GGCTAGAACTAGTT
GG-----CTAGTT GGCTAGTT GG[CTAGAA]CTAGTT
```

Presumed repair mechanism leading to this:

```
GGCTAGAACTAGTT
```

=>

GGCTAG TT CC GATCAA

=>

GGCTAGTT CCGATCAA

Variant-caller software can represent the same deletion in several different, but completely equivalent, ways.

```
GGC----TAGTT GGCTAGTT GGC[TAGAAC]TAGTT

* --- * ---

GGCT----AGTT GGCTAGTT GGCT[AGAACT]AGTT

** -- ** --

GGCTA-----GTT GGCTAGTT GGCTA[GAACTA]GTT
```

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```
*** - *** -

GGCTAG----TT GGCTAGTT GGCTAG[AACTAG]TT

**** ****
```

This function finds:

1. The maximum match of undeleted sequence to the left of the deletion that is identical to the right end of the deleted sequence, and

2. The maximum match of undeleted sequence to the right of the deletion that is identical to the left end of the deleted sequence.

The microhomology sequence is the concatenation of items (1) and (2).

#### Warning

A deletion in a *repeat* can also be represented in several different ways. A deletion in a repeat is abstractly equivalent to a deletion with microhomology that spans the entire deleted sequence. For example;

```
GACTAGCTAGTT
GACTAGTT GACTA[GCTA]GTT

*** -*** -

is really a repeat

GACTAG----TT GACTAGTT GACTAG[CTAG]TT

**** ----

GACT----AGTT GACTAGTT GACT[AGCT]AGTT
```

This function only flags these "cryptic repeats" with a -1 return; it does not figure out the repeat extent.

#### Value

The length of the maximum microhomology of del. sequence in context.

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```
# and
#
# TAAATTATTTATTATTG
# TAAA----TTATTAATTTATTG = TAAATTATTATTG

FindDelMH("TAAATTATTTATTAATTTATTG", "TTTA", 8, warn.cryptic = FALSE) # -1
```

FindMaxRepeatDel

Return the number of repeat units in which a deletion is embedded.

## **Description**

Return the number of repeat units in which a deletion is embedded.

## Usage

```
FindMaxRepeatDel(context, rep.unit.seq, pos)
```

#### **Arguments**

context A string that embeds rep.unit.seq at position pos

rep.unit.seq A substring of context at pos to pos + nchar(rep.unit.seq) -1, which is the

repeat unit sequence.

pos The position of rep.unit.seq in context.

#### **Details**

This function is primarily for internal use, but we export it to document the underlying logic.

For example FindMaxRepeatDel("xyaczt", "ac", 3) returns 0.

If substr(context,pos,pos + nchar(rep.unit.seq) -1) != rep.unit.seq then stop.

If this functions returns 0, then it is necessary to look for microhomology using the function FindDelMH.

#### Warning

This function depends on the variant caller having "aligned" the deletion within the context of the repeat.

For example, a deletion of CAG in the repeat

**GTCAGCAGCATGT** 

can have 3 "aligned" representations as follows:

```
CT---CAGCAGGT
CTCAG---CAGGT
CTCAGCAG---GT
```

In these cases this function will return 2. (Please not that the return value does not include the rep.uni.seq in the count.)

However, the same deletion can also have an "unaligned" representation, such as

```
CTCAGC---AGGT
```

(a deletion of AGC).

In this case this function will return 1 (a deletion of AGC in a 2-element repeat of AGC).

GetVAF

#### Value

The number of repeat units in which rep.unit.seq is embedded, not including the input rep.unit.seq in the count.

## **Examples**

```
FindMaxRepeatDel("xyACACzt", "AC", 3) # 1
FindMaxRepeatDel("xyACACzt", "CA", 4) # 0
```

GetVAF

Extract the VAFs (variant allele frequencies) from a VCF file.

## Description

Extract the VAFs (variant allele frequencies) from a VCF file.

## Usage

```
GetStrelkaVAF(vcf)
GetMutectVAF(vcf)
```

## **Arguments**

vcf

Said VCF as a data.frame.

#### Value

A vector of VAFs, one for each row of vcf.

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ICAMS: In-depth Characterization and Analysis of Mutational Signatures

#### **Description**

Analysis and visualization of experimentally elucidated mutational signatures – the kind of analysis and visualization in Boot et al., "In-depth characterization of the cisplatin mutational signature in human cell lines and in esophageal and liver tumors",

Genome Research 2018, https://doi.org/10.1101/gr.230219.117. "ICAMS" stands for In-depth Characterization and Analysis of Mutational Signatures. "ICAMS" has functions to read in variant call files (VCFs) and to collate the corresponding catalogs of mutational spectra and to analyze and plot catalogs of mutational spectra and signatures. Handles both "counts-based" and "density-based" catalogs of mutational spectra or signatures.

#### **Details**

"ICAMS" can read in VCFs generated by Strelka or Mutect, and collate the mutations into "catalogs" of mutational spectra. "ICAMS" can create and plot catalogs of mutational spectra or signatures for single base substitutions (SBS), double base substitutions (DBS), and small insertions and deletions (ID). It can also read and write these catalogs.

#### **Catalogs**

A key data type in "ICAMS" is a "catalog" of mutation counts, of mutation densities, or of mutational signatures.

Catalogs are S3 objects of class matrix and one of several additional classes that specify the types of the mutations represented in the catalog. The possible additional class is one of

- SBS96Catalog (strand-agnostic single base substitutions in trinucleotide context)
- SBS192Catalog (transcription-stranded single-base substitutions in trinucleotide context)
- SBS1536Catalog
- DBS78Catalog
- DBS144Catalog
- DBS136Catalog
- IndelCatalog

as.catalog is the main constructor.

Conceptually, a catalog also has one of the following types, indicated by the attribute catalog.type:

- 1. Matrix of mutation counts (one column per sample), representing (counts-based) mutational spectra (catalog.type = "counts").
- 2. Matrix of mutation densities, i.e. mutations per occurrences of source sequences (one column per sample), representing (density-based) mutational spectra (catalog.type = "density").
- 3. Matrix of mutational signatures, which are similar to spectra. However where spectra consist of counts or densities of mutations in each mutation class (e.g. ACA > AAA, ACA > AGA, ACA > ATA, ACC > AAC, ...), signatures consist of the proportions of mutations in each class (with all the proportions summing to 1). A mutational signature can be based on either:

- mutation counts (a "counts-based mutational signature", catalog.type = "counts.signature"),
   or
- mutation densities (a "density-based mutational signature", catalog.type = "density.signature").

Catalogs also have the attribute abundance, which contains the counts of different source sequences for mutations. For example, for SBSs in trinucleotide context, the abundances would be the counts of each trinucleotide in the human genome, exome, or in the transcribed region of the genome. See TransformCatalog for more information. Abundances logically depend on the species in question and on the part of the genome being analyzed.

In "ICAMS" abundances can sometimes be inferred from the catalog class attribute and the function arguments region, ref.genome, and catalog.type. Otherwise abundances can be provided as an abundance argument. See all.abundance for examples.

Possible values for region are the strings genome, transcript, exome, and unknown; transcript includes entire transcribed regions, i.e. the introns as well as the exons.

If you need to create a catalog from a source other than this package (i.e. other than with ReadCatalog or StrelkaSBSVCFFilesToCatalog, MutectVCFFilesToCatalog, etc.), then use as.catalog.

#### **Creating catalogs from variant call files (VCF files)**

- 1. StrelkaSBSVCFFilesToCatalog creates 3 SBS catalogs (96, 192, 1536) and 3 DBS catalogs (78, 136, 144) from the Strelka SBS VCFs.
- 2. StrelkaIDVCFFilesToCatalog creates an ID (small insertion and deletion) catalog from the Strelka ID VCFs.
- 3. MutectVCFFilesToCatalog creates 3 SBS catalogs (96, 192, 1536), 3 DBS catalogs (78, 136, 144) and ID (indel) catalog from the Mutect VCFs.

#### Plotting catalogs

The PlotCatalog functions plot mutational spectra for **one** sample or plot **one** mutational signature.

The PlotCatalogToPdf functions plot catalogs of mutational spectra or of mutational signatures to a PDF file.

## Wrapper functions to create catalogs from VCFs and plot the catalogs to PDF files

- 1. StrelkaSBSVCFFilesToCatalogAndPlotToPdf creates all type of SBS and DBS catalogs from Strelka SBS VCFs and plots the catalogs.
- 2. StrelkaIDVCFFilesToCatalogAndPlotToPdf creates an ID (indel) catalog from Strelka ID VCFs and plot it.
- 3. MutectVCFFilesToCatalogAndPlotToPdf creates all types of SBS, DBS, and ID catalogs from Mutect VCFs and plots the catalogs.

## The ref.genome (reference genome) argument

Many functions take the argument ref.genome.

To create a mutational spectrum catalog from a VCF file, ICAMS needs the reference genome sequence that matches the VCF file. The ref. genome argument provides this.

ref.genome must be one of

1. A variable from the Bioconductor BSgenome package that contains a particular reference genome, for example BSgenome. Hsapiens. 1000genomes. hs37d5.

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- 2. The strings "hg38" or "GRCh38", which specify BSgenome. Hsapiens. UCSC. hg38.
- 3. The strings "hg19" or "GRCh37", which specify BSgenome. Hsapiens. 1000genomes. hs37d5.
- 4. The strings "mm10" or "GRCm38", which specify BSgenome. Mmusculus. UCSC. mm10.

All needed reference genomes must be installed separately by the user. Further instructions are at https://bioconductor.org/packages/release/bioc/html/BSgenome.html.

Use of ICAMS with reference genomes other than the 2 human genomes and 1 mouse genome specified above is restricted to catalog.type of counts or counts.signature unless the user also creates the necessary abundance vectors. See all.abundance.

Use available.genomes() to get the list of available genomes.

## Writing catalogs to files

The WriteCatalog functions write a catalog to a file.

#### Reading catalogs

The ReadCatalog functions read a file that contains a catalog in standardized format.

#### **Transforming catalogs**

The TransformCatalog function transforms catalogs of mutational spectra or signatures to account for differing abundances of the source sequence of the mutations in the genome.

For example, mutations from ACG are much rarer in the human genome than mutations from ACC simply because CG dinucleotides are rare in the genome. Consequently, there are two possible representations of mutational spectra or signatures. One representation is based on mutation counts as observed in a given genome or exome, and this approach is widely used, as, for example, at https://cancer.sanger.ac.uk/cosmic/signatures, which presents signatures based on observed mutation counts in the human genome. We call these "counts-based spectra" or "counts-based signatures".

Alternatively, mutational spectra or signatures can be represented as mutations per source sequence, for example the number of ACT > AGT mutations occurring at all ACT 3-mers in a genome. We call these "density-based spectra" or "density-based signatures".

This function can also transform spectra based on observed genome-wide counts to "density"-based catalogs. In density-based catalogs mutations are expressed as mutations per source sequences. For example, a density-based catalog represents the proportion of ACCs mutated to ATCs, the proportion of ACGs mutated to ATGs, etc. This is different from counts-based mutational spectra catalogs, which contain the number of ACC > ATC mutations, the number of ACG > ATG mutations, etc.

This function can also transform observed-count based spectra or signatures from genome to exome based counts, or between different species (since the abundances of source sequences vary between genome and exome and between species).

## Collapsing catalogs

The CollapseCatalog functions

- 1. Take a mutational spectrum or signature catalog that is based on a fined-grained set of features (for example, single-nucleotide substitutions in the context of the preceding and following 2 bases).
- 2. Collapse it to a catalog based on a coarser-grained set of features (for example, single-nucleotide substitutions in the context of the immediately preceding and following bases).

ICAMS.to.SigPro.ID

#### Data

1. CatalogRowOrder Standard order of rownames in a catalog. The rownames encode the type of each mutation. The rownames denote the mutation types. For example, for SBS96 catalogs, the rowname AGAT represents a mutation from AGA > ATA.

2. TranscriptRanges Transcript ranges and strand information for a particular reference genome.

 ${\tt ICAMS.to.SigPro.ID} \qquad {\it Linker from PCAWG(ICAMS)-formatted\ to\ SigProExtractor-formatted\ indel\ names}.$ 

## **Description**

This data is designed for converting ICAMS-formatted indel names to SigProExtractor-formatted indel names.

## Usage

```
ICAMS.to.SigPro.ID
```

#### **Format**

A 83\*1 matrix. Its contents (first column) contain SigProExtractor formatted indel names in SigProExtractor order. Its rownames refer to the corresponding PCAWG(ICAMS)-formatted indel names.

MutectVCFFilesToCatalog

Create SBS, DBS and Indel catalogs from Mutect VCF files

## Description

Create 3 SBS catalogs (96, 192, 1536), 3 DBS catalogs (78, 136, 144) and Indel catalog from the Mutect VCFs specified by files

#### Usage

```
MutectVCFFilesToCatalog(files, ref.genome, trans.ranges = NULL,
  region = "unknown")
```

## **Arguments**

files Character vector of file paths to the Mutect VCF files.

ref.genome A ref.genome argument as described in ICAMS.

trans.ranges a data.table which contains transcript range and strand information. Please

refer to TranscriptRanges for more details.

region A character string designating a genomic region; see as.catalog and ICAMS.

#### **Details**

This function calls VCFsToSBSCatalogs, VCFsToDBSCatalogs and VCFsToIDCatalogs

#### Value

A list of 3 SBS catalogs (one each for 96, 192, and 1536), 3 DBS catalogs (one each for 78, 136, and 144) and ID catalog. If trans.ranges = NULL, SBS 192 and DBS 144 catalog will not be generated. Each catalog has attributes added. See as .catalog for more details.

#### Note

SBS 192 and DBS 144 catalogs include only mutations in transcribed regions. In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

#### **Examples**

 ${\tt MutectVCFFilesToCatalogAndPlotToPdf}$ 

Create SBS, DBS and Indel catalogs from Mutect VCF files and plot them to PDF

## Description

Create 3 SBS catalogs (96, 192, 1536), 3 DBS catalogs (78, 136, 144) and Indel catalog from the Mutect VCFs specified by files and plot them to PDF

#### Usage

```
MutectVCFFilesToCatalogAndPlotToPdf(files, ref.genome,
   trans.ranges = NULL, region = "unknown", output.file)
```

## **Arguments**

files	Character vector of file paths to the Mutect VCF files.
ref.genome	A ref. genome argument as described in ICAMS.
trans.ranges	a data.table which contains transcript range and strand information. Please refer to TranscriptRanges for more details.
region	A character string designating a genomic region; see as.catalog and ICAMS.
output.file	The name of the PDF file to be produced.

## Details

This function calls MutectVCFFilesToCatalog and PlotCatalogToPdf

PlotCatalog 19

#### Value

A list of 3 SBS catalogs (one each for 96, 192, and 1536), 3 DBS catalogs (one each for 78, 136, and 144), Indel catalog and their graphs plotted to PDF with specified file name. If trans.ranges = NULL, SBS 192 and DBS 144 catalog will not be generated and plotted. Each catalog has attributes added. See as.catalog for more details.

#### Note

SBS 192 and DBS 144 catalogs include only mutations in transcribed regions. In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

## **Examples**

PlotCatalog

Plot one spectrum or signature.

## Description

Plot the spectrum of **one** sample or plot **one** signature. The type of graph is based on one attribute("catalog.type") of the input catalog. You can first use TransformCatalog to get different types of catalog and then do the plotting.

#### Usage

```
PlotCatalog(catalog, plot.SBS12 = NULL, cex = NULL, grid = NULL,
    upper = NULL, xlabels = NULL)
```

## **Arguments**

catalog	A catalog as defined in ICAMS with attributes added. See as.catalog for more details.
plot.SBS12	Only meaningful for class SBS192Catalog; if TRUE, generate an abbreviated plot of only SBS without context, i.e. C>A, C>G, C>T, T>A, T>C, T>G each on transcribed and untranscribed strands, rather than SBS in trinucleotide context, e.g. ACA > AAA, ACA > AGA,, TCT > TAT,
cex	A numerical value giving the amount by which mutation class labels, mutation counts(if it exists), y axis and its labels, x axis labels and its annotations(if it exists), sample name and legend(if it exists) should be magnified relative to the default. Only implemented for SBS96Catalog, SBS192Catalog and DBS144Catalog.

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grid A logical value indicating whether to draw grid lines. Only implemented for

SBS96Catalog.

upper A logical value indicating whether to draw horizontal lines and the names of

major mutation class on top of graph. Only implemented for SBS96Catalog.

xlabels A logical value indicating whether to draw x axis labels. Only implemented for

SBS96Catalog.

#### Value

```
invisible(TRUE)
```

#### Note

The sizes of repeats involved in deletions range from 0 to 5+ in the mutational-spectra and signature catalog rownames, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

#### **Examples**

PlotCatalogToPdf

Plot catalog to a PDF file.

## Description

Plot catalog to a PDF file. The type of graph is based on one attribute("catalog.type") of the input catalog. You can first use TransformCatalog to get different types of catalog and then do the plotting.

## Usage

```
PlotCatalogToPdf(catalog, file, plot.SBS12 = NULL, cex = NULL,
   grid = NULL, upper = NULL, xlabels = NULL)
```

## **Arguments**

catalog A catalog as defined in ICAMS with attributes added. See as.catalog for more

details.

file The name of the PDF file to be produced.

plot. SBS12 Only meaningful for class SBS192Catalog; if TRUE, generate an abbreviated plot

of only SBS without context, i.e. C>A, C>G, C>T, T>A, T>C, T>G each on transcribed and untranscribed strands, rather than SBS in trinucleotide context, e.g. ACA > AAA, ACA > AGA, ..., TCT > TAT, ... There are 12 bars in the

graph.

cex	A numerical value giving the amount by which mutation class labels, mutation counts(if it exists), y axis and its labels, x axis labels and its annotations(if it exists), sample name and legend(if it exists) should be magnified relative to the default. Only implemented for SBS96Catalog, SBS192Catalog and DBS144Catalog.
grid	A logical value indicating whether to draw grid lines. Only implemented for SBS96Catalog.
upper	A logical value indicating whether to draw horizontal lines and the names of major mutation class on top of graph. Only implemented for SBS96Catalog.
xlabels	A logical value indicating whether to draw x axis labels. Only implemented for SBS96Catalog.

## Value

```
invisible(TRUE)
```

#### Note

The sizes of repeats involved in deletions range from 0 to 5+ in the mutational-spectra and signature catalog rownames, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

## **Examples**

ReadAndSplitMutectVCFs

Read and split Mutect VCF files.

## Description

Read and split Mutect VCF files.

## Usage

```
ReadAndSplitMutectVCFs(files)
```

## **Arguments**

files Character vector of file paths to the Mutect VCF files.

#### Value

A list with 3 in-memory VCFs and two left-over VCF-like data frames with rows that were not incorporated into the first 3 VCFs, as follows:

- 1. SBS VCF with only single base substitutions.
- 2. DBS VCF with only doublet base substitutions as called by Mutect.
- 3. ID VCF with only small insertions and deletions.
- 4. other.subs VCF like data.frame with rows for coordinate substitutions involving 3 or more nucleotides, e.g. ACT > TGA or AACT > GGTA.
- 5. multiple.alternative.alleles VCF like data.frame with rows for variants with multiple alternative alleles, for example ACT mutated to both AGT and ACT at the same position.

#### See Also

MutectVCFFilesToCatalog

#### **Examples**

ReadAndSplitStrelkaSBSVCFs

Read and split Strelka SBS VCF files.

## **Description**

Read and split Strelka SBS VCF files.

#### Usage

```
ReadAndSplitStrelkaSBSVCFs(files)
```

## **Arguments**

files

Character vector of file paths to the Strelka SBS VCF files.

#### Value

A list of 3 in-memory objects as follows:

- 1. SBS.vcfs List of data.frames of pure SBS mutations no DBS or 3+BS mutations.
- 2. DBS.vcfs List of data.frames of pure DBS mutations no SBS or 3+BS mutations.
- 3. ThreePlus List of data.tables with the key CHROM, LOW.POS, HIGH.POS. containing rows that that in the input that did not represent SBSs or DBSs.

## See Also

StrelkaSBSVCFFilesToCatalog

ReadCatalog 23

#### **Examples**

ReadCatalog

Read catalog.

## **Description**

Read a catalog in standardized format from path.

## Usage

```
ReadCatalog(file, ref.genome = NULL, region = "unknown",
  catalog.type = "counts", strict = TRUE)
```

#### **Arguments**

file Path to a catalog on disk in the standardized format.

ref.genome A ref.genome argument as described in ICAMS.

region region A character string designating a genomic region; see as.catalog and ICAMS.

catalog.type One of "counts", "density", "counts.signature", "density.signature".

strict If TRUE, do additional checks on the input, and stop if the checks fail.

#### **Details**

See also WriteCatalog

#### Value

A catalog as an S3 object; see as.catalog.

#### Note

In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

24 revc

ReadStrelkaIDVCFs

Read Strelka ID (insertion and deletion) VCF files.

## **Description**

Read Strelka ID (insertion and deletion) VCF files.

## Usage

```
ReadStrelkaIDVCFs(files)
```

## **Arguments**

files

Character vector of file paths to the VCF files.

#### Value

A list of vcfs from files.

#### Note

In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

### See Also

```
StrelkaIDVCFFilesToCatalog
```

## **Examples**

revc

Reverse complement every string in string.vec.

## **Description**

```
Based on reverseComplement. Handles IUPAC ambiguity codes but not "u" (uracil). (see <a href="https://en.wikipedia.org/wiki/Nucleic_acid_notation">https://en.wikipedia.org/wiki/Nucleic_acid_notation</a>).
```

## Usage

```
revc(string.vec)
```

#### **Arguments**

string.vec

A character vector.

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#### Value

A character vector with the reverse complement of every string in string.vec.

#### **Examples**

```
revc("aTgc") # GCAT

# A vector and strings with ambiguity codes
revc(c("ATGC", "aTGc", "wnTCb")) # GCAT GCAT VGANW

## Not run:
revc("ACGU") # An error
## End(Not run)
```

SigPro.to.ICAMS.ID

Linker from SigProExtractor-formatted to PCAWG(ICAMS)-formatted indel names.

## **Description**

This data is designed for converting SigProExtractor-formatted indel names to ICAMS-formatted indel names.

## Usage

```
SigPro.to.ICAMS.ID
```

## **Format**

A 83\*1 matrix. Its contents (first column) contain PCAWG(ICAMS)- formatted indel names in PCAWG(ICAMS) order. Its rownames refer to the corresponding SigProExtractor indel names.

```
StrelkaIDVCFFilesToCatalog
```

Create ID (indel) catalog from Strelka ID VCF files

## Description

Create ID (indel) catalog from the Strelka ID VCFs specified by files

## Usage

```
StrelkaIDVCFFilesToCatalog(files, ref.genome, region = "unknown")
```

## **Arguments**

files Character vector of file paths to the Strelka ID VCF files.

ref.genome A ref.genome argument as described in ICAMS.

region A character string designating a genomic region; see as.catalog and ICAMS.

#### **Details**

This function calls VCFsToIDCatalogs

#### Value

An ID (indel) catalog with attributes added. See as. catalog for more details.

#### Note

In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

## **Examples**

StrelkaIDVCFFilesToCatalogAndPlotToPdf

Create ID (indel) catalog from Strelka ID VCF files and plot them to PDF

#### **Description**

Create ID (indel) catalog from the Strelka ID VCFs specified by files and plot them to PDF

## Usage

```
StrelkaIDVCFFilesToCatalogAndPlotToPdf(files, ref.genome,
  region = "unknown", output.file)
```

#### **Arguments**

files Character vector of file paths to the Strelka ID VCF files.

ref.genome A ref.genome argument as described in ICAMS.

region A character string designating a genomic region; see as.catalog and ICAMS.

output.file The name of the PDF file to be produced.

#### **Details**

This function calls StrelkaIDVCFFilesToCatalog and PlotCatalogToPdf

#### Value

An ID (indel) catalog and its graph plotted to PDF with specified file name. The ID (indel) catalog has attributes added. See as.catalog for more details.

#### Note

In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

## **Examples**

StrelkaSBSVCFFilesToCatalog

Create SBS and DBS catalogs from Strelka SBS VCF files.

## **Description**

Create 3 SBS catalogs (96, 192, 1536) and 3 DBS catalogs (78, 136, 144) from the Strelka SBS VCFs specified by files

## Usage

```
StrelkaSBSVCFFilesToCatalog(files, ref.genome, trans.ranges = NULL,
  region = "unknown")
```

#### **Arguments**

files Character vector of file paths to the Strelka SBS VCF files.

ref.genome A ref.genome argument as described in ICAMS.

trans.ranges a data.table which contains transcript range and strand information. Please

refer to TranscriptRanges for more details.

region A character string designating a genomic region; see as.catalog and ICAMS.

## **Details**

This function calls VCFsToSBSCatalogs and VCFsToDBSCatalogs.

## Value

A list of 3 SBS catalogs (one each for 96, 192, and 1536) and 3 DBS catalogs (one each for 78, 136, and 144). If trans.ranges = NULL, SBS 192 and DBS 144 catalog will not be generated. Each catalog has attributes added. See as .catalog for more details.

#### Note

SBS 192 and DBS 144 catalog only contains mutations in transcribed regions.

#### **Examples**

 ${\tt StrelkaSBSVCFFilesToCatalogAndPlotToPdf}$ 

Create SBS and DBS catalogs from Strelka SBS VCF files and plot them to PDF

## Description

Create 3 SBS catalogs (96, 192, 1536) and 3 DBS catalogs (78, 136, 144) from the Strelka SBS VCFs specified by files and plot them to PDF

## Usage

```
StrelkaSBSVCFFilesToCatalogAndPlotToPdf(files, ref.genome,
  trans.ranges = NULL, region = "unknown", output.file)
```

## **Arguments**

files	Character vector of file paths to the Strelka SBS VCF files.
ref.genome	A ref. genome argument as described in ICAMS.
trans.ranges	a data.table which contains transcript range and strand information. Please refer to TranscriptRanges for more details.
region	A character string designating a genomic region; see as.catalog and ICAMS.
output.file	The name of the PDF file to be produced.

## **Details**

 $This \ function \ calls \ {\tt StrelkaSBSVCFFilesToCatalog} \ and \ {\tt PlotCatalogToPdf}$ 

### Value

A list of 3 SBS catalogs (one each for 96, 192, and 1536), 3 DBS catalogs (one each for 78, 136, and 144) and their graphs plotted to PDF with specified file name. If trans.ranges = NULL, SBS 192 and DBS 144 catalog will not be generated and plotted. Each catalog has attributes added. See as.catalog for more details.

## Note

SBS 192 and DBS 144 catalogs include only mutations in transcribed regions.

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## **Examples**

TranscriptRanges

Transcript ranges data

## **Description**

Transcript ranges and strand information for a particular reference genome.

## Usage

```
trans.ranges.GRCh37
trans.ranges.GRCh38
trans.ranges.GRCm38
```

## **Format**

A data.table which contains transcript range and strand information for a particular reference genome. colnames are chrom, start, end, strand, gene.symbol. It uses one-based coordinates.

#### Details

This information is needed to generate catalogs that depend on transcriptional strand information, for example catalogs of class SBS192Catalog.

```
trans.ranges.GRCh37: Human GRCh37. trans.ranges.GRCh38: Human GRCh38. trans.ranges.GRCm38: Mouse GRCm38.
```

For these two tables, only genes that are associated with a CCDS ID are kept for transcriptional strand bias analysis.

```
This information is needed for StrelkaSBSVCFFilesToCatalog, StrelkaSBSVCFFilesToCatalogAndPlotToPdf, MutectVCFFilesToCatalog, MutectVCFFilesToCatalogAndPlotToPdf, VCFsToSBSCatalogs and VCFsToDBSCatalogs.
```

30 TransformCatalog

#### Source

```
ftp://ftp.ebi.ac.uk/pub/databases/gencode/Gencode_human/release_30/GRCh37_mapping/
gencode.v30lift37.annotation.gff3.gz
ftp://ftp.ebi.ac.uk/pub/databases/gencode/Gencode_human/release_30/gencode.v30.annotation.
gff3.gz
ftp://ftp.ebi.ac.uk/pub/databases/gencode/Gencode_mouse/release_M21/gencode.vM21.
annotation.gff3.gz
```

## **Examples**

```
trans.ranges.GRCh37
# chrom
                 end strand gene.name
       start
        65419
              71585 +
                             OR4F5
    1
    1 367640 368634
                             OR4F29
   1 621059 622053
                           0R4F16
   1 859308 879961
                           SAMD11
   1 879583 894689
                             NOC2L
  . . .
         . . .
                 . . .
                               . . .
                      . . .
```

TransformCatalog

Transform between counts and density spectrum catalogs and counts and density signature catalogs.

## Description

Transform between counts and density spectrum catalogs and counts and density signature catalogs.

## Usage

```
TransformCatalog(catalog, target.ref.genome = NULL,
  target.region = NULL, target.catalog.type = NULL,
  target.abundance = NULL)
```

## **Arguments**

catalog

An SBS or DBS catalog as described in ICAMS; must **not** be an ID (indel) catalog.

target.ref.genome

A ref.genome argument as described in ICAMS. If NULL, then defaults to the ref.genome attribute of catalog.

target.region A region argument; see as.catalog and ICAMS. If NULL, then defaults to the region attribute of catalog.

target.catalog.type

A character string acting as a catalog type identifier, one of "counts", "density", "counts.signature", "density.signature"; see as.catalog. If NULL, then defaults to the catalog. type attribute of catalog.

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target.abundance

A vector of counts different source K-mer sequences for mutations. See all.abundance. If NULL, then the function attempt to infer the target.abundace from the class of catalog and the values of the target.ref.genome, target.region, and target.catalog.type. It is an error if the inferred abundance is different from an non-NULL target.abundance.

#### **Details**

Only the following transformations are legal:

- 1. counts -> counts (used to transform between the source abundance and target.abundance)
- 2. counts -> density
- 3. counts -> (counts.signature,density.signature)
- 4. density -> counts (the semantics are to infer the genome-wide or exome-wide counts based on the densities)
- 5. density -> density (a null operation, generates a warning)
- 6. density -> (counts.signature, density.signature)
- 7. counts.signature -> counts.signature (used to transform between the source abundance and target.abundance)
- 8. counts.signature -> density.signature
- 9. counts.signature -> (counts, density) (generates an error)
- 10. density.signature -> density.signature (a null operation, generates a warning)
- 11. density.signature -> counts.signature
- 12. density.signature -> (counts, density) (generates an error)

## Value

A catalog as defined in ICAMS.

32 VCFsToDBSCatalogs

VCFsToDBSCatalogs

Create DBS catalogs from VCFs

## **Description**

Create a list of 3 catalogs (one each for DBS78, DBS144 and DBS136) out of the contents in list.of.DBS.vcfs. The VCFs must not contain any type of mutation other then DBSs.

## Usage

```
VCFsToDBSCatalogs(list.of.DBS.vcfs, ref.genome, trans.ranges = NULL,
   region = "unknown")
```

## **Arguments**

list.of.DBS.vcfs

List of in-memory data frames of pure DBS mutations – no SBS or 3+BS mutations. The list names will be the sample ids in the output catalog.

ref.genome A ref.genome argument as described in ICAMS.

trans.ranges a data.table which contains transcript range and strand information. Please

refer to TranscriptRanges for more details.

region A character string designating a genomic region; see as.catalog and ICAMS.

## Value

A list of 3 DBS catalogs, one each for 78, 144, 136: catDBS78 catDBS144 catDBS136. If trans.ranges = NULL, DBS 144 catalog will not be generated. Each catalog has attributes added. See as .catalog for more details.

## Note

DBS 144 catalog only contains mutations in transcribed regions.

VCFsToIDCatalogs 33

VCF	cT0	TD	C 2 +	217	nac
VU.F	SIO	111	Lai.	a 1 (	195

Create ID (insertion and deletion) catalog from ID VCFs

## **Description**

Create ID (insertion and deletion) catalog from ID VCFs

## Usage

```
VCFsToIDCatalogs(list.of.vcfs, ref.genome, region = "unknown")
```

#### **Arguments**

list.of.vcfs List of in-memory VCFs. The list names will be the sample ids in the output

catalog.

ref.genome A ref.genome argument as described in ICAMS.

region A character string acting as a region identifier, one of "genome", "exome".

#### Value

An S3 object containing an ID (indel) catalog with class "catalog". See as.catalog for more details.

#### Note

In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

#### **Examples**

VCFsToSBSCatalogs

Create SBS catalogs from SBS VCFs

## **Description**

Create a list of 3 catalogs (one each for 96, 192, 1536) out of the contents in list.of.SBS.vcfs. The SBS VCFs must not contain DBSs, indels, or other types of mutations.

## Usage

```
VCFsToSBSCatalogs(list.of.SBS.vcfs, ref.genome, trans.ranges = NULL,
  region = "unknown")
```

34 WriteCatalog

#### **Arguments**

list.of.SBS.vcfs

List of in-memory data frames of pure SBS mutations - no DBS or 3+BS muta-

tions. The list names will be the sample ids in the output catalog.

ref.genome A ref.genome argument as described in ICAMS.

trans.ranges a data.table which contains transcript range and strand information. Please

refer to TranscriptRanges for more details.

region A character string designating a genomic region; see as.catalog and ICAMS.

#### Value

A list of 3 SBS catalogs, one each for 96, 192, 1536: catSBS96 catSBS192 catSBS1536. If trans.ranges = NULL, SBS 192 catalog will not be generated. Each catalog has attributes added. See as.catalog for more details.

## Note

SBS 192 catalogs only contain mutations in transcribed regions.

#### **Examples**

WriteCatalog

Write a catalog

## **Description**

Write a catalog to a file.

## Usage

```
WriteCatalog(catalog, file, strict = TRUE)
```

## **Arguments**

catalog A catalog as defined in ICAMS; see also as.catalog.

file The path to the file to be created.

strict If TRUE, do additional checks on the input, and stop if the checks fail.

## Details

See also ReadCatalog.

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## Note

In ID (insertion and deletion) catalogs, deletion repeat sizes range from 0 to 5+, but for plotting and end-user documentation deletion repeat sizes range from 1 to 6+.

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