# Package 'ICAMS'

April 30, 2019

```
Maintainer Steve Rozen <steverozen@gmail.com>
Description A toolkit for analysis and visualization of experimentally
      elucidated mutational signatures -- the kind of analysis and visualization
      presented in Boot et al., "In-depth characterization of the cisplatin
      mutational signature in human cell lines and in esophageal and liver
      tumors", 2018, https://genome.cshlp.org/content/28/5/654.short. This
      package has functions to read in variant call files and to collate the
      corresponding catalog of mutational spectra and to plot catalogs of
      mutational spectra or signatures.
License GPL-3
Encoding UTF-8
LazyData true
Language en-US
biocViews
Imports Biostrings,
      BSgenome,
      BSgenome. Hsapiens. 1000 genomes. hs37d5,
      BSgenome. Hsapiens. UCSC. hg38,
      data.table,
      dplyr,
      GenomicRanges,
      graphics,
      grDevices,
      IRanges,
      RColorBrewer,
      stats,
      stringi,
      utils
Depends R (>= 3.5),
RoxygenNote 6.1.1
Suggests testthat
```

Title In-depth Characterization and Analysis of Mutational Signatures

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

Type Package

Version 2.0.0.9007

2 as.catalog

# **R** topics documented:

	as.catalog	2
	CatalogRowOrder	3
	CollapseCatalog	3
	FindDelMH	4
	GetVAF	6
	ICAMS	$\epsilon$
	MutectVCFFilesToCatalog	ç
	PlotCatalog	
	PlotCatalogToPdf	11
	PreserveCatalogAttribute	12
	ReadAndSplitMutectVCFs	12
	ReadAndSplitStrelkaSNSVCFs	13
	ReadCatalog	14
	ReadStrelkaIDVCFs	14
	revc	15
	StrelkaIDVCFFilesToCatalog	15
	StrelkaSNSVCFFilesToCatalog	16
	TranscriptRanges	
	TransformCatalog	17
	VCFsToDNSCatalogs	18
	VCFsToIDCatalogs	19
	VCFsToSNSCatalogs	19
	WriteCatalog	20
Index		21
inaex		_

as.catalog *Create attributes of a catalog* 

## Description

Create attributes of a catalog

## Usage

```
as.catalog(catalog, ref.genome, region, catalog.type)
```

## Arguments

catalog A catalog as defined in ICAMS.

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

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

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

## Value

The original catalog with the following attributes added: ref.genome, region, type, abundance, class.

CatalogRowOrder 3

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 SNS96 catalogs, the rowname AGAT represents a mutation from AGA > ATA.

## Usage

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

#### **Format**

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

#### Note

In the ID (insertion and deletion) catalog, deletion repeat size is in the range from 0 to 5+, but for plotting and end user documentation it ranges from 1 to 6+.

CollapseCatalog

"Collapse" a catalog.

## **Description**

"Collapse" a catalog. Do not use this function for signature catalogs.

#### Usage

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

## **Arguments**

catalog

A catalog as defined in ICAMS.

## **Details**

```
Collapse192CatalogTo96 Collapse an SNS 192 catalog to an SNS 96 catalog. Collapse1536CatalogTo96 Collapse an SNS 1536 catalog to an SNS 96 catalog. Collapse144CatalogTo78 Collapse a DNS 144 catalog to a DNS 78 catalog.
```

4 FindDelMH

#### Value

A catalog as defined in ICAMS.

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)
```

## **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, cat various messages.

#### **Details**

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

## Example:

 ${\tt GGCTAGTT}\ aligned\ to\ {\tt GGCTAGAACTAGTT}\ with\ a\ deletion\ represented\ as:$ 

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

Presumed repair mechanism leading to this:

```
GGCTAGAACTAGTT
CCGATCTTGATCAA

=>
GGCTAG TT
CC GATCAA
....
```

GGCTAGTT CCGATCAA FindDelMH 5

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

```
GGCTAGTT GGCTAGTT GGC[TAGAAC]TAGTT

* --- * ---

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

** -- ** --

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

*** - *** -

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

**** ****
```

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

This function only flags this case with a -1 return; it does not figure out the repeat extent.

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).

## Value

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

6 ICAMS

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.

**ICAMS** 

ICAMS: In-depth Characterization and Analysis of Mutational Signatures

## **Description**

A toolkit for analysis and visualization of experimentally elucidated mutational signatures – the kind of analysis and visualization presented 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://genome.cshlp.org/content/28/5/654.short.

#### **Details**

ICAMS can read in variant call files (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 nucleotide substitutions (SNS), double nucleotide substitutions (DNS), 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.

A catalog has one of the following types:

- 1. Matrix of mutation counts (one column per sample), representing (count-based) mutational spectra.
- 2. Matrix of mutation densities, i.e. mutations per occurrences of source sequences (one column per sample), representing (density-based) mutational spectra.

ICAMS 7

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:

- 3.1 mutation counts (a "count-based mutational signature"), or
- 3.2 mutation densities (a "density-based mutational signature").

Many functions take the argument catalog.type, with possible values "counts", "density", "counts.signature", or "density.signature", corresponding to the types of catalogs in items 1, 2, 3.1, and 3.2, above.

Catalogs are implemented as S3 objects of class matrix and one of the ICAMS classes SNS96Catalog, SNS192Catalog, SNS1536Catalog, DNS78Catalog, DNS144Catalog, DNS136Catalog, IndelCatalog.

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

Nanhai: put this in the doc for as.catalog: you must ensure that the rows are in the expected order and have the expected rownames.

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

- 1. StrelkaSNSVCFFilesToCatalog creates 3 SNS catalogs (96, 192, 1536) and 3 DNS catalogs (78, 136, 144) from the Strelka SNS VCFs.
- 2. StrelkaIDVCFFilesToCatalog creates ID (indel) catalog from the Strelka ID VCFs.
- 3. MutectVCFFilesToCatalog creates 3 SNS catalogs (96, 192, 1536), 3 DNS catalogs (78, 136, 144) and ID (indel) catalog from the Mutect VCFs.

#### The ref. genome argument

Many functions take the argument ref. genome. This can be either

- 1. A variable from the Bioconductor BSgenome package that contains a particular reference genome, for example BSgenome. Hsapiens. 1000genomes. hs37d5. BSgenome::available.genomes() returns the available genomes.
- 2. The strings "hg38" or "GRCh38" are shorthand for BSgenome. Hsapiens. UCSC. hg38, and the strings "hg19" or "GRCh37" are shorthand for BSgenome. Hsapiens. 1000genomes. hs37d5.

## The Bioconductor BSgenome package

This package will be installed automatically if ICAMS is installed with devtools::install\_local or with devtools::install\_github. Otherwise you must manually install BSgenome and the necessary genomes, e.g.

BSgenome. Hsapiens. 1000genomes. hs37d5.

See instructions at

https://bioconductor.org/packages/release/bioc/html/BSgenome.html.

Genomes other than the two human genomes mentioned above must be installed manually.

Use available genomes to get the list of available genomes.

8 ICAMS

#### 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.

#### Writing catalogs

The WriteCatalog functions write a catalog of mutational spectra or of mutational signatures to a file.

#### Reading catalogs

The ReadCatalog functions read a file that contains a catalog of mutational spectra or of signatures 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, 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 "count-based spectra" or "count-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 count-based 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), and
- 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).

## Data

1. CatalogRowOrder Standard order of rownames in a catalog. The rownames of encode the type of each mutation. The rownames denote the mutation types. For example, for SNS96 catalogs, the rowname AGAT represents a mutation from AGA > ATA.

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

MutectVCFFilesToCatalog

Create SNS, DNS and Indel catalogs from Mutect VCF files

## Description

Create 3 SNS catalogs (96, 192, 1536), 3 DNS catalogs (78, 136, 144) and Indel catalog from the Mutect VCFs specified by vector.of.file.paths

## Usage

```
MutectVCFFilesToCatalog(vector.of.file.paths, ref.genome, trans.ranges,
    region)
```

#### **Arguments**

vector.of.file.paths

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

### **Details**

This function calls VCFsToSNSCatalogs, VCFsToDNSCatalogs and VCFsToIDCatalogs

#### Value

A list of 3 SNS catalogs (one each for 96, 192, and 1536), 3 DNS catalogs (one each for 78, 136, and 144) and ID catalog. Each catalog has attributes added. See as catalog for more details.

## Note

SNS 192 and DNS 144 catalogs include only mutations in transcribed regions.

10 PlotCatalog

PlotCatalog

Plot one spectrum or signature.

### **Description**

Plot the spectrum of **one** sample or plot **one** signature.

## Usage

```
PlotCatalog(catalog, strandbias = FALSE, ...)
## S3 method for class 'SNS96Catalog'
PlotCatalog(catalog, cex = 0.8, grid = TRUE,
 upper = TRUE, xlabels = TRUE)
## S3 method for class 'SNS192Catalog'
PlotCatalog(catalog, cex = 0.8)
## S3 method for class 'SNSClassStrandBias'
PlotCatalog(catalog, strandbias = TRUE,
 cex = 1)
## S3 method for class 'SNS1536Catalog'
PlotCatalog(catalog)
## S3 method for class 'DNS78Catalog'
PlotCatalog(catalog)
## S3 method for class 'DNSClassStrandBias'
PlotCatalog(catalog, strandbias = TRUE,
 cex = 1)
## S3 method for class 'DNS136Catalog'
PlotCatalog(catalog)
## S3 method for class 'IndelCatalog'
PlotCatalog(catalog)
```

## **Arguments**

catalog	A catalog as defined in ICAMS with attributes added. See as.catalog for more details.
strandbias	If TRUE, plot strand bias graph for SNS192 or DNS144 catalog. Leave out this parameter if you don't intend to plot strand bias graph.
	Additional arguments to be passed to methods.

## Value

```
invisible(TRUE)
```

PlotCatalogToPdf 11

#### Note

The sizes of repeats involved in deletions range from 0 to 5+ in the catalog rownames, but for plotting and end user documentation they ranges from 1 to 6+.

PlotCatalogToPdf

Plot catalogs to a PDF file.

### **Description**

Plot catalogs to a PDF file.

#### Usage

```
PlotCatalogToPdf(catalog, filename, strandbias = FALSE, ...)
## S3 method for class 'SNS96Catalog'
PlotCatalogToPdf(catalog, filename, grid = TRUE,
 upper = TRUE, xlabels = TRUE)
## S3 method for class 'SNS192Catalog'
PlotCatalogToPdf(catalog, filename)
## S3 method for class 'SNSClassStrandBias'
PlotCatalogToPdf(catalog, filename,
  strandbias = TRUE)
## S3 method for class 'SNS1536Catalog'
PlotCatalogToPdf(catalog, filename)
## S3 method for class 'DNS78Catalog'
PlotCatalogToPdf(catalog, filename)
## S3 method for class 'DNSClassStrandBias'
PlotCatalogToPdf(catalog, filename,
  strandbias = TRUE, cex = 1)
## S3 method for class 'DNS136Catalog'
PlotCatalogToPdf(catalog, filename)
## S3 method for class 'IndelCatalog'
PlotCatalogToPdf(catalog, filename)
```

## **Arguments**

catalog	A catalog as defined in ICAMS with attributes added. See as $.$ catalog for more details.
filename	The name of the PDF file to be produced.
strandbias	If TRUE, plot strand bias graph for SNS192 or DNS144 catalog. Leave out this parameter if you don't intend to plot strand bias graph.
	Additional arguments to be passed to methods.

#### Value

```
invisible(TRUE)
```

#### Note

The sizes of repeats involved in deletions range from 0 to 5+ in the catalog rownames, but for plotting and end user documentation they ranges from 1 to 6+.

PreserveCatalogAttribute

Preserve attributes of the input catalog

## Description

Preserve attributes of the input catalog

## Usage

```
PreserveCatalogAttribute(pre.catalog, new.catalog)
```

## **Arguments**

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

details.

new.catalog A new catalog which needs to inherit the necessary attributes from pre.catalog.

### Value

The new catalog that has inherited the necessary attributes from pre.catalog

ReadAndSplitMutectVCFs

Read and split Mutect VCF files.

## **Description**

Read and split Mutect VCF files.

### Usage

```
ReadAndSplitMutectVCFs(vector.of.file.paths)
```

## **Arguments**

```
vector.of.file.paths
```

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. SNS VCF with only single nucleotide substitutions.
- 2. DNS VCF with only doublet nucleotide 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

Read And Split Strelka SNSVCFs

Read and split Strelka SNS VCF files.

## **Description**

Read and split Strelka SNS VCF files.

## Usage

ReadAndSplitStrelkaSNSVCFs(vector.of.file.paths)

#### **Arguments**

vector.of.file.paths

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

## Value

A list of 3 in-memory objects as follows:

- 1. SNS.vcfs List of data.frames of pure SNS mutations no DNS or 3+BS mutations.
- 2. DNS.vcfs List of data.frames of pure DNS mutations no SNS 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 SNSs or DNSs.

#### See Also

StrelkaSNSVCFFilesToCatalog

14 ReadStrelkaIDVCFs

ReadCatalog	Read catalog.
-------------	---------------

## Description

Read a catalog in standardized format from path.

## Usage

```
ReadCatalog(path, ref.genome, region, catalog.type, strict = TRUE)
```

## **Arguments**

path Path to a catalog on disk in the standardized format. ref.genome A ref.genome argument as described in ICAMS.

region One of "genome", "exome".

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 in standard in-memory format with attributes added. See as.catalog for more details.

## Note

In the ID (insertion and deletion) catalog, deletion repeat size ranges from 0 to 5+, but for plotting and end user documentation it ranges from 1 to 6+.

ReadStrelkaIDVCFs Read Strelka ID (insertion and deletion) VCF files	
--	--

#### **Description**

Read Strelka ID (insertion and deletion) VCF files.

## Usage

```
ReadStrelkaIDVCFs(vector.of.file.paths)
```

## Arguments

```
vector.of.file.paths
```

Character vector of file paths to the VCF files.

revc 15

#### Value

A list of vcfs from vector.of.file.paths.

#### Note

In the ID (insertion and deletion) catalog, deletion repeat size ranges from 0 to 5+, but for plotting and end user documentation it ranges from 1 to 6+.

#### See Also

StrelkaIDVCFFilesToCatalog

revc

Reverse complement every string in string.vec.

## **Description**

Reverse complement every string in string.vec.

#### Usage

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

## **Arguments**

string.vec

a vector of type character.

## Value

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

```
StrelkaIDVCFFilesToCatalog
```

Create ID (indel) catalog from Strelka ID VCF files

## Description

Create ID (indel) catalog from the Strelka ID VCFs specified by vector.of.file.paths

#### Usage

```
StrelkaIDVCFFilesToCatalog(vector.of.file.paths, ref.genome, region)
```

### **Arguments**

```
vector.of.file.paths
```

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

#### **Details**

This function calls VCFsToIDCatalogs

### Value

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

#### Note

In the ID (insertion and deletion) catalog, deletion repeat size ranges from 0 to 5+, but for plotting and end user documentation it ranges from 1 to 6+.

 ${\tt StrelkaSNSVCFFilesToCatalog}$ 

Create SNS and DNS catalogs from Strelka SNS VCF files.

## **Description**

Create 3 SNS catalogs (96, 192, 1536) and 3 DNS catalogs (78, 136, 144) from the Strelka SNS VCFs specified by vector.of.file.paths

#### Usage

```
StrelkaSNSVCFFilesToCatalog(vector.of.file.paths, ref.genome, trans.ranges,
  region)
```

## **Arguments**

vector.of.file.paths

Character vector of file paths to the Strelka SNS 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 acting as a region identifier, one of "genome", "exome".

#### **Details**

This function calls VCFsToSNSCatalogs and VCFsToDNSCatalogs.

### Value

A list of 3 SNS catalogs (one each for 96, 192, and 1536) and 3 DNS catalogs (one each for 78, 136, and 144). Each catalog has attributes added. See as .catalog for more details.

### Note

SNS 192 and DNS 144 catalog only contains mutations in transcribed regions.

TranscriptRanges 17

TranscriptRanges

Transcript ranges data

### **Description**

Transcript ranges and strand information for a particular reference genome.

### Usage

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

#### **Format**

A data.table which contains transcript range and strand information for a particular reference genome. It contains chromosome name, start, end position, strand information and gene name and is keyed by chrom, chromStart, and chromEnd. It uses one-based coordinate system.

#### Details

trans.ranges.GRCh37 A data.table which contains transcript range and strand information for **Human** GRCh37. It is derived from a raw **GFF3** format file (ftp://ftp.ebi.ac.uk/pub/databases/gencode/Gencode\_human/releftrom which only the following four gene types are kept to facilitate transcriptional strand bias analysis: protein\_coding, retained\_intron, processed\_transcript and nonsense\_mediated\_decay. Needed for StrelkaSNSVCFFilesToCatalog, MutectVCFFilesToCatalog, VCFsToSNSCatalogs and VCFsToDNSCatalogs.

trans.ranges.GRCh38 A data.table which contains transcript range and strand information for **Human** GRCh38. It is derived from a raw **GFF3** format file (ftp://ftp.ebi.ac.uk/pub/databases/gencode/Gencode\_human/releftom which only the following four gene types are kept to facilitate transcriptional strand bias analysis: protein\_coding, retained\_intron, processed\_transcript and nonsense\_mediated\_decay. Needed for StrelkaSNSVCFFilesToCatalog, MutectVCFFilesToCatalog, VCFsToSNSCatalogs and VCFsToDNSCatalogs.

TransformCatalog

Transform between count and density catalogs and signatures.

## **Description**

Transform between count and density catalogs and signatures.

## Usage

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

18 VCFsToDNSCatalogs

#### **Arguments**

```
catalog An SNS or DNS catalog as described in ICAMS; must not be an ID (indel) catalog.

target.ref.genome

A ref.genome argument as described in ICAMS.

target.region One of "genome", "exome".

target.catalog.type

A character string acting as a catalog type identifier, one of "counts", "density", "counts.signature", "density.signature".
```

#### **Details**

Only the following transformations are legal:

```
1. counts -> counts
```

- 2. counts -> density
- 3. counts -> (counts.signature,density.signature)
- 4. density -> counts (in which case the semantics are to infer the genome-wide or exome-wide counts based on the densities.)
- 5. density -> (counts.signature, density.signature)
- 6. (counts.signature, density.signature) ->
   (counts.signature, density.signature)
   (density.signature -> density.signature is a null operation.)
- 7. density -> density (A null operation.)

## Value

A catalog as defined in ICAMS.

VCFsToDNSCatalogs

Create DNS catalogs from VCFs

### **Description**

Create a list of 3 catalogs (one each for DNS78, DNS144 and DNS136) out of the contents in list.of.DNS.vcfs. The VCFs must not contain any type of mutation other then DNSs.

## Usage

```
VCFsToDNSCatalogs(list.of.DNS.vcfs, ref.genome, trans.ranges, region)
```

#### **Arguments**

list.of.DNS.vcfs

List of in-memory data frames of pure DNS mutations – no SNS 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 acting as a region identifier, one of "genome", "exome".

VCFsToIDCatalogs 19

#### Value

A list of 3 DNS catalogs, one each for 78, 144, 136: catDNS78 catDNS144 catDNS136. Each catalog has attributes added. See as.catalog for more details.

## Note

DNS 144 catalog only contains mutations in transcribed regions.

VCFsToIDCatalogs

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)
```

## **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.

VCFsToSNSCatalogs

Create SNS catalogs from SNS VCFs

#### **Description**

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

## Usage

```
VCFsToSNSCatalogs(list.of.SNS.vcfs, ref.genome, trans.ranges, region)
```

20 WriteCatalog

### **Arguments**

list.of.SNS.vcfs

List of in-memory data frames of pure SNS mutations - no DNS or 3+BS mu-

tations. 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 acting as a region identifier, one of "genome", "exome".

#### Value

A list of 3 SNS catalogs, one each for 96, 192, 1536: catSNS96 catSNS192 catSNS1536. Each catalog has attributes added. See as .catalog for more details.

#### Note

SNS 192 catalog only contains mutations in transcribed regions.

WriteCatalog Write a catalog

## **Description**

Write a catalog to a file.

## Usage

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

## Arguments

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

path 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.

## Note

In the ID (insertion and deletion) catalog, deletion repeat size ranges from 0 to 5+, but for plotting and end user documentation it ranges from 1 to 6+.

# **Index**

17

```
*Topic datasets
                                                  trans.ranges.GRCh38 (TranscriptRanges),
    CatalogRowOrder, 3
    TranscriptRanges, 17
                                                  TranscriptRanges, 9, 16, 17, 18, 20
                                                  TransformCatalog, 8, 17
as.catalog, 2, 7, 9–12, 14, 16, 19, 20
available.genomes, 7
                                                  VCFsToDNSCatalogs, 9, 16, 17, 18
                                                  VCFsToIDCatalogs, 9, 16, 19
BSgenome, 7
                                                  VCFsToSNSCatalogs, 9, 16, 17, 19
catalog.row.order(CatalogRowOrder), 3
                                                  WriteCatalog, 8, 14, 20
CatalogRowOrder, 3, 8
Collapse144CatalogTo78
        (CollapseCatalog), 3
Collapse1536CatalogTo96
         (CollapseCatalog), 3
Collapse192CatalogTo96
         (CollapseCatalog), 3
CollapseCatalog, 3, 8
FindDelMH, 4
GetMutectVAF (GetVAF), 6
GetStrelkaVAF (GetVAF), 6
GetVAF, 6
ICAMS, 2-4, 6, 7, 9-12, 14-16, 18-20
ICAMS-package (ICAMS), 6
MutectVCFFilesToCatalog, 7, 9, 13, 17
PlotCatalog, 8, 10
PlotCatalogToPdf, 8, 11
PreserveCatalogAttribute, 12
ReadAndSplitMutectVCFs, 12
ReadAndSplitStrelkaSNSVCFs, 13
ReadCatalog, 7, 8, 14, 20
{\tt ReadStrelkaIDVCFs}, \textcolor{red}{14}
revc, 15
StrelkaIDVCFFilesToCatalog, 7, 15, 15
StrelkaSNSVCFFilesToCatalog, 7, 13, 16,
         17
trans.ranges.GRCh37 (TranscriptRanges),
```