# **ICAMS**

# January 14, 2019

```
Type Package
Title In-depth Characterization and Analysis of Mutational Signatures
Version 0.0.0.9000
Author Steve Rozen, Nanhai Jiang, Arnoud Boot
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Description This package has functions to read in VCF files from Strelka and GATK,
      create SNS, DNS, ID catalogs and do different types of plotting.
License GPL-3
Encoding UTF-8
LazyData true
Imports data.table,
      dplyr,
      Biostrings,
      BSgenome,
      BSgenome. Hsapiens. 1000 genomes. hs37d5,
      graphics,
      grDevices,
      GenomicRanges,
      methods,
      RColorBrewer,
      RCurl,
      stringr,
      utils
Depends R (>= 2.10)
RoxygenNote 6.1.1
Suggests knitr,
      rmarkdown,
      testthat
VignetteBuilder knitr
Collate 'utility_functions.R'
      'VCF_related_functions.R'
      'DNS_related_functions.R'
      'INDELS_related_functions.R'
      'SNS_related_functions.R'
      'catalog_related_functions.R'
      'plot_DNS_catalog.R'
```

2 R topics documented:

'plot\_INDELS\_catalog.R'
'plot\_SNS\_catalog.R'
'read\_write\_catalog.R'
'test\_functions.R'

# R topics documented:

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AddSequence

Add sequence context to a data frame with mutation records

# Description

Add sequence context to a data frame with mutation records

## Usage

AddSequence(df, seq = BSgenome.Hsapiens.1000genomes.hs37d5)

# Arguments

df An input data frame storing mutation records of a VCF file.

seq A particular reference genome.

# Value

A data frame with a new column added to the input data frame, which contains sequence context information.

4 AddTranscript

Add Sequence ID Add sequence context to a data frame with mutation records
--

### **Description**

Add sequence context to a data frame with mutation records

#### Usage

```
AddSequenceID(df, seq = BSgenome.Hsapiens.1000genomes.hs37d5)
```

#### **Arguments**

df A data frame storing mutation records of a VCF file. IMPORTANT: The rep-

resentation of indels in df must have been canonicalized, so that context bases (which are added by some indel callers) are placed in a column "Left.context.base" and so that, for deletions, ALT is the empty string, and, for insertions, REF is

the empty string.

seq A particular reference genome.

#### Value

A data frame with 2 new columns added to the input data frame. One column contains sequence context information and the other column contains the length of the "context" string to the left of the site of the variant.

AddTranscript Add transcript information to a data frame with mutation records	
--	--

## **Description**

Add transcript information to a data frame with mutation records

## Usage

```
AddTranscript(df, trans.ranges)
```

#### **Arguments**

df A data frame storing mutation records of a VCF file.

trans.ranges A data.table with the genomic ranges and strands of transcripts.

#### Value

A data frame with new columns added to the input data frame, which contain the mutated gene's name, range and strand information.

Canonicalize1DEL 5

Canonicalize1DEL Can

Canonicalize1DEL

# Description

Canonicalize1DEL

# Usage

```
Canonicalize1DEL(ref, alt, context)
```

# **Arguments**

ref TODO alt TODO context TODO

## Value

**TODO** 

Canonicalize1ID

Canonicalize1ID

# Description

Canonicalize1ID

# Usage

```
Canonicalize1ID(ref, alt, context)
```

# Arguments

ref TODO alt TODO context TODO

### Value

6 CanonicalizeDNS

Canonicalize1INS

Canonicalize1INS

# Description

Canonicalize1INS

# Usage

```
Canonicalize1INS(ref, alt, context)
```

## Arguments

ref TODO alt TODO context TODO

### Value

TODO

CanonicalizeDNS

CanonicalizeDNS

# Description

CanonicalizeDNS

# Usage

```
CanonicalizeDNS(ref.vec, alt.vec)
```

# Arguments

ref.vec TODO alt.vec TODO

#### Value

CanonicalizeID 7

CanonicalizeID

 ${\it Canonicalize ID}$ 

# Description

CanonicalizeID

# Usage

```
CanonicalizeID(ref, alt, context)
```

# Arguments

ref TODO alt TODO context TODO

# Value

TODO

CanonicalizeQUAD

Canonicalize QUAD

# Description

CanonicalizeQUAD

# Usage

CanonicalizeQUAD(quad)

# Arguments

quad

TODO

# Value

8 Cat192StrandToPdf

Plot the 1536 mutation catalog of >= 1 samples to a PDF file

### **Description**

Plot the 1536 mutation catalog of >= 1 samples to a PDF file

#### Usage

```
Cat1536ToPdf(catalog, name, id = colnames(catalog), abundance)
```

#### **Arguments**

catalog A matrix whose rownames indicate the 1536 SNS mutation types while its

columns contain the counts of each mutation type from different samples. The mutation types are in six-letters like CATTAT, first 2-letters CA refers to (-2, -1) position, third letter T refers to the base which has mutation, next second 2-letters TA refers to (+1, +2) position, last letter T refers to the base after mu-

tation.

name Name of the PDF file to be produced.

id A vector containing the identifier of each sample.

abundance A matrix containing pentanucleotide abundance information.

#### Value

invisible(TRUE)

Cat192StrandToPdf Plot the transcription strand bias graph of 6 SNS mutation types

 $("C>A", "C>G", "C>T", "T>A", "T>C", "T>G") \ of \ different \ samples$ 

to a PDF file.

### **Description**

Plot the transcription strand bias graph of 6 SNS mutation types ("C>A", "C>G", "C>T", "T>A", "T>C", "T>G") of different samples to a PDF file.

# Usage

```
Cat192StrandToPdf(catalog, name, id = colnames(catalog),
  type = "counts", cex = 1, abundance = NULL)
```

Cat192ToPdf 9

#### **Arguments**

catalog A matrix whose rownames indicate the 192 SNS mutation types while its columns

contain the counts of each mutation type from different samples.

name The name of the PDF file to be produced.

The ID information of the sample which has mutations.

type A vector of values indicating the type of graph for each sample. If type =

"counts", the graph will plot the occurrences of the 192 mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the sample. If type = "density", the graph will plot the rates of mutations per million trinucleotides for each mutation type. The default value for type is "counts".

cex A numerical value giving the amount by which mutation class labels, y axis

labels, sample name and legend should be magnified relative to the default.

abundance A matrix containing trinucleotide abundance and strand information, to be used

only when type = "density".

#### Value

invisible(TRUE)

Cat192ToPdf

Plot the SNS 192 mutation catalog of different samples to a PDF file

#### Description

Plot the SNS 192 mutation catalog of different samples to a PDF file

#### Usage

```
Cat192ToPdf(catalog, name, id = colnames(catalog), type = "counts",
  cex = 0.8, abundance = NULL)
```

#### **Arguments**

catalog A matrix whose rownames indicate the 192 SNS mutation types while its columns

contain the counts of each mutation type from different samples.

name The name of the PDF file to be produced.

id The ID information of the sample which has mutations.

type A vector of values indicating the type of graph for each sample. If type =

"counts", the graph will plot the occurrences of the 192 mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the sample. If type = "density", the graph will plot the rates of mutations per million trinucleotides for each mutation type. The default value for type is "counts".

cex A numerical value giving the amount by which mutation class labels on top of

graph, y axis labels and sample name should be magnified relative to the default.

abundance A matrix containing trinucleotide abundance and strand information, to be used

only when type = "density".

#### Value

invisible(TRUE)

10 CatDNS144ToPdf

Cat96ToPdf	Plot the SNS 96 mutation catalog of different samples to a PDF file

#### **Description**

Plot the SNS 96 mutation catalog of different samples to a PDF file

### Usage

```
Cat96ToPdf(catalog, name, id = colnames(catalog), type = "density",
 abundance = NULL)
```

### Arguments

catalog A matrix whose rownames in	dicate the 96 SNS mutation types while its columns
------------------------------------	--

contain the counts of each mutation type from different samples.

The name of the PDF file to be produced. name

id A vector containing the ID information of different samples.

A vector of values indicating the type of plot for each sample. If type = "dentype

> sity", the graph will plot the rates of mutations per million trinucleotides for each mutation type. If type = "counts", the graph will plot the occurrences of the 96 mutation types in the sample. If type = "signature", the graph will plot

mutation signatures of the sample. The default value for type is "density".

abundance A matrix containing trinucleotide abundance information. To be used only when

type = "density".

#### Value

invisible(TRUE)

CatDNS144ToPdf Plot the transcription strand bias graph of 10 major DNS muta-

tion types ("AC>NN", "AT>NN", "CC>NN", "CG>NN", "CT>NN", "GC>NN", "TA>NN", "TC>NN", "TG>NN", "TT>NN") of different

samples to a PDF file.

#### **Description**

Plot the transcription strand bias graph of 10 major DNS mutation types ("AC>NN", "AT>NN", "CC>NN", "CG>NN", "CT>NN", "GC>NN", "TA>NN", "TC>NN", "TG>NN", "TT>NN") of different samples to a PDF file.

### Usage

```
CatDNS144ToPdf(catalog, name, id = colnames(catalog), type = "counts",
  cex = 1, abundance = NULL)
```

CatDNS78ToPdf 11

#### **Arguments**

catalog A matrix whose rownames indicate the 144 DNS mutation types while its columns

contain the counts of each mutation type from different samples.

name The name of the PDF file to be produced.

id The ID information of the sample which has mutations.

type A vector of values indicating the type of graph for each sample. If type =

"counts", the graph will plot the occurrences of the 10 major DNS mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the 10 major DNS mutation types in the sample. If type = "density", the graph will plot the rates of mutations per million dinucleotides for each of the

10 major DNS mutation types. The default value for type is "counts".

cex A numerical value giving the amount by which mutation class labels, y axis

labels, sample name and legend should be magnified relative to the default.

abundance A matrix containing dinucleotide abundance and strand information, to be used

only when type = "density".

#### Value

invisible(TRUE)

CatDNS78ToPdf Plot the DNS 78

Plot the DNS 78 mutation catalog of different samples to a PDF file

## Description

Plot the DNS 78 mutation catalog of different samples to a PDF file

#### Usage

```
CatDNS78ToPdf(catalog, name, id = colnames(catalog), type = "density",
  abundance = NULL)
```

#### Arguments

catalog A matrix whose rownames indicate the 78 DNS mutation types while its columns

contain the counts of each mutation type from different samples.

name The name of the PDF file to be produced.

id A vector containing the ID information of different samples.

type A vector of values indicating the type of plot for each sample. If type = "den-

sity", the graph will plot the rates of mutations per million nucleotides for each mutation type. If type = "counts", the graph will plot the occurrences of the 78 mutation types in the sample. If type = "signature", the graph will plot mutation

signatures of the sample. The default value for type is "density".

abundance A matrix containing dinucleotide abundance information, to be used only when

type = "density".

# Value

invisible(TRUE)

file	CatIDToPdf	Plot the insertion and deletion catalog of different samples to a PDF file
------	------------	--

## **Description**

Please take note that the deletions Repeat Size ranges from 0 to 5+ in the catalog, but for plotting and end user documentation it ranges from 1 to 6+.

#### Usage

```
CatIDToPdf(catalog, name, id = colnames(catalog), type = "counts")
```

### **Arguments**

catalog A matrix whose rownames indicate the insertion and deletion mutation types

while its column contains the counts of each mutation type from different sam-

ples.

name The name of the PDF file to be produced.

id A vector containing the ID information of different samples.

type A vector of values indicating the type of plot for each sample. If type = "counts",

the graph will plot the occurrences of the insertion and deletion mutation types in the sample. If type = "signature", the graph will plot mutation signatures of

the sample. The default value for type is "counts".

## Value

invisible(TRUE)

CheckSeqContextInVCF Check that the sequence context information is consistent with the

value of the column REF.

### **Description**

Check that the sequence context information is consistent with the value of the column REF.

## Usage

```
CheckSeqContextInVCF(vcf, column.to.use)
```

### **Arguments**

vcf In-memory VCF as a data.frame; must be an SNS or DNS VCF.

column.to.use The column name as a string of the column in the VCF with the context infor-

mation

Collapse144to78 13

#### Value

Throws error with location information if the value of REF is inconsistent with the value of seq.21context. Assumes the first base of the reference allele is at position (size(<context string>)-1)/2, and generates error if this is not an integer. Indices are 1-based.

Collapse144to78

Collapse a DNS 144 catalog matrix to a DNS 78 catalog matrix

### **Description**

Collapse a DNS 144 catalog matrix to a DNS 78 catalog matrix

#### Usage

Collapse144to78(catDNS144)

#### **Arguments**

catDNS144

A DNS 144 catalog matrix whose row names indicate the 192 mutation types while its columns show the occurrences of each mutation type of different samples.

#### Value

A DNS 78 catalog matrix whose row names indicate the 96 mutation types while its columns show the occurrences of each mutation type of different samples.

Collapse1536to96

Collapse a SNS 1536 catalog matrix to a 96 catalog matrix

## Description

Collapse a SNS 1536 catalog matrix to a 96 catalog matrix

### Usage

Collapse1536to96(cat1536)

#### **Arguments**

cat1536

A SNS 1536 catalog matrix whose row names indicate the 1536 mutation types while its columns show the occurrences of each mutation type of different samples.

#### Value

A SNS 96 catalog matrix whose row names indicate the 96 mutation types while its columns show the occurrences of each mutation type of different samples.

Collapse192to96

Collapse a SNS 192 catalog matrix to a 96 catalog matrix

#### **Description**

Collapse a SNS 192 catalog matrix to a 96 catalog matrix

### Usage

Collapse192to96(cat192)

### **Arguments**

cat192

A SNS 192 catalog matrix whose row names indicate the 192 mutation types while its columns show the occurrences of each mutation type of different samples.

#### Value

A SNS 96 catalog matrix whose row names indicate the 96

CreateOneColDNSCatalog

Create double nucleotide catalog for \*one\* sample from a Variant Call Format (VCF) file

### **Description**

Create double nucleotide catalog for \*one\* sample from a Variant Call Format (VCF) file

#### Usage

```
CreateOneColDNSCatalog(vcf, sample.id = "count")
```

# **Arguments**

vcf An in-memory VCF file annotated by the AddSequence and AddTranscript func-

tions. It must \*not\* contain indels and must \*not\* contain SNS (single nu-

cleotide substituions), or triplet base substituions etc.

sample.id Usually the sample id, but defaults to "count".

### Value

A list of three matrices containing the DNS catalog: catDNS78, catDNS144, catQUAD136 respectively.

CreateOneColIDCatalog Create an indel (ID) mutation catalog for \*one\* sample from a Variant Call Format (VCF) file

#### **Description**

Create an indel (ID) mutation catalog for \*one\* sample from a Variant Call Format (VCF) file

#### Usage

CreateOneColIDCatalog(ID.vcf, SBS.vcf)

#### **Arguments**

ID.vcf

An in-memory VCF as a data.frame annotated by the AddSequence and AddTranscript functions. It must only contain indels and must \*not\* contain SBS (single base substituions), DBS, or triplet base substituions etc.

\* Sequence must already have been added to ID.vcf

One design decision for variant callers is the representation of "complex indels", e.g. mutations e.g. CAT > GC. Some callers represent this as C>G, A>C, and T>\_. Others might represent it as CAT > CG. Multiple issues can arise. In PCAWG, overlapping indel/SBS calls from different callers were included in

the indel VCFs.

SBS.vcf

An in-memory VCF as a data frame. Because we have to work with some PCAWG data, we will look for neigboring indels and indels adjoining SBS. That means this functions takes an SBS VCF and an ID VCF from the same sample.

## Value

A list with two elemsents: ID.cat: A 1-column matrix containing the mutation catalog information. problems: Locations of neighboring indels or indels neighboring SBS. In the future we might handle these depending on what we find in the indel calls from different variant callers. TODO(steve) Is problems implemented?

CreateOneColSNSCatalog

Create single nucleotide mutation catalog for \*one\* sample from a Variant Call Format (VCF) file.

## **Description**

Create single nucleotide mutation catalog for \*one\* sample from a Variant Call Format (VCF) file.

#### **Usage**

```
CreateOneColSNSCatalog(vcf, sample.id = "count")
```

16 DNSVCFsToCatalogs

#### **Arguments**

vcf An in-memory VCF file annotated by the AddSequence and AddTranscript func-

tions. It must \*not\* contain indels and must \*not\* contain DNS (double nucleotide substituions), or triplet base substituions etc., even if encoded as neigh-

boring SNS.

sample.id Usually the sample id, but defaults to "count".

#### Value

A list of three matrices containing the SNS mutation catalog: 96, 192, 1536 catalog respectively.

CreateTransRange

Create a Transcript Range file from the raw GFF3 File

#### **Description**

Create a Transcript Range file from the raw GFF3 File

### Usage

CreateTransRange(path)

### **Arguments**

path

The name/path of the raw GFF3 File, or a complete URL.

#### Value

A data frame which contains chromosome name, start, end position, strand information and gene name. Only the following four gene types are kept to facilitate transcriptional strand bias analysis: protein\_coding, retained\_intron, processed\_transcript and nonsense\_mediated\_decay.

DNSVCFsToCatalogs

Create a list of 3 catalogs (one each for DNS78, DNS144 and QUAD136) out of the contents of the VCFs in list.of.vcfs

# Description

Create a list of 3 catalogs (one each for DNS78, DNS144 and QUAD136) out of the contents of the VCFs in list.of.vcfs

#### Usage

DNSVCFsToCatalogs(list.of.vcfs, genome, trans.ranges)

## Arguments

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

output catalog.

genome Name of a particular reference genome (without quotations marks).

trans.ranges A data frame containing transcript ranges.

FindDelMH 17

#### Value

A list of 3 catalogs, one each for DNS78, DNS144, QUAD136: catDNS78 catDNS144 catQUAD136

|--|

## Description

Microhomology can be alligned in multiple equivalent ways. Example:

### Usage

```
FindDelMH(context, q, pos)
```

### **Arguments**

context	TODO
q	TODO
pos	TODO

#### **Details**

GGCTAGTT aligned to

All the same pairs of sequence, aligned 5 different ways. 4 bp of microhomology.

Need to find:

- (1) The maxium match of undeleted sequence on left that is identical to the right end of deleted sequence, and
- (2) The maxium match of undeleted sequence on right that is identical to the left end of deleted sequence.

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

#### Value

18 FindMaxRepeatIns

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

TODO(Steve): check this statement; what if there is no repeat?

## Description

```
e.g. q = ac pos = 3 context = xyaczt pos ^ Return 1
```

## Usage

FindMaxRepeatDel(context, q, pos)

### **Arguments**

context A string that embeds q at position pos

q A substring of context at pos to pos + nchar(q) - 1

pos The position of q

### **Details**

```
If substr(context, pos, pos + nchar(q) - 1) != q then stop
```

#### Value

The number of repeat units in which q is embedded.

FindMaxRepeatIns FindMaxRepeatIns

### **Description**

If q is an insertion into context between pos and pos+1 if q is repeated in context it might start at pos+1:

## Usage

FindMaxRepeatIns(context, q, pos)

# Arguments

context	TODO
q	TODO
pos	TODO

#### **Details**

```
e.g. q = ac pos = 4 context = abxyac pos ^ start ^
or q might start at pos + 1 - len(q)
e.g. q = ac pos = 4 context = xyaczz pos ^ start ^
```

GetStrelkaVAF

#### Value

**TODO** 

GetStrelkaVAF Extract the VAFs (variant allele frequencies) from a VAF created by Strelka version 1

## **Description**

Extract the VAFs (variant allele frequencies) from a VAF created by Strelka version 1

### Usage

```
GetStrelkaVAF(strelka.vcf)
```

#### **Arguments**

strelka.vcf said VCF as a data.frame

#### Value

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

MakeVCFDNSdf

Take DNS ranges and the original VCF and generate a VCF with dinucleotide REF and ALT alleles. The output VCF has minimal columns: just CHROM, POS, ID, REF, ALT.

## Description

Take DNS ranges and the original VCF and generate a VCF with dinucleotide REF and ALT alleles. The output VCF has minimal columns: just CHROM, POS, ID, REF, ALT.

## Usage

```
MakeVCFDNSdf(DNS.range.df, SNS.vcf.dt)
```

# Arguments

 ${\tt DNS.range.df} \qquad {\tt Data\ frame\ with\ columns\ CHROM,\ LOW,\ HIGH}$ 

SNS.vcf.dt TODO

#### Value

20 PlotCat192

PlotCat1536	Plot the pentanucleotide sequence contexts for one sample, normalized by pentanucleotide occurrence in the genome.
	by peniunucieotide occurrence in the genome.

## **Description**

Plot the pentanucleotide sequence contexts for one sample, normalized by pentanucleotide occurrence in the genome.

## Usage

```
PlotCat1536(catalog, id, scale = TRUE, abundance)
```

#### **Arguments**

catalog A matrix whose rownames indicate the 1536 SNS mutation types while its col-

umn contains the counts of each mutation type. The mutation types are in sixletters like CATTAT, first 2-letters CA refers to (-2, -1) position, third letter T refers to the base which has mutation, next second 2-letters TA refers to (+1, +2)

position, last letter T refers to the base after mutation.

id The id of the sample to be displayed on top of the graph.

scale A logical value indicating whether to do color scaling for all mutation types.

abundance A matrix containing pentanucleotide abundance information.

#### Value

invisible(TRUE)

PlotCat192	Plot the SNS 192 mutation catalog of one sample
------------	---

#### **Description**

Plot the SNS 192 mutation catalog of one sample

#### Usage

```
PlotCat192(catalog, id, type = "counts", cex = 0.8, abundance = NULL)
```

## Arguments

catalog A matrix whose rownames in	indicate the 192 SNS mutation types while its col-
------------------------------------	--

umn contains the counts of each mutation type.

id The ID information of the sample which has mutations.

type A value indicating the type of the graph. If type = "counts", the graph will plot

the occurrences of the 192 mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the sample. If type = "density", the graph will plot the rates of mutations per million trinucleotides for each mutation

type. The default value for type is "counts".

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cex A numerical value giving the amount by which mutation class labels on top of

graph, y axis labels and sample name should be magnified relative to the default.

abundance A matrix containing trinucleotide abundance and strand information, to be used

only when type = "density".

#### Value

invisible(TRUE)

PlotCat192Strand Plot the transcription strand bias graph of 6 SNS mutation types

("C>A", "C>G", "C>T", "T>A", "T>C", "T>G") in one sample

## **Description**

Plot the transcription strand bias graph of 6 SNS mutation types ("C>A", "C>G", "C>T", "T>A", "T>C", "T>G") in one sample

## Usage

```
PlotCat192Strand(catalog, id, type = "counts", cex = 1,
  abundance = NULL)
```

## **Arguments**

catalog A matrix whose rownames indicate the 192 SNS mutation types while its col-

umn contains the counts of each mutation type.

id The ID information of the sample which has mutations.

type A value indicating the type of the graph. If type = "counts", the graph will plot

the occurrences of the 6 SNS mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the 6 SNS mutation types in the sample. If type = "density", the graph will plot the rates of mutations per million trinucleotides for each of the 6 SNS mutation types. The default value for type

is "counts".

cex A numerical value giving the amount by which mutation class labels, y axis

labels, sample name and legend should be magnified relative to the default.

abundance A matrix containing trinucleotide abundance and strand information, to be used

only when type = "density".

#### Value

invisible(TRUE)

PlotCatDNS144

PlotCat96	Plot the SNS 96 mutation catalog of one sample	
-----------	--	--

### **Description**

Plot the SNS 96 mutation catalog of one sample

## Usage

```
PlotCat96(catalog, id, type = "density", abundance = NULL)
```

## **Arguments**

catalog A matrix whose rownames indicate the 96 SNS mutation types while its colum	catalog	A matrix whose rownames indicate the 96 SNS mutation types while its columns
--	---------	--

contain the counts of each mutation type.

id The ID information of the sample which has mutations.

type A value indicating the type of the graph. If type = "density", the graph will

plot the rates of mutations per million trinucleotides for each mutation type. If type = "counts", the graph will plot the occurrences of the 96 mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the

sample. The default value for type is "density".

abundance A matrix containing trinucleotide abundance information. To be used only when

type = "density".

## Value

invisible(TRUE)

PlotCatDNS144	Plot the transcription strand bias graph of 10 major DNS muta-
	tion types ("AC>NN", "AT>NN", "CC>NN", "CG>NN", "CT>NN",
	"GC>NN", "TA>NN", "TC>NN", "TG>NN", "TT>NN") in one sam-

ple.

### **Description**

Plot the transcription strand bias graph of 10 major DNS mutation types ("AC>NN", "AT>NN", "CC>NN", "CG>NN", "CT>NN", "GC>NN", "TA>NN", "TC>NN", "TG>NN", "TT>NN") in one sample.

## Usage

```
PlotCatDNS144(catalog, id, type = "counts", cex = 1,
  abundance = NULL)
```

PlotCatDNS78 23

#### **Arguments**

catalog A matrix whose rownames indicate the 144 DNS mutation types while its col-

umn contains the counts of each mutation type.

id The ID information of the sample which has mutations.

type A value indicating the type of the graph. If type = "counts", the graph will

plot the occurrences of the 10 major DNS mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the 10 major DNS mutation types in the sample. If type = "density", the graph will plot the rates of mutations per million dinucleotides for each of the 10 major DNS mutation

types. The default value for type is "counts".

cex A numerical value giving the amount by which mutation class labels, y axis

labels, sample name and legend should be magnified relative to the default.

abundance A matrix containing dinucleotide abundance and strand information, to be used

only when type = "density".

#### Value

invisible(TRUE)

PlotCatDNS78 Plot the DNS 78 mutation catalog of one sample

#### **Description**

Plot the DNS 78 mutation catalog of one sample

#### Usage

```
PlotCatDNS78(catalog, id, type = "density", abundance = NULL)
```

#### **Arguments**

catalog A matrix whose rownames indicate the 78 DNS mutation types while its columns

contain the counts of each mutation type from different samples.

id The ID information of the sample which has mutations.

type A value indicating the type of the graph. If type = "density", the graph will plot

the rates of mutations per million nucleotides for each mutation type. If type = "counts", the graph will plot the occurrences of the 78 mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the

sample. The default value for type is "density".

abundance A matrix containing dinucleotide abundance information, to be used only when

type = "density".

#### Value

invisible(TRUE)

24 PyrPenta

PlotCatID	Plot the insertion and deletion catalog of one sample.

## Description

Please take note that the deletions Repeat Size ranges from 0 to 5+ in the catalog, but for plotting and end user documentation it ranges from 1 to 6+.

### Usage

```
PlotCatID(catalog, id, type = "counts")
```

## Arguments

catalog A matrix whose rownames indicate the insertion and deletion mutation types

while its column contains the counts of each mutation type.

id The ID information of the sample which has mutations.

type A value indicating the type of the graph. If type = "counts", the graph will plot

the occurrences of the insertion and deletion mutation types in the sample. If type = "signature", the graph will plot mutation signatures of the sample. The

default value for type is "counts".

#### Value

invisible(TRUE)

PyrPenta	PyrPenta

## Description

PyrPenta

#### Usage

PyrPenta(mutstring)

### **Arguments**

mutstring a mutation string

## Value

a mutation string

PyrTri 25

PyrTri PyrTri

## Description

PyrTri

# Usage

PyrTri(mutstring)

## Arguments

mutstring

a mutation string

## Value

a mutation string

ReadAbundance3Bp

Read data from a nucleotide abundance file with 3 base pairs

# Description

Read data from a nucleotide abundance file with 3 base pairs

## Usage

ReadAbundance3Bp(path)

## Arguments

path

Path to the file with the nucleotide abundance information with 3 base pairs.

#### Value

A matrix whose row names indicate 32 different types of 3 base pairs combinations while its column contains the occurrences of each type.

26 ReadAbundance5Bp

ReadAbundance4Bp

Read data from a nucleotide abundance file with 4 base pairs

## Description

Read data from a nucleotide abundance file with 4 base pairs

### Usage

ReadAbundance4Bp(path)

### **Arguments**

path

Path to the file with the nucleotide abundance information with 4 base pairs.

#### Value

A matrix whose row names indicate 10 different types of 2 base pairs combinations while its column contains the occurrences of each type.

ReadAbundance5Bp

Read data from a nucleotide abundance file with 5 base pairs

## **Description**

Read data from a nucleotide abundance file with 5 base pairs

## Usage

ReadAbundance5Bp(path)

# Arguments

path

Path to the file with the nucleotide abundance information with 5 base pairs.

## Value

A matrix whose row names indicate 512 different types of 5 base pairs combinations while its column contains the occurrences of each type.

ReadBedTranscriptRanges

Read transcript ranges and strands from a bed format file. Mostly for testing.

### Description

Read transcript ranges and strands from a bed format file. Mostly for testing.

#### Usage

ReadBedTranscriptRanges(path)

#### **Arguments**

path

Path to the file with the transcript information (in bed format).

#### Value

A data.table keyed by chrom, chromStart, and chromEnd.

ReadCat

Read Catalog Functions

## Description

Read a catalog in PCAWG7 format from path

## Usage

```
ReadCat96(path, strict = TRUE)
ReadCat192(path, strict = TRUE)
ReadCat1536(path, strict = TRUE)
ReadCatDNS78(path, strict = TRUE)
ReadCatDNS144(path, strict = TRUE)
ReadCatQUAD136(path, strict = TRUE)
ReadCatID(path, strict = TRUE)
```

### **Arguments**

path Path to a catalog on disk in the "PCAWG7" format.

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

28 ReadListOfVCFs

#### **Details**

ReadCat96 Read a 96 SNS catalog from path

ReadCat192 Read a 192 SNS catalog from path

ReadCat1536 Read a 1536 SNS catalog from path

ReadCatDNS78 Read a 78 DNS catalog from path

ReadCatDNS144 Read a 144 DNS catalog from path

ReadCatQUAD136 Read a 136 QUAD catalog from path

ReadCatID Read a ID (insertion/deletion) catalog from path

Please take note that the deletions Repeat Size ranges from 0 to 5+ in the catalog, but for plotting and end user documentation it ranges from 1 to 6+.

#### Value

A catalog in canonical in-memory format.

ReadListOfVCFs

Read a list of VCF files from path

#### **Description**

Read a list of VCF files from path

### Usage

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

## Arguments

vector.of.file.paths

A vector containing the paths of the VCF files.

#### Value

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

ReadStrelkaVCF 29

ReadStrelkaVCF

Read in the data lines of a Variant Call Format (VCF) file

### **Description**

Read in the data lines of a Variant Call Format (VCF) file

### Usage

ReadStrelkaVCF(path)

#### **Arguments**

path

The name/path of the VCF file, or a complete URL.

#### Value

A data frame storing mutation records of a VCF file.

 ${\tt ReadTranscriptRanges}$ 

Read transcript ranges and strands from a bed format file. Use this one for the new, cut down gff3 file (2018 11 24)

# Description

Read transcript ranges and strands from a bed format file. Use this one for the new, cut down gff3 file (2018 11 24)

## Usage

ReadTranscriptRanges(path)

## **Arguments**

path

Path to the file with the transcript information with 1-based start end positions of genomic ranges.

## Value

A data.table keyed by chrom, chromStart, and chromEnd.

RevcDNS144

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 of every string in string.vec.

RevcDNS144

RevcDNS144

# Description

RevcDNS144

## Usage

RevcDNS144(mutstring)

# Arguments

mutstring

TODO

### Value

RevcSNS96

# Description

RevcSNS96

### Usage

RevcSNS96(mutstring)

## **Arguments**

mutstring

a mutation string

### Value

a mutation string

SNSVCFsToCatalogs

Create a list of 3 catalogs (one each for 96, 192, 1536) out of the contents of the VCFs in list.of.vcfs

## **Description**

Create a list of 3 catalogs (one each for 96, 192, 1536) out of the contents of the VCFs in list.of.vcfs

## Usage

```
SNSVCFsToCatalogs(list.of.vcfs, genome, trans.ranges)
```

## **Arguments**

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

output catalog.

genome Name of a particular reference genome (without quotations marks).

trans.ranges A data frame containing transcript ranges.

### Value

A list of 3 catalogs, one each for 96, 192, 1536: cat96 cat192 cat1536

32 StandardChromName

SplitSNSVCF Split an in-memory VCF into SNS, DNS, and variants involving > 2 consecutive bases	SplitSNSVCF	
--	-------------	--

### **Description**

SNSs are single nucleotide substitutions, eg C>T, A<G,.... DNSs are double nucleotide substitutions, eg CC>TT, AT>GG, ... Variants involving > 2 consecutive bases are rare, so this function just records them. These would be variants such ATG>CCT, AGAT > TCTA, ...

#### Usage

```
SplitSNSVCF(vcf.df, max.vaf.diff = 0.02)
```

## Arguments

vcf.df An in-memory data frame containing a VCF file contents.

max.vaf.diff The maximum difference of VAF, default value is 0.02.

#### Value

A list of 3 in-memory objects with the elements:

StandardChromName	Standardize the Chromosome name annotations for a data frame
-------------------	--

# Description

Standardize the Chromosome name annotations for a data frame

## Usage

StandardChromName(df)

#### **Arguments**

df

A data frame whose first column contains the Chromosome name.

## Value

A data frame whose Chromosome names are only in the form of 1:22, "X" and "Y".

TestDNSCatalog 33

TestDNSCatalog	This function is to test whether the predefined functions are working correctly to produce the desired DNS catalogs

## Description

This function is to test whether the predefined functions are working correctly to produce the desired DNS catalogs

### Usage

TestDNSCatalog(vcf.df)

## **Arguments**

vcf.df

An in-memory data frame containing a VCF file contents.

 ${\tt TestMakeCatalogFromSNSVCFs}$ 

This function is to make catalogs from the sample VCF files to compare with the expected catalog information

## Description

This function is to make catalogs from the sample VCF files to compare with the expected catalog information

## Usage

TestMakeCatalogFromSNSVCFs()

TestSNSandDNSCat	This function is to test whether the predefined functions are working
	correctly to produce the desired SNS and DNS catalogs

# Description

This function is to test whether the predefined functions are working correctly to produce the desired SNS and DNS catalogs

# Usage

TestSNSandDNSCat()

34 VCFFiles2Catalog

TestSNSCatalog	This function is to test whether the predefined functions are working correctly to produce the desired SNS catalogs

## Description

This function is to test whether the predefined functions are working correctly to produce the desired SNS catalogs

## Usage

```
TestSNSCatalog(vcf.df)
```

### **Arguments**

vcf.df An in-memory data frame containing a VCF file contents.

VCFFiles2Catalog	Create 3 SNS catalogs (96, 192, 1536) and 3 DNS catalogs (78, 136,
	144) in the VCFs specified by vector.of.file.paths

# Description

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

### Usage

```
VCFFiles2Catalog(vector.of.file.paths, genome, trans.ranges)
```

## **Arguments**

vector.of.file.paths

A vector containing the paths of the VCF files.

genome Name of a particular reference genome (without quotations marks).

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

### 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)

WriteCat 35

WriteCat	Write a matrix of mutation catalog to a file on disk

## Description

Write a matrix of mutation catalog to a file on disk

## Usage

```
WriteCat(ct, path, num.row, row.order, row.header, strict)
```

## Arguments

ct	A matrix of mutation catalog.
CC	11 mania of matanon catalog.

path The path of the file to be written on disk.

num.row The number of rows in the file to be written.

row.order The row order to be used for writing the file.

row.header The row header to be used for writing the file.

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

write a Sivs 1330 mulation calalog to a file on alsk	WriteCat1536	Write a SNS 1536 mutation catalog to a file on disk	
--	--------------	---	--

# Description

Write a SNS 1536 mutation catalog to a file on disk

# Usage

```
WriteCat1536(ct, path, strict = TRUE)
```

# Arguments

ct A matrix of SNS 1536 mutation catalog.

path The path of the file to be written on disk.

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

36 WriteCatDNS144

WriteCat192	Write a SNS 192 mutation	catalog to a file on disk
-------------	--------------------------	---------------------------

## Description

Write a SNS 192 mutation catalog to a file on disk

### Usage

```
WriteCat192(ct, path, strict = TRUE)
```

## Arguments

ct A matrix of SNS 192 mutation catalog.
path The path of the file to be written on disk.

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

Write a SNS 96 mutation catalog to a file on disk

### **Description**

Write a SNS 96 mutation catalog to a file on disk

#### Usage

```
WriteCat96(ct, path, strict = TRUE)
```

#### **Arguments**

ct A matrix of SNS 96 mutation catalog.
path The path of the file to be written on disk.

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

WriteCatDNS144 Write a DNS 144 mutation catalog to a file on disk

### **Description**

Write a DNS 144 mutation catalog to a file on disk

## Usage

```
WriteCatDNS144(ct, path, strict = TRUE)
```

### **Arguments**

ct A matrix of DNS 144 mutation catalog.
path The path of the file to be written on disk.

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

WriteCatDNS78 37

WriteCatDNS78	Write a DNS 78 mutation catalog to a file on disk
---------------	---

# Description

Write a DNS 78 mutation catalog to a file on disk

## Usage

```
WriteCatDNS78(ct, path, strict = TRUE)
```

# Arguments

ct	A matrix of DNS 78 mutation catalog.
path	The path of the file to be written on disk.
strict	If TRUE, do additional checks on the input, and stop if the checks fail.

WriteCatID	Write a ID (insertion/deletion) catalog to a file on disk	
------------	---	--

# Description

Please take note that the deletions Repeat Size ranges from 0 to 5+ in the catalog, but for plotting and end user documentation it ranges from 1 to 6+.

# Usage

```
WriteCatID(ct, path, strict = TRUE)
```

# Arguments

ct	A matrix of ID (insertion/deletion) catalog.
path	The path of the file to be written on disk.
strict	If TRUE, do additional checks on the input, and stop if the checks fail.

38 WriteCatQUAD136

W 11 0 10H1D126	***
WriteCatOUAD136	W

Write a QUAD 136 catalog to a file on disk

# Description

Write a QUAD 136 catalog to a file on disk

# Usage

```
WriteCatQUAD136(ct, path, strict = TRUE)
```

## **Arguments**

ct A matrix of QUAD 136 catalog.

path The path of the file to be written on disk.

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

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