# Package 'humarray'

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Type Package

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<b>Description</b> Utilises GRanges, data.frame or IRanges objects. Integrates gene annotation for ImmunoChip (or your custom chip) with function calls. Intuitive wrappers for annotation lookup (gene lists, exon ranges, etc) and conversion (e.g, between build 36 and 37 coordinates). Conversion between ensembl and HGNC gene ids, chip ids to rs-ids for SNP-arrays. Retrieval of chromosome and position for gene, band or SNP-ids, or reverse lookup. Simulation functions for ranges objects.
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humarray-package

Simplify Analysis and Annotation of Human Microarray Datasets

# Description

Utilises GRanges, data.frame or IRanges objects. Integrates gene annotation for ImmunoChip (or your custom chip) with function calls. Intuitive wrappers for annotation lookup (gene lists, exon ranges, etc) and conversion (e.g, between build 36 and 37 coordinates). Conversion between ensembl and HGNC gene ids, chip ids to rs-ids for SNP-arrays. Retrieval of chromosome and position for gene, band or SNP-ids, or reverse lookup. Simulation functions for ranges objects.

## **Details**

Package: humarray
Type: humarray
Version: 1.0.0
Date: 2016-03-03
License: GPL (>= 2)

This package helps to simplify common tasks in human genetics research, such as annotation lookup, conversion and labelling for GWAS analysis. Functions are provided that utilise GRanges,

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IRanges and data.frame (snpStats) objects for input and output. The new ChipInfo object, based on GRanges, once established, can provide seamless and automatic lookup for SNPs and their features within all functions, with no need to keep passing an object explicitly with each function call. By default, the annotation for immunoChip is built-in, but you can provide your own annotation for any chip to take its place. Intuitive wrappers are provided for annotation lookup (gene lists, exon ranges, cytobands, telomeres, etc) which take care of database lookup and download seamlessly in the background. Conversion between build 36 and 37 coordinates, with parameters required in most cases. New functions allow simple conversion between ensembl and HGNC gene labels, and chip specific ids to rs-ids for SNP-arrays. Simple function allow fast retrieval of chromosome and position for gene labels, karyotype bands or SNP-ids, or reverse lookup to see which features appear at, or nearest to a given location. There are randomisation functions to generate simulated GRanges and RangedData objects, lists of sample IDs and lists of SNP-ids.

#### List of functions ANNOTATION LOOKUP:

- compact.gene.list Make a compact version of gene annotation
- ENS.to.GENE Convert ensembl ids to HGNC gene ids
- GENE.to.ENS Convert gene ids to ensembl ids
- get.immunobase.snps Download GWAS hits from immunobase.org
- get.centromere.locs Return Centromere locations across the genome
- get.chr.lens Get chromosome lengths from build database
- get.cyto Return Cytoband/Karyotype locations across the genome
- get.exon.annot Get exon names and locations from UCSC
- get.gene.annot Get human gene names and locations from biomart
- get.genic.subset return subset of a ranged object that overlaps genes
- get.GO.for.genes Retreive GO terms from biomart for a given gene list
- get.immunog.locs Retrieve locations of Immunoglobin regions across the genome
- get.recombination.map Get HapMap recombination rates for hg18 (build 36)
- get.telomere.locs Derive Telomere locations across the genome
- get.t1d.regions Obtain a listing of known T1D associated genomic regions
- get.tld.subset return subset of a ranged object that overlaps ichip dense mapped regions
- nearest.gene Retrieve the 'n' closest GENE labels or positions near specified locus
- plotGeneAnnot Plot genes to annotate figures with genomic axes
- ucsc.sanitizer Standardize genome build string

## RANGED DATA:

- makeGRanges Wrapper to construct GRanges object from chr,pos or chr,start,end
- conv.37.36 Convert from build 37 to build 36 SNP coordinates
- conv.36.37 Convert from build 36 to build 37 SNP coordinates
- conv.37.38 Convert from build 37 to build 38 SNP coordinates
- conv.38.37 Convert from build 38 to build 37 SNP coordinates
- recomWindow Extend an interval or SNP by distance in centimorgans (recombination distance)
- ranges.to.txt Convert GRanges/RangedData to chr:pos1-pos2 vector
- select.autosomes Select ranges only within the 22 autosomes in a ranged data object

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- ranges.to.data.frame Convert RangedData/GRanges to a data.frame
- data.frame.to.GRanges Convert a data.frame with positional information to GRanges
- data,frame.to.ranges Convert a data.frame with positional information to RangedData/GRanges
- chrSel Select chromosome subset of GRanges or RangedData object
- rranges Simulate a GRanges or RangedData object
- chrNums Extract chromosome numbers from GRanges/RangedData
- expand.nsnp Expand genomic locations to the ranges covering the 'n' closest SNPs
- endSnp Find closest SNPs to the ends of ranges
- rangeSnp Find closest SNPs to the starts and ends of ranges
- startSnp Find closest SNPs to the starts of ranges
- force.chr.pos Force a valid genomic range, given the inputted coordinates
- in.window Select all ranges lying within a chromosome window
- plotRanges Plot the locations specified in a GRanges or RangedData object
- set.chr.to.char Change the chromosome labels in a RangedData or GRanges object to string codes
- set.chr.to.numeric Change the chromosome labels in a RangedData or GRanges object to numbers
- invGRanges Invert a ranged object

## CHIP SUPPORT:

- convert.textpos.to.data Convert a chr:pos1-pos2 vector to a matrix
- chip.support Retrieve current ChipInfo annotation object
- ids.by.pos Order rs-ids or ichip ids by chrosome and position
- id.to.rs Convert from chip ID labels to dbSNP rs-ids
- rs.to.id Convert from dbSNP rs-ids to chip ID labels
- get.nearby.snp.lists Obtain nearby SNP-lists within a recombination window
- nearest.snp Retrieve the 'n' closest SNP ids or positions near specified locus
- snps.in.range Retrieve SNP ids or positions in specified range
- AB Returns the A and B allele for SNP ids
- Band Retrieve the cytoband(s) for snp ids, genes or locations
- Band.gene Retrieve the cytoband(s) for genes labels
- Band.pos Find the cytoband(s) overlapping a chromosome location
- Chr Find chromosome for SNP ids, gene name or band
- Gene.pos Find the gene(s) overlapping a chromosome location
- Pos Find the chromosome position for SNP ids, gene name or band
- Pos.gene Find the chromosome, start and end position for gene names
- Pos.band Find the chromosome, start and end position for cytoband names

# Author(s)

Nicholas Cooper

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#### See Also

```
NCmisc ~~
```

#### **Examples**

```
#' randomly generated GRanges object
rranges()
```

Α1

Access alleles for ChipInfo

## **Description**

A1/A2: Returns the letter for the A1/A2 alleles for the chip object, e.g, 'A','C','G','T', etc Only if these are annotated internally, or else a vector of NAs

A1<-/A2<-: Allows user to set the allele codes for each SNP of the chip object, e.g, A,C,G,T,K, etc. If you are using allele codes this is likely to necessary as each genotyping produces a different set of allele codes. If using in conjunction with snpStats, remember that allele codes are always flipped to be alphabetical, so the reference allele is the later letter in the alphabet. Note, assignment to A2 needs to be done separately.

# Usage

```
A1(x)

## S4 method for signature 'ChipInfo'
A1(x)

A2(x)

## S4 method for signature 'ChipInfo'
A2(x)

A1(x) <- value

## S4 replacement method for signature 'ChipInfo'
A1(x) <- value

A2(x) <- value

## S4 replacement method for signature 'ChipInfo'
A2(x) <- value
```

# **Arguments**

```
x a ChipInfo object
value new allele codes, e.g, A,C,G,T
```

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#### Value

character vector of allele codes (or NAs)

A1<-: updates the ChipInfo object specified with new allele codes for the 'A1' slot

A2<-: updates the ChipInfo object specified with new allele codes for the 'A2' slot

AΒ

Returns the A and B allele for SNP ids

## **Description**

For a set of chip ids or rs ids, returns a two column matrix containing the A and B allele. For snpStats objects the default is that A,B are coded in alphabetical order, so A,C; A,T; C,T; C,G are possible A,B pairs. Allele codes are specific to each dataset, so you should upload your allele codes into the current ChipInfo object to make the alleles produced by this function meaningful.

## Usage

AB(ids)

## **Arguments**

ids

character, a list of chip ids or rs-ids as contained in the current ChipInfo object

#### Value

Returns a two column matrix containing the A and B allele.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

### See Also

```
Chr, Pos, Pos. band, Band, Band. gene, Band. pos, Gene. pos
```

## **Examples**

```
snp.ids <- c("rs3842724","rs9729550","rs1815606","rs114582555","rs1240708","rs6603785")
AB(snp.ids)</pre>
```

8 Band

as As("ChipInfo", "GRanges")

## **Description**

```
As("ChipInfo", "GRanges")
As("ChipInfo", "GRanges")
As("ChipInfo", "GRanges")
As("GRanges", "ChipInfo")
As("RangedData", "ChipInfo")
As("data.frame", "ChipInfo")
As("data.frame", "RangedData")
As("data.frame", "GRanges")
```

Note that for automatic conversion of a data.frame to RangedData/GRanges, a column named 'chr' or 'seqnames' in the data.frame is expected/required to make the conversion effectively. Otherwise use 'ranged.to.data.frame()'

As("GRanges", "data.frame")

Band

Retrieve the cytoband(s) for snp ids, genes or locations

#### **Description**

Allows retrieval of the the cytoband/karyotype label, based on multiple possible input featues, including SNP chip or rs-ids, HGNC gene labels, GRanges or RangedData object, chromosome and position vectors. The most robust way to use the function is to use the parameter names to imply the type of input, e.g, use the 'genes' parameter to input gene labels, the 'snps' parameter to enter SNP ids, etc. However, if you enter the first argument as a GRanges or RangedData object instead of using the 'ranges' argument, this will be detected and automatically moved to the 'ranges' parameter.

#### Usage

```
Band(genes = NULL, chr = NULL, ranges = NULL, snps = NULL,
build = NULL, dir = NULL, ...)
```

## **Arguments**

genes	character, an optional vector of gene ids, or RangedData/GRanges object
chr	character, an optional vector of chromosomes to combine with 'pos' or 'start'+'end' (enter in) to describe positions to retrieve the band from
ranges	optional GRanges or RangedData object describing positions for which we want bands
snps	optional SNP ids, e.g, chip ids or rs-ids, to retrieve the band they fall within

Band.gene 9

build	character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The default when build is NULL is to use the build from the current ChipInfo annotation
dir	character, 'dir' is the location to download cyto annotation information; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.
• • •	further arguments to Band.gene if entering gene names, or further arguments to Band.pos if entering ranges, or chr, pos/start/end

#### Value

Returns a vector of bands, if any entries span more than one band, the bands will be concatenated as character type, delimited by semicolons (;)

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

```
Chr, Pos, Pos.gene, Band, Band.gene, Band.pos, Gene.pos
```

## **Examples**

```
setwd(tempdir())
Band(chr=1,pos=1234567) # using chr,pos vectors
rd <- RangedData(ranges=IRanges(start=87654321,end=87654321),space=1)
gr <- as(rd,"GRanges")
Band(rd) # using RangedData, autodetects this parameter should be 'ranges' not 'genes'
Band(ranges=gr) # using GRanges
Band("SLC6A4") # serotonin gene [5-HTT]
a.few.snps <- c("rs3842724","imm_11_2147527","rs9467354")
Band(a.few.snps) # using SNP ids in the 'genes' parameter (still works!)
Band(snps=a.few.snps) # using SNP ids with the dedicated 'snps' parameter is quicker
Band(chr="X",pos=8000000)
# Band() with longer ranges #
Band(chr=12,start=40000000,end=500000000,build="hg19") # concatenates if range spans multiple bands
Band(chr=12,start=40000000,end=500000000,build="hg18") # one extra band in the older annotation</pre>
```

Band.gene

Retrieve the cytoband(s) for genes labels

## **Description**

Allows retrieval of the the cytoband/karyotype label for HGNC gene labels.

## Usage

```
Band.gene(genes, build = NULL, dir = getwd(), append.chr = TRUE,
  data.frame = FALSE, warnings = TRUE)
```

Band.gene

## **Arguments**

genes	character, an optional vector of gene ids, or RangedData/GRanges object
build	character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The default when build is NULL is to use the build from the current ChipInfo annotation
dir	character, 'dir' is the location to download cyto annotation information; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.
append.chr	logical, it is typical that the chromosome character preceeds cytoband labels, but if this parameter is set to FALSE, it will be left off.
data.frame	logical, if data.frame is true, instead of returning a vector of full cytoband labels, a data.frame will be returned.
warnings	logical, if warnings=FALSE and SNP ids are entered instead of Gene labels, then the function will automatically detect this and return the result of Band(snps='genes')

#### Value

Returns a vector of bands, if any entries span more than one band, the bands will be concatenated as character type, delimited by semicolons (;). If data.frame is true, instead of returning a vector of full cytoband labels, a data.frame will be returned with a 'chr' [chromosome] column, 'band' cytoband label without the chromosome prefix, and rownames equal to 'genes'

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

Chr, Pos, Pos. gene, Band, Band. gene, Band. pos, Gene. pos

## **Examples**

```
setwd(tempdir())
a.few.snps <- c("rs3842724","imm_11_2147527","rs9467354")
Band.gene("HLA-C") # using chr,pos vectors
Band.gene(a.few.snps) # fails with warning as these are SNPs, not genes
Band.gene(a.few.snps,warnings=FALSE) # with warnings=FALSE this continues with snps entered
Band.gene("SLC6A4") # serotonin gene [5-HTT]
Band.gene("SLC6A4",append.chr=FALSE)
Band.gene("SLC6A4",data.frame=TRUE)</pre>
```

Band.pos 11

Band.pos Find the cytoband(s) overlapping a chromosome location	
---	--

## **Description**

Allows retrieval of cytobands/karyotypes intersected by a chromosome and position, which can be entered using chr, pos/start/end vectors, or a RangedData or GRanges object

## Usage

```
Band.pos(chr = NA, pos = NA, start = NA, end = NA, ranges = NULL,
build = NULL, dir = NULL, bioC = FALSE, one.to.one = TRUE)
```

## **Arguments**

chr	character, an optional vector of chromosomes to combine with 'pos' or 'start'+'end (enter in) to describe positions to retrieve the possible overlapping cytoband(s)
pos	integer, an optional vector of chromosome positions (for SNPs), no need to enter start or end if this is entered, and vice-versa
start	integer, an optional vector of start points for chromosome ranges
end	integer, an optional vector of end points for chromosome ranges
ranges	optional GRanges or RangedData object describing positions for which we want bands, removing the need to enter chr, pos, start or end
build	character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The default when build is NULL is to use the build from the current ChipInfo annotation
dir	character, 'dir' is the location to download gene annotation information to; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.
bioC	logical, if true then return position information as a GRanges object, or Ranged-Data if 'ranges' is RangedData, else a data.frame
one.to.one	logical, whether to concatenate multiple hits for the same range into one result, or spread the result over multiple lines, one for each cytoband overlapped

## Value

Returns a set of cytobands separated by semicolons (if more than one) for each range entered. If bioC=TRUE, returns the equivalent as a GRanges object, unless a RangedData object was used for the ranges parameter, in which case a RangedData object would be returned. If one.to.one is FALSE, then instead of concatenating multiple cytobands into one line per range, each is listed separately as a new row, with an index added to correspond to the original input order of ranges, if bioC=TRUE; or just adds additional elements to the resulting vector if bioC=FALSE.

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

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#### See Also

```
Chr, Pos, Pos. band, Band, Band. gene, Band. pos, Gene. pos
```

## **Examples**

```
setwd(tempdir())
Band.pos(chr=6, start=31459636, end=31462760)
Band.pos(chr=22, pos=3452345)
Band.pos(Chr("rs689"),Pos("rs689")) # combine Chr(), Pos() to find the cytoband for SNP rs689
Band.pos(chr=1,start=110000000,end=1200000000,build="hg19") # multiple cytobands in range
Band.pos(chr=1,start=110000000,end=1200000000,one.to.one=FALSE) # list separately
Band.pos(Pos.band(c("13q21.31","1p13.2"),bioC=TRUE)) # use ranges object returned by Pos.band()
# note that 3 ranges are returned for each entry as the start/end overlap the adjacent ranges
```

chip

Retrieve the Chip name for ChipInfo

# Description

Simply returns the name of the chip, e.g, 'ImmunoChip'

## Usage

```
chip(x)
## S4 method for signature 'ChipInfo'
chip(x)
```

#### **Arguments**

Χ

a ChipInfo object

## Value

character string

chip.support

Retrieve current ChipInfo annotation object

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## **Description**

This function returns the current 'ChipInfo' annotation object, containing chromosome, id, position, strand, 'rs' id, allele 1, allele 2 for each SNP of a microarray chip, in either hg18 or hg19 (build 36/37) coordinates. Can also be used to update the current object to a new object. This package makes extensive use of this class of annotation object for the working microarray chip, e.g, default is ImmunoChip, but Metabochip is also built-in, and you can also load your own annotation if using a different chip. The class of the object used is 'ChipInfo' which is a GRanges object, modified to always have columns for A1, A2 (alleles), rs.id, and a quality control flag. The default display is tidier than GRanges, it has nice coersion to and frame data.frame and indexing by chromosome using [[n]] has been added, in addition to normal [i,j] indexing native to GRanges. A1 and A2 values are usually specific to each dataset so for immunochip you may need to manually update these values to reflect the allele coding in your own dataset.

## Usage

```
chip.support(build = NULL, refresh = FALSE, alternate.file = NULL,
  warn.build = TRUE)
```

#### **Arguments**

build character, either "hg18", "hg19" or "hg38". Will also accept build numbers, 36,

37 or 38.

refresh logical, FALSE to just load whatever object is already in memory (except when

first using a function in this package, there should be a ChipInfo object loaded), or TRUE to reload from the original source. For instance you may wish to do this when you want to use a different chip, different build, or if the annotation

has been modifed via a manual correction).

alternate.file character, name of an alternative RData file containing a ChipInfo object to use

instead of the object found in getOption("chip.info"). This will replace the cur-

rent ChipInfo object.

warn.build logical, whether to warn if the 'build' argument does not match the current value

of getOption("ucsc"). The default is to display this warning, but if you set this

argument to FALSE this can be suppressed.

#### Value

returns the current ChipInfo object [S4]. This may be slow first time, but subsequent lookups should be much faster. Builds 36/38 are not stored explicitly so will take a little while to convert the first time, but subsequent lookups should be fast. To increase the speed save the object locally and use option(chip.info=<PATH>) to set a custom path for future chip.support() calls [which are also made internally by many of the function in this package]. This is also the option to set if you want to add a ChipInfo object for a different chip, e.g, metabochip, exomechip, etc.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

## See Also

```
ChipInfo, build, rs.id, QCfail, convTo36, convTo37, A1, A2
```

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#### **Examples**

```
chip.support() # shows the current ChipInfo object (default is 'ImmunoChip' build 37)
#/donttest{
chip.support(build=36) # gives warning as hg19 version is currently loaded
chip.support(build=36,refresh=TRUE)
getOption("chip.info") # shows the object is now saved in the tmp directory for subsequent calls
chip.support(build=38,refresh=TRUE)
#}
```

chipId

Access chip-ids for ChipInfo

## **Description**

Returns the chip-ids for the chip object, e.g, "imm\_1\_898835", etc Only if these are annotated internally, or else a vector of NAs Note that the main purpose of this is because sometimes chip-ids do not satisfy conditions to be an R column/row name, e.g, start with a number, illegal characters, etc. So this allows certain functions to return the actual chip names that would match the official manifest. These will largely be the same as the rownames, but the rownames will always be valid R column names, converted from the original using clean.snp.ids() [internal function]

#### Usage

```
chipId(x)
## S4 method for signature 'ChipInfo'
chipId(x)
```

## **Arguments**

Х

a ChipInfo object

#### Value

chip ids: character vector of IDs (or NAs)

ChipInfo

Constructor (wrapper) for ChipInfo annotation object

#### **Description**

This class annotates a microarray SNP chip with data for each SNP including chromosome, id, position, strand, 'rs' id, allele 1, allele 2 for each SNP of a microarray chip, in either hg18, hg19 or hg38 (build 36/37/38) coordinates. This package makes extension use of this class of annotation object for the working microarray chip, e.g, default is ImmunoChip, but Metabochip is also built-in, and you can also load your own annotation if using a different chip. The class is basically a GRanges object, modified to always have columns for A1, A2 (alleles), rs.id, and a quality control flag. The default display is tidier than GRanges, it has nice coersion to and frame data.frame and subsetting by chromosome using [[n]] has been added, in addition to normal [i,j] indexing native to GRanges.

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# Usage

```
ChipInfo(GRanges = NULL, chr = NULL, pos = NULL, ids = NULL,
  chip = "unknown chip", build = "", rs.id = NULL, chip.id = NULL,
  A1 = NULL, A2 = NULL, QCcode = NULL)
```

# **Arguments**

GRanges	a GRanges object containing chromosome, start/end = position, and strand information for the chip object to be created, also rownames should be used to code the chip-ids for each SNP.
chr	optional, alternative to using 'GRanges' to input SNP locations, enter here a vector of chromosome numbers/letters for each SNP. The recommended coding is: 1:22, X, Y, XY, MT
pos	optional, vector of positions (integers), use in conjunction with 'chr' and 'ids' as an alternative way to input SNP position information instead of GRanges.
ids	optional, vector of SNP chip-ids, use in conjunction with 'chr' and 'pos' as an alternative way to input SNP position information instead of GRanges.
chip	character, name of the chip you are making this annotation for (only used for labelling purposes)
build	character, either "hg18" or "hg19". Will also accept build number, 36 or 37. This indicates what coordinates the object is using, and will be taken into account by conversion functions, and annotation lookup functions throughout this package.
rs.id	'rs' ids are standardized ids for SNPs, these usually differ from each chips' own IDs for each snp. If you don't know these, or can't find them, they can be left blank, but will render the functions 'rs.to.id()' and 'id.to.rs()' useless for this ChipInfo object.
chip.id	chip ids are the chip-specific ids for SNPs, these usually differ between chips' even for the same snp. If you don't know these, or can't find them, they can be left blank, but will render the function 'chip.id()' useless for this ChipInfo object. The main purpose of this parameter is for when the real chip ids are not valid R row/column name strings, and by using this column, some functions can return the real chip ids instead of the sanitized version
A1	the first allele letter code for each SNP, e.g, usually "A", "C", "G", or "T", but you can use any scheme you like. Can be left blank.
A2,	as for A1, but for allele 2.
QCcode	optional column to keep track of SNPs passing and failing QC. You can completely ignore this column. It works based on integer codes, 0,1,2, you may wish to use simple 0 and 1, for pass and fail respectively, or else 0 can be pass, and 1,2, can indicate failure for different criteria. 0 will always be treated as a pass and anything else as a fail, so you can code fails however you wish.

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## **Description**

This class annotates a microarray SNP chip with data for each SNP including chromosome, id, position, strand, 'rs' id, allele 1, allele 2 for each SNP of a microarray chip, in either hg18, hg19 or hg38 (build 36/37/38) coordinates. This package makes extension use of this class of annotation object for the working microarray chip, e.g, default is ImmunoChip, and you can also load your own annotation if using a different chip. The class is basically a GRanges object, modified to always have columns for A1, A2 (alleles), rs.id, and a quality control flag. The default display is tidier than GRanges, it has nice coersion to and frame data.frame and subsetting by chromosome using [[n]] has been added, in addition to normal [i,j] indexing native to GRanges. Note that with this package the first time annotation is used it might be slow, but subsequent calls should be fast. METHODS "[[", show, print, length, dim, rownames, initialize build, chip, rs.id, A1, A2, QCcode, QCcode<-, QCpass, QCfail convTo36, convTo37, convTo38 COERCION can use 'as' to convert to and from: GRanges, RangedData, data.frame

Use the 'ChipInfo()' wrapper to construct ChipInfo objects from scratch

#### Usage

```
## S4 method for signature 'ChipInfo'
initialize(.Object, ...)
```

#### Arguments

.0bject An object generated from the ChipInfo class prototype, see methods:initialize

. . . Additional arguments to initialize. None recommended.

## **Fields**

```
seqnames: Object of class "Rle", containing chromosomes for each range, see GRanges.
```

ranges: Object of class "IRanges", containing genomic start and end, see GRanges.

strand: Object of class "Rle", containing plus or minus coding for forward or reverse strand, see GRanges.

seqinfo: Object of class "Seqinfo", containing chromosome listing, see GRanges.

chip: Name, class "character", containing user description of the chip, e.g, 'immunoChip'.

build: Object of class "character", annotation version, e.g, hg18, hg19, hg38, etc.

elementMetaData: Object of class "DataFrame", see GRanges, but with specific column names: A1, A2, QCcode and rs.id.

# Author(s)

Nick Cooper

Chr 17

Chr

Find chromosome for SNP ids, gene name or band

#### **Description**

Allows retrieval of the chromosome associated with a SNP-id, HGNC gene label, karyotype band, or vector of such ids. For SNPs the ids can be either chip ids, or rs-ids, but must be contained in the current annotation. Default behaviour is to assume 'id' are SNP ids, but if none are found in the SNP annotation, the id's will be passed to functions Pos.gene() and Pos.band() to see whether a result is found. This latter step will only happen if no SNP ids are retreived in the first instance, and if snps.only=TRUE, then genes and bands will not be searched and NA's returned. If you are repeatedly searching for chromosomes for genes/bands, using the dedicated Pos.gene and Pos.band functions would be slightly faster than relying on the fallback behaviour of the Chr() function. See documentation for these functions for more information. The build used will be that in the current ChipInfo object.

## Usage

```
Chr(ids, dir = NULL, snps.only = FALSE)
```

## **Arguments**

ids character, a vector of rs-ids or chip-ids representing SNPs in the current Chip-

Info annotation, or gene ids, or karyotype bands. Can also be a SnpMatrix

object.

dir character, only relevant when gene or band ids are entered, in this case 'dir' is the

location to download gene and cytoband information; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted

after use.

snps.only logical, if TRUE, only search SNP ids, ignore the possibility of genes/cytobands.

## Value

A character vector of Chromosomes for each ids, with NA values where no result was found.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

Pos

## **Examples**

```
setwd(tempdir())
Chr(c("rs689","rs9467354","rs61733845"))
Chr("CTLA4")
Chr("13q21.31")
Chr(c("CTLA4","PTPN22"),snps.only=TRUE) # fails as these are genes
```

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```
Chr(c("rs689","PTPN22","13q21.31")) # mixed input, will default to SNPs, as at least 1 was found
```

```
chrIndices-methods
```

~~ Methods for Function chrIndices ~~

## **Description**

```
~~ Methods for function chrIndices ~~
```

#### Methods

```
signature(object = "RangedData")
```

```
chrInfo-methods
```

~~ Methods for Function chrInfo ~~

# Description

```
~~ Methods for function chrInfo ~~
```

#### Methods

```
signature(object = "RangedData")
```

chrm

Chromosome method for RangedData objects

# Description

Return the list of chromosome values from a RangedData object

# Usage

```
chrm(object)
## S4 method for signature 'RangedData'
chrm(object)
## S4 method for signature 'GRanges'
chrm(object)
## S4 method for signature 'ChipInfo'
chrm(object)
```

## **Arguments**

object

RangedData object

## Value

vector of chromosome values for each range/SNP

chrNames-methods 19

chrNames-methods	~~ Methods for Function chrNames ~~	

# Description

~~ Methods for function chrNames ~~

# Methods

```
signature(object = "RangedData")
```

chrNums

Extract chromosome numbers from GRanges/RangedData

# Description

Sometimes chromosomes are codeds as 1:22, sometimes there is also X,Y, etc, sometimes it's chr1, ch2, etc. This function extracts the set of chromosome labels used by a ranged object (ie, GRanges or RangedData) and converts the labels to numbers in a consistent way, so 1:22, X, Y, XT, MT ==> 1:26, and optionally you can output the conversion table of codes to numbers, then input this table for future conversions to ensure consistency.

## Usage

```
chrNums(ranged, warn = FALSE, table.out = FALSE, table.in = NULL)
```

# Arguments

ranged	GRanges or RangedData object
warn	logical, whether to display a warning when non autosomes are converted to numbers
table.out	logical, whether to return a lookup table of how names matched to integers
table.in	data.frame/matrix, col 1 is the raw text names, col 2 is the integer that should be assigned, col 3 is the cleaned text (of col 1) with 'chr' removed. the required form is outputted by this function if you set 'table.out=TRUE', so the idea is that to standardize coding amongst several RangedData objects you can save the table each time and ensure future coding is consistent with this. Note that chromosomes 1-22, X, Y, XY, and MT are always allocated the same integer, so table is only useful where there are extra NT, COX, HLA regions, etc.

# Value

a set of integers of length equal to the number of unique chromosomes in the ranged data.

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## **Examples**

```
require(genoset)
gg <- rranges(1000)
chrNames(gg); chrNums(gg)
gg <- rranges(1000,chr.pref=TRUE) # example where chromosomes are chr1, chr2, ...
chrNames(gg); chrNums(gg)
lookup <- chrNums(gg,table.out=TRUE)
lookup
gg2 <- rranges(10)
chrNums(gg2,table.in=lookup) # make chromosome numbers using same table as above</pre>
```

chrSel

Select chromosome subset for ranged objects

## **Description**

Returns the object filtered for specific chromosomes for a ranged object
Returns the object filtered for specific chromosomes for a RangedData object
Returns the object filtered for specific chromosomes for a GRanges object
Returns the object filtered for specific chromosomes for a GRanges object

## Usage

```
chrSel(object, chr)
## S4 method for signature 'RangedData'
chrSel(object, chr)
## S4 method for signature 'GRanges'
chrSel(object, chr)
## S4 method for signature 'ChipInfo'
chrSel(object, chr)
```

## **Arguments**

object a ChipInfo, GRanges or RangedData object

chr vector, string or numeric of which chromosome(s) to select

# Value

vector of chromosome values for each range/SNP

chrSelect 21

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Select chromosome subset of GRanges or RangedData object

## **Description**

One of the main differences between RangedData and GRanges is the way of selecting the subset for a chromosome. RangedData just uses [n] where 'n' is the chromosome name or number. Whereas GRanges, does not have a method like this, so need to select using [chr(X)==chr.num,] This wrapper allows selection of a chromosome or chromosomes regardless of whether the object is RangedData or GRanges type.

#### Usage

```
chrSelect(X, chr, index = FALSE)
```

# **Arguments**

X A GRanges or RangedData object

chr Vector, the chromosome(s) (number(s) or name(s)) to select

index logical, if FALSE, will assume 'chr' is a string, indicating the chromosome

name, if TRUE, if 'chr' is numeric, will assume it refers to the chromosome index, which if there are some chromosomes not represented, may be different to the name. E.g., an object with data for chromosomes 1,2,4,5 would select

chromosome 5 with chr=4, if index=TRUE.

#### Value

returns an object of the same type as X, with only the chromosome subset specified.

#### **Examples**

```
some.ranges <- rranges(100,chr.range=1:10)
chrSelect(some.ranges,6)
more.ranges <- rranges(10, chr.range=21:25)
chrSelect(more.ranges,1:22) # gives warning
select.autosomes(more.ranges)</pre>
```

coerce-methods

~~ Methods for Function coerce ~~

## **Description**

```
~~ Methods for function coerce ~~
```

22 compact.gene.list

#### Methods

```
signature(from = "ChipInfo", to = "data.frame")
signature(from = "ChipInfo", to = "GRanges")
signature(from = "ChipInfo", to = "RangedData")
signature(from = "data.frame", to = "ChipInfo")
signature(from = "data.frame", to = "GRanges")
signature(from = "data.frame", to = "RangedData")
signature(from = "GRanges", to = "ChipInfo")
signature(from = "GRanges", to = "data.frame")
signature(from = "RangedData", to = "ChipInfo")
signature(from = "RangedData", to = "data.frame")
```

coerce<--methods

~~ Methods for Function coerce<- ~~

## **Description**

```
~~ Methods for function coerce<- ~~
```

#### Methods

```
signature(from = "ChipInfo", to = "GRanges")
```

compact.gene.list

Make a compact version of gene annotation

## **Description**

When adding gene annotation to genomic ranges, sometimes there are many genes associated with a single feature, so that compiling a table becomes awkward, if some rows contain hundreds of genes. This function takes a character vector of gene lists delimited by some separator and provides a compact representation of the gene labels

# Usage

```
compact.gene.list(x, n = 3, sep = ";", others = FALSE)
```

# Arguments

X	is a character vector of gene label listings, where multiple hits are delimited by 'sep'	
n	number of genes to list before abbreviating	
sep	character, separator used to delimit genes in elements of x	
others logical, TRUE to abbreviate with '+ # others' or FALSE to append just		

number of genes not listed.

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#### Value

```
a character vector with the form: gene-1, gene-2, ..., gene-n, + length(gene-n) - n [others]
```

#### **Examples**

```
my.genes <- c("ERAP1","HLA-C;CTLA4;IFIH","INS;MYC","AGAP1;APOE;DRDB1;FUT2;HCP5;BDNF;COMT")
compact.gene.list(my.genes)
compact.gene.list(my.genes,n=2,others=TRUE)</pre>
```

conv.36.37

Convert from build 36 to build 37 SNP coordinates

## **Description**

Convert range or SNP coordinates between builds using a chain file. Depending on the chain file this can do any conversion, but the default will use the hg18 to hg19 (36–>37) chain file built into this package. The positions to convert can be entered using using chr, pos vectors, or a RangedData or GRanges object. This function is a wrapper for liftOver() from rtracklayer, providing more control of input and output and 'defensive' preservation of order and length of the output versus the input ranges/SNPs.

# Usage

```
conv.36.37(ranges = NULL, chr = NULL, pos = NULL, ..., ids = NULL,
    chain.file = NULL, include.cols = TRUE)
```

## **Arguments**

ranges	optional GRanges or RangedData object describing positions for which conversion should be performed. No need to enter chr, pos if using ranges	
chr	character, an optional vector of chromosomes to combine with 'pos' to describe positions to convert to an alternative build	
pos	integer, an optional vector of chromosome positions (for SNPs), no need to enter a ranges object if this is provided along with 'chr'	
	additional arguments to makeGRanges(), so in other words, can use 'start' and 'end' to specify ranges instead of 'pos'.	
ids	if the ranges have ids (e.g, SNP ids, CNV ids), then by including this parameter when using chr, pos input, the output object will have these ids as rownames. For ranges input these ids would already be in the rownames of the GRanges or RangedData object, so use of this parameter should be unnecessary	
chain.file	character, a file location for the liftOver chain file to use for the conversion. If this argument is left NULL the default UCSC file that converts from hg18 to hg19 will be used. Can also use a 'Chain' object from rtracklayer created using import.chain(). Alternate chain files for other conversions are available from http://crossmap.sourceforge.net/, and you could also customize these or create your own. So this function can be used for conversion between any in-out build combination, using this argument, not just 36–37.	
include.cols	logical, whether to include any extra columns (e.g, in addition to positional information) in the output object.	

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#### Value

Returns positions converted from build 36 to 37 (or equivalent for alternative chain files). If using the 'ranges' parameter for position input, the object returned will be of the same format. If using chr and pos to input, then the object returned will be a data frame with columns, chr and pos with rownames 'ids'. Output will be the same length as the input, which is not necessarily the case for liftOver() which does the core part of this conversion. Using vector or GRanges input will give a resulting data frame or GRanges object respectively that has the same order of rownames as the original input. Using RangedData will result in an output that is sorted by genome order, regardless of the original order. If ranges has no rownames, or if 'ids' is blank when using chr, pos, ids of the form rngXXXX will be generated in order to preserve the original ordering of locations.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### References

http://crossmap.sourceforge.net/

#### See Also

```
conv. 37. 36, conv. 37. 38, conv. 38. 37, convTo37, convTo36
```

#### **Examples**

```
# various chain files downloadable from http://crossmap.sourceforge.net/ #
options(ucsc="hg18")
gene.labs <- c("CTLA4","IL2RA","HLA-C")
snp.ids <- c("rs3842724","rs9729550","rs1815606","rs114582555","rs1240708","rs6603785")
pp <- Pos(snp.ids); cc <- Chr(snp.ids)
conv.36.37(chr=cc,pos=pp,ids=snp.ids)
pp <- Pos(gene.labs)
gg <- GRanges(ranges=IRanges(start=pp$start,end=pp$end),seqnames=pp$chr)
conv.36.37(gg) # order of output is preserved
rr <- as(gg,"RangedData")
conv.36.37(rr) # note the result is same as GRanges, but in genome order</pre>
```

conv.37.36

Convert from build 37 to build 36 SNP coordinates

## **Description**

Convert range or SNP coordinates between builds using a chain file. Depending on the chain file this can do any conversion, but the default will use the hg19 to hg18 (37–>36) chain file built into this package. The positions to convert can be entered using using chr, pos vectors, or a RangedData or GRanges object. This function is a wrapper for liftOver() from rtracklayer, providing more control of input and output and 'defensive' preservation of order and length of the output versus the input ranges/SNPs.

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#### Usage

```
conv.37.36(ranges = NULL, chr = NULL, pos = NULL, ..., ids = NULL)
```

## **Arguments**

ranges	optional GRanges or RangedData object describing positions for which conversion should be performed. No need to enter chr, pos if using ranges	
chr	character, an optional vector of chromosomes to combine with 'pos' to describe positions to convert from build hg19 to hg18	
pos	integer, an optional vector of chromosome positions (for SNPs), no need to enter a ranges object if this is provided along with 'chr'	
• • •	additional arguments to makeGRanges(), so in other words, can use 'start' and 'end' to specify ranges instead of 'pos'.	
ids	if the ranges have ids (e.g, SNP ids, CNV ids), then by including this parameter when using chr, pos input, the output object will have these ids as rownames. For ranges input these ids would already be in the rownames of the GRanges or RangedData object, so use of this parameter should be unnecessary	

#### Value

Returns positions converted from build 37 to 36. If using the 'ranges' parameter for position input, the object returned will be of the same format. If using chr and pos to input, then the object returned will be a data.frame with columns, chr and pos with rownames 'ids'. Output will be the same length as the input, which is not necessarily the case for liftOver() which does the core part of this conversion. Using vector or GRanges input will give a resulting data.frame or GRanges object respectively that has the same order of rownames as the original input. Using RangedData will result in an output that is sorted by genome order, regardless of the original order.

#### Author(s)

```
Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>
```

#### See Also

```
conv. 36. 37, conv. 37. 38, conv. 38. 37, convTo37, convTo36
```

# **Examples**

```
gene.labs <- c("CTLA4","IL2RA","HLA-C")
pp <- Pos.gene(gene.labs,build=37)
gg <- GRanges(ranges=IRanges(start=pp$start,end=pp$end),seqnames=pp$chr)
conv.37.36(gg) # order of output is preserved ### HERE!!! ###
rr <- as(gg,"RangedData")
conv.37.36(rr) # note the result is same as GRanges, but in genome order</pre>
```

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conv.37.38

Convert from build 37 to build 38 SNP coordinates

#### **Description**

Convert range or SNP coordinates between builds using a chain file. Depending on the chain file this can do any conversion, but the default will use the hg19 to hg38 (37–>38) chain file built into this package. The positions to convert can be entered using using chr, pos vectors, or a RangedData or GRanges object. This function is a wrapper for liftOver() from rtracklayer, providing more control of input and output and 'defensive' preservation of order and length of the output versus the input ranges/SNPs.

## Usage

```
conv.37.38(ranges = NULL, chr = NULL, pos = NULL, ..., ids = NULL)
```

#### **Arguments**

ranges	optional GRanges or RangedData object describing positions for which conversion should be performed. No need to enter chr, pos if using ranges	
chr	character, an optional vector of chromosomes to combine with 'pos' to describe positions to convert from build hg19 to hg38	
pos	integer, an optional vector of chromosome positions (for SNPs), no need to enter a ranges object if this is provided along with 'chr'	
•••	additional arguments to makeGRanges(), so in other words, can use 'start' and 'end' to specify ranges instead of 'pos'.	
ids	if the ranges have ids (e.g, SNP ids, CNV ids), then by including this parameter when using chr, pos input, the output object will have these ids as rownames. For ranges input these ids would already be in the rownames of the GRanges or RangedData object, so use of this parameter should be unnecessary	

#### Value

Returns positions converted from build 37 to 38. If using the 'ranges' parameter for position input, the object returned will be of the same format. If using chr and pos to input, then the object returned will be a data.frame with columns, chr and pos with rownames 'ids'. Output will be the same length as the input, which is not necessarily the case for liftOver() which does the core part of this conversion. Using vector or GRanges input will give a resulting data.frame or GRanges object respectively that has the same order of rownames as the original input. Using RangedData will result in an output that is sorted by genome order, regardless of the original order.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

## See Also

```
conv. 36. 37, conv. 37. 36, conv. 37. 38, convTo37, convTo36
```

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## **Examples**

```
gene.labs <- c("CTLA4","IL2RA","HLA-C")
pp <- Pos.gene(gene.labs,build=37)
gg <- GRanges(ranges=IRanges(start=pp$start,end=pp$end),seqnames=pp$chr)
conv.37.38(gg) # order of output is preserved ### HERE!!! ###
rr <- as(gg,"RangedData")
conv.37.38(rr) # note the result is same as GRanges, but in genome order</pre>
```

conv.38.37

Convert from build 38 to build 37 SNP coordinates

### **Description**

Convert range or SNP coordinates between builds using a chain file. Depending on the chain file this can do any conversion, but the default will use the hg38 to hg19 (38–>37) chain file built into this package. The positions to convert can be entered using using chr, pos vectors, or a RangedData or GRanges object. This function is a wrapper for liftOver() from rtracklayer, providing more control of input and output and 'defensive' preservation of order and length of the output versus the input ranges/SNPs.

#### Usage

```
conv.38.37(ranges = NULL, chr = NULL, pos = NULL, ..., ids = NULL)
```

## **Arguments**

ranges	optional GRanges or RangedData object describing positions for which conversion should be performed. No need to enter chr, pos if using ranges	
chr	character, an optional vector of chromosomes to combine with 'pos' to describe positions to convert from build hg38 to hg19	
pos	integer, an optional vector of chromosome positions (for SNPs), no need to enter a ranges object if this is provided along with 'chr'	
• • •	additional arguments to makeGRanges(), so in other words, can use 'start' and 'end' to specify ranges instead of 'pos'.	
ids	if the ranges have ids (e.g, SNP ids, CNV ids), then by including this parameter when using chr, pos input, the output object will have these ids as rownames. For ranges input these ids would already be in the rownames of the GRanges or RangedData object, so use of this parameter should be unnecessary	

#### Value

Returns positions converted from build 38 to 37. If using the 'ranges' parameter for position input, the object returned will be of the same format. If using chr and pos to input, then the object returned will be a data.frame with columns, chr and pos with rownames 'ids'. Output will be the same length as the input, which is not necessarily the case for liftOver() which does the core part of this conversion. Using vector or GRanges input will give a resulting data.frame or GRanges object respectively that has the same order of rownames as the original input. Using RangedData will result in an output that is sorted by genome order, regardless of the original order.

28 convert.textpos.to.data

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

```
conv. 36. 37, conv. 37. 36, conv. 37. 38, convTo37, convTo36
```

#### **Examples**

```
gene.labs <- c("CTLA4","IL2RA","HLA-C")
pp <- Pos.gene(gene.labs,build=38)
gg <- GRanges(ranges=IRanges(start=pp$start,end=pp$end),seqnames=pp$chr)
conv.38.37(gg) # order of output is preserved ### HERE!!! ###
rr <- as(gg,"RangedData")
conv.38.37(rr) # note the result is same as GRanges, but in genome order</pre>
```

```
convert.textpos.to.data
```

Convert a chr:pos1-pos2 vector to a matrix

## **Description**

Takes standard text positions, such as what you might see on the UCSC genome browser, such as chr1:10,000,234-11,000,567 for a range, or chrX:234,432 for a SNP, and converts to with cols: chr, start, end.

# Usage

```
convert.textpos.to.data(text)
```

## **Arguments**

text

character vector, format like chr:pos1-pos2

#### Value

a matrix of the same length as 'ranges' with columns chr, start and end, and rownames will be the same as the original text vector.

#### See Also

```
ranged.to.txt
```

# **Examples**

```
txt <- ranged.to.txt(rranges())
convert.textpos.to.data(txt)</pre>
```

convTo37

convTo37

Convert ChipInfo between build 36/37/38 coordinates

## **Description**

Returns the a ChipInfo object with positions updated to build 36/37/38 coordinates, assuming that the build() slot was entered correctly. Ensure that the value of ucsc(x) is correct before running this function for conversion; for instance, if the coordinates are already build 37/hg19, but ucsc(x)!="hg19" (incorrect value), then these coordinates will be transformed in a relative manner rendering the result meaningless.

## Usage

```
convTo37(x)

## S4 method for signature 'ChipInfo'
convTo37(x)

convTo36(x)

## S4 method for signature 'ChipInfo'
convTo36(x)

## S4 method for signature 'ChipInfo'
convTo38(x)
```

## **Arguments**

Х

a ChipInfo object

#### Value

convTo37: Returns a ChipInfo object with the build updated to hg19 coordinates convTo36: Returns a ChipInfo object with the build updated to hg18 coordinates convTo38: Returns a ChipInfo object with the build updated to hg38 coordinates

df.to.GRanges

Convert a data.frame with positional information to GRanges

#### **Description**

Convert a data.frame containing chromosome and position information to a GRanges object. Assumes the position information is contained in columns named 'chr', 'start' and 'end' respectively (not case sensitive) although you can enter alternative column names for each as parameters. 'seqnames' will be automatically detected as an alternative to 'chr' if present. Column names that are default GRanges slot names such as 'seqnames', 'ranges', 'strand', 'seqlevels', etc, will be removed during conversion, so rename these if you want them to be translated into the resulting GRanges

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objects' column metadata. If there is a column 'pos' but no columns 'start' and 'end' this will be detected automatically without needing to change the default parameters and start will equal end equals pos (ie., SNPs).

## Usage

```
df.to.GRanges(dat, ...)
```

## **Arguments**

a data.frame with chromosome and position informationadditional arguments to df.to.ranged(), namely: ids, start, end, width, chr, exclude and build

#### Value

A RangedData or GRanges object. If 'dat' doesn't use the default column names, specify these using parameters ids, start, and end or width. Exclude will remove prevent any column names of 'dat' specified not to be translated to the returned GRanges object. 'build' specifies the 'genome' slot of the resulting object. 'ids' allows specification of a column to be converted to the rownames of the new object.

## See Also

```
ranged.to.data.frame, df.to.ranged
```

## **Examples**

```
chr <- sample(1:22,10)
start <- end <- sample(1000000,10)
df1 <- cbind(chr,start,end)</pre>
df.to.GRanges(df1) # basic conversion
width <- rep(0.10)
df2 <- cbind(chr,start,width)</pre>
df.to.GRanges(df2,end=NULL,width="width") # define ranges with start and width
id.col <- paste0("ID",1:10)
rs.id <- paste0("rs", sample(10000,10))
df3 <- cbind(chr,start,end,id.col,rs.id)</pre>
df.to.GRanges(df3) # additional columns kept
df4 <- cbind(chr,start,end,id.col,rs.id, ranges=1:10)</pre>
df.to.GRanges(df4) # 'ranges' column excluded as illegal name
df.to.GRanges(df4, exclude="rs.id") # manually exclude column
df5 <- cbind(chr,start,end,rs.id)</pre>
rownames(df5) <- paste0("ID",1:10)</pre>
df.to.GRanges(df5) # rownames are kept
df.to.GRanges(df4,ids="id.col") # use column of 'dat' for rownames
```

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df.to.ranged	Convert a data.frame with positional information to Ranged-Data/GRanges
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## **Description**

Convert a data.frame containing chromosome and position information to a RangedData or GRanges object. Assumes the position information is contained in columns named 'chr', 'start' and 'end' respectively (not case sensitive) although you can enter alternative column names for each as parameters. 'seqnames' will be automatically detected as an alternative to 'chr' if present. If there is a column 'pos' but no columns 'start' and 'end' this will be detected automatically without needing to change the default parameters and start will equal end equals pos (ie., SNPs). Column names that are default GRanges slot names such as 'seqnames', 'ranges', 'strand', 'seqlevels', etc, will be removed during conversion, so rename these if you want them to be translated into the resulting object.

# Usage

```
df.to.ranged(dat, ids = NULL, start = "start", end = "end",
  width = NULL, chr = "chr", exclude = NULL, build = NULL,
  GRanges = FALSE, fill.missing = TRUE)
```

#### **Arguments**

dat	a data.frame with chromosome and position information
ids	character string, an optional column name containing ids which will be used for rownames in the new object, as long as the ids are unique. If not, this option is overridden and the ids will simply be a normal column in the new object.
start	character, the name of a column in the data.frame contain the start point of each range. Not case sensitive. In the case of SNP data, a column called 'pos' will also be automatically detected without modifying 'start' or 'end', and will be used for both start and end.
end	character, the name of a column in the data.frame containing the end point of each range, can also use 'width' as an alternative specifier, in which case 'end' should be set to NULL. Not case sensitive. In the case of SNP data, a column called 'pos' will also be automatically detected without modifying 'start' or 'end', and will be used for both start and end.
width	the name of a column in the data.frame containing 'width' of ranges, e.g, SNPs would be width=0. This is optional, with 'start' and 'end' being the default way to specify an interval. If using 'width' you must also set 'end' to NULL. Not case sensitive.
chr	character, the name of the column in the data.frame containing chromosome values. The default is 'chr' but 'seqnames' will also be detected automatically even when chr='chr'. Not case sensitive.
exclude	character string, and column names from the data.frame to NOT include in the resulting S4 object.
build	the ucsc build for the result object which will apply to the 'universe' (Ranged-Data) or 'genome' slot (GRanges) of the new object.

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GRanges logical, whether the resulting object should be GRanges (TRUE), or Ranged-Data (FALSE)

fill.missing logical, GRanges/RangedData objects cannot handle missing chrs/positions, so

if fill missing is selected, will insert values of chr99, and start=end=1, and if FALSE, will exclude any row with a missing value from the resulting object.

#### Value

A RangedData or GRanges object. If 'dat' doesn't use the default column names 'chr', 'start'/end' or 'pos', specify these using parameters 'ids', 'start', and 'end' or 'width'. Exclude will remove prevent any column names of 'dat' specified not to be translated to the returned GRanges object. 'build' specifies the 'genome' slot of the resulting object. 'ids' allows specification of a column to be converted to the rownames of the new object.

#### See Also

```
ranged.to.data.frame, df.to.ranged
```

## **Examples**

```
chr <- sample(1:22,10)
start <- end <- sample(1000000,10)
df1 <- cbind(CHR=chr,Start=start,enD=end)</pre>
print(df1)
df.to.GRanges(df1) # not case sensitive!
width \leftarrow rep(0,10)
df2 <- cbind(chr,start,width)</pre>
df.to.GRanges(df2,end=NULL,width="width") # define ranges with start and width
id.col <- paste0("ID",1:10)</pre>
rs.id <- paste0("rs", sample(10000,10))
df3 <- cbind(chr,start,end,id.col,rs.id)</pre>
df.to.GRanges(df3) # additional columns kept
df4 <- cbind(chr,start,end,id.col,rs.id, ranges=1:10)</pre>
df.to.GRanges(df4) # 'ranges' column excluded as illegal name
df.to.GRanges(df4, exclude="rs.id") # manually exclude column
df5 <- cbind(chr,start,end,rs.id)</pre>
rownames(df5) <- paste0("ID",1:10)</pre>
df.to.GRanges(df5) # rownames are kept
df.to.GRanges(df4,ids="id.col") # use column of 'dat' for rownames
```

endSnp

Find closest SNPs to the ends of ranges

## **Description**

For given genome ranges (GRanges/RangedData) will try to find the closest snps to the end of the ranges.

# Usage

```
endSnp(ranged = NULL, snp.info = NULL, chr = NULL, pos = NULL,
nearest = T)
```

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# Arguments

ranged	A GRanges or RangedData object specifying the range(s) you wish to find SNPs near the ends of. Alternatively leave this parameter as NULL and specify ranges using chr, pos
snp.info	ChipInfo/GRanges/Ranged data object describing the SNPs relevant to your query, e.g, SNPs on the chip you are using. If left NULL, the SNP set used will be that retrieved by chip.support() which will depend on your options() settings, see ?chip.support for more info
chr	optional alternative to 'ranged' input, use in conjunction with 'pos' to specify the ranges to find the SNPs near the ends of.
pos	matrix with 2 columns for start, end positions, or a single column if all ranges are SNPs. An optional alternative to 'ranged' input, use in conjunction with 'chr' to specify the ranges to find the SNPs near the ends of.
nearest	will preferably find an exact match but if nearest=TRUE, will fall-back on nearest match, even if slightly outside the range.

## Value

a list of SNP-ids (rownames of 'snp.info') fulfilling the criteria, the output vector (character) should be the same length as the number of ranges entered.

# **Examples**

```
endSnp(chr=c(1:3),pos=cbind(c(100000,200000,300000),c(30000000,4000000,10000000)))
endSnp(rranges())
```

ENS.to.GENE

Convert ensembl ids to HGNC gene ids

# Description

Retrieve the gene IDs (HGNC) corresponding to a list of ensembl gene ids. Note that this will not find all IDs found on ensembl.org, as it uses bioMart which seems to be incomplete, but this only pertains to a small minority of genes, so this function should have general utility for most applications. This is of course the case at the time of writing - bioMart is likely to be updated at some point.

## Usage

```
ENS.to.GENE(ens, dir = NULL, build = NULL, name.dups = FALSE,
   name.missing = TRUE, ...)
```

## **Arguments**

ens	character, a list of ensembl gene ids, of the form ENSG00xxxxxxxxx
dir	character, 'dir' is the location to download gene and cytoband information; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.

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build	character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The default when build is NULL is to use the build from the current ChipInfo annotation
name.dups	logical, if TRUE then duplicates will have a suffix appended to force the list to be unique (e.g, so it would be usable as rownames, or in a lookup table). Otherwise duplicate entries will just appear in the list multiple times
name.missing	logical, if TRUE then missing values will be named as MISSING_n (n=1 to # of missing), ensuring a valid unique name if the results are to be used as rownames, etc. If FALSE then these will be left as NA.
	further arguments to get gene annot()

#### Value

Returns a vector of HGNC gene ids corresponding to the 'ens' ensembl ids entered, any ids not found will be returned as MISSING\_n (n=1 to # of missing), if name.missing=TRUE. If name.missing is FALSE then missing will be set to NA. Similarly with 'name.dups', if duplicates are found and name.dups is true, each will be appended with suffix \_n; else their names will be left as is.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

```
GENE.to.ENS, rs.to.id, id.to.rs; eg2sym, sym2eg from package 'gage'
```

# Examples

```
\label{eq:setwd} setwd(tempdir()) \\ ENS.ids <- c("ENSG00000183214", "ENSG00000163599", "ENSG00000175354", "ENSG00000134460") \\ ENS.to.GENE(ENS.ids) \\ gene.ids <- c("HLA-B","IFIH1","fake_gene!","FUT2") \\ ENS.to.GENE(GENE.to.ENS(gene.ids)) # lookup fails for the fake id, gives warning \\ \\ Setward the setward that the setward th
```

expand.nsnp

Expand genomic locations to the ranges covering the 'n' closest SNPs

#### **Description**

Sometimes for chip data we want to create windows around some locus, and fixed distance [see flank()], recombination distance [see recomWindow()] or a number of SNPs might be used. This function allows expansion of regions according to a set number of SNPs. The result gives two regions for each row of a GRanges or RangedData object describing the start and end of the left flanking 'nsnp' region, and right flanking 'nsnp' region respectively.

## Usage

```
expand.nsnp(ranged, snp.info = NULL, nsnp = 10, add.chr = FALSE)
```

## **Arguments**

ranged	a GRanges or RangedData object describing the locations for which we want to find regions encompassing 'nsnps' closest SNPs.	
snp.info	An object of type: ChipInfo, RangedData or GRanges, describing the set of SNPs you are using (e.g, chip annotation). If left as null the ChipInfo object from chip.support() with default options() will be used	
nsnp	Number of nearest SNPs to return for each location	
add.chr	add.chr logical, whether to add a chromosome column for the output object	

## Value

Two regions for each row of a the 'ranged' object describing the start and end of the left flanking 'nsnp' region, and right flanking 'nsnp' region respectively. If 'ranged' has rownames these should stay in the same order in the resulting object. Chromosome will be the final column if you set add.chr=TRUE.

## See Also

```
nearest.snp, chip.support, recomWindow
```

# **Examples**

```
rngs <- rranges()
# not run - slow ~5 seconds # expand.nsnp(rngs)
# not run - slow ~5 seconds # expand.nsnp(rngs,add.chr=TRUE)</pre>
```

```
extraColumnSlotNames2-methods
```

~~ Methods for Function extraColumnSlotNames2 ~~

# Description

~~ Methods for function extraColumnSlotNames2 ~~

## Methods

```
signature(x = "ANY")
```

36 Gene.pos

_	- 1	
force	chr	$n \cap S$

Force a valid genomic range, given the inputted coordinates

#### **Description**

Enter a pair of genomic locations representing a range for a given chromosome and this function will ensure that no position is less than 1 or greater than the relevant chromosome lengths. Anything below will be coerced to 1, and anything above to the chromosome length.

## Usage

```
force.chr.pos(Pos, Chr, snp.info = NULL, build = NULL, dir = NULL)
```

# Arguments

Pos	must be numeric, length 2, e.g, c(20321,30123)
Chr	chromosome label
snp.info	optional object to take boundaries from, the maxima and minima for each chromosome within this object will take the place of the chromsome lengths $/\ 1$ .
build	ucsc build, only need to enter if this differs from getOption("ucsc")
dir	directory to use for download of chromosome lengths (only if you wish to keep

the chromosome length file)

## **Examples**

```
pss <- ps <- c(345035,345035); ch <- 1
force.chr.pos(ps,ch)
pss[1] <- 0
force.chr.pos(pss,ch) # won't allow zero
pss[1] <- -1
force.chr.pos(pss,ch) # won't allow negative
pss[1] <- 645035012
force.chr.pos(pss,ch) # won't allow pos > chromosome length
```

Gene.pos

Find the gene(s) overlapping a chromosome location

# **Description**

Allows retrieval of genes intersected by a chromosome and position, which can be entered using chr, pos/start/end vectors, or a RangedData or GRanges object

# Usage

```
Gene.pos(chr = NA, pos = NA, start = NA, end = NA, ranges = NULL,
build = NULL, dir = NULL, bioC = FALSE, one.to.one = TRUE)
```

Gene.pos 37

#### **Arguments**

chr	character, an optional vector of chromosomes to combine with 'pos' or 'start'+'end' (enter in) to describe positions to retrieve the possible overlapping gene(s)
pos	integer, an optional vector of chromosome positions (for SNPs), no need to enter start or end if this is entered, and vice-versa
start	integer, an optional vector of start points for chromosome ranges
end	integer, an optional vector of end points for chromosome ranges
ranges	optional GRanges or RangedData object describing positions for which we want genes, removing the need to enter chr, pos, start or end
build	character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The default when build is NULL is to use the build from the current ChipInfo annotation
dir	character, 'dir' is the location to download gene annotation information to; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.
bioC	logical, if true then return position information as a GRanges object, or Ranged- Data if 'ranges' is RangedData, else a data.frame
one.to.one	logical, whether to concatenate multiple hits for the same range into one result, or spread the result over multiple lines, one for each gene overlapped

#### Value

Returns a set of genes separated by semicolons (if more than one) for each range entered. If bioC=TRUE, returns the equivalent as a GRanges object, unless a RangedData object was used for the ranges parameter, in which case a RangedData object would be returned. If one.to.one is FALSE, then instead of concatenating multiple genes into one line per range, each is listed separately as a new row, with an index added to correspond to the original input order of ranges, if bioC=TRUE; or just adds additional elements to the resulting vector if bioC=FALSE.

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

Chr, Pos, Pos. band, Band, Band. gene, Band. pos, Gene. pos

```
setwd(tempdir())
Gene.pos(chr=6, start=31459636, end=31462760)
Gene.pos(chr=22, pos=3452345) # no gene here
Gene.pos(chr("rs689"),Pos("rs689")) # combine with Chr() and Pos() to find gene(s) for SNP rs689
Gene.pos(chr=1,start=114000000,end=1150000000,build="hg19") # multiple genes in range
Gene.pos(chr=1,start=114000000,end=1150000000,one.to.one=FALSE) # list separately
ii <- Pos.gene(c("CTLA4","PTPN22"))
Gene.pos(ii$chr,ii$start,ii$end,bioC=FALSE) # returns same genes inputted on line above</pre>
```

38 GENE.to.ENS

GENE.to.ENS

Convert gene ids to ensembl ids

# **Description**

Retrieve the ensembl IDs corresponding to a list of common gene names (HGNC format).

## Usage

```
GENE.to.ENS(genes, dir = NULL, ...)
```

# Arguments

genes	character, gene labels, e.g, "APOE"
dir	character, 'dir' is the location to download gene and cytoband information; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.
	further arguments to get.gene.annot()

## Value

Returns a vector of HGNC gene ids corresponding to the 'ens' ensembl ids entered, any ids not found will be returned as NA.

## Author(s)

```
Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>
```

## See Also

```
GENE.to.ENS, rs.to.id, id.to.rs; eg2sym, sym2eg from package 'gage'
```

```
setwd(tempdir())
gene.ids <- c("MYC","PTPN2","IL2RA","APOE")
GENE.to.ENS(gene.ids)</pre>
```

get.centromere.locs 39

## Description

Returns the locations of centromeres in the human genome, for a given build, as a list by chromosome, text vector, or GRanges/RangedData object.

## Usage

```
get.centromere.locs(dir = NULL, build = NULL, bioC = TRUE,
    GRanges = TRUE, text = FALSE, autosomes = FALSE)
```

## **Arguments**

dir	character, location to store file with the this annotation. If NULL then getOption("save.annot.in.current")>=1 will result in this file being stored in the current directory, or if <=0, then this file will not be stored.
build	string, currently 'hg18' or 'hg19' to specify which annotation version to use. Default is build-36/hg-18. Will also accept integers 36,37 as alternative arguments.
bioC	logical, whether to return the annotation as a ranged S4 object (GRanges or RangedData), or as a data.frame.
GRanges	logical, whether to return a GRanges object, or FALSE to return RangedData
text	logical, whether to return locations as a text vector of the form: chrN:xxxx-xxxx
autosomes	logical, if TRUE, only return results for autosomes, if FALSE, also include $\boldsymbol{X}$ and $\boldsymbol{Y}$ .

## Value

Returns a list, GRanges or RangedData object, depending on input parameters. Contained will be centromere chromosome and start and end positions.

```
setwd(tempdir())
get.centromere.locs()
get.centromere.locs(bioC=FALSE,autosomes=TRUE)
get.centromere.locs(text=TRUE)
```

40 get.chr.lens

## Description

Quick and easy way to retrieve human chromosome lengths. Can select from hg18/hg19 (ie, build 36/37), or any future builds (hg20, etc) stored in the same location on the build website. Default is to return lengths for 22 autosomes, but can also retrieve X,Y and Mitochondrial DNA lengths by 'autosomes=FALSE' or n=1:25. Even if not connected to the internet can retrieve hard coded lengths for hg18 or hg19.

## Usage

```
get.chr.lens(dir = NULL, build = NULL, autosomes = FALSE,
  len.fn = "humanChrLens.txt", mito = FALSE, names = FALSE,
  delete.after = FALSE, verbose = FALSE)
```

## **Arguments**

dir	directory to retrieve/download the annotation from/to (defaults to current getwd()) if dir is NULL then will automatically delete the annotation text file from the local directory after downloading
build	string, currently 'hg17', 'hg18' or 'hg19' to specify which annotation version to use. Default is getOption("ucsc"). Will also accept integers 17,18,19,35,36,37 as alternative arguments.
autosomes	logical, if TRUE, only load the lengths for the 22 autosomes, else load $X,Y,[MT]$ as well
len.fn	optional file name to keep the lengths in
mito	logical, whether to include the length of the mitochondrial DNA (will not include unless autosomes is also FALSE)
names	logical, whether to name the chromosomes in the resulting vector
delete.after	logical, if TRUE then delete the text file that these lengths were downloaded to.
verbose	logical, if TRUE display extra information on progress of chromsome retrieval If FALSE, then the file will be kept, meaning future lookups will be faster, and available offline.

```
setwd(tempdir())
get.chr.lens(delete.after=TRUE) # delete.after simply deletes the downloaded txt file after reading
get.chr.lens(build=35,autosomes=TRUE,delete.after=TRUE) # only for autosomes
get.chr.lens(build="hg19",mito=TRUE,delete.after=TRUE) # include mitochondrial DNA length
```

get.cyto 41

get.cyto	Return Cytoband/Karyotype locations across the genome	

## Description

Returns the locations of cytobands/karyotype-bands in the human genome, for a given build, as a data.frame, or GRanges/RangedData object.

## Usage

```
get.cyto(build = NULL, dir = NULL, bioC = TRUE, GRanges = TRUE,
  refresh = FALSE)
```

#### **Arguments**

build	string, currently 'hg18' or 'hg19' to specify which annotation version to use. Default is build-36/hg-18. Will also accept integers 36,37 as alternative arguments.
dir	character, location to store file with the this annotation. If NULL then getOption("save.annot.in.current")>=1 will result in this file being stored in the current directory, or if <=0, then this file will not be stored.
bioC	logical, whether to return the annotation as a ranged S4 object (GRanges or RangedData), or as a data.frame
GRanges	logical, whether to return a GRanges object, or FALSE to return RangedData
refresh	logical, whether to re-download the file if the existing file has become corrupted

## Value

Returns a list, GRanges or RangedData object, depending on input parameters. Contained will be centromere chromosome and start and end positions.

## **Examples**

```
require(BiocInstaller)
setwd(tempdir())
get.cyto()
cyto.frame <- get.cyto(bioC=FALSE)
prv(cyto.frame)
get.cyto(build=36)</pre>
```

get.exon.annot

Get exon names and locations from UCSC

### **Description**

Various R packages assist in downloading exonic information but often the input required is complex, or several lines of code are required to initiate, returning an object that might require some manipulation to be useful. This function simplifies the job considerably, not necessarily requiring any arguments. The object returned can be a standard data.frame or a bioconductor GRanges/RangedData object. The raw annotation file downloaded will be kept in the working directory so that subsequent calls to this function run very quickly, and also allow use offline.

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#### Usage

```
get.exon.annot(dir = NULL, build = NULL, bioC = T, transcripts = FALSE,
    GRanges = TRUE)
```

#### **Arguments**

dir character, location to store file with the gene annotation. If NULL then getOp-

tion("save.annot.in.current")>=1 will result in this file being stored in the current

directory, or if <=0, then this file will not be stored.

build string, currently 'hg18' or 'hg19' to specify which annotation version to use.

Default is build-36/hg-18. Will also accept integers 36,37 as alternative argu-

ments.

bioC logical, whether to return the annotation as a ranged S4 object (GRanges or

RangedData), or as a data.frame

transcripts logical, if TRUE, return transcripts rather than exons

GRanges logical, if TRUE and bioC is also TRUE, then returned object will be GRanges,

otherwise it will be RangedData

#### Value

Returns a data.frame, GRanges or RangedData object, depending on input parameters. Contained will be HGNC gene labels, chromosome, start and end positions, transcript id number and name

### **Examples**

```
setwd(tempdir())
get.exon.annot()
```

get.gene.annot

Get human gene names and locations from biomart

## Description

Various R packages assist in downloading genomic information but often the input required is complex, or several lines of code are required to initiate, returning an object that might require some manipulation to be useful. This function simplifies the job considerably, not necessarily requiring any arguments. The object returned can be a standard data.frame or a bioconductor GRanges/RangedData object. The raw annotation file downloaded will be kept in the working directory so that subsequent calls to this function run very quickly, and also allow use offline.

```
get.gene.annot(dir = NULL, build = NULL, bioC = TRUE,
  duplicate.report = FALSE, one.to.one = FALSE, remap.extra = FALSE,
  discard.extra = TRUE, only.named = FALSE, ens.id = FALSE,
  refresh = FALSE, GRanges = TRUE, host.txt = "")
```

get.gene.annot 43

# Arguments

dir	character, location to store file with the gene annotation. If NULL then getOption("save.annot.in.current")>=1 will result in this file being stored in the current directory, or if <=0, then this file will not be stored.
build	string, currently 'hg18' or 'hg19' to specify which annotation version to use. Default is build-36/hg-18. Will also accept integers 36,37 as alternative arguments.
bioC	logical, whether to return the annotation as a ranged S4 object (GRanges or RangedData), or as a data.frame
duplicate.repor	t
	logical, whether to provide a report on the genes labels that are listed in more than 1 row - this is because some genes span ranges with substantial gaps within them
one.to.one	logical, as per above, some genes have duplicate entries, sometimes for simplicity you want just one range per gene, if this parameter is set TRUE, one range per gene is enforced, and only the widest range will be kept by default for each unique gene label
remap.extra	logical, whether to remap chromosome annotation for alternative builds and unconnected segments to the closest regular chromosome, e.g, mapping MHC mappings to chromosome 6
discard.extra	logical, similar to above, but if TRUE, then any non-standard chromosome genes will just be discarded
only.named	logical, biomart annotation contains some gene segments without names, if TRUE, then such will not be included in the returned object (note that this will happen also if one.to.one is TRUE)
ens.id	logical, whether to include the ensembl id in the dataframe
refresh	logical, if you already have the file in the current directory, this argument will let you re-download and re-generate this file, e.g, if the file is modified or corrupted this will make a new one without having to manually delete it
GRanges	logical, if TRUE and bioC is also TRUE, then returned object will be GRanges, otherwise it will be RangedData
host.txt	character, the argument to pass to biomaRt::useMart(). Default for build 36 is 'may2009.archive.ensembl.org', and for build 37, "feb2014.archive.ensembl.org" but for recent builds the recommended link is 'www.ensembl.org'

# Value

Returns a data.frame, GRanges or RangedData object, depending on input parameters. Contained will be HGNC gene labels, chromosome and start and end positions, other information depends on specific parameters documented above

```
setwd(tempdir())
get.gene.annot()
```

44 get.GO.for.genes

get.genic.subset

Obtain subset of ranged object overlapping human genes

## **Description**

Return subset of a ranged object that overlaps human genes (or custom ranges/exons). A wrapper for subsetByOverlaps that tries to ensure equivalent builds and chromosome labelling are taken care of automatically, and provides the default case of genic subsetting where no explicit ref parameter is required.

### Usage

```
get.genic.subset(X, ref = NULL, build = NULL)
```

## Arguments

Χ	GRanges or RangedData object, ranged object for which you want the genic
	subset of ranges/SNPs.

ref GRanges or RangedData object, only use if you need to provide for a build other

than 36 or 37 (hg18/hg19), or to use this function for another reference set, for

instance you could provide an object with exons

build e.g, 36/hg18 or 37/hg19, if left as NULL current getOption('ucsc') will be used.

## Value

a GRanges object with the specified genic (or custom) ranges

#### See Also

```
get.gene.annot, get.exon.annot, subsetByOverlaps
```

## **Examples**

```
# all.reg <- rranges(1000) # random set of 1000 regions
# genic <- get.genic.subset(all.reg) # gene regions from the random set
# exonic <- get.genic.subset(all.reg,ref=get.exon.annot()) # exonic regions from the random set</pre>
```

get.GO.for.genes

Retreive GO terms from biomart for a given gene list

## Description

Gene-ontology terms (GO-terms) are commonly used for testing for simple functional enrichment for pathways, etc. This function can retrieve biological function, cellular component, or molecular description, depending on the parameters chosen.

get.immunobase.snps 45

#### Usage

```
get.GO.for.genes(gene.list, bio = T, cel = F, mol = F,
host.txt = "may2009.archive.ensembl.org")
```

## **Arguments**

gene.list

bio	logical, whether to return biological process GO terms
cel	logical, whether to return cellular component GO terms
mol	logical, whether to return molecular function GO terms
host.txt	character, the argument to pass to biomaRt::useMart(). Default is 'may2009.archive.ensembl.org', but more recently the recommended link is 'www.ensembl.org'

a list of gene, use HGNC names, like COMT, HLA-C, CTLA4, etc.

## Value

data.frame containing the gene name in the first column, chromosome in the second column, and the GO terms in the third column, where one gene has multiple GO terms, this will produce multiple rows, so there will usually be more rows than genes entered. The data.frame can have 3,4 or 5 columns depending on how many GO terms are selected.

## **Examples**

```
get.GO.for.genes(c("CTLA4","PTPN2","PTPN22")) # biological terms (default)
get.GO.for.genes(c("CTLA4","PTPN2","PTPN22"),cel=TRUE) # add cellular GO terms
```

## Description

Retrieve human disease top GWAS hits from t1dbase in build hg19 coords (37). 28 Diseases currently available

## Usage

```
get.immunobase.snps(disease = "T1D", snps.only = TRUE, show.codes = FALSE)
```

# Arguments

disease	integer (1-28), or character (abbreviation), or full name of one of the listed diseases. A full list of options can be obtained by setting show.codes=TRUE.
snps.only	logical, default is just to return a list of rs-ids. Setting FALSE gives a table
show.codes	logical, if set to TRUE, instead of looking up t1dbase, will simply return a table of available diseases with their index numbers and abbreviations.

## Value

A character vector of SNP rs-ids

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#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### References

PMID: 20937630

### **Examples**

```
get.immunobase.snps(disease="CEL") # get SNP ids for celiac disease
get.immunobase.snps(disease="AS") # get SNP ids for Ankylosing Spondylitis in build-37/hg19
get.immunobase.snps(show.codes=TRUE) # show codes/diseases available to download
get.immunobase.snps(disease=27) # get SNP ids for Alopecia Areata
get.immunobase.snps("Vitiligo")
```

get.immunog.locs

Retrieve locations of Immunoglobin regions across the genome

## Description

Returns the locations of immunoglobin regions in the human genome, for a given build, as a list by chromosome, text vector, or GRanges/RangedData object. For instance, for CNV research, these regions are known to be highly structurally complex and can lead to false positive CNV-calls, so are often excluded.

## Usage

```
get.immunog.locs(build = NULL, bioC = TRUE, text = FALSE,
    GRanges = TRUE)
```

## **Arguments**

build	string, currently 'hg18' or 'hg19' to specify which annotation version to use. Default is build-36/hg-18. Will also accept integers 36,37 as alternative arguments.
bioC	logical, whether to return the annotation as a ranged S4 object (GRanges or RangedData), or as a data.frame
text	logical, whether to return locations as a text vector of the form: chrN:xxxx-xxxx
GRanges	logical, whether to return a GRanges object, or FALSE to return RangedData

### Value

Returns a list, GRanges or RangedData object, depending on input parameters. Contained will be immunoglobin chromosome, start and end positions.

```
get.immunog.locs()
get.immunog.locs(bioC=FALSE)
get.immunog.locs(text=TRUE,build=37)
```

get.nearby.snp.lists 47

#### **Description**

For a snp.id (or list), extend a window around that chromosome location in recombination units (centimorgans) and return the list of SNPs from the current ChipInfo object that lie in this window. This is a way of extracting SNPs in linkage disequilibrium with an index SNP, that could also be plausible causal candidates. Runs fastest for build 36, otherwise internal conversion takes place (runs using build based on getOptions('ucsc')).

### Usage

```
get.nearby.snp.lists(snpid.list, cM = 0.1, bp.ext = 0, excl.snps = NULL,
  name.by.bands = TRUE)
```

## **Arguments**

snpid.list	character, list of snp-ids (e.g, rs-id or chip id) to obtain lists for. SNPs must all be from the same chromosome - if ranges for SNPs spanning multiple ranges are desired, you must use multiple calls. A warning will be given if SNPs from the same karyotype band are entered as index SNPs, as in a typical GWAS analysis only one SNP would be used like this from each region, ignore the warning if this is not the case for your application.
сМ	numeric, the number of centimorgans to extend the window either side of each SNP
bp.ext	numeric, optional number of base-pairs to extend the window by in addition to the centimorgan extension
excl.snps	character, a list of rs-id or chip-ids of SNPs to exclude from the list returned, as, for instance, they may have failed quality control such as call-rate.
name.by.bands	give labels to each sublist returned by the karotype/cytoband name, but faster not to do this

## Value

Returns a list of vectors of snp-ids falling within the window(s) specified and not in 'excl.snps'. Each snp in 'snpid.list' will correspond to an element in the list returned. If name.by.bands is TRUE, then these list elements will each be named using the local karyotype/cytoband location

# See Also

```
snps.in.range, get.recombination.map, recomWindow, conv.37.36, conv.36.37, expand.nsnp
```

```
# examples not run as too slow

result <- get.nearby.snp.lists("rs900569")
# trick below to extract SNPs within 0.1-0.2cM
get.nearby.snp.lists("rs900569",cM=0.2,excl.snps=result[[1]])
# note that the same query can return a different set with build 36 versus 37</pre>
```

```
get.nearby.snp.lists(c("rs689","rs4909944"),cM=0.001,name.by.bands=FALSE)
```

get.recombination.map Get HapMap recombination rates for hg18 (build 36)

### **Description**

Recombination rate files can be used to calculate recombination distances for genome locations, in centimorgans. This function downloads these reference files from the hapmap NCBI website. At the time of writing they were only available for build 36. If using a more recent build I suggest using the conversion function conv.37.36(), then recomWindow(), then conv.36.37() to get recombination distances for other builds. If getOption("save.annot.in.current") is <=0 then no files will be kept. Otherwise an object containing this mapping data will be saved in the local directory if dir=NULL, or else in the directory specified. Allowing this reference to be saved will greatly increase the speed of this function for subsequent lookups

### Usage

```
get.recombination.map(dir = NULL, verbose = TRUE, refresh = FALSE,
   compress = FALSE)
```

### **Arguments**

dir	character, location to store binary file with the recombination maps for chromosomes 1-22. If NULL then getOption("save.annot.in.current")>=1 will result in this file being stored in the current directory, or if <=0, then this file will not be stored.
verbose	logical, if the binary file is not already downloaded, when verbose is TRUE, there will be some output to the console indicating the progress of the download. If FALSE, all output is suppressed.
refresh	logical, if you already have the binary file in the current directory, this argument will let you re-download and re-generate this file, e.g, if the file is modified or corrupted this will make a new one without having to manually delete it
compress	logical, this argument is passed to 'save' and will result in a larger binary file size, but quicker loading times, so 'FALSE' is recommended for faster retrieval.

## Value

Returns a list object of length 22, containing the recombination map files as 22 separate data.frame's.

```
## not run as it takes roughly 2 minutes to download and read-in ##
setwd(tempdir())
rec.map <- get.recombination.map(getwd())
file.on.disk <- "rrates_genetic_map_chr_1_22_b36.RData"
if(file.exists(file.on.disk)) { unlink(file.on.disk) } # remove the downloaded file</pre>
```

get.t1d.regions 49

ge	et.t1d.regions	Obtain a listing of known T1D associated genomic regions

## Description

This function uses a full list of ichip dense regions combined with a list of t1d SNPs to get the t1d regions. For type 1 diabetes researchers.

## Usage

```
get.t1d.regions(dense.reg = NULL, build = NULL, invert = FALSE)
```

## **Arguments**

dense.reg	GRanges or RangedData object, only use if you need to provide for a build other than 36 or 37 (hg18/hg19).
build	e.g, $36/hg18$ or $37/hg19$ , if left as NULL current getOption('ucsc') will be used.
invert	logical, set to TRUE if you wish to get the set of NON-T1D regions.

## Value

a GRanges object with the specified type 1 diabetes (or inverse) ranges

## Examples

```
# t1d.reg <- get.t1d.regions()
# non.t1d <- get.t1d.regions(build=36,invert=TRUE)</pre>
```

get.t1d.subset	Obtain subset of ranged object overlapping known T1D associated
	genomic regions

## Description

Return subset of a ranged object that overlaps ichip dense mapped regions. For type 1 diabetes and autoimmune disesase researchers.

```
get.t1d.subset(X, T1D.only = TRUE, build = NULL, ichip.regions = NULL,
   T1D.regions = NULL, invert = FALSE)
```

50 get.telomere.locs

## **Arguments**

X	GRanges or RangedData object, ranged object for which you want the T1D subset of ranges/SNPs.
T1D.only	logical, standard is to return type 1 diabetes (T1D) regions subset, but if this parameter is set to FALSE, will return the subset for all 12 autoimmune diseases mapped by the ImmunoChip consortium. (Cortes and Brown, 2010).
build	e.g, $36/hg18$ or $37/hg19$ , if left as NULL current getOption('ucsc') will be used.
ichip.regions	GRanges or RangedData object, only use if you need to provide for a build other than 36 or 37 (hg18/hg19), or for multiple lookups to avoid reloading each time
T1D.regions	GRanges or RangedData object, only use if you need to provide for a build other than 36 or 37 (hg18/hg19), or for multiple lookups to avoid reloading each time.
invert	logical, set to TRUE if you wish to get the set of NON-T1D regions, or non-immune dense regions when T1D.only=FALSE.

## Value

a GRanges object with the specified type 1 diabetes/autoimmune (or inverse) ranges

## **Examples**

```
# all.reg <- rranges(10000)
# t1d <- get.t1d.subset(all.reg) # T1D regions
# non.autoimmune <- get.t1d.subset(T1D.only=FALSE,build=36,invert=TRUE) # non-autoimmune regions</pre>
```

get.telomere.locs

Derive Telomere locations across the genome

## **Description**

Returns the locations of telomeres in the human genome, for a given build, as a list by chromosome, text vector, or GRanges/RangedData object.

# Usage

```
get.telomere.locs(dir = NULL, kb = 10, build = NULL, bioC = TRUE,
    GRanges = TRUE, text = FALSE, autosomes = FALSE, mito.zeros = FALSE)
```

## Arguments

dir	character, location to store file with the this annotation. If NULL then getOption("save.annot.in.current")>=1 will result in this file being stored in the current directory, or if <=0, then this file will not be stored.
kb	The number of base pairs at the start and end of a chromosome that are defined as belonging to the telomere can be a little arbitrary. This argument allows specification of whatever threshold is required.
build	string, currently 'hg18' or 'hg19' to specify which annotation version to use. Default is build-36/hg-18. Will also accept integers 36,37 as alternative arguments.

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bioC	logical, whether to return the annotation as a ranged S4 object (GRanges or RangedData), or as a data.frame
GRanges	logical, whether to return a GRanges object, or FALSE to return RangedData
text	logical, whether to return locations as a text vector of the form: chrN:xxxx-xxxx
autosomes	logical, if TRUE, only return results for autosomes, if FALSE, also include $\boldsymbol{X}$ and $\boldsymbol{Y}$ .
mito.zeros	logical, Mitochondria have no telomeres (are circular) but for some purposes you might want zero values in order to match with other annotation that includes all chromosomes and MT. TRUE adds zeros for chrMT, and FALSE excludes chrMT

#### Value

Returns a text vector, GRanges or RangedData object, depending on input parameters. Contained will be telomere chromosome and start and end positions.

### **Examples**

```
setwd(tempdir())
get.telomere.locs()
get.telomere.locs(bioC=FALSE)
get.telomere.locs(text=TRUE)
```

iChipRegionsB36

Autoimmune enriched regions as mapped on ImmunoChip

### **Description**

Dataset. A consortium of 12 autoimmune diseases (Type 1 diabetes, Celiac disease, Multiple Sclerosis, Crohns Disease, Primary Billiary Cirrhosis, Psoriasis, Rheumatoid Arthritis, Systemic Lupus Erytematosus, Ulcerative Colitis, Ankylosing Spondylitis, Autoimmune Thyroid Disease, Juvenile Idiopathic Arthritis) created the ImmunoChip custom Illumina iSelect microarray in order to investigate known regions from GWAS associating with a p value < 5\*10-8 with any of these diseases, using dense mapping of locii. This object specifies the boundaries of these regions, defined roughly as 0.1 centimorgan recombination distance either side of the top marker in each. The data is in the original build 36 coordinates as a GRanges object, but using functions in the humarray package can easily be converted to build 37, 38 or RangedData/data.frame.

#### **Format**

An object of class GRanges

#### References

Cortes and Brown, Promise and pitfalls of the Immunochip (2011). Arthritis Research and Therapy 2011, 13:101

### See Also

```
conv.36.37 get.t1d.subset get.t1d.regions get.immunobase.snps
```

52 id.to.rs

## **Examples**

```
data(iChipRegionsB36)
prv(iChipRegionsB36)
iChipRegionsB37 <- conv.36.37(iChipRegionsB36)</pre>
```

id.to.rs

Convert from chip ID labels to dbSNP rs-ids

## **Description**

Most SNPs will have an 'rs-id' from dbSNP/HapMap, and these are often the standard for reporting or annotation lookup. These can differ from the IDs used on the chip. This functions looks at the current snp support (ChipInfo object) and returns rs-ids in place of chip IDs. Currently rs-ids are always from build37.

## Usage

```
id.to.rs(ids)
```

# Arguments

ids

character, meant to be a list of chip ids, but if rs-ids are present they will not be altered.

### Value

A character vector of SNP rs-ids, where the input was chip ids, rs-ids or a mixture, any text other than this will result in NA values being returned in the character vector output.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

### See Also

```
rs.to.id, GENE.to.ENS, ENS.to.GENE
```

```
id.to.rs(c("imm\_11\_2138800","rs9467354","vh\_1\_1108138")) \ \ \# \ middle \ one \ is \ already \ a \ rs.id
```

ids.by.pos 53

ids.by.pos

Order rs-ids or ichip ids by chrosome and position

#### **Description**

Simple function to sort a character list of SNP ids into genome order.

## Usage

```
ids.by.pos(ids)
```

## **Arguments**

ids

character, vector of SNP rs-ids or chip-ids, see rs.to.id()

#### Value

the same vector 'ids', sorted by genome position

#### See Also

```
rs.to.id, id.to.rs, Chr, Pos
```

### **Examples**

```
snp.ids <- c("rs3842724","imm_11_2147527","rs689","rs9467354","rs61733845")
Chr(snp.ids) # shows each is on a different chromosome
Pos(snp.ids)
ids.by.pos(snp.ids)
Chr(ids.by.pos(snp.ids))
Pos(ids.by.pos(snp.ids))</pre>
```

ImmunoChipB37

ImmunoChip annotation object (built-in)

## **Description**

Dataset. This object contains chromosome, position, chip SNP labels, SNP rs-ids, for immunochip, plus allele codes based on a UVA/Sanger T1D dataset, which should be updated for use with a different dataset with different allele coding (however the position and rs-id information should still be applicable. The data is in build 37 coordinates as a ChipInfo object, but using functions convTo36 or convTo38 from the humarray package can easily convert this to build 36 or 38.

## Format

An object of class GRanges

54 in.window

#### See Also

```
convTo38 convTo36 A1
```

#### **Examples**

```
data(ImmunoChipB37)
ImmunoChipB36 <- convTo36(ImmunoChipB37)</pre>
```

in.window

Select all ranges lying within a chromosome window

## Description

Input a ranged object (ie., GRanges or RangedData) and this function will return the subset from chromosome 'chr' and within the base-pair range specified by 'pos', in units of 'unit'. By default ranges with ANY overlap are returned, but it can be specified that it must be full overlap. Duplicates can be removed.

## Usage

```
in.window(ranged, chr, pos, full.overlap = F, unit = c("b", "kb", "mb",
    "gb"), rmv.dup = FALSE)
```

## **Arguments**

ranged	GRanges or RangedData object
chr	a chromosome, e.g, 1,2,3,,22,X,Y,XY,MT or however chromosomes are annotated in 'ranged'
pos	a numeric range (length 2), with a start (minima) and end (maxima), specifying the window on the chromosome to select ranges from, base-pair units are specified by 'unit'.
full.overlap	logical, the default is to return objects with ANY overlap with the window, whereas setting this as TRUE, will only return those that fully overlap
unit	the unit of base-pairs that 'pos' is using, eg, "b", "kb", "mb", "gb"
rmv.dup	logical, whether to remove duplicate ranges from the return result. The default is not to remove duplicates.

#### Value

an object of the same type as 'ranged', but only containing the rows that were within the specified bounds

```
require(GenomicRanges)
iG <- get.immunog.locs()[2,] # select the 2nd iG region
ciG <- chrm(iG) # get the chromosome
posiG <- c(start(iG),end(iG)) # get the region start and end
rr <- rranges(10000) # create a large random GRanges object
in.window(rr,chr=ciG,pos=posiG) # set with ANY overlap of iG
in.window(rr,chr=ciG,pos=posiG,TRUE) # set with FULL overlap of iG
in.window(rr,chr=6,pos=c(25,35),unit="mb") # look between 25 - 35 MB on chr6 [ie, MHC]</pre>
```

invGRanges 55

invGRanges	Invert a ranged object Select the empty space between ranges for the whole genome, for instance you may want to overlap with everything NOT in a set of ranges.

## **Description**

Invert a ranged object Select the empty space between ranges for the whole genome, for instance you may want to overlap with everything NOT in a set of ranges.

### Usage

```
invGRanges(X, inclusive = FALSE, build = NULL,
  pad.missing.autosomes = TRUE)
```

## **Arguments**

X a ranged object, GRanges, RangedData or ChipInfo

inclusive logical, TRUE if the ends of ranges should be in the inverted object

build character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The

default when build is NULL is to use the build from the current ChipInfo anno-

tation

pad.missing.autosomes

logical, whether to add entire chromosomes to the inverted range object when

they are not contained within X

### Value

a ranged object of the same type as X, but with the inverse set of human genomic ranges selected

## **Examples**

```
X <- rranges()
invGRanges(X,inclusive=TRUE)
invGRanges(X)
invGRanges(X,pad.missing.autosomes=FALSE)</pre>
```

lambda\_1000

Normalize Lambda inflation factors to specific case-control count

## Description

Lambda inflation statistics are influenced by the size of the generating datasets. To facilitate comparison to other studies, this function calculates then converts a given lambda from n cases and m controls, to be equivalent to 1000 cases and 1000 controls.

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#### Usage

```
lambda_1000(p.values, n = 1000, m = 1000)
```

### **Arguments**

p.values	numeric, a vector of analysis p.values, generated from n cases and m controls (although order switching n/m makes no difference to this function)
n	integer, original number of cases that p.values were derived from
m	integer, original number of controls that p.values were derived from

#### Value

A normalized Lambda coefficient

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### References

Freedman M.L., et al. Assessing the impact of population stratification on genetic association studies. Nat. Genet. 2004;36:388-393.

### **Examples**

```
# create some p-values with clear 'inflation' (divergence from uniform[0,1])
p.vec <- c(runif(3000)/200,runif(7000))
# let's imagine these p values come from 3000 cases and 5000 controls
L1000_a <- lambda_1000(p.vec,3000,5000)
# alternatively, imagine the sample sizes are 10 times larger
L1000_b <- lambda_1000(p.vec,30000,50000)
plot(sort(p.vec),type="l")
L1000_a; L1000_b</pre>
```

makeGRanges

Wrapper to construct GRanges object from chr,pos or chr,start,end

### **Description**

Slightly simplifies the creation of a GRanges object, allowing flexible input of chr, pos, or chr, start, end, and specification of rownames and the 'genome' parameter for specifying the build/coordinate type, e.g, hg18, build 37, etc. Designed for a simplified GRanges object without metadata, and where the 'strand' data is of no interest, so if strand/metadata is to be used, use the original GRanges() constructor.

```
makeGRanges(chr, pos = NULL, start = NULL, end = NULL, row.names = NULL,
build = NULL, ...)
```

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### **Arguments**

chr	character, an optional vector of chromosomes to combine with 'pos' or 'start'+'end' (enter in) to describe positions for the GRanges object
pos	integer/numeric, for SNPs, can enter positions just once in 'pos' instead of entering the same value for start and end
start	integer/numeric, specify the start position of ranges to encode in the new GRanges object, alongside 'end' (do not use 'pos' if using start+end)
end	integer/numeric, specify the end position of ranges to encode in the new GRanges object, alongside 'start' (do not use 'pos' if using start+end)
row.names	character, rownames for the output object, e.g, unique IDs describing the ranges
build	character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The default when build is NULL is to use the build from the current ChipInfo annotation
	further arguments to df.to.GRanges, such as 'fill.missing'

#### Value

Returns a GRanges object with the ranges, build and rownames specified. Rownames will be 1:nrow if the 'row.names' parameter is empty. The strand information will default to '+' for all entries, and the metadata will be empty (this function is only for creation of a very basic GRanges object).

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

## See Also

```
Chr, Pos, Pos.gene, Band, Band.gene, Band.pos, Gene.pos, zlink{df.to.GRanges}
```

## **Examples**

manifest

Convert from chip/rs-ids to manifest chip ID labels

## **Description**

Some SNP-ids aren't legal R row/column names. In order to match datafiles and store annotation as objects, this package converts SNP-names to sanitized versions if necessary, that are legal row/column names. This function converts from such 'legal' versions of the IDs back to the proper names, as per the chip manifest document (or whatever is stored in the chip.id field of the chip.support() object, accessible using chipId()).

```
manifest(ids)
```

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## Arguments

ids

character, either a list of rs-ids or chip-ids. chip ids are preferable as they are unique, and rs.ids are not. Using this function is not recommended for rs.id lists that might have entries that map to multiple chip ids, because entries other than the first will be ignored. For such cases, use 'rs.to.id(manifest=TRUE,multi.list=TRUE,...)'.

#### Value

A character vector of SNP chip-ids matching the manifest format.

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

```
id.to.rs, GENE.to.ENS, ENS.to.GENE
```

#### **Examples**

```
test.ids <- c("imm_1_898835","rs61733845","rs115005664","rs114582555",
    "chr1_20131940","chr1_20133829","rs150992667","rs138231315","rs111577708","rs187104718")
manifest(c("chr1_20131940","ccc_1_67429655_A_G"))
manifest(test.ids)  # even when some are rs-id, still works
data.frame(rs.id=test.ids,legal.id=rs.to.id(test.ids),manifest.id=manifest(test.ids))</pre>
```

meta.me

Meta-analysis using odds ratio and standard error from 2 datasets

#### **Description**

This function calculates meta analysis odds ratios, standard errors and p-values using results from a table containing odds ratio and standard error data for analyses of 2 different datasets (typically logistic regression, but other analyses can be incorporated if an odds-ratio and SE can be derived, for instance one analysis might be a case control logistic regression GWAS and the other a family TDT analysis).

## Usage

# Arguments

X	A data.frame with column names which should be entered in the parameters: OR1, OR2, SE1, SE2, and optionally N1, N2.
OR1	The column name of X containing odds ratios from the first analysis
OR2	Same as OR1 above but pertaining to the second analysis
SE1	The column name of X containing standard errors from the first analysis
SE2	Same as SE1 above but pertaining to the second analysis

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Z1	Only use if method="sample.size" or "z.score". The column name in X with the z.scores in the first analysis.
Z2	Same as Z1 above but pertaining to analysis 2
N1	Only required if method="sample.size". Either the column name in X with the number of samples in the first analysis, of a vector of the same, or if N's is the same for all rows, a scalar' value can be entered for each.
N2	Only required if method="sample.size". Same as N1 above but pertaining to analysis 2
method	character, can be either 'beta', 'z.score' or 'sample.size', and upper/lower case does not matter. 'Beta' is the default and will calculate meta-analysis weights using the inverse variance method (based on standard errors), and will calculate the p-values based on the weighted beta coefficients of the two analyses. 'Z.score' also uses inverse variance but calculates p-values based on the weighted Z scores of the two analyses. 'Sample.size' uses the sqrt of the sample sizes to weight the meta analysis and uses Z scores to calculate p values like 'Z.score' does.#'

#### Value

The object returned should have the same number of rows and rownames as the data.frame X but columns are the meta analysis stastistics, namely: OR.meta, beta.meta, se.meta, z.meta, p.meta, which will contain the meta analysis odds-ratio, beta-coefficient, standard error, z-score, and p-values respectively for each row of X.

### **Examples**

```
X <- data.frame(OR_CC=c(1.8,1.15),OR_Fam=c(1.33,0.95),SE_CC=c(0.02,0.12),SE_Fam=c(0.07,0.5))
rownames(X) <- c("rs689","rs23444")
X
meta.me(X)
X <- data.frame(OR_CC=c(1.8,1.15),OR_CC2=c(1.33,0.95),
SE_CC=c(0.02,0.12),SE_CC2=c(0.02,0.05),
n1=c(5988,5844),n2=c(1907,1774))
# even with roughly the same number of samples the standard error will determine the influence of
# each analysis on the overall odds ratio, note here that the second SE for dataset goes
# from 0.5 to 0.05 and as a result the estimate of the odds ratio goes from 1.137 to 0.977,
# i.e, from very close to OR1, changing to very close to OR2.
meta.me(X,OR2="OR_CC2",SE2="SE_CC2")
# sample size and z-score methods give similar (but distinct) results
meta.me(X,OR2="OR_CC2",SE2="SE_CC2",N1="n1",N2="n2",method="sample.size")
meta.me(X,OR2="OR_CC2",SE2="SE_CC2",N1="n1",N2="n2",method="z.score") # N's will be ignored</pre>
```

nearest.gene

Retrieve the 'n' closest GENE labels or positions near specified locus

## **Description**

Retrieve the 'n' closest GENE labels or positions near specified locus

```
nearest.gene(chr, pos, n = 1, side = c("either", "left", "right"),
  ids = TRUE, limit = NULL, build = NULL, ga = NULL)
```

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### **Arguments**

chr	integer, chromosome, should be a number from 1 to 25, where 23,24,25 are X,Y,MT
pos	integer, genomic position, should be between 1 and the length of the chromosome 'chr'
n	integer, the number of nearest GENEs to seek, if there aren't enough in the annotation then NAs will fill the gaps to force the return value length to equal 'n'
side	character, can be 'either', 'left' or 'right' and specifies which side of the 'pos' to look for nearest genes (where left is decreasing genomic position and right is increasing)
ids	logical, if TRUE will return GENE labels, or if FALSE will return the chromosome positions of the genes
limit	integer, a limit on the maximum distance from the position 'pos' can be specified
build	integer whether to use build 36/37 parameters, 36/37 is preferred, but can enter using any form recognised by ucsc.sanitizer()
ga	RangedData object, e.g, result of get.gene.annot(); gene annotation to save download time if repeatedly calling this function

#### Value

Set of GENE ids (when ids=TRUE), or otherwise genomic positions within chromosome 'chr'. If the number of gemes on the chromosome or the bounds of the 'side' and 'limit' parameters restrict the number returned to less than 'n' then the return value will be padded with NAs.

## See Also

```
\verb|expand.nsnp|, \verb|nearest.snp|, \verb|get.gene.annot||\\
```

## **Examples**

```
nearest.gene(1,159000000,n=10) # return ids
nearest.gene(1,159000000,n=10,build=37)
nearest.gene(1,159000000,n=10,build=36,ids=FALSE) # return positions
nearest.gene(1,159000000,n=10,build=37,ids=FALSE)
nearest.gene(6,25000000,n=10,build=37,ids=FALSE,side="left") # only genes to the left of the locus
nearest.gene(6,25000000,n=10,build=37,ids=FALSE,side="right") # only genes to the right of the locus
```

nearest.snp

Retrieve the 'n' closest SNP ids or positions near specified locus

## Description

Retrieve the 'n' closest SNP ids or positions near specified locus

```
nearest.snp(chr, pos, n = 1, side = c("either", "left", "right"),
  ids = TRUE, limit = NULL, build = NULL)
```

## **Arguments**

chr	integer, chromosome, should be a number from 1 to 25, where 23,24,25 are $X,Y,MT$
pos	integer, genomic position, should be between 1 and the length of the chromosome 'chr' $$
n	integer, the number of nearest SNPs to seek, if there aren't enough in the annotation then NAs will fill the gaps to force the return value length to equal 'n'
side	character, can be 'either', 'left' or 'right' and specifies which side of the 'pos' to look for nearest snps (where left is decreasing genomic position and right is increasing)
ids	logical, if TRUE will return snp ids (chip ids, for rs-ids, use id.to.rs on the output), or if FALSE will return the chromosome positions of the SNPs.
limit	integer, a limit on the maximum distance from the position 'pos' can be specified
build	integer whether to use build 36/37 parameters, 36/37 is preferred, but can enter using any form recognised by ucsc.sanitizer()

## Value

Set of SNP ids (when ids=TRUE), or otherwise genomic positions within chromosome 'chr'. If the number of SNPs on the chromosome or the bounds of the 'side' and 'limit' parameters restrict the number returned to less than 'n' then the return value will be padded with NAs.

## See Also

```
expand.nsnp, nearest.gene
```

## **Examples**

```
nearest.snp(1,159000000,n=10) # return ids
nearest.snp(1,159000000,n=10,build=37)
nearest.snp(1,159000000,n=10,build=36,ids=FALSE) # return positions

nearest.snp(1,159000000,n=10,build=37,ids=FALSE)
nearest.snp(6,25000000,n=10,build=37,ids=FALSE,side="left") # only SNPs to the left of the locus
nearest.snp(6,25000000,n=10,build=37,ids=FALSE,side="right") # only SNPs to the right of the locus
```

```
plot, GRanges, ANY-method
```

Plot method for GRanges objects

## Description

See plotRanges()

Plot method for RangedData objects

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#### Usage

```
## S4 method for signature 'GRanges,ANY'
plot(x, y, ...)
## S4 method for signature 'RangedData,ANY'
plot(x, y, ...)
```

## **Arguments**

x a GRanges or RangedData objecty not used for plotRanges

... further arguments, see plotRanges()

## See Also

plotRanges

plotGeneAnnot

Plot genes to annotate figures with genomic axes

## **Description**

Quite often it is helpful to visualize genomic locations in the context of Genes in the same region. This function makes it simple to overlay genes on plots where the x-axis is chromosomal location.

## Usage

```
plotGeneAnnot(chr = 1, scl = c("b", "kb", "mb", "gb"), y.ofs = 0,
  width = NA, txt = T, chr.pos.offset = 0, gs = NULL, build = NULL,
  dir = NULL, box.col = "green", txt.col = "black", join.col = "red",
   ...)
```

## Arguments

chr	chromosome number/name that the plot-range lies on	
scl	character, the scale that the x axis uses, ie, "b", "kb", "mb", or "gb", meaning base-pairs, kilobases, megabases or gigabase-pairs.	
y.ofs	numeric, y-axis-offset, depending on what units are on your y-axis, you may prefer to specify an offset so that the gene annotation is drawn at an appropriate level on the vertical axis, this value should be the centre of annotation	
width	depending on the range of your y-axis, you might want to expand or reduce the vertical width of the gene annotation (in normal graph units), default when width=NA is 10 percent of the y-axis size.	
txt	logical, TRUE to include the names of genes on top of their representation on the plot, or if FALSE, genes are drawn without labels.	
chr.pos.offset	if for some reason zero on the x-axis is not equal to 'zero' on the chromsome, then this offset can correct the offset. For instance if you were using a graph of the whole genome and you were plotting genes on chromosome 10, you would set this offset to the combined lengths of chromosomes 1-9 to get the start point	

in the correct place.

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gs	GRanges or RangedData object, this is annotation for the location of genes.
	This will be retrieved using get.gene.annot() if 'gs' is NULL. THere may be
	several reasons for passing an object directly to 'gs'; firstly speed, if making
	many calls then you won't need to load the annotation every time; secondly, if
	you want to use an alternative annotation you can create your own so long as
	it is a GRanges/RangedData object and contains a column called 'gene' (which
	doesn't strictly have to contain gene labels, it could be any feature you require,
	eg., transcript names, etc).
build	string, currently 'hg18' or 'hg19' to specify which annotation version to use.
	Default is build-36/hg-18. Will also accept integers 36,37 as alternative argu-
	ments.
dir	character, location to store file with the gene annotation. If NULL then getOp-
	tion("save.annot.in.current")>=1 will result in this file being stored in the current
	directory, or if <=0, then this file will not be stored.
box.col	genes are drawn as boxes, this sets the colour of the boxes
txt.col	this sets the colour of the label text (Gene names)
join.col	for exons, or multipart genes, joins are made between the sections with a central
-	line, this sets the colour of that line.
	further arguments to 'rect', the graphics function used to plot the 'genes'.
	- · · · · · · · · · · · · · · · · · · ·

#### Value

Returns a data.frame, GRanges or RangedData object, depending on input parameters. Contained will be HGNC gene labels, chromosome and start and end positions, other information depends on specific parameters documented above

# Examples

```
# EXAMPLE PLOT OF SOME SIMULATED SNPS on chr21-p11.1 #
# do we need to require(GenomicRanges)? #
setwd(tempdir())
loc <- c(9.9,10.2)
Band(chr=21,pos=loc*10^6)
rr <- in.window(rranges(50000),chr=21,pos=loc,unit="mb") # make some random MHC ranges
# create some SNPs and plot
rr3 <- rr; end(rr3) <- start(rr3)
rownames(rr3) <- paste0("rs",sample(10^6,nrow(rr3)))
plotRanges(rr3,col="blue",scl="mb",xlim=loc,xlab="Chr21 position (Mb)",ylab="")
# NOW add UCSC hg18 GENE annotation to the plot #
plotGeneAnnot(chr=21,pos=c(9.95,10.1),scl="mb",y.ofs=1,build=36)</pre>
```

plotRanges

Plot the locations specified in a GRanges or RangedData object

### **Description**

GRanges and RangedData objects are used in bioconductor to store genomic locations and ranges, such as transcripts, genes, CNVs and SNPs. This function allows simple plotting of this data directly from the ranged object. SNPs will be plotted as dots and ranges as lines. Either can be plotted using vertical bars at the start/end of each range. There are options for labelling and other graphical parameters. This package also creates a generic 'plot' method for GRanges and RangedData that calls this function.

plotRanges

## Usage

```
plotRanges(ranged, labels = NULL, do.labs = T, skip.plot.new = F,
  lty = "solid", alt.y = NULL, v.lines = FALSE, ylim = NULL,
  xlim = NULL, scl = c("b", "Kb", "Mb", "Gb"), col = NULL, srt = 0,
  pos = 4, pch = 1, lwd = 1, cex = 1, ...)
```

# **Arguments**

ranged	GRanges or RangedData object with genomic ranges. Should only contain one chromosome, but if not, the first will be used	
labels	by default labels for each range are taken from the rownames of 'ranged', but if you want to use another column in the ranged object, specify the column name or number to use to label these ranges on the plot. Or else input a character vector the same length as ranged for custom labels.	
do.labs	logical, whether or not to display these labels	
skip.plot.new	logical, whether to append to an existing plot (TRUE), or start a new plot (FALSE -> default)	
lty	line type to use, see '?lines()' - not used for SNP data when v.lines=FALSE	
alt.y	alternative y-axis values (other than the default ordering from the input) Th can be a vector of length 1 or length(ranged), or else a column name in range to take the values from	
v.lines	TRUE will plot the ranges as pairs of vertical lines, occupying the full vertical extent of the plot, whereas FALSE will plot the ranges as individual horizontal lines	
ylim	numeric, length 2, the y-axis limits for the plot, same a 'ylim' for 'plot()	
xlim	numeric, length 2, the x-axis limits for the plot, same a 'xlim' for 'plot(), This shouldn't usually be needed as the automatic x-limits should work well, however is here in case fine tuning is required.	
scl	character, the scale that the x axis uses, ie, 'b', 'kb', 'mb', or 'gb', meaning basepairs, kilobases, megabases or gigabase-pairs.	
col	character, colour, same as 'col' argument for plot(), etc.	
srt	integer, text rotation in degrees (see par) for labels	
pos	integer, values of '1', '2', '3' and '4', respectively indicate positions below, to the left of, above and to the right of the specified coordinates. See 'pos' in graphics:text()	
pch	point type, see '?points()' - not used for ranged data	
lwd	line width, see '?lines()' - not used for SNP data when v.lines=FALSE	
cex	font/symbol size, see '?plot()' - passed to plot, points if using SNP data	
	further arguments to 'plot', so long as skip.plot.new==FALSE.	

## Value

Plots the ranges specified in 'ranged' to the current plot, or to a new plot

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#### **Examples**

Pos

Find the chromosome position for SNP ids, gene name or band

## Description

Allows retrieval of the the chromosome position associated with a SNP-id, HGNC gene label, kary-otype band, or vector of such ids. For SNPs the ids can be either chip ids, or rs-ids, but must be contained in the current annotation. Default behaviour is to assume 'id' are SNP ids, but if none are found in the SNP annotation, the id's will be passed to functions Pos.gene() and Pos.band() to see whether a result is found. This latter step will only happen if no SNP ids are retreived in the first instance, and if snps.only=TRUE, then genes and bands will not be searched and NA's returned. If you are repeatedly searching for positions for genes/bands, using the dedicated Pos.gene() and Pos.band() functions would be slightly faster than relying on the fallback behaviour of the Pos() function. Note that the position for genes and bands are not a single point, so the result will be a range with start and end, see 'values' below. See documentation for these functions for more information.

#### Usage

```
Pos(ids, dir = NULL, snps.only = FALSE)
```

## **Arguments**

ids	character, a vector of rs-ids or chip-ids representing SNPs in the current Chip-Info annotation, or gene ids, or karyotype bands. Can also be a SnpMatrix object.
dir	character, only relevant when gene or band ids are entered, in this case 'dir' is the location to download gene and cytoband information; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.
snps.only	logical, if TRUE, only search SNP ids, ignore the possibility of genes/cytobands.

66 Pos.band

#### Value

When ids are SNP ids, returns a numeric vector of positions for each id, with NA values where no result was found. When ids are genes or karyotype bands, will return a data.frame with columns 'chr' [chromosome], 'start' [starting position of feature], 'end' [end position of feature], and the band without the chromosome prefix, if ids are bands. Note that this function cannot retrieve multiple ranges for a single gene (e.g, OR2A1 in build 38), which means you'd need to use Pos.gene(). The coordinates used will be of version getOption(ucsc="hg18"), or ucsc(chip.support()), which should be equivalent.

### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

Chr

### **Examples**

```
setwd(tempdir())
Pos(c("rs689","rs9467354","rs61733845"))
Pos("CTLA4") # returns a range
Pos("13q21.31") # returns a range
Pos(c("CTLA4","PTPN22"),snps.only=TRUE) # fails as these are genes
Pos(c("rs689","PTPN22","13q21.31")) # mixed input, will default to SNPs, as at least 1 was found
```

Pos.band

Find the chromosome, start and end position for cytoband names

## **Description**

Allows retrieval of the chromosome position of a karyotype/cytoband label, or vector of such labels. Note that the position returned for bands is not a single point as for SNPs, so the result will be a chromosome, then a position range with start and end, and lastly the band without the chromosome prefix

### Usage

```
Pos.band(bands, build = NULL, dir = NULL, bioC = FALSE)
```

## **Arguments**

bands character, a vector of cytoband labels, chromosome[p/q]xx.xx; e.g, 13q21.31,

Yq11.221, 6p23, etc

build character, "hg18" or "hg19" (or 36/37) to show which reference to retrieve. The

default when build is NULL is to use the build from the current ChipInfo anno-

tation

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dir character, 'dir' is the location to download cyto annotation information; if left

as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent

lookups, or deleted after use.

bioC logical, if true then return position information as a GRanges object, else a

data.frame

#### Value

Returns a data.frame with columns 'chr' [chromosome], 'start' [starting position of the gene], 'end' [end position of the gene] and 'band' [band without the chromosome prefix], or if bioC=TRUE, then returns a GRanges object with equivalent information. If returning a data.frame, then it will be in the same order as 'bands'. If bioC=TRUE, then the result will be in genome order, regardless of the order of 'bands'.

#### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

```
Chr, Pos, Pos. gene, Band, Band. gene, Band. pos, Gene. pos
```

### **Examples**

```
setwd(tempdir())
Pos.band("1p13.2")
Pos.band("Yq11.221",build=36)
Pos.band("Yq11.221",build=37)
Pos.band(c("13q21.31","1p13.2","2q33.2","6p23"),bioC=TRUE)
```

Pos.gene

Find the chromosome, start and end position for gene names

### **Description**

Allows retrieval of the chromosome position associated with a HGNC gene label, or vector of such labels. Note that the position returned for genes is not a single point as for SNPs, so the result will be a chromosome, then a position range with start and end.

```
Pos.gene(genes, build = NULL, dir = NULL, bioC = FALSE, band = FALSE,
  one.to.one = TRUE, remap.extra = FALSE, discard.extra = TRUE,
  warnings = TRUE)
```

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# Arguments

genes	character, a vector of gene ids
build	character, "hg18" or "hg19" (or $36/37$ ) to show which reference to retrieve. The default when build is NULL is to use the build from the current ChipInfo annotation
dir	character, 'dir' is the location to download gene annotation information to; if left as NULL, depending on the value of getOption("save.annot.in.current"), the annotation will either be saved in the working directory to speed-up subsequent lookups, or deleted after use.
bioC	logical, if true then return position information as a GRanges object, else a data.frame
band	logical, whether to include band/stripe in returned object
one.to.one	logical, some genes have split ranges, TRUE merges these to give only 1 range per gene, NB: this is the default behaviour when using the more general Pos() function
remap.extra	logical, if TRUE genes with chromosome annotation 'c6_cox' and 'c6_QBL' will be mapped to chromosome 6, and 'NT_xxxx' chromosome labels will all be mapped to 'Z_NT', etc
discard.extra	logical, if TRUE then any gene hit with chromosome not in 1:22, X, Y, XY, MT, will be discarded.
warnings	logical, whether to show warnings when some/all ids are not matched to the reference

#### Value

Returns a data.frame with columns 'chr' [chromosome], 'start' [starting position of the gene], 'end' [end position of the gene], or if bioC=TRUE, then returns a GRanges object with equivalent information, and if band=TRUE, then an extra column is added with band information If returning a data.frame, then it will be in the same order as 'genes'. If bioC=TRUE, then the result will be in genome order, regardless of the order of 'genes'.

### Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

### See Also

Chr, Pos, Pos. band, Band, Band. gene, Band. pos, Gene. pos

```
setwd(tempdir())
Pos.gene(c("CTLA4","PTPN22"))
Pos.gene("MYC",build=36)
Pos.gene("MYC",build=37)
Pos.gene(c("CTLA4","PTPN22"),bioC=TRUE,band=TRUE)
Pos.gene(c("CTLA4","PTPN22"),one.to.one=TRUE,build=38) # OR2A1 is split over two ranges
Pos.gene(c("CTLA4","OR2A1"),one.to.one=FALSE,build=38)
Pos.gene("RNU2-1",one.to.one=FALSE,bioC=TRUE,build=38) # RNU2-1 is split over multiple ranges
```

QCcode 69

QCcode

Access quality control pass or fail codes for ChipInfo

#### **Description**

Returns the pass or fail codes for each SNP of the chip object, e.g, 0,1,..,n etc Only if these are added manually, or else all will be 'pass' (=0)

QCcode<-: Allows user to set the pass or fail codes for each SNP of the chip object, e.g, 0,1,...,n etc. 0 is always pass, >0 is always fail, but each integer can be used to represent a different failure type, or for simplicity, stick to 0 and 1, ie, just pass and fail.

QCpass: Returns the subset of the ChipInfo object for which SNPs pass quality control, according to the QCcodes() slot == 0.

QCfail: Returns the subset of the ChipInfo object for which SNPs fail quality control, according to the QCcodes() slot > 0.

## Usage

```
QCcode(x)
## S4 method for signature 'ChipInfo'
QCcode(x)

QCcode(x) <- value
## S4 replacement method for signature 'ChipInfo'
QCcode(x) <- value

QCpass(x)

QCfail(x, type = NA)
## S4 method for signature 'ChipInfo'
QCpass(x)

## S4 method for signature 'ChipInfo'
QCpass(x)

## S4 method for signature 'ChipInfo'
QCfail(x, type = NA)</pre>
```

## Arguments

x a ChipInfo object

value new pass/fail codes, e.g, 0,1,...,n

type integer between 1 and 100, failure type (user can assign own coding scheme)

### Value

integer vector of pass/fail codes

QCcode<-: updates the object specified with new pass/fail codes for the 'QCcode' slot

QCpass: ChipInfo object for which SNPs pass quality control QCfail: ChipInfo object for which SNPs fail quality control

70 ranged.to.data.frame

## **Description**

Convert a RangedData/GRanges object to a data.frame with columns chr, start and end. Default is to only translate the chromosome and position information, which is faster. Using 'include.cols'=TRUE allows all the columns from 'ranged' to be taken across to the resulting data.frame.

## Usage

```
ranged.to.data.frame(ranged, include.cols = FALSE, use.names = TRUE)
```

#### **Arguments**

ranged A RangedData or GRanges object

include.cols logical, whether to also bring across non-positional columns to the resulting

data.frame

use.names logical, whether to keep the rownames from the original object for the output.

Only has an effect when include.cols=FALSE, otherwise original rownames are

always kept.

### Value

A data frame with columns chr, start and end, and depending on chosen parameters, the same rownames as the input, and optionally the same additional columns.

#### See Also

```
df.to.ranged, df.to.GRanges
```

```
rd <- rranges(9,GRanges=FALSE, fakeids=TRUE)
rd[["fakecol"]] <- sample(nrow(rd))
rd[["rs.id"]] <- paste0("rs",sample(10000,9))
ranged.to.data.frame(rd)
ranged.to.data.frame(rd,,FALSE)
ranged.to.data.frame(rd,TRUE) # keep all the columns
df.to.GRanges(ranged.to.data.frame(rd,TRUE)) # inverse returns original</pre>
```

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ranged.to.txt

Convert GRanges/RangedData to chr:pos1-pos2 vector

## **Description**

Takes a RangedData or GRanged object from some annotation lookup functions and converts to standard text positions, such as what you might see on the UCSC genome browser, such as chr1:10,000,234-11,000,567 for a range, or chrX:234,432 for a SNP. Useful for printing messages, concatenating positions to a single vector, or creating queries for databastes.

## Usage

```
ranged.to.txt(ranges)
```

### **Arguments**

ranges

A RangedData or GRanges object

## Value

a text vector of the same length as 'ranges' with notation as described above representing each position in the 'ranges' object

## See Also

```
convert.textpos.to.data
```

## **Examples**

```
ranged.to.txt(rranges())
```

rangeSnp

Find closest SNPs to the starts and ends of ranges

## **Description**

For given genome ranges (GRanges/RangedData) will try to find the closest snps to the starts and ends of the ranges.

```
rangeSnp(ranged = NULL, snp.info = NULL, chr = NULL, pos = NULL,
nearest = T)
```

72 recomWindow

### **Arguments**

ranged	A GRanges or RangedData object specifying the range(s) you wish to find SNPs near the starts/ends of. Alternatively leave this parameter as NULL and specify ranges using chr, pos
snp.info	ChipInfo/GRanges/Ranged data object describing the SNPs relevant to your query, e.g, SNPs on the chip you are using. If left NULL, the SNP set used will be that retrieved by chip.support() which will depend on your options() settings, see ?chip.support for more info
chr	optional alternative to 'ranged' input, use in conjunction with 'pos' to specify the ranges to find the SNPs near the starts/ends of.
pos	matrix with 2 columns for start, end positions, or a single column if all ranges are SNPs. An optional alternative to 'ranged' input, use in conjunction with 'chr' to specify the ranges to find the SNPs near the starts/ends of.
nearest	will preferably find an exact match but if nearest=TRUE, will fall-back on nearest match, even if slightly outside the range.

#### Value

a list of SNP-ids (rownames of 'snp.info') fulfilling the criteria, the output will be a matrix which should have the same number of rows as the number of ranges entered.

# **Examples**

```
rangeSnp(chr=c(1:3),pos=cbind(c(100000,200000,300000),c(30000000,40000000,10000000)))\\ rangeSnp(rranges())
```

recomWindow	Extend an interval or SNP by distance in centimorgans (recombination distance)

## Description

It is straightforward to extend a genomic interval or position by a number of basepairs, or a percentage, but extending by recombination units of centimorgans is more involved, requiring annotation lookup. This function streamlines this process. This function makes use of recombination rate hapmap reference files to calculate recombination distances for genome locations, in centimorgans. For a given position (or vector), a window can be returned of a given extension on either side of the position, for instance, 1 centimorgan to the left, and to the right of a SNP, giving a 2 centimorgan range as a result. Warning - this function only uses build hg18/36, so please convert to build 36 coordinates before using this function.

```
recomWindow(ranges = NULL, chr = NA, start = NA, end = start,
  window = 0.1, bp.ext = 0, rec.map = NULL, info = TRUE)
```

## **Arguments**

ranges	optional GRanges or RangedData object describing positions for which we want to generate windows, removing the need to enter chr, start and end
chr	character, an optional vector of chromosomes to combine with 'start' and 'end' to describe positions for which to generate recombination windows
start	integer, an vector of start points for chromosome ranges
end	integer, an vector of end points for chromosome ranges
window	numeric, number of centimorgans to extend the window either side of the range or location (can be a fraction)
bp.ext	numeric, optional number of base-pairs to extend the window by in addition to the centimorgan extension
rec.map	recombination map object (list of 22 data.frames) generated using 'get.recombination.map()'; if you are performing many of these operations, loading this object into your workspace and passing it on to this function will save loading it each time, and provide a speed advantage. Only use an object generated by get.recombination.map(), as otherwise the results will almost certainly be meaningless.
info	logical, whether to display the derived window size and number of hapmap SNPs within the window for each window derived

## Author(s)

Chris Wallace and Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

#### See Also

```
get.recombination.map, get.nearby.snp.lists, expand.nsnp
```

## Examples

```
# not run, as initial download of the recombination map takes nearly a minute #
recomWindow(chr=11, start=10000000, end=10000000, window=1, bp.ext=10000)
rd <- RangedData(ranges=IRanges(start=c(1.5,10.1)*10^7, end=c(1.55,10.1)*10^7), space=c(2,10))
rd # show original data
recomWindow(rd) # now extended by the interval
recomWindow(as(rd, "GRanges"), info=FALSE) # also works for GRanges</pre>
```

rownames, ChipInfo-method

rownames method for ChipInfo objects

## Description

rownames: Returns the row names.

dim: Returns the dimension

length: Returns the number of rows show: Displays a preview of the object

print: See 'show' as the behaviour is very similar and ... are just arguments of 'show'. The key difference with 'print' instead of 'show' is that by default the parameter 'up.to' is set to 50, so that any ChipInfo object (or subset) of less than or equal to 50 rows will be displayed in its entirety, rather than just the top/bottom 5 rows.

## Usage

```
## S4 method for signature 'ChipInfo'
rownames(x)

## S4 method for signature 'ChipInfo'
dim(x)

## S4 method for signature 'ChipInfo'
length(x)

## S4 method for signature 'ChipInfo'
show(object)

## S4 method for signature 'ChipInfo'
print(x, ...)
```

## **Arguments**

x a ChipInfo object object a ChipInfo object

... further arguments to showChipInfo()

## Value

rownames: Character vector of row names (SNP IDs).

dim: same as length

length: integer, number of rows, same as inherited nrow()

show: Displays a preview of the object

print: Prints the object to terminal using 'showChipInfo()'.

rownames, GRanges-method

rownames method for GRanges objects

#### **Description**

rownames: Returns the row names.

## Usage

```
## S4 method for signature 'GRanges'
rownames(x)
```

rownames<-methods 75

## **Arguments**

x a GRanges object

#### Value

rownames: Character vector of row names (e.g, SNP IDs).

rownames<--methods ~~ *Methods for Function* rownames<- ~~

## **Description**

```
~~ Methods for function rownames<- ~~
```

#### Methods

```
signature(x = "GRanges")
```

rranges

Simulate a GRanges or RangedData object

## **Description**

For testing purposes, this function will generate a S4 ranged object based on the human genome. The default is to produce ranges selected from chromosomes, with probability of a position in each chromosome equal to the length of that chromosome versus the whole genome. The maximum position allocated within each chromosome will be within the length bounds of that chromosome. You can specify SNPs (ie., start =end), but the default is for random ranges. You can alter the UCSC build to base the chromosome lengths on, and you can specify whether chromosomes should appear as chr1,chr2,... versus 1,2,..

## Usage

```
rranges(n = 10, SNP = FALSE, chr.range = 1:26, chr.pref = FALSE,
  order = TRUE, equal.prob = FALSE, GRanges = TRUE, build = NULL,
  fakeids = FALSE)
```

# Arguments

n	integer, number of rows to simulate
SNP	logical, whether to simulate SNPs (width 1, when SNPs=TRUE) or just ranges (when SNP=FALSE)
chr.range	integer vector of values from 1 to 26, to specify which chromosomes to include in the simulated object. 23-26 are X,Y,XY,MT respectively.
chr.pref	logical, if TRUE chromosomes will be coded as $chr1, chr2,, versus 1, 2,$ when $chr.pref=FALSE$
order	logical, if TRUE the object returned will be in genomic order, otherwise the order will be randomized

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equal.prob logical, when FALSE (default), random positions will be selected on chromosomes chosen randomly according to the their length (i.e, assuming every point on the genome has equal probability of being chosen. If equal.prob=TRUE, then chromosomes will be selected with equal probability, so you could expect just as many MT (mitochondrial) entries as Chr1 entries. logical, if TRUE the returned object will be GRanges format, or if FALSE, then **GRanges** RangedData format build character, to specify the UCSC version to use, which has a small effect on the

chromosome lengths. Use either "hg18" or "hg19". Will also accept build num-

ber, e.g, 36 or 37.

fakeids logical, whether to add rownames with random IDs (TRUE) or leave rownames

blank (FALSE). If SNP=TRUE, then ids will be fake rs-ids.

#### Value

returns a ranged object (GRanges or RangedData) containing data for 'n' simulated genomic ranges, such as SNPs or CNVs across chromosomes in 'chr.range', using UCSC 'build'.

## **Examples**

```
rranges()
rr <- rranges(SNP=TRUE,chr.pref=TRUE,fakeids=TRUE)</pre>
width(rr) # note all have width 1
tt <- table(chrm(rranges(1000)))</pre>
print(tt/sum(tt)) # shows frequencies at which the chr's were sampled
tt <- table(chrm(rranges(1000,equal.prob=TRUE)))</pre>
print(tt/sum(tt)) # shows frequencies at which the chr's were sampled
```

rs.id

Access rs-ids for ChipInfo

## **Description**

Returns the rs-ids for the chip object, e.g, "rs689", etc Only if these are annotated internally, or else a vector of NAs

# Usage

```
rs.id(x, b = TRUE)
## S4 method for signature 'ChipInfo'
rs.id(x, b = TRUE)
```

## **Arguments**

a ChipInfo object Х

logical, whether to show 'b' suffixes on rs.ids which are created in the backb ground to allow duplicate ids to be uniquely represented for lookup and reference purposes.

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#### Value

rs-ids: character vector of IDs (or NAs)

rs.to.id

Convert from dbSNP rs-ids to chip ID labels

#### **Description**

Most SNPs will have an 'rs-id' from dbSNP/HapMap, and these are often the standard for reporting or annotation lookup. These can differ from the IDs used on the chip. This functions looks at the current snp support (ChipInfo object) and looks up chip IDs based on rs-ids.

# Usage

```
rs.to.id(rs.ids, manifest = FALSE, multi.list = TRUE, force.flat = TRUE)
```

## Arguments

rs.ids	character, meant to be a list of rs-ids, but if chip-ids are present they will not be
	altered.

manifest logical, if TRUE return the ids as specified by the manifest/official set, or as

stored in the column 'chip.id'. If FALSE return the IDs as stored in the rownames of the ChipInfo object, which can differ when the chip.id is an illegal

format for an R row/column name.

multi.list logical, some rs-ids could map to multiple chip ids. It is recommended that if

that is the case then a letter should be appended to duplicate rs-ids to make them unique in the ChipInfo object, e.g, rs1234, rs1234b, rs1234c, etc. If multi.list is TRUE, then the id list will be returned as a list, and any time an rs-id is entered

without a letter suffix, all possible corresponding chip ids will be listed.

force.flat logical, if 'multi.list' is true, then some rs-ids might map to more than one SNP.

If force.flat is TRUE, then multiple SNP listings will be concatenated with a comma. If FALSE, then a list will be returned, with multiple entries where

applicable.

## Value

A character vector of SNP chip-ids, where the input was rs-ids, chip-ids or a mixture, any text other than this will result in NA values being returned in the character vector output. Or, if multi-list is true, then returns a list instead, which takes more than 1 value where there are multiple chip-ids with the same rs-id; if there are no such rs-id duplicates the result will still be a list. Currently rs-ids are always from build37.

## Author(s)

Nicholas Cooper < nick.cooper@cimr.cam.ac.uk>

## See Also

```
id.to.rs, GENE.to.ENS, ENS.to.GENE
```

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#### **Examples**

select.autosomes

Select ranges only within the 22 autosomes in a ranged data object

## **Description**

Select only data from autosomes from a GRanges/RangedData object. Will exclude X,Y, mitochondrial chromosome rows, and can automatically detect whether chromosomes are coded as 'chr1' or just '1', etc.

## Usage

```
select.autosomes(ranges, deselect = FALSE)
```

## **Arguments**

ranges A RangedData or GRanges object

deselect logical, if TRUE, then will select non-autosomes

#### Value

an object of the same format as the input (ranges), except with non-autosomal ranges removed.

## **Examples**

```
rand.ranges <- rranges(chr.range=20:26)
rand.ranges # should include some non-autosomes
select.autosomes(rand.ranges) # only autosomes remain</pre>
```

set.chr.to.char

Change the chromosome labels in a RangedData or GRanges object to string codes

## **Description**

Change the chromosome labels in a RangedData or GRanges object to string codes

## Usage

```
set.chr.to.char(ranged, do.x.y = T, keep = T)
```

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#### **Arguments**

ranged	A GRanges or RangedData object
do.x.y	logical, if TRUE then the usual numbers allocated to chromosomes, X,Y,XY,
	MT will be allocated as 23,24,25,26 respectively. If false, these will just have

'chr' appended as a prefix

keep logical, whether to keep additional metadata columns in the new object

#### Value

returns the 'ranged' object, but wherever a chromosome number was previously, a character label, e.g, 'chr1', or 'X', will returned to replace the number, e.g, 1 or 23 respectively. If table.out is TRUE will return a list where the first element is the resulting object, and the second element is a table showing which numbers were converted to what label This table can then be used for future conversions via the parameter 'table.in' to ensure consistency of coding.

#### See Also

```
set.chr.to.numeric
```

#### **Examples**

```
x <- rranges()
x
x <- set.chr.to.numeric(x) # make entirely numeric
x <- rranges(chr.range=20:26)
# next two will give warning about X, Y, etc
set.chr.to.char(x) # 23 = chrX, etc
set.chr.to.char(x,do.x.y=FALSE) # 23=chr23, etc</pre>
```

set.chr.to.numeric

Change the chromosome labels in a RangedData or GRanges object to numbers

#### **Description**

Change the chromosome labels in a RangedData or GRanges object to numbers

## Usage

```
set.chr.to.numeric(ranged, keep = T, table.in = NULL, table.out = FALSE)
```

## Arguments

ranged A GRanges or RangedData object

keep logical, whether to keep additional metadata columns in the new object

table.in matrix/data.frame object, usually a result of a prior run of set.chr.to.numeric(table.out=TRUE),
 which shows for each label (column 1), what chromosome number should correspond. A way of ensuring consistent coding in different sets.

table.out logical, if FALSE, the output will just be the object with updated chromosome

labels. If TRUE, then the output will be a list, where the first element is the updated object and the second object is a table describing the coding scheme

used to convert from labels to numeric indices.

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#### Value

returns the 'ranged' object, but wherever a chromosome label was previously a character label, e.g, 'chr1', or 'X', will return as a number, e.g, 1 or 23 respectively. If table.out is TRUE will return a list where the first element is the resulting object, and the second element is a table showing which labels were converted to what number. This table can then be used for future conversions via the parameter 'table.in' to ensure consistency of coding.

#### See Also

```
set.chr.to.char
```

#### **Examples**

```
char <- rranges(chr.pref=TRUE)
char
set.chr.to.numeric(char)
# behaviour with X, Y, etc
char <- rranges(chr.range=c(20:26))
#' char
set.chr.to.numeric(char)
tab <- set.chr.to.numeric(char,table.out=TRUE)[[2]]
tab # codes used in conversion #
char <- rranges(chr.range=c(20:26))
set.chr.to.numeric(char, table.in=tab) # code using codes from 'tab'</pre>
```

showChipInfo

Display a ChipInfo object

#### **Description**

Returns a preview of a ChipInfo object to the console. This is similar to a GRanges preview, but the seqlevels are hidden, the UCSC build and chip name are displayed, start and end are merged to the virtual label 'position' (as it's assume we are dealing with SNPs, not ranges), the strand by default is hidden, and the integer codes for pass/fail in QCcodes() are displayed as 'pass' or 'fail', even though this is not how they are represented internally. This is called by the default 'show' method for ChipInfo objects.

#### Usage

```
showChipInfo(x, margin = "", with.classinfo = FALSE,
   print.seqlengths = FALSE, ...)
```

#### **Arguments**

```
x a ChipInfo object
margin margin for display, usually ""
with.classinfo logical, whether to display class information
print.seqlengths
```

logical, whether to display sequence lengths below the main output listing (e.g, chromsomes). Usually tidier when this is FALSE.

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. . .

hidden arguments including: 'head.tail'; number of SNPs to display at start/end (only the head and tail are shown as these objects are generally very large with >100K SNPs); 'up.to'; only SNPs at the start and end are generally displayed, however this parameter specifies that when there are <= 'up.to' SNPs, then all SNPs will be displayed; 'show.strand'; logical, by default the strand is hidden, particularly given that the strand can vary between different datasets of the same chip. Setting to TRUE will display the strand.

#### Value

print compact preview of the object to the standard output (terminal)

#### See Also

ChipInfo

snps.in.range

Retrieve SNP ids or positions in specified range

## **Description**

This function will always use the build in getOption('ucsc'), so use options() if it needs to change.

#### Usage

```
snps.in.range(chr, start = NA, end = start, ids = TRUE)
```

## Arguments

chr	integer, chromosome, should be a number from 1 to 25, where 23,24,25 are X,Y,MT Alternatively chr can be a RangedData or GRanges object in which case SNP lists will be returned in a list for each row of the ranges object.
start	integer, genomic position to define the start of the range to look for SNPs, should be between 1 and the length of the chromosome 'chr'
end	integer, genomic position to define the end of the range to look for SNPs, should be between 1 and the length of the chromosome 'chr', and >= start
ids	logical, if TRUE will return snp ids (chip ids, for rs-ids, use id.to.rs on the output), or if FALSE will return the chromosome positions of the SNPs.

## Value

Set of SNP ids (when ids=TRUE), or otherwise genomic positions within chromosome 'chr', that fall within the genomic range described by the chr, start, and end parameters. Alternatively, if chr is a RangedData or GRanges object then multiple SNP lists will be returned in a list for each row of the ranges object.

## **Examples**

```
snps.in.range(1,9000000,10000000)
snps.in.range(10,19000000,20000000,ids=TRUE)
snps.in.range(10,19000000,20000000,ids=FALSE) # return positions instead of rs-ids
```

startSnp

startSnp	Find closest SNPs to the starts of ranges	

## **Description**

For given genome ranges (GRanges/RangedData) will try to find the closest snps to the starts of the ranges.

## Usage

```
startSnp(ranged = NULL, snp.info = NULL, chr = NULL, pos = NULL,
    start = T, end = F, nearest = T)
```

## **Arguments**

ranged	A GRanges or RangedData object specifying the range(s) you wish to find SNPs near the starts of. Alternatively leave this parameter as NULL and specify ranges using chr, pos
snp.info	ChipInfo/GRanges/Ranged data object describing the SNPs relevant to your query, e.g, SNPs on the chip you are using. If left NULL, the SNP set used will be that retrieved by chip.support() which will depend on your options() settings, see ?chip.support for more info
chr	optional alternative to 'ranged' input, use in conjunction with 'pos' to specify the ranges to find the SNPs near the starts of.
pos	matrix with 2 columns for start, end positions, or a single column if all ranges are SNPs. An optional alternative to 'ranged' input, use in conjunction with 'chr' to specify the ranges to find the SNPs near the starts of.
start	logical whether to return the SNP nearest the range starts
end	logical whether to return the SNP nearest the range ends
nearest	will preferably find an exact match but if nearest=TRUE, will fall-back on nearest match, even if slightly outside the range.

## Value

a list of SNP-ids (rownames of 'snp.info') fulfilling the criteria, the output will be a vector which will have the same length as the input. Unless start=TRUE and end=TRUE, then will return a matrix which should have the same number of rows as the number of ranges entered. Note that endSnp() is equivalent to using this function when end=TRUE and start=FALSE, and rangeSnp() is the same as setting start=TRUE and end=TRUE.

# **Examples**

```
startSnp(chr=c(1:3),pos=cbind(c(100000,200000,300000),c(30000000,4000000,10000000)))\\ startSnp(rranges())
```

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```
toGenomeOrder-methods ~~ Methods for Function toGenomeOrder ~~
```

## Description

```
~~ Methods for function toGenomeOrder ~~
```

#### Methods

```
signature(ds = "RangedData")
```

ucsc

Retrieve the UCSC build for a ChipInfo object

# Description

Returns the UCSC build of the chip object, e.g, 'hg18', 'hg19', or 'hg38'

## Usage

```
ucsc(x)
## S4 method for signature 'ChipInfo'
ucsc(x)
```

## Arguments

Х

a ChipInfo object

#### Value

```
character, 'hg18', 'hg19', or 'hg38'
```

```
[[,ChipInfo,ANY,ANY-method
```

Subset ChipInfo by chromosome

## **Description**

Returns the subset of the ChipInfo object for which SNPs are on the chromosome specified, by either number or character.

## Usage

```
## S4 method for signature 'ChipInfo,ANY,ANY' x[[i, j, ...]]
```

# Arguments

Χ	a ChipInfo object
i	a chromosome number or letter, i.e, one of $seqlevels(x)$
j	always leave missing, not applicable for this method.
	further arguments - again there should not be any

# Value

ChipInfo object for the subset of SNPs on chromosome i

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