# Package 'gt.db'

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Title GT.DB: Genotype Data Management and Analysis

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_	<b>on</b> Framework for storing and manipulating genotype data, phenotype data, and association ly results
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hwe.test		20
ibd.dataset	:	21
ibd.plot		23
ibd.summary		24
ibs.gt.data		25
if.na		26
jt.test		27
keep.attr		 28
ld.gt.data		29
ld.plot		20 30
Id.prune		31
ls.assay		31 32
ls.assay.group		32 33
		33 34
ls.assay.position		
ls.dataset		35
ls.mapping		36
ls.platform		37
ls.prcomp		38
ls.project		39
ls.sample		40
ls.subject		41
ls.test		41
mask.gt.data		42
mask.str		43
match.gt.data		44
mk.assay	•	45
mk.assay.data		46
mk.assay.group	•	47
mk.assay.position	•	48
mk.attr	•	49
mk.dataset	:	50
mk.mapping		51
mk.platform		52
mk.project		52
mk.sample		53
mk.sample.attr		54
mk.subject		55
mk.subject.attr		56
na.if		57
nsubstr		57
nw.align		58
nw.orient.assay		59
orient.gt.data		60
panel.cluster		61
panel.qqpval		62
panel.qqthin		62
prcomp.gt.data		63
progress.bar		64
qqprcomp		65
qqpval		66
qqthin		60 67
hexToRaw		о <i>т</i> 68
IIIA I UNAW		υo

adjust.gt.calls 3

	reshape.gt.data	69
	revcomp	70
	score.and.store	70
	score.chisq.2x2	71
	score.chisq	72
	score.fisher	73
	score.glm	74
	score.glm.general	75
	score.glm.groups	76
	score.gt.data	77
	score.jt	<b>7</b> 9
	score.kruskal	80
	score.lm	81
	score.lm.general	82
	score.lm.groups	83
	score.prcomp	84
	score.trend	85
	set.hidden	86
	snp.loadings	87
	sql.query	88
	store.prcomp	89
	store.sample.data	90
	store.subject.data	91
	store.test.scores	92
	summary.gt.data	93
	unpack.gt.matrix	94
	use.gt.db	95
Index		96

# Description

Interactively assign genotype calls by inspection of a plot of reference versus alternate intensities.

# Usage

```
adjust.gt.calls(data, ..., radius=6)
```

# Arguments

```
a data frame returned by reshape.gt.data.
... additional arguments passed to gt.cluster.plot.
radius the radius around the mouse pointer selected by clicking, in points.
```

4 apply.gt.dataset

#### **Details**

The intensity data is first plotted, then the user is iteratively prompted for a genotype value (in a/h/b/n notation) in the console window. Once a value has been entered, the plot window becomes active and the mouse pointer can be used to select points whose genotypes should be changed to that value. Each left click causes a zone around the mouse pointer position to be updated. The process is terminated by right clicking and selecting 'Stop' from the menu. The user is then prompted for another genotype value in the console window. To finish here, enter q.

#### Value

A data frame like data but with updated genotype calls. If a column called original unmodified genotype does not already exist, then it will be created and populated with the original unmodified genotypes.

#### See Also

```
fetch.gt.data, reshape.gt.data, gt.cluster.plot.
```

#### **Examples**

```
## Not run:
gt <- fetch.gt.data('Demo_2',raw.data=TRUE)
d <- reshape.gt.data(gt[32,], na.codes='n')
adjust.gt.calls(d, scales=list(log=TRUE))
## End(Not run)</pre>
```

```
apply.gt.dataset Apply a Function to a Genotype Dataset
```

#### **Description**

Apply a function to a genotype dataset, processed in manageable chunks, and aggregate the results.

#### **Usage**

```
apply.gt.dataset(gt.dataset, part.fn, aggr.fn, ..., GT.DATA='gt.data')
```

#### **Arguments**

```
gt.dataset a dataset description from gt.dataset.

part.fn a function to apply to each part of the dataset.

aggr.fn an aggregator function to accumulate results.

... additional arguments passed to part.fn.

GT.DATA the name to use for passing the genotype argument to part.fn.
```

#### **Details**

Based on the specification in gt.dataset, the dataset is loaded in chunks defined by parts binsz, and gt.filter. part.fn is applied iteratively to each chunk. The genotype data is passed as the GT.DATA argument to part.fn. The aggregator function aggr.fn should accept two arguments: an accumulated result, initially set to NULL; and a result for a single chunk.

apply.loadings 5

#### Value

Depends on form of part.fn and aggr.fn.

#### See Also

```
fetch.gt.data,gt.dataset.
```

## **Examples**

apply.loadings

Apply PCA SNP Loadings to a New Genotype Set

#### **Description**

Takes a table of SNP loadings from a principal components analysis, and applies them to another set of genotypes, yielding a new set of sample loadings.

#### Usage

```
apply.loadings(x, data)
```

## Arguments

```
x SNP loadings from snp.loadings.
data either a data frame of genotype data from fetch.gt.data, or a dataset description from gt.dataset.
```

#### **Details**

If the SNP loadings were computed on the same platform as the new dataset, then corresponding assays are identified by name. Across platforms, assays are matched up by genomic position.

# Value

A data frame of sample loadings, with one column per principal component, with samples ordered as they are in the supplied genotype strings.

```
prcomp, snp.loadings.
```

6 as.mask

## **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_1')
# compute PCA for unrelated individuals
p1 <- prcomp(gt.dataset('Demo_1'), is.na(pt$father))
# now compute sample loadings for all individuals
s <- snp.loadings(p1)
p2 <- apply.loadings(s, gt.dataset('Demo_1'))</pre>
```

as.mask

Convert To/From Character Masks

# Description

Convert between logical vectors, and strings of T/F characters.

#### Usage

```
as.mask(x)
un.mask(s)
```

## **Arguments**

```
x a logical vector.
```

s a string of T/F characters.

## Value

For as.mask, a string of T/F characters with one character per element of x.

For un.mask, a logical vector with one element per character in s

#### See Also

```
mask.str.
```

```
as.mask(c(TRUE,FALSE,TRUE,FALSE,TRUE))
un.mask('TFFTTF')
```

big.loadings 7

la d' au - 1 a a al d'ac aca	Identify I am a Comple on CND I and diver from a DCA Analysis
big.loadings	Identify Large Sample or SNP Loadings from a PCA Analysis

# **Description**

Takes either a PCA result, or SNP loadings from PCA, and returns a data frame describing large loadings for each principal component.

### Usage

```
big.loadings(x, sigma=6)
```

# **Arguments**

X	either a PCA result structure, or SNP loadings.
sigma	number of standard deviations to qualify as "big".

#### Value

A data frame with sample or assay identifiers, and the following additional columns:

```
name the principal component with high loading.

value the loading for this sample and component.

zscore the standardized loading for this sample.

var the fraction of variance in this component accounted for by this sample.
```

#### See Also

```
prcomp.gt.data, prcomp.gt.dataset, snp.loadings.
```

```
gt.demo.check()
g <- fetch.gt.data('Demo_1')
pc <- prcomp(subset(g, (ploidy=='A')))
big.loadings(pc,5)
sl <- snp.loadings(pc, g)
big.loadings(sl,5)</pre>
```

8 demo\_01

ch.table

Character Based Contingency Table

#### **Description**

Build contingency tables of character counts.

#### Usage

```
ch.table(s1, s2, chars)
```

#### **Arguments**

a vector of character strings.

an optional vector of character strings, with lengths matching strings in \$1.

chars a vector of individual characters to tabulate.

#### Value

If s2 is missing, a matrix of character counts with one row per element of s1 and one column per element of chars.

If s2 is present, a three dimensional array where the first dimension indicates a position within s1 and s2 (with recycling if necessary, and the second and third dimensions form contingency tables of corresponding characters in these strings.

# **Examples**

```
s1 <- c('AACAGCTACAGT','TTGTCGATGTCA')
s2 <- 'AACAGCTACAGT'
ch.table(s1, chars=c('A','C','G','T'))
x <- ch.table(s1, s2, chars=c('A','C','G','T'))
x[1,,]
x[2,,]</pre>
```

demo\_01

Genotype Data for 5000 SNPs on 270 HapMap Samples

# **Description**

Genotype data for 5000 SNPs distributed uniformly across the genome, for 270 samples from the International HapMap Project. This is a subset of data generated by Perlegen for the GAIN collaboration, using a set of SNP arrays designed to tag LD bins across the genome.

#### Usage

```
data(demo_01)
```

#### Source

Perlegen Sciences, Inc., publicly released data.

demo\_02

#### References

The GAIN Collaborative Research Group (2007) New models of collaboration in genome-wide association studies: the Genetic Association Information Network. *Nat. Genet.* **39**: 1045-1051.

The Database of Genotypes and Phenotypes. http://www.ncbi.nlm.nih.gov/sites/entrez?db=gap.

The International HapMap Project. http://www.hapmap.org.

demo\_02

Genotype Data for 371 Chr21 SNPs on 275 HapMap Samples

## **Description**

Genotype data for 371 SNPs distributed across a 300kb segment of human chromosome 21, for 275 samples from the International HapMap Project. The data was generated for the Phase II HapMap by Perlegen Sciences, using a custom array set developed for that project.

# Usage

```
data(demo_02)
```

#### **Source**

Perlegen Sciences, Inc., publicly released data.

#### References

The International HapMap Consortium (2007). A second generation human haplotype map of over 3.1 million SNPs. *Nature* **449**: 851-861.

The International HapMap Project. http://www.hapmap.org.

fetch.gt.data

Load Genotype Data for a Genotyping Dataset

## **Description**

Returns a data frame of assay information and genotypes for the specified dataset.

# Usage

10 fetch.gt.data

#### **Arguments**

dataset.name the unique identifier for the dataset.

mapping.name an identifier for the assay mapping to use.

assay.name a vector of assay names.

dbsnp.rsid a vector of integer dbSNP rsID values.

part a vector of subsets 1..parts.

parts the number of subsets to split the dataset into.

by specifies how to construct subsets. See details.

binsz specifies how to construct subsets. See details.

where additional SQL WHERE clauses to limit the data returned.
show.ids logical: indicates whether to include values of database keys.

genotype logical: indicates if genotypes should be fetched.

gscore logical: indicates if quality scores should be fetched.

raw.data logical: indicates if underlying raw data should be fetched, if available.

#### **Details**

If an assay mapping is not explicitly specified, and there is just one visible mapping for this dataset's platform, that one will be used by default.

The assay.name, dbsnp.rsid, and part arguments are mutually exclusive ways of specifying a set of SNPs for which data should be retrieved.

If part and parts are specified, then the dataset is divided into parts roughly similarly sized subsets, and part specifies which subset to retrieve. The chunking can be done either by primary key (by='assay.id') or by position (by='position'). To preserve some locality, assay subsets are interleaved such that trunc(by/binsz)%parts==(part-1).

#### Value

A data frame with one row per SNP. If any flags have been defined for this dataset, each flag is expanded to a logical column.

assay.data.id

the unique integer identifier for this entry in the ASSAY\_DATA table.

assay.id if show.ids is set: the unique key for this assay.

assay.name the name of this assay.

alleles a slash-separated list of alleles for this assay.

scaffold an identifier for the scaffold to which this assay has been mapped.

position the 1-based genomic position in the specified scaffold. strand the strand (+/-) to which the assay was mapped.

ploidy one of "A" (autosomal), "X" (X linked), "Y" (Y linked), or "M" (mitochon-

drial).

dbsnp.rsid the dbSNP rsID to which this assay is mapped.
dbsnp.orient the relative orientation of the dbSNP refsnp cluster.

genotype a string of a/h/b/n/x genotypes.
qscore a string of packed quality scores.

raw data a string containing packed raw data (such as intensities or read counts).

fetch.prcomp 11

#### See Also

```
summary.gt.data, unpack.gt.matrix.
```

# **Examples**

```
gt.demo.check()
gt <- fetch.gt.data('Demo_1')
str(gt)
nrow(fetch.gt.data('Demo_1', part=1, parts=4))
nrow(fetch.gt.data('Demo_1', part=1, parts=100))
summary.gt.data(gt[1:10,])</pre>
```

fetch.prcomp

Load Principal Components Results for a Genotyping Dataset

## **Description**

Load principal components analysis results for a Genotyping Dataset.

#### Usage

```
fetch.prcomp(dataset.name, prcomp.name, nc)
```

## **Arguments**

```
dataset.name the unique identifier for the dataset.

prcomp.name the unique identifier for the analysis.

nc the number of components to retrieve.
```

# **Details**

If prcomp.name is not present, then if there is just one visible analysis for this dataset, that one will be loaded.

# Value

The same sort of structure as returned by prcomp.gt.data.

#### See Also

```
store.prcomp, prcomp.gt.data, prcomp.gt.dataset.
```

```
## Not run:
pc <- prcomp(gt.dataset('Demo_1', gt.filter=(ploidy=='A')))
store.prcomp(pc, 'demo_pc_1', 'Demo PCA results')
fetch.prcomp('Demo', 'demo_pc_1', nc=4)
## End(Not run)</pre>
```

12 fetch.pt.data

fetch.pt.data

Load Phenotype Data for a Genotype Dataset

## **Description**

Returns a data frame of merged sample and subject phenotypes for the specified genotype dataset, mapped to the appropriate R datatypes.

#### Usage

```
fetch.pt.data(dataset.name, cols, pca=FALSE, show.all=FALSE)
```

# **Arguments**

dataset.name the short unique identifier for the dataset.

cols a vector of either sample or subject attributes to be included.

pca a logical indicating whether the current principal components analysis results

should be included, or a list of arguments to be passed to fetch.prcomp to

specify what to load.

show.all logical: indicates if hidden attributes should be included in the output.

#### Value

A data frame with one row per sample, and one column per attribute defined for this dataset, combining sample data from fetch.sample.data and subject data from fetch.subject.data. The column datatypes are determined using the information in the SAMPLE\_ATTR and SUBJECT\_ATTR tables. The row order conforms to the order of samples in the genotype data, and row names are set to the sample names. The result also includes the predefined sample attributes: subject name, gender, and dataset position.

#### See Also

```
fetch.sample.data.fetch.subject.data.fetch.prcomp.
```

```
gt.demo.check()
head(fetch.pt.data('Demo_1'))
head(fetch.pt.data('Demo_1', cols='plate'))
```

fetch.sample.data 13

```
fetch.sample.data Load Sample Data for a Genotype Dataset
```

### **Description**

Returns a data frame of sample phenotypes for the specified dataset, mapped to the appropriate R datatypes.

# Usage

```
fetch.sample.data(dataset.name, cols, show.all=FALSE)
```

#### **Arguments**

>

```
dataset.name the short unique identifier for the dataset.

cols a vector of column names to be included.
```

show.all logical: indicates if hidden attributes should be included in the output.

#### Value

A data frame with one row per sample, and one column per attribute defined for this dataset. The column datatypes are determined using the information in the SAMPLE\_ATTR table. The row order is arbitrary, but row names are set to the sample names. The result also includes the predefined sample attributes: subject name, gender, and dataset position.

#### See Also

```
mk.sample, ls.sample, fetch.pt.data, store.sample.data.
```

# Examples

```
gt.demo.check()
head(fetch.sample.data('Demo_1'))
```

```
fetch.subject.data Load Subject Data for a Genotyping Project
```

#### **Description**

Returns a data frame of subject phenotypes for the specified project, mapped to the appropriate R datatypes.

# Usage

```
fetch.subject.data(project.name, cols, show.all=FALSE)
```

14 fetch.test.scores

#### **Arguments**

project.name the short unique identifier for the project.

cols a vector of column names to be included.

show.all logical: indicates if hidden attributes should be included in the output.

#### Value

A data frame with one row per subject, and one column per attribute defined for this project. The column datatypes are determined using the information in the SUBJECT\_ATTR table. The row order is arbitrary, but row names are set to the sample names.

## See Also

```
mk.subject, ls.subject, store.subject.data.
```

# **Examples**

```
gt.demo.check()
head(fetch.subject.data('Demo'))
```

fetch.test.scores Load Association Test Results for a Genotyping Dataset

## **Description**

Load association test results for a Genotyping Dataset.

#### Usage

```
fetch.test.scores(dataset.name, test.name, term, max.pvalue)
```

## **Arguments**

dataset.name the unique identifier for a dataset.

test.name the unique identifier for an analysis.

term if present, for result sets with multiple results per assay, specifies which set of

results to fetch.

max.pvalue if present, specifies an upper bound for P values to be fetched.

#### Value

The same sort of structure as returned by score.gt.data.

```
store.test.scores, score.gt.data
```

gplot 15

#### **Examples**

```
## Not run:
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
x <- score.gt.data(status~genotype, pt, gt)
store.test.scores(x, 'status_1', 'Test analysis')
y <- fetch.test.scores('Demo_2', 'status_1')
str(y)
## End(Not run)</pre>
```

gplot

Genome-Wide Level Plot

#### **Description**

Generates a level plot of genome-wide data organized by chromosome and position.

# Usage

```
gplot(formula, data, aggr.fn=max, rescale=FALSE, binsz=1e6,
    subset=TRUE, col.regions=rev(heat.colors(100)[10:100]),
    scales=list(x=list(at=seq(0,250,20), draw=TRUE)),
    shrink=list(x=1,y=0.75), colorkey=list(height=0.25),
    xlab='Position, Mb', ylab='Chromosome', ...)
```

# **Arguments**

```
formula
                 a one-sided formula (i.e. ~ x) describing the values to be plotted.
                 a data frame containing values required to evaluate formula, as well as chromosome
data
                 and position columns.
aggr.fn
                 an aggregating function to apply to data within each bin.
                 logical: indicates if results should be scaled based on the genome-wide average
rescale
                 bin value.
binsz
                 bin size in base pairs
subset
                 a logical or integer vector identifying rows of data to be included in the plot.
col.regions, scales, shrink, colorkey, xlab, ylab
                 see levelplot.
                 additional arguments passed to levelplot.
. . .
```

#### **Details**

The supplied formula is first evaluated across the input data, and then the aggregating function aggr.fn is applied to results in bins of genomic coordinates. Appropriate aggregating functions include max, min, sum, etc.

#### Value

```
A plot object of class "trellis".
```

16 gplot.prcomp

#### See Also

```
levelplot.
```

## **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_1')
gt <- fetch.gt.data('Demo_1')
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
r <- score.gt.data(status~genotype, pt, gt, score.chisq)
gplot(~-log10(pvalue), merge(r,gt))</pre>
```

gplot.prcomp

Genome-Wide Level Plots of SNP Loadings from PCA

## **Description**

Generates level plots of genome-wide SNP loadings from a principal components analysis.

# Usage

## **Arguments**

```
    a data structure returned by snp.loadings.
    a vector of component numbers to plot.
    an aggregating function to apply to data within each bin.
    logical: indicates if results should be scaled based on the genome-wide average bin value.
    xlab, ylab axis labels passed to gplot.
    additional arguments passed to gplot.
```

#### **Details**

The output of gplot.prcomp is a vertically stacked series of genome-wide plots of SNP loadings for a specified set of principal components. The default aggregating function results in a plot of variance binned by genomic interval.

#### Value

```
A plot object of class "trellis".
```

```
prcomp.gt.data, prcomp.gt.dataset, snp.loadings, gplot.
```

gt.cluster.plot 17

#### **Examples**

gt.cluster.plot

Plot Genotype Cluster Data

#### **Description**

Plot raw genotype cluster data (i.e. intensities), with predicted ellipsoid boundary regions.

## Usage

#### **Arguments**

```
an unpacked data frame of genotype information from reshape.gt.data.

x a data frame of genotypes from fetch.gt.data.

rescale logical: indicates if each data in each panel should be scaled to fill the frame.

bounds contours at which to draw ellipsoid boundaries.

min.points the minimum number of points for which to compute ellipsoids.

between, scales, xlab, ylab, par.settings, ...

arguments passed to xyplot.
```

#### Details

gt.cluster.plot is a wrapper around xyplot to facilitate plotting raw data underlying genotype calls. A custom panel function draws minimal-area ellipsoid contours to capture proportions of genotype cluster density given by bounds.

xyplot.gt.data is a method for packed genotype data.

#### Value

```
An object of class "trellis".
```

```
panel.cluster, xyplot, ellipsoidPoints, fetch.gt.data, reshape.gt.data,
adjust.gt.calls.
```

18 gt.dataset

#### **Examples**

```
gt.demo.check()
gt <- fetch.gt.data('Demo_2',raw.data=TRUE)
head(reshape.gt.data(gt))
d <- reshape.gt.data(gt[seq(32,232,40),], na.codes='n')
gt.cluster.plot(d)
gt.cluster.plot(d, scales=list(log=TRUE))
xyplot(gt[seq(32,232,40),])</pre>
```

gt.dataset

Genotype Dataset Specification

#### **Description**

Describes a genotype dataset in the database and how to process it.

#### Usage

#### **Arguments**

```
dataset.name the unique identifier for the dataset.

gt.filter an expression to use for subsetting the genotype data.

parts, by, binsz

specify how to construct chunks. See fetch.gt.data.

progress logical: indicates whether to display a progress bar when processing the dataset.
```

#### **Details**

The specified dataset is loaded in chunks defined by parts and binsz, and part.fn is applied iteratively to each chunk. The genotype data is passed as the gt.data argument to part.fn. The aggregator function aggr.fn should accept two arguments: an accumulated result, initially set to NULL; and a result for a single chunk.

# Value

A list of class gt.dataset, with elements representing all the arguments of gt.dataset.

#### See Also

```
fetch.gt.data, apply.gt.dataset.
```

gt.dist

gt.dist

Calculate Pairwise Genotype Distances

#### **Description**

Calculate genotype-based distances (broadly defined) between pairs of samples, using a choice of distance operators and aggregator functions.

# Usage

```
gt.dist(gt1, gt2=gt1, operation='==', aggregator='sum', na.value=NA)
```

#### **Arguments**

```
vectors of packed diploid genotypes as from fetch.gt.data, with 1:1 match-
ing assays.

operation one of 'n','==','!=','ibs==0','ibs==1', or'ibs'.

aggregator one of 'sum','min', or'max'.

na.value an integer value to use as the result of comparisons involving missing genotypes.
The default excludes these comparisons.
```

#### **Details**

For each genotype assay, an operation is performed on each pairwise combination of samples:

```
'n' 1 if both genotypes are present, else 0.
'==' 1 if genotypes are identical, else 0.
'!=' 1 if genotypes are not identical, else 0.
'ibs==0' 1 if genotypes are identical by state (IBS) for 0 alleles, else 0.
'ibs==1' 1 if genotypes are IBS for 1 allele, else 0.
'ibs==2' 1 if genotypes are IBS for 2 alleles, else 0.
'ibs' the number of alleles IBS, 0..2.
```

The matrices of results are then aggregated across genotype assays, using one of the available aggregation functions.

# Value

A matrix of integers with length (gt1) rows and length (gt2) columns, with elements giving the aggregated distance for the corresponding pair of samples.

#### See Also

```
match.gt.data, ibd.dataset.
```

```
gt <- fetch.gt.data('Demo_2')
gt <- substr(gt$genotype[1:20],1,10)
gt.dist(gt)
gt.dist(gt, operation='ibs', aggregator='min')</pre>
```

20 gt.split

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Convert between Packed Genotype Strings and Genotype Vectors

## **Description**

 ${\tt gt.split}$  takes a string of a/h/b/n genotypes and returns a vector with one genotype per element;  ${\tt gt.paste}$  reverses this conversion.

#### Usage

#### **Arguments**

S	a character string of packed a/h/b/n genotypes.
V	a vector of unpacked genotypes. See details.
convert	how genotypes should be represented.
alleles	a vector of length 2 specifying A and B alleles, for $\texttt{convert='char'}$ .
na.codes	a vector of missing genotype codes: see details.
strand	for convert='char', strand to report.
sep	for convert=' char', a string for separating alleles.

#### **Details**

If convert='score.a', then unpacked genotypes are coded as numeric scores, where "a"=2, "h"=1, and "b"=0. If convert='score.b', then unpacked genotypes are coded as numeric scores, where "a"=0, "h"=1, and "b"=2. if convert='char', then genotypes are coded as factors with levels formed from concatenated pairs of alleles. If convert='none', then genotypes are coded as factors with three levels, 'a', 'h', and 'b'.

Missing genotypes are by default coded as NA in the unpacked format. If na.codes is not empty, then the specified single character codes are instead reported as separate factor levels in the unpacked format.

#### Value

For  ${\tt gt.split}$ , a vector of genotypes coded as specified by convert. For  ${\tt gt.paste}$ , a packed genotype string.

```
fetch.gt.data,unpack.gt.matrix.
```

hapmap 21

#### **Examples**

hapmap

Data from the International HapMap Project

# **Description**

hapmap.subjects describes the sample plate and panel membership, and parent/child relationships, for the 270 individuals genotyped in the Phase I and Phase II HapMap Projects.

#### Usage

```
data(hapmap)
```

#### **Source**

The International HapMap Project

#### References

The International HapMap Project. http://www.hapmap.org.

hwe.test

Tests for Hardy Weinberg Equilibrium

## **Description**

Asymtotic and exact tests for Hardy Weinberg equilibrium conditional on observed marginal allele frequencies, for biallelic genotype data.

# Usage

22 ibd.dataset

#### **Arguments**

```
aa, ab, bb biallelic genotype counts (may be vectors).test the type of test to perform. See details.tail See details.
```

#### **Details**

Three tests are implemented: the likelihood ratio test, the traditional chi-squared test without continuity correction, and the conditional exact test. All are defined as in Weir (1996). All the tests are vectorized for efficiency.

The default is to perform a two-sided test. If tail='lower', then the result is the probability of seeing no more than the observed number of heterozygotes under the hypothesis of Hardy Weinberg equilibrium. If tail='upper', then the result is the probability of seeing at least as many as the observed number of heterozygotes.

#### Value

A vector of P values.

#### References

Weir, B.S. (1996) Genetic Data Analysis II. Sinauer, Sunderland, MA.

#### **Examples**

```
# Table 3.1 in Weir (1996)
d \leftarrow data.frame(aa=0:9, ab=seq(19,1,-2), bb=21:30)
d$p.exact <- with(d, hwe.test(aa,ab,bb,'exact'))</pre>
d$p.chisq <- with(d, hwe.test(aa,ab,bb,'chisq'))</pre>
d$chisq <- qchisq(d$p.chisq,df=1,lower.tail=FALSE)</pre>
d[order(d$p.chisq),]
qt.demo.check()
gt <- fetch.gt.data('Demo_1')</pre>
pt <- fetch.pt.data('Demo_1')</pre>
s <- summary.gt.data(gt, pt$plate=='J+C')</pre>
d <- with(s,data.frame(</pre>
    lratio=hwe.test(AA,AB,BB,'lratio'),
    chisq=hwe.test(AA, AB, BB, 'chisq'),
    exact=hwe.test(AA,AB,BB,'exact')
p < -1ist(limits=c(-10,0), at=c(-2,-4,-6,-8),
           labels=c('1E-2','1E-4','1E-6','1E-8'))
splom(\sim log(d, 10), pscales=list(p,p,p))
```

ibd.dataset

Calculate Identity by Descent for a Genotype Dataset

#### **Description**

Estimates identity by descent for all sample pairs in a genotype dataset, from observed identity by state information.

ibd.dataset 23

#### Usage

#### **Arguments**

```
dataset.name the identifier for the dataset to analyze.

binsz, part, parts
    passed to fetch.gt.data, to identify the regions to be analyzed.

maf.min minimum minor allele frequency to be included in the analysis.

hw.p.min minimum Hardy-Weinberg equilibrium P value for SNPs to be included in the analysis.

gt.rate.min minimum call rate for SNPs to be included.

... additional arguments for ibd.gt.data.
```

#### **Details**

Pairwise identity-by-descent (IBD) is estimated using data from a series of intervals of size binsz, equally spaced across the genome. The algorithm determines the minimum number of alleles identical by state (IBS) for SNPs within each bin, and averages these values across bins. The minimum IBS value gives an upper limit on IBD for that bin, and approaches IBD as the number of assayed SNPs increases.

Determining IBD for close relatives requires only a fraction of the available data from a whole genome scan. Very accurate estimates of IBD are not particularly useful, because of intrinsic variability in the recombination process. The default values for binsz, part, and parts should not need to be changed.

Bins with few SNPs may not be sufficiently informative for IBD, if there is a substantial probability of unrelated samples sharing the same haplotype by chance. Rare genotyping errors can also impact apparent IBD, since a single error within a bin will change the minimum IBS. The min.snps setting ensures reasonable informativeness, and max.snps, maf.min, hw.p.min, and gt.rate.min control the expected error rate.

The estimated genomic proportion with IBD=1 is biased upwards because a small proportion of bins are not sufficiently informative to distinguish IBD=0 from IBD=1. The estimate for IBD=2 seems to be less biased. The following table shows expected genomic IBD proportions for common familial relationships.

IBD=0	IBD=1	IBD=2	Relationship
1.00	0.00	0.00	unrelated
0.75	0.25	0.00	first cousins
0.50	0.50	0.00	half siblings
0.00	1.00	0.00	parent-child
0.25	0.50	0.25	full siblings
0.00	0.00	1.00	duplicate samples

It is important to note that the actual IBD proportions for most relative pairs (except for parent-child) show substantial natural variation, independent of the variation due to the estimation procedure.

24 ibd.plot

#### Value

A list with three elements. The first two elements are matrices, containing the estimated proportions of the genome with IBD=1 and IBD=2, for all pairs of samples in the dataset. The third element is an ordered list of sample names from the GTPUB dataset.

#### See Also

```
ibd.plot, ibd.summary, ibd.outliers.
```

#### **Examples**

```
gt.demo.check()
ibd <- ibd.dataset('Demo_1', min.snps=10, binsz=10e6, parts=1)
ibd.plot(ibd, jitter=0.005)</pre>
```

ibd.plot

Plot Identity by Descent Data

# **Description**

Plot identity by descent data for all sample pairs.

# Usage

#### **Arguments**

#### Details

Produces a two-panel plot, with the upper panel showing pairwise IBD=1 versus IBD=2 proportions, and the lower panel showing a histogram of the IBD=1 values. In the upper panel, outliers are colored by whether they are outliers on IBD=1, on IBD=2, or both. Outliers on IBD=1 are defined by a Bonferroni corrected P value of less than 0.05, assuming a normal distribution for unrelated individuals.

# Value

```
A plot object of class "trellis".
```

```
ibd.dataset, ibd.summary, ibd.outliers.
```

ibd.summary 25

#### **Examples**

```
gt.demo.check()
ibd <- ibd.dataset('Demo_1', min.snps=10, binsz=10e6, parts=1)
ibd.plot(ibd, jitter=0.005)</pre>
```

ibd.summary

Summarize Identity by Descent Analysis Results

#### **Description**

Summarize results from an identity-by-descent analysis.

#### Usage

```
ibd.summary(ibd, alpha=0.05)
ibd.outliers(ibd, alpha=0.05)
```

#### **Arguments**

ibd results from ibd.dataset.
alpha a P value threshold for identifying IBD=1 outliers.

#### **Details**

ibd.summary and ibd.outliers both identify sample pairs with elevated genome-wide IBD=1 or IBD=2 proportions. For IBD=1, a threshold is selected assuming a normal distribution for unrelated individuals, using the specified alpha threshold with a Bonferroni correction. For IBD=2, an (arbitrary) threshold of 0.05 is used.

Unrelated pairs should have neither elevated; parent-sibling pairs, cousins and half sibs should have only IBD=1 elevated; duplicates should have only IBD=2 elevated; siblings should have both elevated.

### Value

For ibd.summary, a table of counts of distinct sample pairs, indicating how many have neither IBD=1 or IBD=2 elevated; only elevated IBD=1; only elevated IBD=2; or both elevated.

For ibd.outliers, a data frame with 7 columns:

```
rds.id.1, sample.name.1
identifiers for sample 1.

rds.id.2, sample.name.2
identifiers for sample 2.

grp the type of outlier: one of 'IBD=1', 'IBD=2', 'Both'.
ibd.1 the inferred proportion of the genome with IBD=1.
ibd.2 the inferred proportion of the genome with IBD=2.
```

```
ibd.dataset, ibd.plot.
```

26 ibs.gt.data

#### **Examples**

```
gt.demo.check()
ibd <- ibd.dataset('Demo_1', min.snps=10, binsz=10e6, parts=1)
ibd.summary(ibd)
ibd.outliers(ibd)</pre>
```

ibs.gt.data

Calculate Pairwise Identity by State for Genotype Data

## **Description**

Calculates identity-by-state (IBS) for all sample pairs in a set of genotype data.

#### Usage

```
ibs(x, sample.mask=TRUE)
## S3 method for class 'gt.data':
ibs(x, sample.mask=TRUE)
## S3 method for class 'gt.dataset':
ibs(x, sample.mask=TRUE)
grr(x, sample.mask=TRUE)
```

#### **Arguments**

```
either a data frame of genotypes from fetch.gt.data, or a dataset description from gt.dataset.sample.maskan optional mask identifying a subset of samples to be included in the results.
```

# Details

ibs.gt.data and ibs.gt.dataset assemble counts of markers versus the number of alleles IBS. grr computes the mean and standard deviation of IBS, as described in Abecasis *et al.* (2001).

#### Value

For ibs.gt.data and ibs.gt.dataset, a list of three square matrices of counts of markers with IBS=0, IBS=1, and IBS=2, for each sample pair.

For grr, a list of two square matrices giving the mean and standard deviation of the number of alleles IBS, for each sample pair.

#### References

Abecasis, G.R., Cherny, Stacey, S.S., Cookson, W.O.C., and Cardon, L.R. (2001) GRR: graphical representation of relationship errors. *Bioinformatics* 17: 742-743.

```
ibd.summary, gt.dataset.
```

if.na 27

#### **Examples**

```
gt.demo.check()
ibs <- ibs(gt.dataset('Demo_1'))
str(ibs)
pt <- fetch.pt.data('Demo_1')
g <- grr(gt.dataset('Demo_1'), (pt$plate=='CEU'))
with(g, xyplot(sd[lower.tri(sd)]~mu[lower.tri(mu)]))</pre>
```

if.na

Conditional Element Selection for Missing Values

# Description

if.na returns a value with the same shape as val, populated from either yes or no depending on whether the element of is.na(val) is TRUE or FALSE.

# Usage

```
if.na(val, yes, no=val)
```

## **Arguments**

```
val an object to be tested for NA values.

yes return values for NA elements of val.

return values for non-NA elements of val.
```

#### **Details**

The result is equivalent to ifelse (is.na(val), yes, no).

#### Value

A vector of the same length as val, where missing values in are taken from yes, and non-missing values are taken from no (or val itself, if no is missing).

# References

Inspired by Oracle's nvl() and nvl2() functions.

## See Also

```
is.na, ifelse.
```

```
x <- c(1:3,NA,NA)
if.na(x, 4)
if.na(x, 0, x+1)</pre>
```

28 jt.test

jt.test	Jonckheere-Terpstra Nonparametric Test for Trend

# **Description**

Test for association between genotypes and a quantitative outcome, using the nonparametric Jonckheere-Terpstra test for ordered differences among genotype classes.

## Usage

#### **Arguments**

x vector of quantitative response values.

y group membership.

alternative alternative hypothesis to be tested.

asymp logical: use asymptotic formula for variance, or don't bother.

correct logical: apply continuity correction?

perm number of repetitions for a permutation test.

na.action what to do with missing data.

permgraph logical: draw a histogram of the permutations?

permreps logical: return the permutations?

### **Details**

The Jonckheere-Terpstra test computes Mann-Whitney rank sum statistics for ordered group labels, and does a two-sided test on the sum of those statistics. This tests for a monotonic trend in response as a function of group membership. The model formula should be of the form outcome~genotype without additional terms.

#### Value

A list of class htest, with the following components:

```
statistic the observed J-T statistic. alternative same as input.
```

method the string: "Jonckheere-Terpstra test".

data.name the names of the input data.

EH the expected test statistic based on sample size.

VH variance (adjusted for ties if necessary).

p.value asymptotic p-value.

#### References

```
http://tolstoy.newcastle.edu.au/R/help/06/06/30112.html.
```

keep.attr 29

#### See Also

```
kruskal.test, score.kruskal.
```

#### **Examples**

```
x \leftarrow rnorm(30) + c(rep(1,10), rep(2,20))

y \leftarrow c(rep(1,10), rep(2,10), rep(3,10))

jt.test(x, y)
```

keep.attr

Keep User Attributes

# **Description**

This function creates objects with the property that they more systematically preserve user attributes when they are indexed.

# Usage

```
keep.attr(.Data, ..., .Attr=NULL)
kept.attr(x)
## S3 method for class 'keep.attr':
x[...]
## S3 replacement method for class 'keep.attr':
x[...] <- value</pre>
```

# Arguments

```
.Data an R object for which attributes are to be kept.
... attributes in key=value form to be attached to the object.
.Attr additional attributes to attach, collected into a list.
x an object of class 'keep.attr'.
value a suitable replacement value.
```

# Value

For keep.attr, the original object with additional class 'keep.attr', with additional attributes attached. Indexing on this object will propagate all user specified attributes, so long as the indexing operation returns an object of the same class. Thus, for a data frame, attributes will be preserved for operations that return a new data frame.

For kept.attr, a list of all the user defined attributes associated with the object.

```
attributes.
```

30 ld.gt.data

#### **Examples**

```
d <- data.frame(a=c(1,2,3), b=c(4,5,6), c=c(7,8,9))
d <- structure(d, xyz='something')
e <- keep.attr(d, abc='more')
d[3] <- 5
e[3] <- 5
str(d)
str(d[1])
str(d[,-1])
str(d[,1])
str(e)
str(e[1])</pre>
```

ld.gt.data

Compute Pairwise Linkage Disequilibrium

#### **Description**

Takes genotype data as returned by fetch.gt.data and computes a matrix of pairwise linkage disequilibrium values.

## Usage

# Arguments

```
g1, g2 genotype data from fetch.gt.data.

outer logical: specifies whether result should be an outer product of g1 and g2.

measure the measure of LD to report. See details.

method the method to use for calculating LD.

max.it the maximum number of EM iterations to perform.

epsilon convergence criterion for the EM algorithm.
```

#### **Details**

LD is computed between unphased genotypes in g1 and g2 under an assumption of Hardy Weinberg equilibrium. Two algorithms are implemented: an iterative EM solution, and an exact solution.

All markers should have consistent ploidy status. The LD calculations account for haploid and diploid genotypes by using gender information at sex-linked loci.

With the iterative algorithm, a warning is printed if some pairwise LD calculations do not converge in the specified number of iterations. If measure=' failed', then the function returns an array of logical values indicating which cases failed to converge.

For the exact algorithm, the most-likely result is reported. With extreme deviations from HWE, it is possible for more than one solution to have the same likelihood, and in these cases, the one that is reported is essentially arbitrary.

Id.plot 31

#### Value

If outer is TRUE, then the result is a matrix of LD values with one row per element of g1 and one column per element of g2. If outer is FALSE, then the result is a vector of values obtained by comparing corresponding elements of g1 with g2 with recycling of the shorter argument. This will usually only make sense when the lengths of g1 and g2 are either equal or equal to 1.

The number of pairwise calculations that failed to converge is returned in an attribute with the name 'failed'.

#### References

Weir, B.S. (1996) Genetic Data Analysis II. Sinauer, Sunderland, MA.

Gaunt, T. R., Rodriguez, S., & Day, I. N. M. (2007) Cubic exact solutions for the estimation of pairwise haplotype frequencies: implications for linkage disequilibrium analyses and a web tool 'CubeX'. *BMC Bioinformatics* **8**: 428.

```
CubeX. http://www.oege.org/software/cubex/.
```

#### See Also

```
fetch.gt.data, ld.plot.
```

## **Examples**

```
gt.demo.check()
gt <- fetch.gt.data('Demo_2')[1:6,]
ld.gt.data(gt, measure='rsqr')
ld.gt.data(gt, measure='dprime')
ld.gt.data(gt, measure='pvalue')
ld.gt.data(gt[1:3,], gt[3:5,], outer=TRUE)
ld.gt.data(gt[1:3,], gt[3:5,], outer=FALSE)</pre>
```

ld.plot

Pairwise Linkage Disequilibrium Plot

# Description

Generates a level plot of the strength of pairwise linkage disequilibrium among a set of SNPs.

# Usage

#### **Arguments**

```
gt.data genotype data from fetch.gt.data.

col a color gradient.

measure the measure(s) of LD to be plotted. See details.

rotate logical: indicates if the plot should be rotated clockwise 45 degrees.
```

32 Id.prune

```
logical: indicates if the LD matrix should be displayed with equal SNP spacing,
or in genomic coordinates.

colorkey, scales
    passed to levelplot.
... additional arguments to pass to levelplot.
```

#### **Details**

SNPs are sorted by accession and contig position. Values of measure are as in ld.gt.data. If measure has two elements, then the first value is plotted in the upper left triangle and the second value is plotted in the lower right triangle.

#### Value

```
A plot object of class "trellis".
```

#### See Also

```
ld.gt.data levelplot.
```

#### **Examples**

ld.prune

Prune SNP List to Limit Linkage Disequilibrium

## **Description**

Takes genotype data as returned by fetch.gt.data and eliminates SNPs in high LD across a sliding window.

# Usage

```
ld.prune(gt.data, min.maf=0.01, max.rsqr=0.2, span=20, subsets=TRUE)
```

# **Arguments**

```
genotype data for a single contiguous sequence, from fetch.gt.data.

min.maf a minimum minor allele frequency for inclusion.

max.rsqr the maximum allowed R-squared value.

span the number of sequential SNPs to check for LD.

subsets an optional list of masks identifying groups of subjects within which LD should be checked.
```

Is.assay 33

#### **Details**

LD is computed over a sliding window and SNPs are iteratively eliminated to guarantee that each remaining SNP has the specified maximum R-squared value for at least span SNPs on either side, ordered by sequence position.

If multiple subsets are specified, then the inclusion criteria are enforced for ALL subsets.

#### Value

A new data frame based on gt.data, containing a subset of SNPs satisfying the LD limitations.

#### See Also

```
fetch.gt.data, ld.gt.data.
```

#### **Examples**

```
gt.demo.check()
gt <- fetch.gt.data('Demo_2')
p <- fetch.subject.data('Demo')
by.plate <- list(p$plate=='CEU', p$plate=='YRI', p$plate=='J+C')
x <- ld.prune(gt, subsets=by.plate)
nrow(gt)
nrow(x)</pre>
```

ls.assay

List Assay Definitions

## **Description**

List definitions of assays associated with a genotyping platform.

#### Usage

```
ls.assay(platform.name, assay.group='%', show.ids=FALSE)
```

# **Arguments**

#### Value

A data frame with one row per assay, and 4 or 6 columns:

```
assay.group.id

if show.ids is set: a unique integer key for the assay group associated with this assay.

assay.group the assay group associated with this assay.

assay.id if show.ids is set: the unique integer ID for this assay.
```

34 ls.assay.group

```
assay.name the assay name.

alleles a slash separated list of allele sequences.

probe.seq the genomic flanking sequence for the assay, with the variant position denoted by an underscore ('_').
```

#### See Also

```
ls.assay.group, ls.assay.position, mk.assay.
```

#### **Examples**

```
gt.demo.check()
head(ls.assay('Demo_Set_1'))
head(ls.mapping('Demo_Set_1', show.ids=TRUE))
```

ls.assay.group

List Assay Groups

#### **Description**

This returns a list of assay groups for the specified genotyping platform.

#### Usage

```
ls.assay.group(platform.name, show.ids=FALSE)
```

#### **Arguments**

#### Value

A data frame with one row per assay group, and 4 or 5 columns:

```
assay.group.id

if show.ids is set: a unique integer key for this assay group.

assay.group a short, unique identifier for the assay group.

description a free-text description of the assay group.

created.by the user name that created the assay group.

created.dt the creation date of the assay group.
```

#### See Also

```
mk.assay.group.
```

```
gt.demo.check()
ls.assay.group('Demo_Set_1')
ls.assay.group('Demo_Set_1', show.ids=TRUE)
```

Is.assay.position 35

```
ls.assay.position List Assay Positions
```

## **Description**

List mapped genomic positions of assays associated with a genotyping platform.

# Usage

```
ls.assay.position(platform.name, mapping.name, show.ids=FALSE)
```

#### **Arguments**

#### **Details**

If mapping.name is missing, it will default to the current (visible) mapping for the specified platform, if that is unique.

# Value

A data frame with one row per assay, and 7 or 8 columns:

```
assay.id
                 if show.ids is set: the unique integer ID for this assay.
assay.name
                 the assay name.
scaffold
                 a string identifying the sequence to which the assay is mapped.
position
                 a one-based position within the specified scaffold.
                 either "+", "-", or NA.
strand
                 describes the expected allele count: see mk.assay.position.
ploidy
dbsnp.rsid
                 the dbSNP refSNP cluster ID for this assay.
dbsnp.orient the orientation of the assay compared to the dbSNP cluster: either "+", "-", or
                 NA.
```

#### See Also

```
ls.assay.mk.assay.position.
```

```
gt.demo.check()
head(ls.assay.position('Demo_Set_1'))
```

36 Is.dataset

ls.dataset	List Genotype Datasets

#### **Description**

This returns a list of genotype datasets defined in the current database.

# Usage

#### Arguments

```
project.name an SQL LIKE expression for matching project names.

dataset.name an SQL LIKE expression for matching dataset names.

show.all logical: indicates if hidden datasets and members of hidden projects should be included in the output.

show.ids logical: indicates whether to include values of database keys.
```

#### Value

A data frame with one row per dataset, and 7 or 10 columns:

```
dataset.id
                  if show.ids is set: the unique integer key for this dataset.
dataset.name a short, unique identifier for the dataset.
project.id
                  if show.ids is set: the unique integer key for the project associated with this
project.name the project associated with this dataset.
                  if show.ids is set: the unique integer key for the platform associated with this
platform.id
                  dataset.
platform.name
                  the genotyping platform associated with this dataset.
                  a free-text description of the dataset.
description
is.hidden
                  logical: indicates if the project is hidden.
created.by
                  the user name that created the dataset.
                  the creation date of the dataset.
created.dt
```

# See Also

```
ls.project, ls.platform, mk.dataset.
```

```
gt.demo.check()
ls.dataset()
ls.dataset(show.ids=TRUE)
```

Is.mapping 37

ls.mapping	List Assay Mapping Sets	

# **Description**

This returns a list of mapping sets for the specified genotyping platform.

# Usage

```
ls.mapping(platform.name, show.all=FALSE, show.ids=FALSE)
```

# **Arguments**

# Value

A data frame with one row per mapping, and 4 or 5 columns:

```
mapping.id if show.ids is set: a unique integer key for this mapping.

mapping a short, unique identifier for the mapping.

description a free-text description of the mapping.

created.by the user name that created the mapping.

created.dt the creation date of the mapping.
```

# See Also

```
mk.mapping.
```

```
gt.demo.check()
ls.mapping('Demo_Set_1')
ls.mapping('Demo_Set_1', show.ids=TRUE)
```

38 Is.platform

ls.platform

List Genotyping Platforms

# Description

This returns a list of genotyping platforms defined in the current database.

# Usage

```
ls.platform(platform.name='%', show.ids=FALSE)
```

# **Arguments**

```
platform.name an SQL LIKE expression for matching platform names. show.ids logical: indicates whether to include values of database keys.
```

# Value

A data frame with one row per platform, and 6 or 7 columns:

# See Also

```
mk.platform.
```

```
gt.demo.check()
ls.platform()
ls.platform(show.ids=TRUE)
```

Is.prcomp 39

ls.prcomp	List Principal Components Result Sets	

# **Description**

This returns a list of principal components analysis result sets in the database for a specified genotype dataset.

# Usage

```
ls.prcomp(dataset.name, prcomp.name='%', show.all=FALSE, show.ids=FALSE)
```

# **Arguments**

dataset.name a genotype dataset name.

prcomp.name an SQL LIKE expression for matching principal components analysis set names.

show.all logical: indicates if hidden result sets should be included in the output.

show.ids logical: indicates whether to include values of database keys.

#### Value

A data frame with one row per result set, and 9 or 10 columns:

prcomp.id if show.ids is set: the unique integer key for this test set.

prcomp.name a short identifier for the result set, which is unique for the specified genotype

dataset.

description a free-text description of the result set.

the text of the call used to perform the analysis.

components the number of components retained in the database.

samples the number of samples included in the analysis.

assays the number of assays included in the analysis.

is.hidden logical: indicates if the result set is hidden.

created.by the user name that created the result set.

created.dt the creation date of the result set.

# See Also

```
fetch.prcomp, store.prcomp, rm.prcomp.
```

```
# FIXME
```

40 ls.project

ls.project List Genotyping Project
------------------------------------

# Description

This returns a list of genotyping projects defined in the current database.

# Usage

```
ls.project(project.name='%', show.all=FALSE, show.ids=FALSE)
```

# **Arguments**

```
project.name an SQL LIKE expression for matching project names.

show.all logical: indicates if hidden projects should be included in the output.

show.ids logical: indicates whether to include values of database keys.
```

#### Value

A data frame with one row per project, and 6 or 7 columns:

```
project.id if show.ids is set: a unique integer key for this project.

project.name a short, unique identifier for the project.

description a free-text description of the project.

datasets a count of the number of datasets under this project.

is.hidden logical: indicates if the project is hidden.

created.by the user name that created the project.

created.dt the creation date of the project.
```

# See Also

```
mk.project.
```

```
gt.demo.check()
ls.project()
ls.project(show.ids=TRUE)
```

Is.sample 41

ls.sample	List Samples in a	Genotype Dataset
is.sampie	Lisi sampies in a	Genorype Daiuser

# Description

This returns a description of all samples defined in the specified genotype dataset.

### Usage

```
ls.sample(dataset.name, show.ids=FALSE)
```

# **Arguments**

```
dataset.name the unique identifier for the dataset.

show.ids logical: indicates whether to include values of database keys.
```

#### Value

A data frame with one row per dataset, and 4 or 6 columns:

```
sample.id if show.ids is set: the unique integer key for this sample.

sample.name a short, unique identifier for the sample.

subject.id if show.ids is set: the unique integer key for the subject associated with this sample.

subject.name a short, unique identifier for the subject.

gender a factor indicating the gender of this sample.

position a 1-based integer indicating the position of this sample's genotyping data in the genotype arrays for this dataset.
```

# See Also

```
ls.dataset, mk.sample.
```

```
gt.demo.check()
head(ls.sample('Demo_1'))
head(ls.sample('Demo_1', show.ids=TRUE))
```

42 Is.test

ls.subject

List Subjects in a Genotyping Project

### **Description**

This returns a list of subjects defined in the specified genotyping project.

#### Usage

```
ls.subject(project.name, show.ids=FALSE)
```

# **Arguments**

```
project.name a unique project identifier.
show.ids logical: indicates whether to include values of database keys.
```

#### Value

A data frame with one row per dataset, and 1 or 2 columns:

```
subject.id if show.ids is set: the unique integer key for this subject. subject.name a short, unique identifier for the subject.
```

#### See Also

```
ls.project, mk.subject.
```

# **Examples**

```
gt.demo.check()
head(ls.subject('Demo'))
head(ls.subject('Demo', show.ids=TRUE))
```

ls.test

List Association Test Result Sets

# **Description**

This returns a list of association test result sets in the database for a specified genotype dataset.

# Usage

```
ls.test(dataset.name, test.name='%', show.all=FALSE, show.ids=FALSE)
```

# Arguments

```
\hbox{\tt dataset.name}\ a\ genotype\ dataset\ name.
```

```
test.name an SQL LIKE expression for matching test set names.
```

show.all logical: indicates if hidden result sets should be included in the output.

show.ids logical: indicates whether to include values of database keys.

mask.gt.data 43

#### Value

A data frame with one row per test set, and 8 or 9 columns:

test.id if show.ids is set: the unique integer key for this test set.

a short identifier for the test set, unique when combined with term for the test.name

specified genotype dataset.

description a free-text description of the test set.

the type of model fit: 'lm', 'glm', etc. fit

the model formula used for scoring each genotype assay. model

used for differentiating among multipe test result sets associated with a single term

analysis

logical: indicates if the test set is hidden. is.hidden the user name that created the test set. created.by

created.dt the creation date of the test set.

#### See Also

```
fetch.test.scores, store.test.scores, rm.test.
```

#### **Examples**

# FIXME

mask.gt.data Mask Sample Genotypes

# **Description**

Given a data frame of genotype information, this masks out a subset of samples based on a logical vector.

#### Usage

```
mask.gt.data(gt.data, sample.mask, repack=FALSE)
```

# **Arguments**

a data frame of genotype information. qt.data

sample.mask either a logical vector, or a string of T/F values, indicating which samples should

repack logical: indicates whether masked-out samples should be entirely removed from

the result, or left in place.

# **Details**

If repack is FALSE, then genotypes of masked-out samples are coded as missing. If repack is TRUE, then masked-out samples are deleted from the result.

44 mask.str

#### Value

An updated version of  $\operatorname{gt}$ . data with genotypes for masked-out samples either marked as missing or deleted.

#### See Also

```
fetch.gt.data, summary.gt.data.
```

#### **Examples**

```
gt.demo.check()
gt <- fetch.gt.data('Demo_2')
pt <- fetch.pt.data('Demo_2')
head(summary.gt.data(gt))
gm <- mask.gt.data(gt, pt$gender=='M')
gf <- mask.gt.data(gt, pt$gender=='F')
head(summary.gt.data(gm))
head(summary.gt.data(gf))</pre>
```

mask.str

Mask Character Strings

#### **Description**

Mask selected positions from elements of a character vector.

#### Usage

```
mask.str(str, mask, ch='x')
```

# **Arguments**

a character vector to be masked.

mask
either a logical vector or character mask, with a 1:1 correspondence to character positions in elements of str.

ch a character that will replace masked positions in str.

#### Value

A character vector with the same form as str, with positions specified as FALSE in mask either replaced by the first character in ch, or squeezed out entirely, if ch is an empty string.

### See Also

```
as.mask.
```

```
mask.str('12345678', 'TFTFTFTF', '_')
mask.str('12345678', 'TFTFTFTF', '')
```

match.gt.data 45

match.gt.data

Identify Equivalent Genotyping Assays

# **Description**

This identifies assays from two genotype datasets that correspond to the same polymorphisms, based either on position or dbSNP rsIDs.

# Usage

```
match.gt.data(gt.data.1, gt.data.2, by=c('position','dbsnp.rsid'))
```

# **Arguments**

```
gt.data.1, gt.data.2
data frames with assay information to be compared.
by the method to use to identify equivalent assays.
```

#### **Details**

This first identifies assays with corresponding positions (or dbSNP rsIDs) across the two datasets. For each matching pair, the reported genomic orientation and alleles are checked for compatibility. Putative matches are rejected if the alleles are inconsistent.

# Value

A data frame with four columns. The first two columns contain corresponding assay names from the two datasets. The remaining two columns are:

```
is.flipped logical: indicates that the assays are in opposing genomic orientations. is.swapped logical: indicates that the A/B alleles are interchanged between assays.
```

# See Also

```
fetch.gt.data, orient.gt.data.
```

```
gt.demo.check()
g1 <- fetch.gt.data('Demo_2')
g2 <- orient.gt.data(g1, flip=(g1$dbsnp.orient == '-'))
head(match.gt.data(g1,g2))</pre>
```

46 mk.assay

mk.assay

Create Genotyping Assay Definitions

# Description

mk.assay defines genotyping assays associated with an already defined genotyping platform.

# Usage

```
mk.assay(platform.name, data)
```

#### **Arguments**

# **Details**

A data frame of assay information should provide four columns:

```
assay.group an assay group defined with mk.assay.group.assay.name a unique name (within this platform) for the assay.alleles a slash separated list of valid alleles.
```

**probe.seq** the genomic flanking sequence for the assay, with the variant position denoted by an underscore ('\_').

# Value

If successful, the number of rows inserted into the assay table.

#### See Also

```
ls.assay,mk.assay.position.
```

```
## Not run:
data(demo_01)
head(assay.def.01)
mk.assay('Demo_Set_1', assay.def.01)
## End(Not run)
```

mk.assay.data 47

mk.assay.data

Create Genotyping Assay Data

# **Description**

mk.assay.data defines genotype data associated with already defined genotype assays.

# Usage

```
mk.assay.data(dataset.name, data)
```

### **Arguments**

```
dataset.name a short unique identifier for the genotype dataset.

data a data frame with one row per assay. See details.
```

#### **Details**

A data frame of genotype data can provide up to five columns of information:

```
assay.id the unique integer ID for this assay.
```

flags an integer value composed of single-bit flags.

genotype a string of a/h/b/n/x genotypes.

qscore a string of packed quality scores.

raw.data a string containing packed raw data (such as intensities or read counts).

An assay.name column can be supplied in place of assay.id.

# Value

If successful, the number of rows inserted into the assay data table.

# See Also

```
mk.assay.position, mk.assay, fetch.gt.data.
```

48 mk.assay.group

```
mk.assay.group
```

Create or Remove a Genotype Assay Group

# Description

These functions insert or remove entries from the genotyping assay group table.

### Usage

```
mk.assay.group(platform.name, assay.group, description)
rm.assay.group(platform.name, assay.group)
```

# **Arguments**

```
the unique platform identifier.

assay.group an identifier for the assay group.

description a free-text description of the assay group.
```

#### **Details**

An assay group and its constituent assays can only be removed if it is not in use. Any mappings or datasets that refer to these assays must be explicitly deleted first.

# Value

If successful, the number of rows inserted in or removed from the assay group table (i.e., typically, 1).

### See Also

```
ls.assay.group, ls.platform, mk.platform, mk.assay.
```

```
## Not run:
mk.platform('My_600K', 'My 600K Array Set')
mk.assay.group('My_600K', 'Des01', '300K Array 1 of 2')
mk.assay.group('My_600K', 'Des02', '300K Array 2 of 2')
rm.assay.group('My_600K', c('Des01','Des02'))
rm.platform('My_600K')
## End(Not run)
```

mk.assay.position 49

```
mk.assay.position Create Genotyping Assay Positions
```

# **Description**

mk.assay.position defines map positions associated with genotyping assays.

# Usage

```
mk.assay.position(platform.name, mapping.name, data)
```

## **Arguments**

#### **Details**

If mapping.name is missing, it will default to the current (visible) mapping for the specified platform, if that is unique. A data frame of assay positions can provide up to seven columns of information:

```
assay.name the assay identifier.
```

**scaffold** a string identifying the sequence to which the assay is mapped.

**position** a one-based position within the specified scaffold.

```
strand either "+", "-", or NA.
```

**ploidy** describes the expected allele count: it can take values "A" (autosomal), "M" (mitochondrial, or haploid), "X" (diploid in females, haploid in males), or "Y" (haploid in males, absent in females). Pseudoautosomal positions are coded as "A".

dbsnp.rsid the dbSNP refSNP cluster ID for this assay.

dbsnp.orient the orientation of the assay compared to the dbSNP cluster: either "+", "-", or NA.

#### Value

If successful, the number of rows inserted into the assay map position table.

#### See Also

```
ls.assay.position, mk.mapping, mk.assay.
```

50 mk.attr

mk.attr

Create, Remove, and List Attribute Definitions

#### **Description**

 ${\tt mk.attr}$  and  ${\tt rm.attr}$  create or remove attribute definitions associated with various types of genotype data objects, such as samples and subjects, and  ${\tt ls.attr}$  lists attribute definitions.

# Usage

# Arguments

target	the type of attribute to be created: i.e., $\prime$ subject $\prime$ or $\prime$ sample $\prime$ .
parent.name	the name of the parent container for the new attribute, i.e. a project or dataset name.
attr.name	a short identifier for the new attribute.
datatype	the datatype for attribute values: 'number', 'string', 'boolean', or 'factor'.
levels	for factors, a string describing the factor levels, formed by concatenating a comma-separated list of single quoted strings.
description	a free text description of the attribute.
is.hidden	logical: indicates if the attribute is hidden.
show.all	logical: indicates if hidden attributes should be included in the output.
show.ids	logical: indicates whether to include values of database keys.

# **Details**

These functions are usually not called directly; instead, there are helper functions for each type of attribute (i.e. mk.subject.attr, rm.subject.attr, ls.subject.attr that are more convenient.

Multiple attributes can be created or removed with single calls to mk.attr and rm.attr. The attr.name, datatype, levels, and description arguments can all be vectors and the usual recycling rules apply.

## Value

For mk.attr and rm.attr, if successful, the number of rows inserted or deleted from the target attribute table.

For ls.attr, a data frame with 7 columns:

```
attr.name the attribute name.

datatype one of 'number', 'string', 'boolean', or 'string'.

levels a single-quoted comma-separated list of factor levels.
```

mk.dataset 51

```
description a free text description.

is.hidden logical: indicates if the attribute is hidden.

created.by the user name that created the attribute.

created.dt the creation date of the attribute.
```

#### See Also

```
mk.subject.attr,mk.sample.attr.
```

# **Examples**

mk.dataset

Create or Remove Genotype Datasets

#### **Description**

These functions insert or delete entries from the genotype dataset table.

### Usage

#### **Arguments**

```
dataset.name a short unique identifier for the dataset.

project.name the project associated with this dataset.

platform.name
the genotyping platform for this dataset.

description a free-text description of the dataset.

is.hidden logical: indicates if the dataset is hidden.
```

# **Details**

While datasets are nested within projects, dataset names must be globally unique.

#### Value

If successful, the number of rows inserted in or deleted from the dataset table (i.e., 1).

52 mk.mapping

#### See Also

```
ls.dataset, set.hidden.
```

#### **Examples**

mk.mapping

Create or Remove a Mapping for a Genotyping Platform

# Description

These functions insert or remove entries from the platform mapping table.

# Usage

```
mk.mapping(platform.name, mapping.name, description, is.hidden=FALSE)
rm.mapping(platform.name, mapping.name)
```

### **Arguments**

```
the unique platform identifier.

mapping.name an identifier for the mapping.

description a free-text description of the mapping.

is.hidden logical: indicates if the mapping is hidden.
```

#### **Details**

Removing a mapping results in deletion of all the associated assay map positions.

# Value

If successful, the number of rows inserted in or removed from the mapping table (i.e., typically, 1).

# See Also

```
ls.mapping, mk.platform, mk.assay.position.
```

```
## Not run:
mk.platform('My_600K', 'My 600K Array Set')
mk.mapping('My_600K', 'b36_1', 'Build 36 Vender Annotations')
rm.mapping('My_600K', 'b36_1')
rm.platform('My_600K')
## End(Not run)
```

mk.platform 53

mk.platform

Create or Remove a Genotyping Platform

# **Description**

These functions insert or remove entries from the genotyping platform table.

# Usage

```
mk.platform(platform.name, description)
rm.platform(platform.name)
```

# **Arguments**

```
platform.name a short unique identifier for the platform. description a free-text description of the platform.
```

# Value

If successful, the number of rows inserted in or removed from the platform table (i.e., 1).

#### See Also

```
ls.platform, mk.assay.group.
```

# **Examples**

```
## Not run:
mk.platform('GT_800K', 'My 800K Genotyping Platform')
rm.platform('GT_800K')
## End(Not run)
```

mk.project

Create or Remove a Genotyping Project

### **Description**

These functions insert or remove entries from the project table.

# Usage

```
mk.project(project.name, description, is.hidden=FALSE)
rm.project(project.name)
```

# **Arguments**

```
project.name a short unique identifier for the project.
description a free-text description of the project.
is.hidden logical: indicates if the project is hidden.
```

54 mk.sample

#### Value

If successful, the number of rows inserted or removed from the project table (i.e., 1).

#### See Also

```
ls.project, set.hidden.
```

#### **Examples**

```
## Not run:
mk.project('Demo_9', 'Demo Genome-Wide Association Study')
rm.project('Demo_9')
## End(Not run)
```

mk.sample

Create or Remove Dataset Samples

#### **Description**

 ${\tt mk.sample}$  and  ${\tt rm.sample}$  insert or remove samples from a genotype dataset.

# Usage

```
mk.sample(dataset.name, data)
rm.sample(dataset.name, sample.name)
```

# **Arguments**

```
data a short unique identifier for the dataset.

data a data frame with one row per sample. See details.

sample.name a vector of sample identifiers.
```

#### **Details**

A data frame of sample information should have (at least) four columns: sample.name, subject.name, gender, and position. This information is important because it affects how various functions interpret genotyping data in this dataset. Sample gender is used to determine ploidy for sex linked assays, and the position is used to parse arrays of packed genotype data.

### Value

If successful, the number of rows inserted or deleted from the sample table.

#### See Also

```
ls.sample, mk.subject.
```

mk.sample.attr 55

### **Examples**

```
## Not run:
s <- data.frame(
    sample.name='NA_12345_1'
    subject.name='NA_12345',
    gender='M',
    position=1
)
mk.sample('Demo_1', s)
rm.sample('Demo_1, s$sample.name)
## End(Not run)</pre>
```

mk.sample.attr

Create, Remove, or List Sample Attribute Definitions

# Description

mk.sample.attr and rm.sample.attr create or remove sample attributes associated with a particular genotype dataset, and ls.sample.attr lists sample attributes for this dataset.

# Usage

```
mk.sample.attr(dataset.name, data, description=names(data))
rm.sample.attr(dataset.name, attr.name)
ls.sample.attr(dataset.name)
```

# **Arguments**

 $\hbox{\tt dataset.name} \ \ the \ unique \ identifier \ for \ a \ dataset.$ 

data a data frame with one or more columns to be used to model the new attributes.

See details.

description a vector of free-text descriptions with one element per column in data.

attr.name a vector of attribute names to be removed.

#### **Details**

These functions are wrappers around mk.attr, rm.attr, and ls.attr. A call to mk.sample.attr results in creation of a new attribute for each column in the data argument, based on the data type, factor levels, etc of that column.

#### Value

For mk.sample.attr and rm.sample.attr, the number of attribute definitions inserted or removed from the sample attribute table.

For ls.sample.attr, a data frame with 7 columns describing subject attributes defined for the specified project, as defined in ls.attr.

### See Also

```
mk.attr, rm.attr, ls.attr.
```

56 mk.subject

#### **Examples**

```
## Not run:
sd <- fetch.sample.data('Demo_1')
sd$stuff.1 <- rep(1:3, length.out=nrow(sd))
sd$stuff.2 <- factor(sd$stuff.1, levels=1:3, labels=c('a','b','c'))
sd$stuff.3 <- as.character(sd$stuff.2)
str(sd)
mk.sample.attr('Demo_1', sd[c('stuff.1','stuff.2','stuff.3')])
ls.sample.attr('Demo_1')
rm.sample.attr('Demo_1', c('stuff.1','stuff.2','stuff.3'))
## End(Not run)</pre>
```

mk.subject

Create or Remove Project Subjects

# **Description**

mk.subject and rm.subject insert or remove subjects from a genotyping project.

# Usage

```
mk.subject(project.name, data)
rm.subject(project.name, subject.name)
```

# **Arguments**

```
project.name a short unique identifier for the project.

data a data frame with a subject.name column.

subject.name a vector of subject identifiers.
```

# Value

If successful, the number of rows inserted or deleted from the subject table.

### See Also

```
ls.subject, mk.sample.
```

```
## Not run:
mk.subject('Demo_1', data.frame(subject.name='NA_12345'))
rm.subject('Demo_1', 'NA_12345')
## End(Not run)
```

mk.subject.attr 57

```
mk.subject.attr
```

Create, Remove, or List Subject Attribute Definitions

#### **Description**

mk.subject.attr and rm.subject.attr create or remove subject attributes associated with a particular genotyping project, and ls.subject.attr lists subject attributes for this project.

# Usage

```
mk.subject.attr(project.name, data, description=names(data))
rm.subject.attr(project.name, attr.name)
ls.subject.attr(project.name)
```

#### **Arguments**

```
\label{eq:project.name} \mbox{ the unique identifier for a project.}
```

data a data frame with one or more columns to be used to model the new attributes.

See details.

description a vector of free-text descriptions with one element per column in data.

attr.name a vector of attribute names to be removed.

#### **Details**

These functions are wrappers around mk.attr and rm.attr, and ls.attr. A call to mk.subject.attr results in creation of a new attribute for each column in the data argument, based on the data type, factor levels, etc of that column.

# Value

For mk.sample.attr and rm.sample.attr, the number of attribute definitions inserted or removed from the sample attribute table.

For ls.sample.attr, a data frame with 7 columns describing subject attributes defined for the specified project, as defined in ls.attr.

#### See Also

```
mk.attr,rm.attr,ls.attr.
```

```
## Not run:
sd <- fetch.subject.data('Demo')
sd$stuff.1 <- rep(1:3, length.out=nrow(sd))
sd$stuff.2 <- factor(sd$stuff.1, levels=1:3, labels=c('a','b','c'))
sd$stuff.3 <- as.character(sd$stuff.2)
str(sd)
mk.subject.attr('Demo', sd[c('stuff.1','stuff.2','stuff.3')])
ls.subject.attr('Demo')
rm.subject.attr('Demo', c('stuff.1','stuff.2','stuff.3'))
## End(Not run)</pre>
```

58 nsubstr

na.if

Conditional Conversion to Missing Values

# Description

na.if returns a value with the same shape as v1, with values equal to v2 set to NA.

# Usage

```
na.if(v1, v2)
```

# Arguments

v1 an object to be tested.

v2 a value to be compared to elements of v1.

# **Details**

The result is equivalent to ifelse (v1==v2, NA, v1).

#### Value

An object of the same shape as v1, where values equal to v2 are set to NA.

# References

Inspired by Oracle's nullif() function.

# See Also

```
if.na, ifelse.
```

# **Examples**

```
x \leftarrow c(1:3,1:3)
na.if(x, 3)
```

nsubstr

Count Substring Instances

# Description

Counts exact instances of a target string in each element of a character vector.

# Usage

```
nsubstr(a,b)
```

nw.align 59

### **Arguments**

a a character vector.

b a character string with positive length.

#### Value

An integer vector consisting of the number of instances of b in each element of a.

### **Examples**

```
nsubstr(c('aabbcc',NA,'aabbbbccbb'), 'bb')
```

nw.align

Needleman and Wunsch Sequence Alignment

#### **Description**

Returns a global or partial-local optimal alignment of two nucleotide sequences, with linear gap penalty. nw.score returns just the alignment score.

#### Usage

```
nw.align(gt1, gt2, gap=-1, pm=1, mm=0, ends=FALSE)
nw.score(gt1, gt2, gap=-1, pm=1, mm=0, ends=FALSE)
```

# **Arguments**

gt1, gt2 nucleotide sequences to be aligned.

gap gap penalty.

pm score for a perfect match.

mm score for a mismatch.

ends logical: specifies whether overhanging ends are not penalized. See details.

# **Details**

The default is to perform a global alignment across the full lengths of both sequences. The ends argument can be used to specify that overhangs at one or both ends of one or both sequences should not be penalized. If ends is a scalar, then it controls all four possible overhangs. It may also be set to a vector of four logical values, to control scoring of each possible overhang: the left and right sides of gt1, and left and right sides of gt2.

The dynamic programming algorithm in nw.align is O(m\*n) in time and space, and hence is not appropriate for aligning very long sequences. The method in nw.score is O(n) in space.

To get the longest common subsequence, use

nw.orient.assay

#### Value

#### A list:

score the optimal alignment score.

pct.matched over the aligned intervals of the two sequences, the percentage identity.

ends the starting and ending base positions of the aligned intervals in gt1 and gt2.

alignment a vector of three strings: the aligned gt1, a representation of the matched posi-

tions, and the aligned gt2.

#### References

Needleman, S. B., & Wunsch, C.D. (1970) A general method applicable to the search for similarities in the amino acid sequence of two proteins. *J. Mol. Biol.* **48**: 443-453.

#### See Also

```
nw.orient.assay.
```

# **Examples**

```
s1 <- 'GAGTTC'
s2 <- 'AACATCGAGGTCTACGGATT'
nw.align(s1, s2, ends=FALSE)
nw.align(s1, s2, ends=TRUE)</pre>
```

nw.orient.assay

Orient Assay Sequences

#### **Description**

Determines the relative orientation of pairs of genotyping assays by sequence alignment.

#### Usage

```
nw.orient.assay(gt1, gt2, delta=1)
```

# Arguments

```
gt1, gt2 assay sequence(s) with a variant position denoted by an underscore ('_').

delta the minimum difference in scores to establish orientation.
```

#### **Details**

Corresponding elements of gt1 and gt2 are split into left and right flanks around the variant position, and then aligned in forward and reverse-complement orientations. The best-scoring orientation is reported if its score exceeds the score of the opposing orientation by at least delta.

### Value

```
A vector of '+', '-', or NA values.
```

orient.gt.data 61

#### See Also

```
nw.align.
```

### **Examples**

```
a1 <- 'ACAAC_ATGCT'
a2 <- 'GCAT_GTTG'
nw.orient.assay(a1,a2)</pre>
```

orient.gt.data

Flip Assay Strands and/or Swap Alleles

# **Description**

Given a data frame of genotype information, this updates genomic orientations and/or swaps allele coding for individual assays.

# Usage

```
orient.gt.data(gt.data, flip=FALSE, swap=FALSE)
```

### **Arguments**

gt.data a data frame of genotype information.

flip logical: indicates if strands should be flipped.

swap logical: indicates if alleles should be swapped.

#### **Details**

Both flip and swap are evaluated for each assay in gt.data, so these can either be single logical values or vectors with one value per row in gt.data.

# Value

An updated version of gt.data with the specified adjustments applied.

## See Also

```
fetch.gt.data, match.gt.data.
```

```
gt.demo.check()
gt <- fetch.gt.data('Demo_2')

# orient to dbSNP strand
gt <- orient.gt.data(gt, flip=(gt$dbsnp.orient == '-'))
with(gt, table(strand,dbsnp.orient))

# orient to '+' strand
gt <- orient.gt.data(gt, flip=(gt$strand=='-'))
with(gt, table(strand,dbsnp.orient))</pre>
```

62 panel.cluster

```
# swap to major/minor allele order
swap <- (summary.gt.data(gt)$freq.a < 0.5)
table(swap)
gt <- orient.gt.data(gt, swap=swap)
table(summary.gt.data(gt)$freq.a < 0.5)

# sort alleles
head(gt$alleles)
swap <- (sapply(strsplit(gt$alleles,'/'), order)[1,] == 2)
gt <- orient.gt.data(gt, swap=swap)
head(gt$alleles)</pre>
```

panel.cluster

Panel Function for Drawing Elliptical Cluster Boundaries

# **Description**

Panel function for plotting two-dimensional clusters of points with elliptical boundary regions.

## Usage

```
panel.cluster(x, y, group.number=1, bounds=c(), min.points=4, ...)
```

### **Arguments**

```
x, y as in panel.xyplot.
group.number as in panel.superpose.
bounds contours at which to draw ellipsoid boundaries.
min.points the minimum number of points for which to compute ellipsoids.
... additional arguments passed to panel.xyplot.
```

# **Details**

Points are first plotted as in panel.xyplot. Then elliptical boundaries are drawn using settings from superpose.line, using trellis.par.get.

# See Also

```
gt.cluster.plot,panel.xyplot,panel.superpose,ellipsoidPoints.
```

panel.qqpval 63

panel.qqpval

Quantile-Quantile Plots for P Values: Panel Functions

#### **Description**

Panel functions used by qqpval for generating Quantile-Quantile plots of log transformed P values.

### Usage

```
prepanel.qqpval(x, n, groups=NULL, subscripts, ...)
panel.qqpval(x, n, max.pts, groups=NULL, ...)
```

### **Arguments**

#### See Also

```
qqpval, prepanel.default.xyplot, panel.qq.
```

panel.qqthin

Sparse Normal Quantile-Quantile Plots: Panel Function

# Description

Panel function used by qqthin for generating quantile-quantile plots of very large numbers of observations.

# Usage

```
panel.qqthin(x, max.pts, groups=NULL, ...)
```

# **Arguments**

```
x a numeric vector.
max.pts the maximum number of discrete quantiles to plot.
groups see xyplot.
... additional arguments passed to panel.qqmath.
```

# See Also

```
qqthin, panel.qqmath.
```

64 prcomp.gt.data

```
prcomp.gt.data Principal Components Analysis of Genotype Data
```

#### **Description**

Performs a principal components analysis of a matrix of genotypes and scores each sample against the most significant components.

#### Usage

```
## S3 method for class 'gt.data':
prcomp(x, sample.mask=TRUE, nc=20, ...)
## S3 method for class 'gt.dataset':
prcomp(x, sample.mask=TRUE, nc=20, ...)
```

#### **Arguments**

```
a data frame of genotypes from fetch.gt.data, or a dataset description
from gt.dataset.
sample.mask a logical vector for subsetting samples.
nc the number of components to return.
... not used.
```

### **Details**

This function is useful for computing principal components in cases where an entire genotype matrix can be accommodated in memory. Genotype vectors are centered and scaled, and missing values are imputed, as in Price *et al.* (2006). The principal components are computed using prcomp.

### Value

A list with the following components:

```
a data frame with one row per row in the data table, and no columns, containing the loadings of each sample onto the top principal components.

sdev the standard deviations of the top no principal components.

assays the number of assays included in the analysis.

dataset.name the name of the genotype dataset.

call the call used to generate the analysis.
```

#### References

Price, A. L., *et al.* (2006) Principal components analysis corrects for stratification in genome-wide association studies. *Nat. Genet.* **38**: 904-909.

#### See Also

```
prcomp, fetch.gt.data, gt.dataset, snp.loadings, apply.loadings.
```

progress.bar 65

### **Examples**

```
gt.demo.check()
g <- fetch.gt.data('Demo_1')
p <- prcomp(subset(g, (ploidy=='A')))
screeplot(p)
with(p$loadings, plot(PC1, PC2))</pre>
```

progress.bar

Console Text-Based Progress Bar

# Description

Displays a text-based progress bar on the R console, to indicate how much of a long-running calculation has been completed. The elapsed time and estimated time remaining are also reported.

# Usage

```
progress.bar(done, total, len = 40)
```

# **Arguments**

done the amount of work already done.

total the total amount of work to do.

len the length of the bar, in characters.

# **Details**

After writing the progress bar, the cursor is repositioned at the start of the line so that successive calls will update the bar in place. The console buffer is flushed after each call. The first call should have done=0 to ensure proper initialization of internal data structures.

### Value

Invisible NULL.

```
fn <- function(x) { Sys.sleep(1); progress.bar(x, 20) } x \leftarrow \text{sapply}(0:20, \text{ fn})
```

66 qqprcomp

qqprcomp

Quantile-Quantile Plots of PCA Loadings

### **Description**

Generates a series of quantile-quantile plots of either sample or SNP loadings from a principal components analysis.

### Usage

```
qqprcomp(x, col=1:6, layout, ...)
```

# **Arguments**

Х	either a PCA result structure returned by prcomp.gt.data or prcomp.gt.dataset, or a table of SNP loadings from snp.loadings.
col	a vector of component numbers to plot.
layout	a vector with two elements giving the numbers of columns and rows to use for arranging the plot panels. If not specified, the plots are arranged to minimize wasted space.
	additional arguments passed to qqthin.

#### **Details**

This function generates a series of normal Q-Q plots of loadings from a principal components analysis. It accepts either a PCA result structure (for sample loadings), or a table of SNP loadings returned by snp.loadings. Sparse Q-Q plots are generated automatically when the number of loadings is very large.

Q-Q plots are useful for distinguishing components that reveal either sample or genomic structure in the data, from components that do not. If a component has flat Q-Q plots for both sample and SNP loadings, it is unlikely to be informative.

# Value

A plot object of class "trellis".

# See Also

```
prcomp.gt.data, prcomp.gt.dataset, snp.loadings, qqthin.
```

qqpval 67

qqpval

Quantile-Quantile Plots for P Values

# **Description**

Quantile-Quantile plots of log transformed P values versus their expected uniform distribution, with support for very large numbers of tests.

# Usage

# **Arguments**

#### **Details**

Conventional Q-Q plots for very large numbers of tests are slow and most of the "ink" is spent plotting uninteresting large P values. This function avoids this problem by limiting the number of quantiles plotted. If the number of supplied P values is larger than max.pts, then a threshold is chosen to switch between plotting each observed value, and plotting evenly spaced quantiles. The threshold is optimized to minimize the quantile spacing in the equal-spaced region.

A valid (truncated) Q-Q plot can also be generated from the smallest P values selected from a larger set of tests, by specifying n, the total number of tests the supplied values were selected from.

A reference line is drawn using settings specified by trellis.par.get('reference.line').

#### Value

```
A plot object of class "trellis".
```

### See Also

```
qqthin, qqmath.
```

68 qqthin

#### **Examples**

qqthin

Sparse Normal Quantile-Quantile Plots

# **Description**

Quantile-Quantile plots of a sample versus the normal distribution, with support for very large numbers of observations.

#### Usage

```
qqthin(x, ..., max.pts=1000, panel=panel.qqthin)
```

# **Arguments**

```
    either a numeric vector, or a formula to be evaluated in the context of a data argument.
    additional arguments passed to qqmath.
    the maximum number of discrete quantiles to plot.
    panel as in qqmath.
```

# **Details**

Conventional Q-Q plots for very large numbers of observations are slow and most of the "ink" is spent plotting uninteresting values. This function avoids this problem by limiting the number of distinct quantiles plotted. If the number of values is larger than max.pts, then a threshold is chosen to switch between plotting each extreme value, and plotting evenly spaced quantiles. The threshold is optimized to minimize the quantile spacing in the equal-spaced region.

A reference line is drawn using settings specified by trellis.par.get('reference.line').

#### Value

```
A plot object of class "trellis".
```

# See Also

```
qqpval, qqmath.
```

hexToRaw 69

#### **Examples**

hexToRaw

Convert between Raw Vectors and Hex Strings

# Description

Convert between raw vectors and hex strings.

# Usage

```
hexToRaw(hex)
rawToHex(raw)
```

# **Arguments**

```
hex a string of hex digits.
raw a raw vector.
```

#### Value

For hexToRaw, a raw vector constructed by converting each consecutive pair of hex digits to one byte. For rawToHex, a character string formed by converting each byte of the input vector to its two-digit hex representation.

# See Also

```
charToRaw, rawToChar.
```

```
hexToRaw('1a3b1a3b')
rawToHex(as.raw(seq(0,100,10)))
```

70 reshape.gt.data

```
reshape.gt.data Reshape Genotype Data
```

# Description

This takes a data frame of genotypes with one row per assay, and returns a "long" data frame with one row per genotype per sample.

# Usage

```
reshape.gt.data(gt.data, ...)
```

#### **Arguments**

```
gt.data a data frame of genotypes from fetch.gt.data.
... additional arguments passed to gt.split.
```

# Value

A data frame with one row per assay per sample, and columns corresponding to available data extracted from gt.data. This normally includes genotypes, and may include quality scores and/or one or more columns of additional underlying raw data.

```
assay.name an assay identifier.

sample.name a sample identifier.

genotype the genotype for the specified sample and assay.

qscore the corresponding quality score.

... additional columns of raw data, depending on the dataset.
```

#### See Also

```
fetch.gt.data, unpack.gt.matrix, gt.split.
```

```
gt.demo.check()
gt <- fetch.gt.data('Demo_2',raw.data=TRUE)
head(reshape.gt.data(gt))
d <- reshape.gt.data(gt[seq(32,232,40),], na.codes='n')
gt.cluster.plot(d)</pre>
```

revcomp 71

revcomp

Reverse Complement DNA Sequences

### **Description**

Reverse complement DNA sequences, including IUPAC ambiguity codes.

### Usage

```
revcomp(x)
```

#### **Arguments**

Х

a character vector of DNA sequences

#### Value

A vector with the same shape as x, where each sequence has been replaced by its reverse complement, preserving case.

# **Examples**

```
revcomp(c('AACAGTAGA','AACCNNRacgt'))
```

score.and.store

Test SNPs for Association and Store Results

# **Description**

Perform a series of single-point SNP association tests on a genotype dataset, using an arbitrary scoring function, and store the results in the  ${\tt TEST}$  and  ${\tt TEST}$ \_RESULT tables.

#### Usage

#### **Arguments**

```
dataset.name the name of the dataset to be analyzed.

test.name a unique identifier to associate with these test results.

description a description of this test result set.

formula a symbolic description of the model to be fit.

score.fn the scoring function to be applied to each SNP.

pt.filter an expression to use for subsetting on the phenotype table.

gt.filter an expression to use for subsetting on the genotype table.
```

72 score.chisq.2x2

pca	a logical indicating whether the current principal components analysis results for this dataset should be loaded, or a list of arguments to be passed to fetch.prcomp to specify an analysis to be loaded.
part	a vector of slices 1parts.
parts	the number of slices to split the dataset into.
dryrun	logical: if TRUE, then do not write results to the database.
	additional arguments to pass to score.gt.data.

#### **Details**

This is essentially a wrapper around score.gt.data, that handles fetching phenotype and genotype data, and storing results back into the database. The formula, score.fn, pt.filter, and gt.filter arguments are passed to score.gt.data. The pca argument is passed to fetch.pt.data. And the part and parts arguments are used with fetch.gt.data. The description argument is used for constructing new entries in the TEST table in GTPUB.

If executed as part of an LSF "job array", this function will automatically initialize parts on each node to divide the computation evenly across the job array.

#### See Also

```
fetch.pt.data, fetch.gt.data, score.gt.data.
```

# **Examples**

score.chisq.2x2 Chi-Squared Test for Allelic Association

# **Description**

Test for allelic association with a binary outcome, using a chi-squared test on a 2x2 table of allele counts.

# Usage

```
score.chisq.2x2(formula, data, ploidy)
```

# **Arguments**

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data and details.
```

score.chisq 73

#### **Details**

The 2x2 chi-squared test is the simplest test for association between genotypes and a binary outcome. The model formula should be of the form  $outcome \sim genotype$  without additional terms. We do not apply a continuity correction because that makes the overall distribution of P values slightly conservative.

Sex linked data is handled by counting males as having a single allele, which while technically correct may not be reasonable.

Use of this test is not recommended as it is sensitive to deviations from Hardy Weinberg equilibrium. Usually, score.trend or score.glm are better choices.

#### Value

A data frame with one row and three columns:

```
pvalue P value for the test.

effect the log odds ratio for the 2x2 table.

stderr the standard error of the log odds ratio.
```

#### See Also

```
chisq.test, score.chisq, score.trend, score.glm, score.gt.data.
```

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
score.gt.data(status~genotype, pt, gt, score.chisq.2x2)
score.gt.data(status~genotype, pt, gt, score.glm)</pre>
```

```
score.chisq
```

Chi-Squared Test for Genotypic Association

#### **Description**

Test for genotype association with a categorical outcome, using a chi-squared test on an NxM table of genotype counts.

# Usage

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

mode genetic mode of action to test.
```

74 score.fisher

#### **Details**

This performs a simple chi squared test of association using the contingency table for a categorical outcome (with any number of levels) versus diploid genotypes. The model formula should be of the form outcome~genotype without additional terms. Empty levels for the outcome or the genotype will be dropped. We do not apply a continuity correction because that makes the overall distribution of P values slightly conservative.

Sex linked data is handled by treating males as diploid homozygotes for the corresponding haploid alleles, which may or may not be a reasonable thing to do.

#### Value

A data frame with one row and three columns:

```
pvalue P value for the test.

effect for 2x2 tables, the log odds ratio.

stderr for 2x2 tables, the standard error of the log odds ratio.
```

#### See Also

```
chisq.test, score.fisher, score.trend, score.chisq.2x2, score.gt.data.
```

# **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
score.gt.data(status~genotype, pt, gt, score.chisq.2x2)
score.gt.data(status~genotype, pt, gt, score.chisq)</pre>
```

score.fisher

Fisher's Exact Test for Genotypic Association

# Description

Performs Fisher's exact test for genotype association with a categorical outcome, on an NxM table of genotype counts.

# Usage

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

mode genetic mode of action to test.

... additional arguments passed to fisher.test.
```

score.glm 75

#### **Details**

This evaluates Fisher's exact test for association on a contingency table for a categorical outcome (with any number of levels) versus diploid genotypes. The model formula should be of the form outcome~genotype without additional terms. Empty levels for the outcome or the genotype will be dropped.

Sex linked data is handled by treating males as diploid homozygotes for the corresponding haploid alleles, which may or may not be a reasonable thing to do.

#### Value

A data frame with one row and two columns:

```
pvalue P value for the test.
effect for 2x2 tables, the log odds ratio.
```

#### See Also

```
fisher.test, score.chisq, score.gt.data.
```

# **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
score.gt.data(status~genotype, pt, gt, score.chisq)
score.gt.data(status~genotype, pt, gt, score.fisher)</pre>
```

score.glm

Test for Association using Logistic Regression

# Description

Test for association with a binary outcome using logistic regression.

#### Usage

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

drop the model term to test for association.

mode genetic mode of action to be tested.
```

76 score.glm.general

#### **Details**

The model formula may include covariates and should contain a single genotype term without interactions. Significance is assessed by ANOVA comparing the full model with a null model constructed by removing this term.

At sex linked loci, haploid males are treated the same as the corresponding diploid female homozygotes.

#### Value

A data frame with one row and three columns. An effect size is not reported if the mode of action is 'general'.

pvalue P value for an F test comparing the full and null models.

effect the regression coefficient (log odds) for term.
stderr the standard error of the effect size estimate.

#### See Also

lm, score.trend, score.chisq, score.glm.general, score.glm.groups, score.gt.data.

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
score.gt.data(status~plate+genotype, pt, gt, score.glm)
score.gt.data(status~plate+genotype, pt, gt, score.glm.general)</pre>
```

score.glm.general Association Test with a Logistic Model and General Mode of Action

## **Description**

Test for association using logistic regression, with a general mode of action.

#### Usage

```
score.glm.general(formula, data, ploidy)
```

#### **Arguments**

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.
```

#### **Details**

This is similar to score.glm, except that it considers a general mode of action with (log) additive and dominance effects. Overall significance is still assessed by ANOVA comparing the full model with a null model constructed by removing this term.

score.glm.groups 77

#### Value

A data frame with four columns. If there are only two distinct genotypes, then results are equivalent to score.glm with additive mode of action. If there are three distinct genotypes, then the first row of results describes the overall ANOVA test; the second describes the fitted additive effect; and the third describes the fitted dominance effect.

#### See Also

```
glm, score.glm, score.gt.data.
```

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
score.gt.data(status~genotype, pt, gt, score.glm)
score.gt.data(status~genotype, pt, gt, score.glm.general)</pre>
```

score.glm.groups Test for Association using a Logistic Model with Subgroup Effects

# Description

Test for association with a binary outcome, using logistic regression assuming a log-additive allelic effect on risk for the genotype term, where effects are allowed to vary by subgroup.

#### Usage

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

quick logical: if set, just return the overall test result.

mode genetic mode of action to test.
```

78 score.gt.data

#### **Details**

The model formula can contain multiple genotype terms with interactions, where the interacting variables are factors. Overall significance is assessed by a likelihood ratio test comparing the full model with a null model constructed by removing all genotype terms. Significance of the interaction term(s) is assessed by a likelihood ratio test comparing the full model with a model with no interactions. Finally, genotype effects are computed for each subgroup. At sex linked loci, haploid males are treated the same as the corresponding diploid female homozygotes.

#### Value

A data frame with four columns and at least two rows. The first two rows describe likelihood ratio tests for the full model versus a null model, and the full model versus a model with no subgroup interaction. Additional rows describe tests for effects within each subgroup.

term	for subgroup effects, the corresponding genotype:subgroup interaction term.
pvalue	for tests on nested models, the P value for a likelihood ratio test comparing the two models. For subgroup effects, the P value of a Wald test on the effects size.
effect	for subgroups, the estimated allelic effect (log odds) from the regression.
stderr	for subgroups, the estimated standard error of the effect size.

#### See Also

```
glm, score.glm, score.gt.data.
```

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:5,]
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
score.gt.data(status~plate+genotype, pt, gt, score.glm)
score.gt.data(status~plate*genotype, pt, gt, score.glm.groups)</pre>
```

score.gt.data

Test SNPs for Association

## **Description**

Perform a series of single-point SNP association tests, using an arbitrary scoring function.

# Usage

score.gt.data 79

### **Arguments**

formula	a symbolic description of the model to be fit.
pt.data	a data frame of phenotypes from fetch.pt.data.
gt.data	a data frame of genotypes from fetch.gt.data.
score.fn	the scoring function to be applied to each SNP.
pt.filter	an expression to use for subsetting on the phenotype table.
gt.filter	an expression to use for subsetting on the genotype table.
progress	logical: specifies if a progress bar should be displayed on the R console.
	additional arguments to pass to score.fn.

#### **Details**

For each row of genotypes in gt.data, a data frame will be constructed by merging columns of pt.data referenced in the model formula with those genotypes. The specified score function will be invoked for each of these data frames in turn.

If no scoring function is specified, then one will be deduced from the form of the model formula. A trend test will be used for simple binary-outcome models of the form "status genotype". Logistic regression will be used for binary outcomes with more complicated model functions. And linear regression will be used for quantitative outcomes.

The filters pt.filter and gt.filter are evaluated in the context of the pt.data and gt.data tables, respectively, similar to how subset works.

It is expected that some tests will fail for some SNPs (say, due to degeneracies where a SNP or trait is invariant across the samples for which data is available). Errors from the scoring function are converted to warnings.

## Value

A data frame with up to five columns, and one or more rows per SNP tested, depending on the form of the test. Some columns may not be populated for certain test types.

assay.name	an identifier for the assay tested.
term	for tests that return more than one result per assay, something specifying the individual results.
pvalue	an uncorrected P value for this test.
effect	an estimated effect size, where that makes sense.
stderr	the estimated standard error of the effect size.

```
score.chisq.2x2, score.chisq, score.fisher, score.trend, score.kruskal,
score.jt, score.lm, score.lm.general, score.lm.groups, score.glm, score.glm.general,
score.glm.groups, score.and.store.
```

score.jt

#### **Examples**

score.jt

Jonckheere-Terpstra Nonparametric Test for Association

# Description

Test for association between genotypes and a quantitative outcome, using the nonparametric Jonckheere-Terpstra test for ordered differences among genotype classes.

#### Usage

```
score.jt(formula, data, ploidy, ...)
```

#### **Arguments**

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

additional arguments passed to jt.test.
```

#### **Details**

The Jonckheere-Terpstra test computes Mann-Whitney rank sum statistics for ordered diploid genotypes, and does a two-sided test on the sum of those statistics. This tests for a monotonic trend in outcomes as a function of genotype. The model formula should be of the form outcome~genotype without additional terms.

The test does not directly provide an estimate of an effect size. Instead, we center and rescale the test statistic to fall in the range -1 to +1. The expected value of this score is independent of sample size and indicates the direction of effect.

#### Value

A data frame with one row and three columns:

pvalue	P value for the test assuming an asymptotic normal distribution for the test statistic.
effect	the test statistic centered and scaled to be in the range $-1$ to $+1$ .
stderr	the estimated standard error of effect.

#### References

```
http://tolstoy.newcastle.edu.au/R/help/06/06/30112.html.
```

score.kruskal 81

#### See Also

```
jt.test, score.kruskal, score.gt.data.
```

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$score <- rnorm(nrow(pt))
score.gt.data(score~genotype, pt, gt, score.kruskal)
score.gt.data(score~genotype, pt, gt, score.jt)</pre>
```

score.kruskal

Kruskal-Wallis Nonparametric Test for Association

## **Description**

Test for association between genotypes and a quantitative outcome, using the nonparametric Kruskal-Wallis test for differences among genotype classes.

#### Usage

#### **Arguments**

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

mode genetic mode of action to test.
```

## **Details**

This tests whether the summed ranks of the outcome are independent of genotype. The model formula should be of the form outcome~genotype without additional terms. The default is to make no assumptions about mode of action.

#### Value

A data frame with one row and one column:

```
pvalue asymptotic chi-squared P value for the test.
```

```
kruskal.test, score.jt, score.gt.data.
```

82 score.lm

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$score <- rnorm(nrow(pt))
score.gt.data(score~genotype, pt, gt, score.kruskal)
score.gt.data(score~genotype, pt, gt, score.lm)</pre>
```

score.lm

Test for Association using a Simple Linear Model

#### **Description**

Test for association using linear regression.

# Usage

#### **Arguments**

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

drop the model term to test for association.

mode genetic mode of action to test.
```

## **Details**

The model formula may include covariates and should contain a single genotype term without interactions. Significance is assessed by ANOVA comparing the full model with a null model constructed by removing this term.

At sex linked loci, haploid males are treated the same as the corresponding diploid female homozygotes.

#### Value

A data frame with one row and three columns. An effect size is not reported if the mode of action is 'general'.

```
pvalue P value for an F test comparing the full and null models.

effect the regression coefficient for term.

stderr the standard error of the effect size estimate.
```

```
lm, score.lm.general, score.lm.groups, score.gt.data.
```

score.lm.general 83

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$score <- rnorm(nrow(pt))
score.gt.data(score~plate+genotype, pt, gt, score.lm)
score.gt.data(score~genotype, pt, gt, score.lm, mode='recessive')
score.gt.data(score~genotype, pt, gt, score.lm, mode='general')
score.gt.data(score~genotype, pt, gt, score.lm.general)</pre>
```

score.lm.general Test for Association using a Linear Model and General

## **Description**

Test for association using linear regression, for a general mode of action.

#### Usage

```
score.lm.general(formula, data, ploidy)
```

## **Arguments**

formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

#### Details

This is similar to score.lm, except that it considers a general mode of action with additive and dominance effects. Overall significance is still assessed by ANOVA comparing the full model with a null model constructed by removing this term.

#### Value

A data frame with four columns. If there are only two distinct genotypes, then results are equivalent to score. 1m with additive mode of action. If there are three distinct genotypes, then the first row of results describes the overall ANOVA test; the second describes the fitted additive effect; and the third describes the fitted dominance effect.

```
lm, score.lm, score.gt.data.
```

84 score.lm.groups

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$score <- rnorm(nrow(pt))
score.gt.data(score~genotype, pt, gt, score.lm, mode='general')
score.gt.data(score~genotype, pt, gt, score.lm.general)</pre>
```

score.lm.groups

Test for Association using a Linear Model with Subgroup Effects

## **Description**

Test for association using linear regression assuming an additive allelic effect for the genotype term, where effects are allowed to vary by subgroup.

## Usage

## **Arguments**

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.

quick logical: if set, just return the overall test result.

mode genetic mode of action to test.
```

#### **Details**

The model formula can contain multiple genotype terms with interactions, where the interacting variables are factors. Overall significance is assessed by ANOVA comparing the full model with a null model constructed by removing all genotype terms. Significance of the interaction term(s) is assessed by ANOVA comparing the full model with a model with no interactions. Finally, genotype effects are computed for each subgroup. At sex linked loci, haploid males are treated the same as the corresponding diploid female homozygotes.

## Value

A data frame with four columns and at least two rows. The first two rows describe ANOVA tests for the full model versus a null model, and the full model versus a model with no subgroup interaction. Additional rows describe tests for effects within each subgroup.

term	for subgroup effects, the corresponding genotype:subgroup interaction term.
pvalue	for ANOVA tests, the P value for an F test comparing the full and null models. For subgroup effects, the P value for a T test on the effect size.
effect	for subgroups, the estimated allelic effect from the regression.
stderr	for subgroups, the estimated standard error of the effect size.

score.prcomp 85

#### See Also

```
lm, score.lm, score.gt.data.
```

## **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:5,]
pt$score <- rnorm(nrow(pt))
score.gt.data(score~plate+genotype, pt, gt, score.lm)
score.gt.data(score~plate*genotype, pt, gt, score.lm.groups)</pre>
```

score.prcomp

Test Phenotypic Association with Principal Components

## **Description**

Evaluates evidence for association between sample loadings from a principal components analysis, and one or more phenotypes.

# Usage

```
score.prcomp(formula, pc.data, pt.data, ...)
```

# Arguments

formula	a symbolic description of the model to be fitted. The left hand side should be given as PC.
pc.data	a PCA result structure returned by $\verb"prcomp.gt.data" or \verb"prcomp.gt.dataset".$
pt.data	phenotype data from fetch.pt.data.
	additional arguments passed to 1m.

#### **Details**

The specified model formula is evaluated by linear regression for each principal component, and results are summarized as in an ANOVA based on the proportion of variance of the principal component explained.

## Value

A data frame with one row per principal component and three columns:

```
R^2 the adjusted multivariate R\hat{2} statistic for the model.
F value an F statistic for the model.
Pr(>F) the tail probability associated with this F statistic.
```

```
prcomp.gt.data, prcomp.gt.dataset, lm.
```

86 score.trend

#### **Examples**

score.trend

Cochran-Armitage Trend Test for Association

# Description

Test for genotype assocation with a binary outcome, using the Cochran-Armitage test for trend in proportions.

#### Usage

```
score.trend(formula, data, ploidy)
```

## **Arguments**

```
formula a symbolic description of the model to be fit.

data a data frame containing the variables in the model.

ploidy see fetch.gt.data. Ignored.
```

## **Details**

The Cochran-Armitage test for trend is a test for a linear dependence of outcome proportions on genotype expressed as an allele count. The model formula should be of the form outcome~genotype without additional terms.

In place of a conventional effect size, we return the slope and standard error for the trend in proportions. This slope is thus the fitted change in outcome proportion per allele.

## Value

A data frame with one row and three columns:

```
pvalue P value for test.

effect the slope of the trend in proportions.

stderr the standard error of the slope.
```

```
\verb|prop.trend.test|, \verb|score.glm|, \verb|score.gt.data|.
```

set.hidden 87

#### **Examples**

```
gt.demo.check()
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')[1:10,]
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
score.gt.data(status~genotype, pt, gt, score.trend)
score.gt.data(status~genotype, pt, gt, score.glm)</pre>
```

set.hidden

Update Hidden Status

#### **Description**

This updates the "hidden" status of a database object (project, dataset, platform mapping, sample attribute, etc).

# Usage

```
set.hidden(table, name, is.hidden=TRUE, ...)
```

## **Arguments**

```
table the type of object to be hidden (or revealed).

name the name of the object to be hidden (or revealed).

is.hidden logical: the new status for the object.

additional qualifiers to identify the target object.
```

#### Value

If successful, the number of rows updated in the corresponding table (i.e., 1).

## See Also

```
mk.project, mk.dataset.
```

```
## Not run:
set.hidden('dataset','Demo_1',is.hidden=TRUE)
ls.dataset('Demo')
set.hidden('dataset','Demo_1',is.hidden=FALSE)
ls.dataset('Demo')
set.hidden('subject_attr','plate',is.hidden=TRUE,project.name='Demo')
## End(Not run)
```

88 snp.loadings

snp.loadings

SNP Loadings from Principal Components Analyses

#### **Description**

Computes loadings of assays onto previously computed principal components. Results can be computed for either a specific set of SNPs or the entire original genotype dataset.

# Usage

```
snp.loadings(x, data)
```

#### **Arguments**

```
x a structure returned by prcomp.

data either a data frame of genotypes from fetch.gt.data, or a dataset description from gt.dataset.
```

#### **Details**

snp.loadings computes SNP loadings across either an entire dataset or a specific set of genotype assays. The genotype data needs to cover the same individuals included in the original principal components analysis.

If the analysis was done with prcomp.gt.dataset and data is missing, then loadings are computed across the same dataset specification used in the original analysis.

#### Value

A list with the following components:

```
a data frame with one row per assay, and no columns, containing the loadings of each assay onto the top principal components.

sdev the standard deviations of the top no principal components.

assays annotations for the assays in loadings.

dataset.name the name of the source genotype dataset.

call the call used to generate the analysis.
```

# See Also

```
prcomp.gt.data, fetch.gt.data, gt.dataset, apply.loadings.
```

```
gt.demo.check()
p <- prcomp(gt.dataset('Demo_1', gt.filter=(ploidy=='A')))
s <- snp.loadings(p, gt.dataset('Demo_1'))
head(s)</pre>
```

sql.query 89

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sal.	anerv
29 - •	query

Simplified SQL Statement Execution

## **Description**

These functions parse and execute SQL statements, and either fetch query results or return the number of affected rows. All result sets are also cleaned up. Some database differences and differences in DBI implementations are also concealed.

## Usage

```
sql.query(db, sql, ...)
sql.exec(db, sql, ..., chunk.kb=256)
```

#### **Arguments**

db	a DBI connection object returned by dbConnect.
sql	a valid SQL statement, optionally with embedded bind variables.
• • •	optionally, a data frame or arguments to be used to construct a data frame of bind variables.
chunk.kb	when processing multiple rows of bind variables, a rough limit on the amount of data to send per query.

#### **Details**

To facilitate database agnostic code, several special elements can be used in SQL statements, which are internally replaced with database specific forms:

:user: the database user name.

:sysdate: the current date.

:unhex:(...) a function to convert from a hex string to binary.

:clob:(...) a wrapper for passing long character data back to R.

:blob:(...) a wrapper for passing long binary data back to R, rendered as a hexadecimal string.

**:fromdual:** a placeholder for the Oracle FROM DUAL idiom for SELECT statements that do not need table data.

Both sql.query and sql.exec emulate prepared statements and bind variables in a consistent way across databases. The emulation uses the Oracle syntax for bind variables (i.e., ':1', ':2', etc in SQL are substituted with values of the bind variables).

A single call to these functions may result in multiple SQL statements. These will be grouped into a single transaction for databases that support that, so that a call to sql.exec should be "all or none". RMySQL currently does not support transactions, so a call to sql.exec may partially fail.

## Value

For sql.query, a data frame constructed from the results returned by the query. Column names are transformed by converting '\_' to '.' and changing to lower case. For sql.exec, the return value is the number of affected rows.

90 store.prcomp

#### See Also

```
dbConnect, fetch.
```

## **Examples**

```
## Not run:
sql.exec(db, 'create table xyzzy (a number, b number)')
sql.exec(db, 'insert into xyzzy values (:1,:2)', a=6, b=1:4)
sql.exec(db, 'insert into xyzzy values (:1,:2)', data.frame(1,2))
sql.exec(db, 'select * from xyzzy')
sql.exec(db, 'drop table xyzzy')
sql.exec(db, 'select :user:, :sysdate: :fromdual:')
## End(Not run)
```

store.prcomp

Store or Remove Principal Components Results

#### **Description**

Store, or remove, principal components analysis results in the database.

## Usage

#### **Arguments**

```
a principal components result returned by prcomp.gt.data or prcomp.gt.dataset.

prcomp.name a short unique identifier for the analysis.

description a free-text description of this analysis.

nc the number of components to store.

is.hidden logical: indicates if the dataset is hidden.

dataset.name the name of a parent dataset for which an analysis is to be removed.
```

#### **Details**

Analysis names only need to be unique within the scope of a particular dataset.

#### Value

The number of principal components stored into the database.

```
fetch.prcomp, prcomp.gt.data, prcomp.gt.dataset.
```

store.sample.data 91

#### **Examples**

```
## Not run:
pc <- prcomp(gt.dataset('Demo_1', gt.filter=(ploidy=='A')))
store.prcomp(pc, 'demo_pc_1', 'Demo PCA results')
fetch.prcomp('Demo', 'demo_pc_1', nc=4)
## End(Not run)</pre>
```

# Description

Stores sample phenotype data into the database from a data frame.

#### Usage

```
store.sample.data(dataset.name, data)
```

#### **Arguments**

```
dataset.name the short unique identifier for the dataset.

data a data frame of sample phenotypes. See details.
```

## **Details**

The data argument should have a sample.name column, with samples previously defined by mk.sample. Additional columns should supply values of sample attributes previously defined by mk.sample.attr. Missing values are not explicitly recorded in the database.

#### Value

A vector of counts of rows inserted in the database for each column of phenotype information in the input data.

## See Also

```
mk.sample, mk.sample.attr, fetch.sample.data, fetch.pt.data.
```

```
## Not run:
p <- ls.sample('Demo_1')['sample.name']
p$stuff <- rnorm(nrow(p))
mk.sample.attr('Demo_1', p['stuff'], 'Some random stuff')
store.sample.data('Demo_1', p)
head(fetch.sample.data('Demo_1')
rm.sample.attr('Demo_1', 'stuff')
## End(Not run)</pre>
```

92 store.subject.data

```
store.subject.data Store Subject Data
```

# **Description**

Stores subject phenotype data into the database from a data frame.

## Usage

```
store.subject.data(project.name, data)
```

## **Arguments**

```
project.name the short unique identifier for the project.

data a data frame of subject phenotypes. See details.
```

#### **Details**

The data argument should have a subject.name column, with subjects previously defined by mk.subject. Additional columns should supply values of subject attributes previously defined by mk.subject.attr. Missing values are not explicitly recorded in the database.

## Value

A vector of counts of rows inserted in the database for each column of phenotype information in the input data.

#### See Also

```
mk.subject, mk.subject.attr, fetch.subject.data, fetch.pt.data.
```

```
## Not run:
p <- ls.subject('Demo')['subject.name']
p$stuff <- rnorm(nrow(p))
mk.subject.attr('Demo', p['stuff'], 'Some random stuff')
store.subject.data('Demo', p)
head(fetch.subject.data('Demo')
rm.subject.attr('Demo', 'stuff')
## End(Not run)</pre>
```

store.test.scores 93

```
store.test.scores Store or Remove Association Test Results
```

# Description

Store, or remove, association test results in the database.

## Usage

```
store.test.scores(x, test.name, description, is.hidden=FALSE)
rm.test(dataset.name, test.name)
```

## **Arguments**

```
a principal components result returned by prcomp.gt.data or prcomp.gt.dataset.

test.name a short unique identifier for the analysis.

description a free-text description of this analysis.

is.hidden logical: indicates if the result set is hidden.

dataset.name the name of a parent dataset for which an analysis is to be removed.
```

#### **Details**

Analysis names only need to be unique within the scope of a particular dataset.

### Value

The number of results stored into the database.

## See Also

```
fetch.test.scores, score.gt.data.
```

```
## Not run:
pt <- fetch.pt.data('Demo_2')
gt <- fetch.gt.data('Demo_2')
pt$status <- as.logical(rbinom(nrow(pt), 1, 0.5))
x <- score.gt.data(status~genotype, pt, gt)
store.test.scores(x, 'status_1', 'Test analysis')
y <- fetch.test.scores('Demo_2', 'status_1')
str(y)
## End(Not run)</pre>
```

94 summary.gt.data

```
summary.gt.data Genotype Data Summary
```

#### **Description**

Computes genotype counts, allele frequencies, call rates, and tests for Hardy Weinberg equilibrium for a packed genotype data structure.

# Usage

```
## S3 method for class 'gt.data':
summary(object, sample.mask, by.sample=FALSE, ...)
## S3 method for class 'gt.dataset':
summary(object, sample.mask, by.sample=FALSE, ...)
```

#### **Arguments**

```
either a data frame of genotypes from fetch.gt.data, or a dataset description from gt.dataset.
sample.mask an optional mask identifying a subset of samples to be included in the summaries.
by.sample logical: indicates if statistics should be computed by sample (as opposed to by SNP).
... not used.
```

#### **Details**

The sample.mask argument may either be a character string or a logical vector with one element per genotype.

#### Value

If by . sample is FALSE: a data frame with one row per genotype assay, and 9 columns:

```
NN count of missing ('n') genotypes.

AA, AB, BB counts of diploid r/h/a genotypes.

A_, B_ counts of haploid r/a genotypes.

gt.rate genotype call rate for this SNP.

freq.a, freq.b allele frequencies.

hw.p.value Hardy-Weinberg equilibrium P value, calculated from a likelihood ratio test.
```

If by sample is TRUE: a data frame with one row per sample and 9 columns, with samples ordered by position in the genotype strings, and row names set to RNA\_DNA\_SOURCE\_ID.

```
fetch.gt.data,gt.dataset.
```

unpack.gt.matrix 95

#### **Examples**

```
gt.demo.check()
g <- fetch.gt.data('Demo_1')[1:10,]
summary(g)
summary(g, attr(g,'gender')[['F']])
summary(g, by.sample=TRUE)[1:10,]</pre>
```

```
unpack.gt.matrix Convert Packed Genotype Strings to a Genotype Matrix
```

## **Description**

This takes a data frame of genotypes with one row per assay, and returns a new data frame with one column per assay and one row per sample.

# Usage

```
unpack.gt.matrix(gt.data, names=gt.data$assay.name, ...)
```

## **Arguments**

```
gt.data a data frame of genotypes from fetch.gt.data.names column names for the resulting data frame.additional arguments passed to gt.split.
```

#### Value

A data frame with one column per row in gt.data, and one row per sample.

## See Also

```
fetch.gt.data,gt.split.
```

```
gt.demo.check()
gt <- fetch.gt.data('Demo_1')
head(unpack.gt.matrix(gt[1:5,]))
head(unpack.gt.matrix(gt[1:5,], convert='char'))</pre>
```

96 use.gt.db

use.gt.db

Define GT.DB Database Connection

# **Description**

Define a DBI connection to be used to access a GT.DB database. This connection is used implicitly by other GT.DB functions that interact with the database.

# Usage

```
use.gt.db(dbConnection)
```

# Arguments

dbConnection a connection object from dbConnect.

#### Value

None.

#### See Also

```
dbDriver, dbConnect.
```

```
## Not run:
library(RSQLite)
tmpname <- tempfile('db')
gt.db <- dbConnect(dbDriver('SQLite'), tmpname)
use.gt.db(gt.db)
## End(Not run)</pre>
```

# Index

*Topic <b>NA</b>	mk.subject,54
if.na, 25	mk.subject.attr,55
na.if, 56	score.and.store, 69
*Topic character	set.hidden,85
as.mask,4	sql.query, 87
ch.table, 6	store.prcomp, 88
gt.dist, 17	store.sample.data,89
mask.str,42	store.subject.data,90
nsubstr, 56	store.test.scores, 91
*Topic <b>classes</b>	use.gt.db, 94
hexToRaw, 67	*Topic datasets
keep.attr, 27	demo_01,6
*Topic database	demo_02, <b>7</b>
apply.gt.dataset,2	hapmap, 19
fetch.gt.data,7	*Topic <b>hplot</b>
fetch.prcomp,9	gplot, 13
fetch.pt.data, 10	gplot.prcomp, 14
fetch.sample.data, 11	gt.cluster.plot, 15
fetch.subject.data, 11	ibd.plot, 22
fetch.test.scores, 12	ld.plot, 29
gt.dataset, 16	panel.cluster, 60
ls.assay,31	panel.qqpval,61
ls.assay.group, 32	panel.qqthin,61
ls.assay.position, 33	qqprcomp, 64
ls.dataset,34	qqpval, 65
ls.mapping, 35	qqthin, <mark>66</mark>
ls.platform, 36	*Topic <b>htest</b>
ls.prcomp, 37	hwe.test, 19
ls.project,38	jt.test, <mark>26</mark>
ls.sample, 39	score.and.store, 69
ls.subject,40	score.chisq,71
ls.test, 40	score.chisq.2x2,70
mk.assay,44	score.fisher,72
mk.assay.data,45	score.glm, 73
mk.assay.group,46	score.glm.general,74
mk.assay.position,47	score.glm.groups,75
mk.attr,48	score.gt.data,76
mk.dataset,49	score.jt,78
mk.mapping,50	score.kruskal,79
mk.platform, 51	score.lm,80
mk.project, 51	score.lm.general,81
mk.sample,52	score.lm.groups, 82
mk.sample.attr,53	score.trend,84

98 INDEX

*Topic <b>iplot</b>	big.loadings,5
adjust.gt.calls,1	
*Topic <b>logic</b>	ch.table,6
if.na, 25	charToRaw,67
na.if,56	chisq.test, 71, 72
*Topic <b>manip</b>	o <del>-</del> 00 04
adjust.gt.calls,1	dbConnect, 87, 88, 94
apply.loadings, $3$	dbDriver,94
big.loadings,5	$demo_01, 6$
gt.split,18	demo_02,7
ibd.dataset,20	11 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1
ibd.summary,23	ellipsoidPoints, $15,60$
ibs.gt.data,24	fetch, 88
ld.gt.data,28	fetch.gt.data, 2, 3, 7, 15-18, 21, 24,
ld.prune, 30	28–31, 42, 43, 45, 59, 62, 68, 70–75,
mask.gt.data,41	77–82, 84, 86, 92, 93
match.gt.data,43	
nw.align, 57	fetch.prcomp, 9, 10, 37, 70, 88
nw.orient.assay,58	fetch.pt.data, 10, 11, 70, 77, 83, 89, 90 fetch.sample.data, 10, 11, 89
orient.gt.data,59	
reshape.gt.data,68	fetch.subject.data, 10, 11, 90
revcomp, 69	fetch.test.scores, 12, 41, 91
score.prcomp,83	fisher.test, 72, 73
summary.gt.data,92	glm, 75, 76
unpack.gt.matrix,93	gplot, 13, 14
*Topic <b>multivariate</b>	gplot.prcomp, 14
prcomp.gt.data,62	grr(ibs.gt.data), 24
snp.loadings,86	gt.cluster.plot, 1, 2, 15, 60
*Topic <b>regression</b>	gt.dataset, 2, 3, 16, 24, 62, 86, 92
score.and.store, 69	gt.dist, 17
score.glm,73	gt.paste(gt.split), 18
score.glm.general,74	gt.split, 18, 68, 93
score.glm.groups,75	90.00110, 10, 00, 70
score.gt.data,76	hapmap, 19
score.lm, 80	hexToRaw, 67
score.lm.general,81	hwe.test, 19
score.lm.groups, 82	
*Topic <b>utilities</b>	ibd.dataset, 17, 20, 22, 23
progress.bar,63	ibd.gt.data,21
[.keep.attr( <i>keep.attr</i> ), 27	ibd.outliers, 22, 23
<pre>[<keep.attr(keep.attr), 27<="" pre=""></keep.attr(keep.attr),></pre>	ibd.outliers(ibd.summary),23
	ibd.plot, 22, 22, 23
adjust.gt.calls, 1, 15	ibd.summary, 22, 23, 23, 24
apply.gt.dataset, 2, 16	ibs(ibs.gt.data),24
apply.loadings, 3, 62, 86	ibs.gt.data,24
as.mask, 4, 42	ibs.gt.dataset( <i>ibs.gt.data</i> ),24
assay.dat.01(demo_01),6	if.na, 25, 56
assay.dat.02(demo_02),7	ifelse, 25, 56
assay.def.01(demo_01),6	is.na, 25
assay.def.02(demo_02),7	
assay.pos.01(demo_01),6	jt.test, 26, 78, 79
assay.pos.02(demo_02),7	1
attributes, 27	keep.attr,27

INDEX 99

kept.attr(keep.attr),27	panel.cluster, $15,60$
kruskal.test, 27, 79	panel.qq, <i>61</i>
	panel.qqmath,61
ld.gt.data, 28, 30, 31	panel.qqpval,61
ld.plot, 29, 29	panel.qqthin,61
ld.prune, 30	panel.superpose, 60
levelplot, 13, 14, 30	panel.xyplot, 60
lm, 74, 80, 81, 83	prcomp, 3, 62, 86
ls.assay, 31, 33, 44	prcomp.gt.data, 5, 9, 14, 62, 64, 83, 86,
ls.assay.group, 32, 32, 46	88, 91
ls.assay.position, <i>32</i> , <i>33</i> , <i>47</i>	prcomp.gt.dataset, 5, 9, 14, 64, 83, 86,
ls.attr, 53, 55	88, 91
ls.attr(mk.attr),48	prcomp.gt.dataset
ls.dataset, 34, 39, 50	(prcomp.gt.data), 62
ls.mapping, 35, 50	prepanel.default.xyplot,61
ls.platform, 34, 36, 46, 51	
	prepanel.qqpval(panel.qqpval),61
ls.prcomp, 37	progress.bar, 63
ls.project, 34, 38, 40, 52	prop.trend.test,84
ls.sample, 11, 39, 52	13 65 66
ls.sample.attr(mk.sample.attr),	qqmath, 65, 66
53	qqprcomp,64
ls.subject, 12, 40, 54	qqpval, 61, 65, 66
ls.subject.attr,48	qqthin, 61, 64, 65, 66
ls.subject.attr	
(mk.subject.attr),55	rawToChar, 67
ls.test, $40$	rawToHex(hexToRaw),67
	reshape.gt.data, $1, 2, 15, 68$
mask.gt.data,41	revcomp, 69
mask.str,4,42	rm.assay.data(mk.assay.data),45
match.gt.data, 17, 43, 59	rm.assay.group(mk.assay.group),
max, <i>13</i>	46
min, 13	rm.assay.position
mk.assay, <i>32</i> , <b>44</b> , <i>45</i> – <i>47</i>	(mk.assay.position),47
mk.assay.data,45	rm.attr, 53, 55
mk.assay.group, 32, 44, 46, 51	rm.attr(mk.attr),48
mk.assay.position, 33, 44, 45, 47, 50	rm.dataset (mk.dataset), 49
mk.attr, 48, 53, 55	rm.mapping (mk.mapping), 50
mk.dataset, 34, 49, 85	rm.platform(mk.platform), 51
nk.mapping, 35, 47, 50	rm.prcomp, 37
nk.platform, 36, 46, 50, 51	rm.prcomp(store.prcomp), 88
mk.practorm, 30, 40, 50, 51 mk.project, 38, 51, 85	rm.project (mk.project), 51
mk.sample, 11, 39, 52, 54, 89	rm.sample(mk.sample), 52
mk.sample.attr, 49, 53, 89	rm.sample.attr(mk.sample.attr),
nk.subject, 12, 40, 52, 54, 90	53
nk.subject.attr,48,49,55,90	rm.subject (mk.subject), 54
15.50	rm.subject.attr,48
na.if,56	rm.subject.attr
nsubstr, 56	(mk.subject.attr),55
nw.align, 57, 59	rm.test,41
nw.orient.assay, 58, 58	rm.test(store.test.scores),91
nw.score(nw.align),57	
	samples.01 ( $demo_01$ ),6
orient.gt.data,43,59	samples.02(demo_02),7

100 INDEX

```
score.and.store, 69,77
score.chisq, 71, 71, 73, 74, 77
score.chisq.2x2, 70, 72, 77
score.fisher, 72, 72, 77
score.glm, 71, 73, 74-77, 84
score.glm.general, 74, 74, 77
score.glm.groups, 74, 75, 77
score.gt.data, 12, 70-75, 76, 76, 79-81,
       83, 84, 91
score.jt, 77, 78, 79
score.kruskal, 27, 77, 79, 79
score.lm, 77, 80, 81, 83
score.lm.general, 77, 80, 81
score.lm.groups, 77, 80, 82
score.prcomp, 83
score.trend, 71, 72, 74, 77, 84
set.hidden, 50, 52, 85
snp.loadings, 3, 5, 14, 62, 64, 86
sql.exec(sql.query), 87
sql.exec, MySQLConnection-method
       (sql.query), 87
sql.exec,OraConnection-method
       (sql.query), 87
sql.exec, SQLiteConnection-method
       (sql.query), 87
sql.exec-methods (sql.query), 87
sql.query, 87
store.prcomp, 9, 37, 88
store.sample.data, 11,89
store.subject.data, 12,90
store.test.scores, 12, 41, 91
sum, 13
summary.gt.data, 9, 42, 92
summary.gt.dataset
       (summary.gt.data), 92
un.mask(as.mask),4
unpack.gt.matrix, 9, 18, 68, 93
use.gt.db,94
xyplot, 15, 61
xyplot.gt.data(gt.cluster.plot),
       15
```