

Package ‘ind1KG’

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Title Data from 1000 Genomes, NA19240 (female) chr6 excerpt

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Description Elements of samtools/bioc workflow for dealing with personal sequence focusing on identification and interpretation of rare variants

Depends R (>= 2.10.1), chopsticks

Suggests Rsamtools, rtracklayer, GenomicFeatures, org.Hs.eg.db, SNPlocs.Hsapiens.dbSNP.20090506, TxDb.Hsapiens.UCSC.hg18.knownGene, lumiHumanIDMapping, GGBase, GGtools, hmyriB36

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biocViews Genetics, HighThroughputSequencing

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c6snp

*SNP metadata used in calling for 1000 genomes pilot data***Description**

SNP metadata used in calling for 1000 genomes pilot data – restricted to chr6

Usage

```
data(c6snp)
```

Format

A data frame with 1143009 observations on the following 16 variables.

chr a numeric vector

chrPosFrom a numeric vector

chrPosTo a numeric vector

rs a factor with levels rs1000 rs1000009 rs1000025 rs10000302 ...

ChrAllele a factor with levels - A AA AAA AAAA AAAAA AAAAAA AAAAAA ...

variantAllele a factor with levels (CA)11/12/13/14/- (G)14/15/16/18/19/20/21/22/23/C
(G)20/21/22/23/24/25/27/-/G/GGG (LAREDELETION)/-/A ...

snpAlleleChrOrien a factor with levels (A)1/13/15/G (A)10/12 (A)10/14 (C)10/11 (CA)10/11/13/14/15
(CA)10/14/15/16/17/18/20/21 (CA)10/17/18/19/20/21/22/23/24/25 (CA)11/12/13/14/-/CACA
(CA)11/12/13/14/15/16/17 ...

snp2chrOrien a numeric vector

snpClassAbbrev a factor with levels MicrosatelliteNamed snp dips mixed multi-base single base

snpClassCode a numeric vector

mapLocType a numeric vector

mapLocCnt a numeric vector

mapWeight a numeric vector

contigLabel a factor with levels DR53 c6_COX c6_QBL reference

unPlacedContig a factor with levels NT_113898.1 NT_113899.1

Details

Column headings: ===== chr,chrPosFrom, chrPosTo, rs,ChrAllele,variantAllele,snpAlleleChrOrien,
snp2chrOrien, snpClassAbbrev,snpClassCode,mapLocType,mapLocCnt,mapWeight, contigLabel,unPlacedContig

Column description: ===== Col1: Chr Col2: chrPosFrom: all chromosome positions are 1 based, that is the first base is counted as 1. Position is for each base, not "interbase". Col3: chrPosTo: Col4: rs Col5: ChrAllele: the base or bases on the chromosomes at the snp position or ranges. Col6: variantAllele: This is the other allele that is not on the chromosome. For ex. Snp is A/C, chromosome has A, variantAllele will be "C". Col7: snpAlleleChrOrien: This is the list of alleles for the snp in the chromosome orientation. Col8: : the alignment orientation between snp flank and the chromosome sequence. #orien: 0 - same; 1 - opposite Col9: snpClassAbbrev: the variation type of the snp. Details at: <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=helpsnpfaq&part=Reports#Reports>. Var

Col10: snpClassCode: The numeric code for snp variation class. possible snpClassCode, SnpClass-Abbrev and descriptions are below:

1 single base Only single base variation.ex.A/G. 2 dips indel or dips: deletion insertion polymorphism.ex.-/T.ex.ss149071 obs=AA/GCCTG 3 HETEROZYGOUS HETEROZYGOUS 4 Microsatellite Microsatellite 5 Named snp observed field starts with '(', and not class 3 an 4.ex.(Alu) 6 NOVARIATION NOVARIATION 7 mixed If the subsnp's in an rs cluster have different snp class. 8 multi-base Multiple Nucleotide Polymorphism, where all alleles are same length, and length > 1.ex.ss2421179 AT/GA

Col11: mapLocType: The alignment type at snp site. Possible values and meanings are: 1 Insertion on contig: snp is always represented as one base and this one base in the snp sequence is substituted with more than one bases on the contig sequence in the alignment. 2 Contig allele is one base long.snp is always represented as one base and this one base in the snp sequence is substituted with exactly one base on the contig. 3 Deletion on the contig: part of the snp flanking sequence including the snp was absent on the contig sequence in the alignment. 4 In the alignment, part of the snp flanking sequence including snp is replaced with the contig sequence of longer length. 5 In the alignment, part of the snp flanking sequence including snp is replaced with the contig sequence of exactly the same length. 6 In the alignment, part of the snp flanking sequence including snp is replaced with the contig sequence of a shorter length.

Col12: mapLocCnt: the total number of locations the snp maps to within the assembly. Col13: Mapweight: A number that codes for the mapping quality of the snp on each assembly: 1 = snp aligns at exactly one location 2 = snp aligns at two locus on same chromosome 3 = snp aligns at two locus on different chromosomes or more than 3 and less than 10 locations 10= snp aligns at 10 or more 10 locations

Col14: ContigLabel: This is used to show when a snp maps to alternative haplotypes or PAR region. Possible values are: DR53 PAR c22_H2 c5_H2 c6_COX c6_QBL mitochondrial genome reference

col15: unPlacedContig: This field only has value when a snp hits an unplaced contig, there is no chromosome positions for the snp. chrPosFrom and chrPosTo will be NULL. In this case, unPlacedContig will have the actually contig accession that is unplaced on a chromosome.

Source

1000 genomes pilot data

Examples

```
data(c6snp)
c6snp[1:3,]
```

gw6c6.snp240

data frame with snp annotation regarding chr6, pertinent to the affy genomewide6 SNP chip

Description

data frame with snp annotation regarding chr6, pertinent to the affy genomewide6 SNP chip

Usage

```
data(gw6c6.snp240)
```

Format

A data frame with 56271 observations on the following 7 variables.

```
man_fsetid a character vector
dbsnp_rs_id a character vector
physical_pos a numeric vector
strand a numeric vector
allele_a a character vector
allele_b a character vector
call240 a numeric vector
```

Details

an extract from the pd.genomewidesnp.6 metadata

Examples

```
data(gw6c6.snp240)
str(gw6c6.snp240)
```

n240

NA19240 – 3 million reads from chromosome 6 derived from 1000 genomes bam file

Description

NA19240 – 3 million reads from chromosome 6 derived from 1000 genomes bam file

Usage

```
data(n240)
```

Format

The format is:

List of 1

\$:List of 13

```
..$ qname : chr [1:3000000] "EAS254_13:7:88:1639:15041" "EAS139_43:2:31:1128:9551" "EAS254_13:8:68:520:6861"
"BGI-FC20AHFAAXX_6_26_477:352" ...
```

```
..$ flag : int [1:3000000] 35 35 35 16 35 35 35 0 35 35 ...
```

```
..$ rname : Factor w/ 1 level "6": 1 1 1 1 1 1 1 1 1 1 ...
```

```
..$ strand: Factor w/ 3 levels "-", "+", "*": 2 2 2 1 2 2 2 2 2 2 ...
```

```
..$ pos : int [1:3000000] 5001 5002 5004 5004 5005 5010 5012 5018 5018 5023 ...
```

```
..$ qwidth: int [1:3000000] 51 51 51 36 51 51 36 36 36 45 ...
```

```
..$ mapq : int [1:3000000] 0 0 0 0 0 0 0 0 0 0 ...
```

```
..$ cigar : chr [1:3000000] "51M" "51M" "51M" "36M" ...
```

```
..$ mrnm : Factor w/ 1 level "6": 1 1 1 NA 1 1 1 NA 1 1 ...
```

```
..$ mpos : int [1:3000000] 5163 5203 5170 NA 5156 5183 5199 NA 5252 5273 ...
```

```
..$ isize : int [1:3000000] 214 253 218 NA 203 225 224 NA 271 296 ...
```

```
..$ seq :Formal class 'DNAStringSet' [package "Biostrings"] with 5 slots
```

```

.. ..@ super :Formal class 'DNAStrng' [package "Biostrings"] with 6 slots
.. ..@ xdata :Formal class 'RawPtr' [package "IRanges"] with 2 slots
.. ..@ xp :<externalptr>
.. ..@ .link_to_cached_object:<environment: 0xc5354b8>
.. ..@ offset : int 0
.. ..@ length : int 118321039
.. ..@ elementMetadata: NULL
.. ..@ elementType : chr "ANYTHING"
.. ..@ metadata : list()
.. ..@ ranges :Formal class 'IRanges' [package "IRanges"] with 6 slots
.. ..@ start : int [1:3000000] 1 52 103 154 190 241 292 328 364 400 ...
.. ..@ width : int [1:3000000] 51 51 51 36 51 51 36 36 36 45 ...
.. ..@ NAMES : NULL
.. ..@ elementMetadata: NULL
.. ..@ elementType : chr "integer"
.. ..@ metadata : list()
.. ..@ elementMetadata: NULL
.. ..@ elementType : chr "ANYTHING"
.. ..@ metadata : list()
..$ qual :Formal class 'PhredQuality' [package "Biostrings"] with 5 slots
.. ..@ super :Formal class 'BString' [package "Biostrings"] with 6 slots
.. ..@ xdata :Formal class 'RawPtr' [package "IRanges"] with 2 slots
.. ..@ xp :<externalptr>
.. ..@ .link_to_cached_object:<environment: 0xc5354b8>
.. ..@ offset : int 0
.. ..@ length : int 118321039
.. ..@ elementMetadata: NULL
.. ..@ elementType : chr "ANYTHING"
.. ..@ metadata : list()
.. ..@ ranges :Formal class 'IRanges' [package "IRanges"] with 6 slots
.. ..@ start : int [1:3000000] 1 52 103 154 190 241 292 328 364 400 ...
.. ..@ width : int [1:3000000] 51 51 51 36 51 51 36 36 36 45 ...
.. ..@ NAMES : NULL
.. ..@ elementMetadata: NULL
.. ..@ elementType : chr "integer"
.. ..@ metadata : list()
.. ..@ elementMetadata: NULL
.. ..@ elementType : chr "ANYTHING"
.. ..@ metadata : list()

```

Details

result of Rsamtools scanBam applied to a 3mm line excerpt from the SLX chr6 aligned reads for NA19240

update october 2010 to reflect modifications to cigar handling in Rsamtools

Source

1000 genomes pilot data

Examples

```
data(n240)
sapply(n240[[1]], class)
```

oregdf	<i>data frame representing some of the OREGanno track of UCSC browser</i>
--------	---

Description

data frame representing some of the OREGanno track of UCSC browser

Usage

```
data(oregdf)
```

Format

A data frame with 86 observations on the following 7 variables.

space a factor with levels chr6

start a numeric vector

end a numeric vector

width a numeric vector

name a factor with levels OREG0004577 OREG0004578 ...

score a numeric vector

strand a factor with levels +

Examples

```
data(oregdf)
oregdf[1:5,]
```

pup240_500k	<i>data frame on a pileup for 500000 variants noted in NA19240</i>
-------------	--

Description

data frame on a pileup for 500000 variants noted in NA19240

Usage

```
data(pup240_500k)
```

Format

A data frame with 500000 observations on the following 5 variables.

V2 a numeric vector
V3 a character vector
V4 a character vector
V5 a numeric vector
V9 a character vector

Examples

```
data(pup240_500k)  
pup240_500k[1:5,]
```

pup240_disc	<i>parsed samtools pileup result excerpt</i>
-------------	--

Description

parsed samtools pileup result excerpt – only locations where the called base differs from the reference

Usage

```
data(pup240_disc)
```

Format

A data frame with 31243 observations on the following 5 variables.

loc location
ref reference base
indiv call for NA19240
depth coverage depth at this base
pileup pileup

Details

asterisks are to denote deletions...

Examples

```
data(pup240_disc)  
pup240_disc[1:4,]
```

`rsgdf`*slice of information on transcripts, for ind1KG vignette*

Description

slice of information on transcripts, for ind1KG vignette

Usage

```
data(rsgdf)
```

Format

A data frame with 73 observations on the following 13 variables.

`space` a factor with levels chr6

`start` a numeric vector

`end` a numeric vector

`width` a numeric vector

`name` a factor with levels NM_000129 NM_000904 NM_001003698 NM_001003699 ...

`score` a numeric vector

`strand` a factor with levels + -

`thickStart` a numeric vector

`thickEnd` a numeric vector

`color` a numeric vector

`blockCount` a numeric vector

`blockSizes` a factor with levels 1013,192,167,230,156,80,739, 1013,192,242,230,156,80,739,
1026, 106,119,123,213,90,164,145,137,190,2911,165,798,2949, ...

`blockStarts` a factor with levels 0, 0,10224,16062,19437,21152,22478,25840,27681,28397,30622,31385,3498
0,107199,143191,169690,283829,351818,509940,, ...

Examples

```
data(rsgdf)  
rsgdf[1:4,]
```

yri240_6

*snp.matrix representation of called SNP on chr6 for NA19240***Description**

snp.matrix representation of called SNP on chr6 for NA19240

Usage

```
data(yri240_6)
```

Format

The format is: List of 2 \$ hm2 :Formal class 'snp.matrix' [package "snpMatrix"] with 1 slots
@ .Data: raw [1, 1:265955] 01 01 03 02- attr(*, "dimnames")=List of 2
\$: chr "NA19240"\$: chr [1:265955] "rs4097465" "rs7754266" "rs9393087"
 "rs12192290" ... \$ supp:'data.frame': 265955 obs. of 5 variables: ..\$ dbSNPalleles: Factor w/
 6 levels "A/C","A/G","A/T",,..: 6 2 5 3 1 4 6 1 4 5\$ Assignment : Factor w/ 6 levels
 "A/C","A/G","A/T",,..: 6 2 5 3 1 4 6 1 4 5\$ Chromosome : Factor w/ 1 level "chr6": 1 1 1
 1 1 1 1 1 1\$ Position : int [1:265955] 37012 94609 94901 95272 96774 97749 98500 99536
 99694 99750\$ Strand : Factor w/ 2 levels "+","-": 2 1 1 1 1 1 2 1 1 2 ...

Details

import of hapmap data using read.hapmap.snps

Examples

```
data(yri240_6)
names(yri240_6)
```

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