XtraSNPlocs.Hsapiens.dbSNP144.GRCh38

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`XtraSNPlocs.Hsapiens.dbSNP144.GRCh38`
The XtraSNPlocs.Hsapiens.dbSNP144.GRCh38 package

Description

Extra SNP locations and alleles for Homo sapiens extracted from NCBI dbSNP Build 144. The source data files used for this package were created by NCBI on May 30, 2015, and contain SNPs mapped to reference genome GRCh38.p2 (a patched version of GRCh38 that doesn't alter chromosomes 1-22, X, Y, MT).

While the **SNPlocs.Hsapiens.dbSNP144.GRCh38** package contains only molecular variations of class *snp*, this package contains molecular variations of other classes (*in-del*, *heterozygous*, *microsatellite*, *named-locus*, *no-variation*, *mixed*, and *multinucleotide-polymorphism*).

Details

SNPs from dbSNP were filtered to keep only those satisfying the 3 following criteria:

- The SNP is NOT a single-base substitution (i.e. its class is NOT *snp*) but is a molecular variation that belongs to any other class supported by dbSNP: *in-del*, *heterozygous*, *microsatellite*, *named-locus*, *no-variation*, *mixed*, or *multinucleotide-polymorphism*.
- The SNP is marked as notwithdrawn.
- A *single* location on the reference genome (GRCh38.p2) is reported for the SNP, and this location is on chromosomes 1-22, X, Y, or MT.

Note

The source data files used for this package are the same as those used for the **SNPlocs.Hsapiens.dbSNP144.GRCh38** package and were created by the dbSNP Development Team at NCBI on May 30, 2015.

Author(s)

H. Pages

References

SNP Home at NCBI: http://www.ncbi.nlm.nih.gov/snp

dbSNP Human BUILD 144 announcement: http://www.ncbi.nlm.nih.gov/mailman/pipermail/dbsnp-announce/2015q2/000163.html

GRCh38.p2 assembly: http://www.ncbi.nlm.nih.gov/assembly/GCF_000001405.28/

hg38 genome at UCSC: http://genome.ucsc.edu/cgi-bin/hgGateway?db=hg38

Note that hg38 and GRCh38 are the same assemblies (i.e. the 455 genomic sequences in both of them are the same), except that they use different conventions to name the sequences (i.e. for the chromosome and scaffold names).

See Also

- The **SNPlocs.Hsapiens.dbSNP144.GRCh38** package for SNPs of class *snp*.
- XtraSNPlocs objects in the **BSgenome** software package for how to access the data stored in this package.
- The GRanges class in the **GenomicRanges** package.
- The **VariantAnnotation** software package to annotate variants with respect to location and amino acid coding.

Examples

```
## ---------------------------------------------------------------------
## A. BASIC USAGE
## ---------------------------------------------------------------------
snps <- XtraSNPlocs.Hsapiens.dbSNP144.GRCh38
snpcount(snps)

## Get the location, RefSNP id, and alleles for all "extra SNPs" on
## chromosome 22 and MT:
my_snps1 <- snpsBySeqname(snps, c("ch22", "chMT"), c("RefSNP_id", "alleles"))
my_snps1

## Get the location and alleles for some RefSNP ids:
my_rsids <- c("rs367617508", "rs398104919", "rs3831697", "rs372470289",
              "rs141568169", "rs34628976", "rs67551854")
my_snps2 <- snpsById(snps, my_rsids, c("RefSNP_id", "alleles"))
my_snps2

## ---------------------------------------------------------------------
## B. COMPUTE AND ADD REFERENCE ALLELE AS AN ADDITIONAL METADATA COLUMN
## ---------------------------------------------------------------------
library(BSgenome.Hsapiens.UCSC.hg38)
genome <- BSgenome.Hsapiens.UCSC.hg38

## Before we can call getSeq(genome, my_snps1), we need to harmonize the
## seqinfo components of 'genome' and 'my_snps1':
seqlevelsStyle(my_snps1)  # dbSNP
seqlevelsStyle(genome)  # UCSC
```

```
seqlevelsStyle(my_snps1) <- seqlevelsStyle(genome)
genome(my_snps1) <- "hg38"

ref_allele1 <- getSeq(genome, my_snps1)
ref_allele1[ref_allele1 == ""] <- "-"
mcols(my_snps1)$ref_allele <- ref_allele1
my_snps1

## ---------------------------------------------------------------------
## C. COMPARE ALLELES REPORTED BY dbSNP WITH REFERENCE ALLELE
## ---------------------------------------------------------------------
alleles1 <- mcols(my_snps1)$alleles
alleles1 <- CharacterList(strsplit(alleles1, "/", fixed=TRUE))
disagrees_idx <- which(all(as.character(ref_allele1) != alleles1))
my_snps1[disagrees_idx]
length(disagrees_idx) / length(my_snps1)  # 0.003261601
## Conclusion: 0.33% of the "extra SNPs" in dbSNP have reported alleles
## that disagree with the computed reference allele :-/
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