SNPlocs.Hsapiens.dbSNP155.GRCh38

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`SNPlocs.Hsapiens.dbSNP155.GRCh38`
The SNPlocs.Hsapiens.dbSNP155.GRCh38 package

Description

Human SNP locations and alleles extracted from dbSNP Build 155 and placed on the GRCh38/hg38 assembly

Details

The 949,021,448 SNPs in this package were extracted from the RefSNP JSON files for chromosomes 1-22, X, Y, and MT, located at `https://ftp.ncbi.nih.gov/snp/archive/b155/JSON/` (these files were created by NCBI in May 2021).

These SNPs are compatible with packages **BSgenome.Hsapiens.NCBI.GRCh38** and **BSgenome.Hsapiens.UCSC.hg38** that is, they can be "injected" in the BSgenome objects contained in these packages.

SNP positions and alleles are reported with respect to the *plus* strand.

Only SNPs of type *snv* (*single-nucleotide* variant a.k.a. *single-base substitution*) were kept. Other variant types supported by dbSNP are: *delins* (indel), *ins* (insertion), *del* (deletion), and *mnv* (multiple nucleotide variation). These other variants are NOT included in **SNPlocs.Hsapiens.dbSNP155.GRCh38** but are available in the **XtraSNPlocs.Hsapiens.dbSNP155.GRCh38** package.

Note

The SNPs in this package can be "injected" in `BSgenome.Hsapiens.NCBI.GRCh38` or `BSgenome.Hsapiens.UCSC.hg38`, and will land at the correct positions.

See `?injectSNPs` in the **BSgenome** software package for more information about the SNP injection mechanism.

Author(s)

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References

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

dbSNP Human Build 155 Release announcement (June 22, 2021): https://www.ncbi.nlm.nih.gov/mailman/pipermail/dbsnp-announce/2021q2/000229.html

The GRCh38.p13 assembly: https://www.ncbi.nlm.nih.gov/assembly/GCF_000001405.39/

The hg38 genome at UCSC (based on GRCh38.p13, as of April 2022, but the UCSC folks could change this in the future and base hg38 on a more recent patch release of GRCh38): http://genome.ucsc.edu/cgi-bin/hgGateway?db=hg38

See Also

- The **XtraSNPlocs.Hsapiens.dbSNP155.GRCh38** package for SNPs of type other than *snv*.
- snpcount in the **BSgenome** software package for how to access the data stored in this package.
- IUPAC_CODE_MAP in the **Biostrings** package.
- The GPos class in the **GenomicRanges** package.
- injectSNPs in the **BSgenome** software package for SNP injection.
- The **VariantAnnotation** software package to annotate variants with respect to location and amino acid coding.

Examples

```
## ---------------------------------------------------------------------
## A. BASIC USAGE
## ---------------------------------------------------------------------

snps <- SNPlocs.Hsapiens.dbSNP155.GRCh38
snpcount(snps)
seqinfo(snps)

## Get the positions and alleles of all SNPs on chromosomes 22 and MT:
snpsBySeqname(snps, seqnames=c("22", "MT"))

## Get the positions and alleles of all SNPs within some regions:
snpsByOverlaps(snps, GRanges(c("Y:230001-232000", "19:88501-89000")))

## ---------------------------------------------------------------------
## B. EXTRACT SNP INFORMATION FOR A SET OF RS IDS
## ---------------------------------------------------------------------

my_rsids <- c("rs2639606",  "rs75264089", "rs73396229", "rs55871206",
              "rs10932221", "rs56219727", "rs73709730", "rs55838886",
              "rs3734153",  "rs79381275", "rs1516535",  "rs74342513")

## Note that the first call to snpsById() takes a long time but
## subsequent calls are faster.
my_snps <- snpsById(snps, my_rsids)
my_snps

## Translate the IUPAC ambiguity codes used to represent the alleles
## into nucleotides:
IUPAC_CODE_MAP[mcols(my_snps)$alleles_as_ambig]
```

```
## ---------------------------------------------------------------------
## C. INJECTION IN THE REFERENCE GENOME
## ---------------------------------------------------------------------

library(BSgenome.Hsapiens.UCSC.hg38)
ref_genome <- BSgenome.Hsapiens.UCSC.hg38
ref_genome

alt_genome <- injectSNPs(ref_genome, "SNPlocs.Hsapiens.dbSNP155.GRCh38")
alt_genome  # note the additional line "with SNPs injected from..."

alphabetFrequency(ref_genome$chr22)
alphabetFrequency(alt_genome$chr22)

## Get the number of nucleotides that were modified by this injection:
neditAt(ref_genome$chr22, alt_genome$chr22)  # 12798921
```

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