SNPlocs.Hsapiens.dbSNP155.GRCh38

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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</pre>
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
"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)</pre>
my_snps
## Translate the IUPAC ambiguity codes used to represent the alleles
## into nucleotides:
IUPAC_CODE_MAP[mcols(my_snps)$alleles_as_ambig]
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