

RSNPper: utilities for SNP data

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1 Introduction

This document describes *RSNPper* version 1.0, added to Bioconductor in October of 2003. This first version focuses on SNP metadata, with functions that retrieve SNP-related data from the Boston Children’s Hospital Informatics Program *SNPper* web service ?.

Earlier non-released versions of this package included considerable code for working with *prettybase* format and for conducting other tasks in SNP discovery projects. That material has been moved to *inst/OLD* and may be re-introduced later. Users seeking legacy support should contact the author.

2 How it works

The core of this package is the XML-RPC service at CHIP accessible through the following URL stub:

```
> print(.SNPperBaseURL)
```

```
[1] "http://snpper.chip.org/bio/rpcserv/dummy?cmd="
```

The `useSNPper` function allows you to work directly with the XML-RPC server by packing up appropriate command and argument strings.

```
> dput(useSNPper)
```

```
function (cmd, parmstring)
```

```
{
```

```
  targ <- url(paste(.SNPperBaseURL, cmd, parmstring, sep = ""))
```

```
  open(targ)
```

```
  on.exit(close(targ))
```

```
  readLines(targ)
```

```
}
```

```
> print(useSNPper("geneinfo", "&name=CRP")[1:7])
```

```
[1] " <SNPPER-RPC SOURCE=\"*RPCSERV-NAME*\" VERSION=\"$$Revision: 1.1.1.1 $\" GENOME=\"h
```

```
[2] " <GENEINFO>"
```

```
[3] " <GENE ID=\"1735\">"
```

```
[4] " <GENEID>1735</GENEID>"
```

```
[5] " <NAME>CRP</NAME>"
```

```
[6] " <CHROM>chr1</CHROM>"
```

```
[7] " <STRAND>-</STRAND>"
```

The main functions of *RSNPper* attend to simplifying specification of parameters and parsing and packaging the XML results.

Note on auditability. All functions return textual information coupled with auditing information as a 'toolInfo' attribute, detailing the SNPper supplied information on the human genome sequence build, the dbSNP version, and the SNPper version from which the results are obtained. At present, there is one exception: when `itemsInRange` is invoked with `item='countsnps'`, no toolInfo data is obtained. This will be corrected once the `countsnps` command at SNPper returns valid XML element tags.

3 Overview of the functions

The current set of functions intended for investigative use is:

- `geneInfo` – general information about location and nomenclature
- `geneLayout` – information about exon locations
- `geneSNPs` – all SNPs associated with a given gene
- `SNPinfo` – detailed information on a SNP
- `itemsInRange` – supports chromosome scanning for genes, SNPs, or counts of SNPs

An omission: for SNP information, I have not collected information on submitter.

4 Demonstrations

4.1 Obtaining information on genes

The `geneInfo` function will collect some basic information on a gene. The gene may be specified by HUGO name, mRNA accession number, or SNPper id.

```
> print(geneInfo("CRP"))
```

```
SNPper Gene metadata:
```

```
There are 1 entries.
```

```
Basic information:
```

GENEID	NAME	CHROM	STRAND	PRODUCT	NSNPS		
1	1735	CRP	chr1	- C-reactive protein, pentraxin-related	141		
	TX.START	TX.END	CODSEQ.START	CODSEQ.END	LOCUSLINK	OMIM	UNIGENE
1	157948704	157951003	157949939	157950899	1401	123260	Hs.76452
	SWISSPROT	MRNAACC	PROTACC	REFSEQACC			
1	P02741	NM_000567	NP_000558	NULL			

```
SNPper info:
```

SOURCE	VERSION	GENOME	DBSNP
[1,] "*RPCSERV-NAME*"	"\$Revision: 1.1.1.1 \$"	"hg18"	"125"

The `geneLayout` function provides information on exon locations.

```
> print(geneLayout("546"))
```

ID	NAME	CHROM	TRANSCRIPT.START
" "	"S100BPB"	"chr1"	"33055763"
CODINGSEQ.START	TRANSCRIPT.END	CODINGSEQ.END	exon1.start
"33064288"	"33097062"	"33094226"	"33055763"
exon1.end	exon2.start	exon2.end	exon3.start
"33055877"	"33063501"	"33063617"	"33064286"
exon3.end	exon4.start	exon4.end	exon5.start
"33065118"	"33066181"	"33066269"	"33068152"
exon5.end	exon6.start	exon6.end	exon7.start
"33068255"	"33091267"	"33091354"	"33094112"
exon7.end			
"33097062"			

```
attr(,"toolInfo")
```

SOURCE	VERSION	GENOME
"*RPCSERV-NAME*"	"\$Revision: 1.1.1.1 \$"	"hg18"

DBSNP
"125"

Information on all the genes catalogued in a certain chromosomal region can be obtained using `itemsInRange`.

```
> print(itemsInRange("genes", "chr1", "156400000", "156500000"))
```

```
[[1]]
              NAME                CHROM
              "CD1D"              "chr1"
              PRODUCT              NSNPS
"CD1D antigen, d polypeptide"    "114"

[[2]]
              NAME                CHROM                PRODUCT
              "CD1A"              "chr1" "CD1A antigen precursor"
              NSNPS
              "53"
```

```
$CHR
[1] "chr1"
```

```
$START
[1] "156400000"
```

```
$END
[1] "156500000"
```

```
$COUNT
[1] "2"
```

```
attr(,"toolInfo")
              SOURCE                VERSION                GENOME
              "*RPCSERV-NAME*" "$Revision: 1.1.1.1 $"          "hg18"
              DBSNP
              "125"
```

4.2 Obtaining information on SNPs

Suppose you want information on the SNP with dbSNP id rs25.

```
> print(SNPinfo("25"))
```

SNPper SNP metadata:

```
DBSNPID CHROMOSOME POSITION ALLELES VALIDATED
```

```
[1,] "rs25" "chr7" "11550667" "A/G" "Y"
```

There are details on 5 populations
and 1 connections to gene features

SNPper info:

```
      SOURCE          VERSION          GENOME DBSNP  
[1,] "*RPCSERV-NAME*" "$Revision: 1.1.1.1 $" "hg18" "125"
```

Suppose instead you want information on all the SNPs cataloged in a certain chromosomal region.

```
> ird <- itemsInRange("snps", "chr1", "156400000", "156500000")  
> print(length(ird))
```

```
[1] 557
```

```
> print(ird[1:3])
```

```
[[1]]
```

DBSNPID	TSCID	CHROMOSOME	POSITION	ALLELES	ROLE
"rs16839876"	" "	"chr1"	"156400131"	"A/T"	" "
RELPOS	AMINO	AMINOPOS			
" "	" "	" "	" "		

```
[[2]]
```

DBSNPID	TSCID	CHROMOSOME	POSITION	ALLELES	ROLE
"rs12117055"	" "	"chr1"	"156400300"	"C/T"	" "
RELPOS	AMINO	AMINOPOS			
" "	" "	" "	" "		

```
[[3]]
```

DBSNPID	TSCID	CHROMOSOME	POSITION	ALLELES	ROLE
"rs17455763"	" "	"chr1"	"156400743"	"A/T"	" "
RELPOS	AMINO	AMINOPOS			
" "	" "	" "	" "		

Note that the start and end locations are supplied as strings. This is to avoid coercion to textual scientific notation.

Additional detail on the count of SNPs can be obtained more briefly:

```
> print(itemsInRange("countsnps", "chr1", "156400000", "156500000"))
```

```
total exonic nonsyn  
553    12    2
```

To see all the SNPs associated with a given gene, use the `geneSNPs` function. This requires knowledge of the SNP per gene id, which can be obtained using `geneInfo`.

```
> gs <- geneSNPs("546")
> print(length(gs))
```

```
[1] 150
```

```
> print(gs[1:3])
```

```
[[1]]
```

DBSNPID	TSCID
"rs11809784"	" "
CHROMOSOME	POSITION
"chr1"	"33046120"
ALLELES	ROLE
"A/C"	"Promoter"
RELPOS	AMINO
"-18168"	" "
AMINOPOS	HUGO
" "	"S100PBP"
LOCUSLINK	NAME
"64766"	"S100P binding protein Riken isoform a"
MRNA	
"NM_022753"	

```
[[2]]
```

DBSNPID	TSCID
"rs4422972"	" "
CHROMOSOME	POSITION
"chr1"	"33046500"
ALLELES	ROLE
"G/T"	"Promoter"
RELPOS	AMINO
"-17788"	" "
AMINOPOS	HUGO
" "	"S100PBP"
LOCUSLINK	NAME
"64766"	"S100P binding protein Riken isoform a"
MRNA	
"NM_022753"	

```
[[3]]
```

DBSNPID	TSCID
"rs3845499"	" "
CHROMOSOME	POSITION
"chr1"	"33047367"
ALLELES	ROLE
"A/G"	"Promoter"
RELPOS	AMINO
"-16921"	" "
AMINOPOS	HUGO
" "	"S100PBP"
LOCUSLINK	NAME
"64766"	"S100P binding protein Riken isoform a"
MRNA	
"NM_022753"	

5 Application: SNP density on chr 1

Human chromosome 1 is approximately 300Mb, and 142,629 SNPs have been recorded as of dbSNP build 106, according to NCBI SNP/maplists/maplist-newmap.html on 13 Sep 03. Let's see if these facilities can recover this sort of information. Counting the number of SNPs on a long chromosomal region seems to take a long time for SNPper, so we will break up the task.

```
> print(itemsInRange("countsnp", "chr1", "1", "100000"))

total exonic nonsyn
  340    23    3

> system("sleep 2")
> print(itemsInRange("countsnp", "chr1", "100001", "200000"))

total exonic nonsyn
   28     0     0

> system("sleep 2")
> print(itemsInRange("countsnp", "chr1", "200001", "300000"))

total exonic nonsyn
   48     0     0

> system("sleep 2")
```

These runs complete in a reasonable amount of time. Here we will just look at the first 2Mb in intervals of .1Mb.

```

> starts <- as.character(as.integer(seq(1, 2000001, 1e+05)))
> ends <- as.character(as.integer(as.integer(starts) + 99999))
> out <- matrix(NA, nr = 20, nc = 3)
> for (i in 1:20) {
+   cat(i)
+   out[i, ] <- itemsInRange("countsnps", "chr1", starts[i],
+     ends[i])
+   system("sleep 2")
+ }

```

1234567891011121314151617181920

```

> print(out)

```

	[,1]	[,2]	[,3]
[1,]	340	23	3
[2,]	28	0	0
[3,]	48	0	0
[4,]	14	0	0
[5,]	4	0	0
[6,]	288	0	0
[7,]	49	0	0
[8,]	683	12	1
[9,]	454	39	14
[10,]	461	45	12
[11,]	359	13	0
[12,]	442	54	16
[13,]	400	75	25
[14,]	340	36	6
[15,]	431	38	12
[16,]	292	10	2
[17,]	385	37	9
[18,]	201	7	1
[19,]	405	20	4
[20,]	414	9	3