

MyVariant.info R Client

Adam Mark, Chunlei Wu

May 19, 2021

Contents

- 1 Overview 1
- 2 Variant Annotation Service 2
 - 2.1 Obtaining HGVS IDs from a VCF file. 2
 - 2.2 `getVariant` 3
 - 2.3 `getVariants` 3
- 3 Variant Query Service 4
 - 3.1 `queryVariant`. 4
 - 3.2 `queryVariants` 4
- 4 References 5

1 Overview

MyVariant.Info is a simple-to-use REST web service to query/retrieve genetic variant annotation from an aggregation of variant annotation resources. *myvariant* is an easy-to-use R wrapper to access MyVariant.Info services and explore variant annotations.

2 Variant Annotation Service

2.1 Obtaining HGVS IDs from a VCF file.

- Use `readVcf` from the VariantAnnotation package to read a Vcf file in. The Vcf object can then be passed to `formatHgvs` to retrieve HGVS IDs. HGVS IDs are based on the GRCh38/hg19 reference genome. Support for hg38 is coming soon.

```
> file.path <- system.file("extdata", "dbSNP_mini.vcf", package="myvariant")
> vcf <- readVcf(file.path, genome="hg19")
> rowRanges(vcf)
```

GRanges object with 240 ranges and 5 metadata columns:

	seqnames	ranges	strand	paramRangeID	REF
	<Rle>	<IRanges>	<Rle>	<factor>	<DNAStringSet>
rs376643643	1	10019-10020	*	NA	TA
rs373328635	1	10055	*	NA	T
rs62651026	1	10108	*	NA	C
rs376007522	1	10109	*	NA	A
rs368469931	1	10139	*	NA	A
...
rs544020171	1	17654	*	NA	T
rs563880190	1	17694	*	NA	C
rs574335987	1	17695	*	NA	G
rs374995955	1	17697	*	NA	G
rs543363182	1	17709	*	NA	T
		ALT	QUAL	FILTER	
		<DNAStringSetList>	<numeric>	<character>	
rs376643643		T	NA	.	
rs373328635		TA	NA	.	
rs62651026		T	NA	.	
rs376007522		T	NA	.	
rs368469931		T	NA	.	
...		
rs544020171		C	NA	.	
rs563880190		T	NA	.	
rs574335987		A	NA	.	
rs374995955		C	NA	.	
rs543363182		G	NA	.	

```
-----
seqinfo: 1 sequence from hg19 genome; no seqlengths
```

MyVariant.info R Client

- You can then use `formatHgvs` to extract HGVS IDs from the Vcf object.

```
> hgvs <- formatHgvs(vcf, variant_type="snp")
> head(hgvs)

[1] "1:g.10108C>T" "1:g.10109A>T" "1:g.10139A>T" "1:g.10150C>T" "1:g.10177A>C"
[6] "1:g.10180T>C"
```

2.2 `getVariant`

- Use `getVariant`, the wrapper for GET query of `"/v1/variant/<hgvsid>"` service, to return the variant object for the given HGVS id.

```
> variant <- getVariant("chr1:g.35367G>A")
> variant[[1]]$dbnsfp$genename

NULL

> variant[[1]]$cadd$phred

[1] 3.726
```

2.3 `getVariants`

- Use `getVariants`, the wrapper for POST query of `"/v1/variant"` service, to return the list of variant objects for the given character vector of HGVS ids.

```
> getVariants(c("chr1:g.35367G>A", "chr16:g.28883241A>G"),
+             fields="cadd.consequence")

DataFrame with 2 rows and 4 columns
      query                X_id                cadd._license
  <character>          <character>          <character>
1 chr1:g.35367G>A    chr1:g.35367G>A http://bit.ly/2TIuab9
2 chr16:g.28883241A>G chr16:g.28883241A>G http://bit.ly/2TIuab9
  cadd.consequence
  <character>
1 NONCODING_CHANGE
2 NON_SYNONYMOUS
```

3 Variant Query Service

3.1 queryVariant

- `queryVariant` is a wrapper for GET query of `"/v1/query?q=<query>"` service, to return the query result. This function accepts wild card input terms and allows you to query for variants that contain a specific annotation. For example, the following query searches for the CADD phred score and consequence for all variants whose genename (dbNSFP) is MLL2.

```
> queryVariant(q="dbnsfp.genename:MLL2", fields=c("cadd.phred", "cadd.consequence"))

$took
[1] 3

$total
[1] 0

$max_score
NULL

$hits
list()
```

- You can also use `queryVariant` to retrieve all annotations that map to a specific rsID.

```
> queryVariant(q="rs58991260", fields="dbSNP.flags")$hits
      _id  _score
1 chr1:g.218631822G>A 15.7897
```

3.2 queryVariants

- `queryVariants` is a wrapper for POST query of `"/v1/query?q=<query>"` service, to return the query result. Query terms include any available field as long as scopes are defined. The following example reads the dbSNP rsIDs from a VCF and queries for all fields. The returned DataFrame can then be easily subsetted to include, for example, those that have not been documented in the Welllderly study.

```
> rsids <- paste("rs", info(vcf)$RS, sep="")
> res <- queryVariants(q=rsids, scopes="dbSNP.rsid", fields="all")
```

Finished

Pass `returnall=TRUE` to return lists of duplicate or missing query terms.

```
> subset(res, !is.na(wellderly.vartype))$query
```

```
[1] "rs145427775" "rs55998931" "rs199606420" "rs58108140" "rs62635284"  
[6] "rs62635286" "rs531730856" "rs527952245" "rs546169444" "rs201055865"  
[11] "rs62635298" "rs199856693" "rs201855936" "rs71252251" "rs201045431"  
[16] "rs201635489" "rs533630043" "rs2691315" "rs572465511" "rs372319358"  
[21] "rs11489794" "rs113141985" "rs148220436" "rs150723783" "rs62636367"  
[26] "rs62636368" "rs199745162" "rs200658479" "rs201833382" "rs199740902"  
[31] "rs200978805" "rs201535981" "rs192890528"
```

4 References

MyVariant.info help@myvariant.info