

MyVariant.info R Client

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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", "dbssnp_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, 10055]	*	<NA>	T
rs62651026	1	[10108, 10108]	*	<NA>	C
rs376007522	1	[10109, 10109]	*	<NA>	A
rs368469931	1	[10139, 10139]	*	<NA>	A
...
rs544020171	1	[17654, 17654]	*	<NA>	T
rs563880190	1	[17694, 17694]	*	<NA>	C
rs574335987	1	[17695, 17695]	*	<NA>	G
rs374995955	1	[17697, 17697]	*	<NA>	G
rs543363182	1	[17709, 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

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

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

```
[1] "chr1:g.10108C>T" "chr1:g.10109A>T" "chr1:g.10139A>T" "chr1:g.10150C>T"
[5] "chr1:g.10177A>C" "chr1: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 5 columns

	X_id	X_score	query	cadd._license	cadd.consequence
	<character>	<numeric>	<character>	<character>	<character>
1	chr1:g.35367G>A	1	chr1:g.35367G>A	http://goo.gl/bkpNhq	NONCODING_CHANGE
2	chr16:g.28883241A>G	1	chr16:g.28883241A>G	http://goo.gl/bkpNhq	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"))
```

```
$hits
list()
```

```
$max_score
NULL
```

```
$took
[1] 6
```

```
$total
[1] 0
```

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

```
> queryVariant(q="rs58991260", fields="dbSNP.flags")$hits
```

	_id	_score	flags
1	chr1:g.218631822G>A	16.93929	ASP, G5, G5A, GN0, KGPhase1, KGPhase3, SLO

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]	"rs367896724"	"rs145427775"	"rs147093981"	"rs56289060"	"rs55998931"	"rs199606420"
[7]	"rs58108140"	"rs62635284"	"rs62635286"	"rs200579949"	"rs531730856"	"rs180734498"
[13]	"rs527952245"	"rs201696125"	"rs546169444"	"rs201055865"	"rs62635298"	"rs79585140"
[19]	"rs199856693"	"rs201855936"	"rs201045431"	"rs71252250"	"rs200030104"	"rs78601809"
[25]	"rs62636497"	"rs62636497"	"rs201635489"	"rs533630043"	"rs2691315"	"rs113442401"
[31]	"rs572465511"	"rs372319358"	"rs200358166"	"rs11489794"	"rs113141985"	"rs62636498"
[37]	"rs148220436"	"rs141130360"	"rs150723783"	"rs201459529"	"rs199676946"	"rs62636367"
[43]	"rs62636368"	"rs200205172"	"rs199745162"	"rs200658479"	"rs201833382"	"rs199740902"
[49]	"rs200978805"	"rs201535981"	"rs192890528"	"rs200046632"	"rs374995955"	

4 References

MyVariant.info help@myvariant.info