

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", "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, 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 4 columns
```

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

```
$max_score
```

```
NULL
```

```
$took
```

```
[1] 2
```

```
$total
```

```
[1] 0
```

```
$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      flags
1 chr1:g.218631822G>A 10.73608 ASP, G5, GNO, 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" "rs140194106" "rs145427775" "rs147093981" "rs56289060" "rs112766696"
 [7] "rs55998931"  "rs199606420" "rs58108140"  "rs62635284"  "rs62635286"  "rs531730856"
[13] "rs527952245" "rs201747181" "rs546169444" "rs201055865" "rs62635298"  "rs199856693"
[19] "rs201855936" "rs71252251"  "rs201045431" "rs201635489" "rs533630043" "rs2691315"
[25] "rs572465511" "rs372319358" "rs11489794"  "rs113141985" "rs148220436" "rs150723783"
[31] "rs62636367"  "rs62636368"  "rs199745162" "rs200658479" "rs201833382" "rs199740902"
[37] "rs200978805" "rs201535981" "rs192890528"
```

4 References

MyVariant.info help@myvariant.info