

# MyVariant.info R Client

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## Contents

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

## 1 Overview

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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", "dbnp_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>	.

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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] "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 gene name (dbNSFP) is MLL2.

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

$max_score
NULL

$took
[1] 3

$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      dbSNP._license
1 chr1:g.218631822G>A 10.36088 https://goo.gl/Ztr5r1
      dbSNP.flags
1 ASP, G5, 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

## MyVariant.info R Client

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="db SNP.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"
[6] "rs112766696" "rs55998931" "rs199606420" "rs58108140" "rs62635284"
[11] "rs62635286" "rs531730856" "rs527952245" "rs201747181" "rs546169444"
[16] "rs201055865" "rs62635298" "rs199856693" "rs201855936" "rs71252251"
[21] "rs201045431" "rs201635489" "rs533630043" "rs2691315" "rs572465511"
[26] "rs372319358" "rs11489794" "rs113141985" "rs148220436" "rs150723783"
[31] "rs62636367" "rs62636368" "rs199745162" "rs200658479" "rs201833382"
[36] "rs199740902" "rs200978805" "rs201535981" "rs192890528"
```

## 4 References

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MyVariant.info [help@myvariant.info](mailto:help@myvariant.info)