
MyVariant.info Documentation

Release v1

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CHAPTER 1

Introduction



MyVariant.info provides simple-to-use REST web services to query/retrieve variant annotation data. It's designed with an emphasis on **simplicity** and **performance**.

MyVariant.info provides two simple web services: one for variant queries and the other for variant annotation retrieval. Both return results in JSON format.

Variant query service

URL

```
http://myvariant.info/v1/query
```

Examples

```
http://myvariant.info/v1/query?q=rs58991260
http://myvariant.info/v1/query?q=chr1:69000-70000
http://myvariant.info/v1/query?q=dbsnp.vartype:snp
http://myvariant.info/v1/query?q=_exists_:dbsnp AND NOT dbsnp.vartype:indel
http://myvariant.info/v1/query?q=dbnsfp.polyphen2.hdiv.score:>0.99 AND chrom:1
http://myvariant.info/v1/query?q=cadd.gene.gene_id:ENSG00000113368&facets=cadd.
↳polyphen.cat&size=0
http://myvariant.info/v1/query?q=_exists_:dbsnp AND _exists_:cosmic
```

Hint: View nicely formatted JSON result in your browser with this handy add-on: [JSON formatter](#) for Chrome or [JSONView](#) for Firefox.

To learn more

- You can read the full description of our query syntax [here](#).

- Try it live on [interactive API page](#).
- Play with our [demo application](#).
- Batch queries? Yes, you can. do it with a POST request.

Variant annotation service

URL

```
http://myvariant.info/v1/variant/<variant_id>
```

“<variant_id>” is an HGVS name based variant id using genomic location based on hg19 human genome assembly..

Examples

```
http://myvariant.info/v1/variant/chr1:g.35367G>A  
http://myvariant.info/v1/variant/chr7:g.55241707G>T  
http://myvariant.info/v1/variant/chr9:g.107620835G>A  
http://myvariant.info/v1/variant/chr1:g.160145907G>T  
http://myvariant.info/v1/variant/chr16:g.28883241A>G  
http://myvariant.info/v1/variant/chr3:g.49721532G>A
```

To learn more

- You can read the full description of our query syntax [here](#).
- Try it live on [interactive API page](#).
- Play with our [demo application](#).
- Yes, batch queries via POST request as well.

Variant annotation data

Data sources

We currently obtain variant annotation data from several data resources and keep them up-to-date, so that you don't have to do it:

Total variants loaded: **334,292,622**

Source	version	# of variants	key name*
dbNSFP	v3.0c	82,030,830	dbnsfp
dbSNP	v144	145,132,257	dbSNP
ClinVar	201601	124,047	clinvar
EVS	v2	1,977,300	evs
CADD	v1.3	226,932,858	cadd
MutDB	-	420,221	mutdb
GWAS Catalog	from UCSC	15,243	gwassnps
COSMIC	v68 from UCSC	1,024,498	cosmic
DOCM	-	1,119	docm
SNPedia	-	5,907	snpedia
EMVClass	-	12,066	emv
Scripps Welllderly	-	21,240,519	welllderly
EXAC	v0.3	10,195,872	exac
GRASP	v2.0.0.0	2,212,148	grasp
UniProt	201703	477,711	uniprot
CIViC	201706	1,559	civic
Cancer Genome Interpreter	2017-07-21	334	cgi
genome Aggregation Database	-	260,657,435	gnomad_genome
genome Aggregation Database	-	17,009,588	gnomad_exome
Geno2MP	2017-06-27	1,857,156	geno2mp

* key name: this is the key for the specific annotation data in a variant object.

The most updated information can be accessed [here](#).

Note: Each data source may have its own usage restrictions (e.g. [CADD](#) data are free for non-commercial use only). Please refer to the data source pages above for their specific restrictions.

Variant object

Variant annotation data are both stored and returned as a variant object, which is essentially a collection of fields (attributes) and their values:

```
{
  "_id": "chr1:g.35367G>A",
  "_version": 2,
  "cadd": {
    "alt": "A",
    "annotype": "NonCodingTranscript",
    "chrom": 1,
    "gene": {
      "cds": {
        "cdna_pos": 476,
        "rel_cdna_pos": 0.4
      },
      "feature_id": "ENST00000417324",
      "gene_id": "ENSG00000237613",
    },
    "ref": "G",
    "type": "SNV"
  },
  "dbnsfp": {
    "aa": {
      "aapos_sift": "ENSP00000409362:P44L",
      "alt": "L",
      "codonpos": 2,
      "pos": 44,
      "ref": "P",
      "refcodon": "CCG"
    },
    "alt": "A",
    "ancestral_allele": "G",
    "chrom": "1",
    "ensembl": {
      "geneid": "ENSG00000237613",
      "transcriptid": "ENST00000417324"
    },
    "genename": "FAM138A",
    "hg19": {
      "end": 35367,
      "start": 35367
    }
  }
}
```

The example above omits many of the available fields. For a full example, check out [this example variant](#), or try the interactive [API page](#).

`_id` field

Each individual variant object contains an “`_id`” field as the primary key. We utilize the recommended nomenclature from [Human Genome Variation Society](#) to define the “`_id`” field in MyVariant.info. Specifically, we use HGVS’s genomic reference sequence notation based on the current reference genome assembly (e.g. hg19 for human). The followings are brief representations of major types of genetic variants. More examples could be found at [HGVS recommendations for the description of DNA sequence variants](#) page.

Note: The default reference genome assembly is always human hg19 in MyVariant.info, so we only use “chr??” to represent the reference genomic sequence in “`_id`” field. The valid chromosomes representations are **chr1**, **chr2**, ..., **chr22**, **chrX**, **chrY** and **chrMT**. Do not use *chr23* for *chrX*, *chr24* for *chrY*, or *chrM* for *chrMT*.

- SNV example:

```
chr1:g.35366C>T
```

The above `_id` represents a C to T SNV on chromosome 1, genomic position 35366.

- Insertion example:

```
chr2:g.17142_17143insA
```

The above `_id` represents that an A is inserted between genomic position 17142 and 17143 on chromosome 2.

- Deletion example:

```
chrMT:g.8271_8279del
```

The above `_id` represents that a nine nucleotides deletion between genomic position 8271 and 8279 on chromosome MT. Note that we don’t include the deleted sequence in the `_id` field in this case.

- Deletion/Insertion example:

```
chrX:g.14112_14117delinsTG
```

The above `_id` represents that six nucleotides between genomic position 14112 and 14117 are replaced by TG.

`_score` field

You will often see a “`_score`” field in the returned variant object, which is the internal score representing how well the query matches the returned variant object. It probably does not mean much in variant annotation service when only one variant object is returned. In variant query service, by default, the returned variant hits are sorted by the scores in descending order.

Available fields

The table below lists all of the possible fields that could be in a variant object, as well as all of their parents (for nested fields). If the field is indexed, it may also be directly queried, e.g.

```
q=dbnsfp.polyphen2.hdiv.score:>0.99
```

All fields can be used with `_exists_` or `_missing_` filters, e.g.

```
q=_exists_:db SNP AND _exists_:cosmic
q=_missing_:welllderly
```

or as inputs to the fields parameter, e.g.

```
q=_exists_:db SNP&fields=db SNP.rsid,db SNP.vartype
```

Data release notes

This page contains metadata about each MyVariant.info data release. Click a link to see more.

MyVariant Releases

Variant query service

This page describes the reference for MyVariant.info variant query web service. It's also recommended to try it live on our [interactive API page](#).

Service endpoint

```
http://myvariant.info/v1/query
```

GET request

Query parameters

q

Required, passing user query. The detailed query syntax for parameter “q” we explained [below](#).

fields

Optional, a comma-separated string to limit the fields returned from the matching variant hits. The supported field names can be found from any variant object (e.g. [here](#)). Note that it supports dot notation, and wildcards as well, e.g., you can pass “dbnsfp”, “dbnsfp.genename”, or “dbnsfp.aa.*”. If “fields=all”, all available fields will be returned. Default: “all”.

size

Optional, the maximum number of matching variant hits to return (with a cap of 1000 at the moment). Default: 10.

from

Optional, the number of matching variant hits to skip, starting from 0. Default: 0

Hint: The combination of “**size**” and “**from**” parameters can be used to get paging for large query:

<code>q=cdk*&size=50</code>	first 50 hits
<code>q=cdk*&size=50&from=50</code>	the next 50 hits

fetch_all

Optional, a boolean, which when TRUE, allows fast retrieval of all unsorted query hits. The return object contains a `_scroll_id` field, which when passed as a parameter to the query endpoint, returns the next 1000 query results. Setting `fetch_all = TRUE` causes the results to be inherently unsorted, therefore the `sort` parameter is ignored. For more information see [examples using fetch_all here](#). Default: FALSE.

scroll_id

Optional, a string containing the `_scroll_id` returned from a query request with `fetch_all = TRUE`. Supplying a valid `scroll_id` will return the next 1000 unordered results. If the next results are not obtained within 1 minute of the previous set of results, the `scroll_id` becomes stale, and a new one must be obtained with another query request with `fetch_all = TRUE`. All other parameters are ignored when the `scroll_id` parameter is supplied. For more information see [examples using scroll_id here](#).

sort

Optional, the comma-separated fields to sort on. Prefix with “-” for descending order, otherwise in ascending order. Default: sort by matching scores in descending order.

facets

Optional, a single field or comma-separated fields to return facets, for example, “`facets=cadd`”, “`facets=cadd,dbsnp.vartype`”. See [examples of faceted queries here](#).

callback

Optional, you can pass a “**callback**” parameter to make a [JSONP](#) call.

email

Optional, if you are regular users of our services, we encourage you to provide us an email, so that we can better track the usage or follow up with you.

Query syntax

Examples of query parameter “**q**”:

Simple queries

search for everything:

```
q=rs58991260 # search for rsid
```

Fielded queries

```
q=chr1:69000-70000 # for a genomic range
q=dbsnfp.vartype:snp # for matching value on a specific field

q=dbnsfp.polyphen2.hdiv.pred:(D P) # multiple values for a field
q=dbnsfp.polyphen2.hdiv.pred:(D OR P) # multiple values for a field using OR

q=_exists_:dbsnfp # having dbsnp field
q=_missing_:exac # missing exac field
```

Hint: For a list of available fields, see [here](#).

Range queries

```
q=dbnsfp.polyphen2.hdiv.score:>0.99
q=dbnsfp.polyphen2.hdiv.score:>=0.99
q=exac.af:<0.00001
q=exac.af:<=0.00001

q=exac.ac.ac_adj:[76640 TO 80000] # bounded (including 76640 and 80000)
q=exac.ac.ac_adj:{76640 TO 80000} # unbounded
```

Wildcard queries

Wildcard character “*” or “?” is supported in either simple queries or fielded queries:

```
q=dbnsfp.genename:CDK?
q=dbnsfp.genename:CDK*
```

Note: Wildcard character can not be the first character. It will be ignored.

Scrolling queries

If you want to return ALL results of a very large query, sometimes the paging method described [above](#) can take too long. In these cases, you can use a scrolling query. This is a two-step process that turns off database sorting to allow very fast retrieval of all query results. To begin a scrolling query, you first call the query endpoint as you normally would, but with an extra parameter **fetch_all** = TRUE. For example, a GET request to:


```

{
  "hits": [
    {
      "_id": "chr1:g.69511A>G",
      "_score": 7.2999496,
      "cadd": {
        "gene": {
          "ccds_id": "CCDS30547.1",
          "cds": {
            "cdna_pos": 421,
            "cds_pos": 421,
            "rel_cdna_pos": 0.46,
            "rel_cds_pos": 0.46
          },
          "feature_id": "ENST00000335137",
          "gene_id": "ENSG00000186092",
          "genename": "OR4F5",
          "prot": {
            "domain": "tmhmm",
            "protpos": 141,
            "rel_prot_pos": 0.46
          }
        }
      }
    },
    {
      "_id": "chr1:g.69538G>A",
      "_score": 0.78757036,
      "cadd": {
        "gene": {
          "ccds_id": "CCDS30547.1",
          "cds": {
            "cdna_pos": 448,
            "cds_pos": 448,
            "rel_cdna_pos": 0.49,
            "rel_cds_pos": 0.49
          },
          "feature_id": "ENST00000335137",
          "gene_id": "ENSG00000186092",
          "genename": "OR4F5",
          "prot": {
            "domain": "ndomain",
            "protpos": 150,
            "rel_prot_pos": 0.49
          }
        }
      }
    }
  ],
  "max_score": 7.2999496,
  "took": 2325,
  "total": 2
}

```

“total” in the output gives the total number of matching hits, while the actual hits are returned under “hits” field. “size” parameter controls how many hits will be returned in one request (default is 10). Adjust “size” parameter and “from” parameter to retrieve the additional hits.

Faceted queries

If you need to perform a faceted query, you can pass an optional “*facets*” parameter. For example, if you want to get the facets on species, you can pass “facets=taxid”:

A GET request like this:

```
http://myvariant.info/v1/query?q=cadd.gene.gene_id:ENSG00000113368&facets=cadd.
↳polyphen.cat&size=0
```

should return hits as:

```
{
  "facets": {
    "cadd.polyphen.cat": {
      "_type": "terms",
      "missing": 797,
      "other": 0,
      "terms": [
        {
          "count": 1902,
          "term": "benign"
        },
        {
          "count": 998,
          "term": "probably_damaging"
        },
        {
          "count": 762,
          "term": "possibly_damaging"
        }
      ],
      "total": 3662
    }
  },
  "hits": [],
  "max_score": 0.0,
  "took": 29,
  "total": 4459
}
```

Batch queries via POST

Although making simple GET requests above to our variant query service is sufficient for most use cases, there are times you might find it more efficient to make batch queries (e.g., retrieving variant annotation for multiple variants). Fortunately, you can also make batch queries via POST requests when you need:

```
URL: http://myvariant.info/v1/query
HTTP method: POST
```

Query parameters

q

Required, multiple query terms separated by comma (also support “+” or white space), but no wildcard, e.g., ‘q=rs58991260,rs2500’

scopes

Optional, specify one or more fields (separated by comma) as the search “scopes”, e.g., “scopes=dbsnp.rsid”, “scopes=dbsnp.rsid,dbnsfp.genename”. The available “fields” can be passed to “scopes” parameter are *listed here*. Default:

fields

Optional, a comma-separated string to limit the fields returned from the matching variant hits. The supported field names can be found from any variant object. Note that it supports dot notation, and wildcards as well, e.g., you can pass “dbnsfp”, “dbnsfp.genename”, or “dbnsfp.aa.*”. If “fields=all”, all available fields will be returned. Default: “all”.

email

Optional, if you are regular users of our services, we encourage you to provide us an email, so that we can better track the usage or follow up with you.

Example code

Unlike GET requests, you can easily test them from browser, make a POST request is often done via a piece of code. Here is a sample python snippet:

```
import httplib2
h = httplib2.Http()
headers = {'content-type': 'application/x-www-form-urlencoded'}
params = 'q=rs58991260,rs2500&scopes=dbsnp.rsid'
res, con = h.request('http://myvariant.info/v1/query', 'POST', params,
↳headers=headers)
```

Returned object

Returned result (the value of “con” variable above) from above example code should look like this:

```
[
{'_id': 'chr1:g.218631822G>A',
'dbsnp': {'allele_origin': 'unspecified',
'alleles': [{'allele': 'G', 'freq': 0.9784},
{'allele': 'A', 'freq': 0.02157}],
'alt': 'A',
'chrom': '1',
'class': 'SNV',
'dbsnp_build': 129,
'flags': ['ASP', 'G5', 'G5A', 'GNO', 'KGPhase1', 'KGPhase3', 'SLO'],
'gmaf': 0.02157,
```

```

'hg19': {'end': 218631823, 'start': 218631822},
'ref': 'G',
'rsid': 'rs58991260',
'validated': True,
'var_subtype': 'ts',
'vartype': 'snp'},
'query': 'rs58991260',
'welllderly': {'alleles': [{'allele': 'A', 'freq': 0.0025},
{'allele': 'G', 'freq': 0.9975}],
'alt': 'A',
'chrom': '1',
'gene': 'TGFB2',
'genotypes': [{'count': 1, 'freq': 0.005, 'genotype': 'G/A'},
{'count': 199, 'freq': 0.995, 'genotype': 'G/G'}],
'hg19': {'end': 218631822, 'start': 218631822},
'pos': 218631822,
'ref': 'G',
'vartype': 'snp'}},
{'_id': 'chr11:g.66397320A>G',
'dbsnp': {'allele_origin': 'unspecified',
'alleles': [{'allele': 'A'}, {'allele': 'G'}],
'alt': 'G',
'chrom': '11',
'class': 'SNV',
'dbsnp_build': 36,
'flags': ['ASP', 'INT', 'RV', 'U3'],
'hg19': {'end': 66397321, 'start': 66397320},
'ref': 'A',
'rsid': 'rs2500',
'validated': False,
'var_subtype': 'ts',
'vartype': 'snp'},
'query': 'rs2500'}
]

```

Tip: “query” field in returned object indicates the matching query term.

If a query term has no match, it will return with “**notfound**” field as “**true**”:

```

[
  ...,
  {'query': '...',
   'notfound': true},
  ...
]

```

Variant annotation service

This page describes the reference for the MyVariant.info variant annotation web service. It’s also recommended to try it live on our [interactive API page](#).

Service endpoint

```
http://myvariant.info/v1/variant
```

GET request

Obtaining the variant annotation via our web service is as simple as calling this URL:

```
http://myvariant.info/v1/variant/<variantid>
```

variantid above is an HGVS name based variant id using genomic location based on hg19 human genome assembly.

By default, this will return the complete variant annotation object in JSON format. See [here](#) for an example and [here](#) for more details. If the input **variantid** is not valid, 404 (NOT FOUND) will be returned.

Optionally, you can pass a “**fields**” parameter to return only the annotation you want (by filtering returned object fields):

```
http://myvariant.info/v1/variant/chr1:g.35367G>A?fields=cadd
```

“**fields**” accepts any attributes (a.k.a fields) available from the variant object. Multiple attributes should be separated by commas. If an attribute is not available for a specific variant object, it will be ignored. Note that the attribute names are case-sensitive.

Just like the variant query service, you can also pass a “**callback**” parameter to make a [JSONP](#) call.

Query parameters

fields

Optional, can be a comma-separated fields to limit the fields returned from the variant object. If “fields=all”, all available fields will be returned. Note that it supports dot notation as well, e.g., you can pass “cadd.gene”. Default: “fields=all”.

callback

Optional, you can pass a “**callback**” parameter to make a [JSONP](#) call.

filter

Alias for “fields” parameter.

email

Optional, if you are regular users of our services, we encourage you to provide us an email, so that we can better track the usage or follow up with you.

Returned object

A GET request like this:

```
http://myvariant.info/v1/variant/chr1:g.35367G>A
```

should return a variant object below:

```
{
  "_id": "chr1:g.35367G>A",
  "_version": 2,
  "cadd": {
    "alt": "A",
    "annotype": "NonCodingTranscript",
    "bstatistic": 994,
    "chmm": {
      "bivflnk": 0.0,
      "enh": 0.0,
      "enhbiv": 0.0,
      "het": 0.0,
      "quies": 1.0,
      "reprpc": 0.0,
      "reprpcwk": 0.0,
      "tssa": 0.0,
      "tssaflnk": 0.0,
      "tssbiv": 0.0,
      "tx": 0.0,
      "txflnk": 0.0,
      "txwk": 0.0,
      "znfrpts": 0.0
    },
    "chrom": 1,
    "consdetail": "non_coding_exon,nc",
    "consequence": "NONCODING_CHANGE",
    "consscore": 5,
    "cpg": 0.03,
    "dna": {
      "helt": -2.04,
      "mgw": 0.01,
      "prot": 1.54,
      "roll": -0.63
    },
    "encode": {
      "exp": 31.46,
      "h3k27ac": 23.44,
      "h3k4me1": 23.8,
      "h3k4me3": 8.6
    },
    "exon": "2/3",
    "fitcons": 0.577621,
    "gc": 0.48,
    "gene": {
      "cds": {
        "cdna_pos": 476,
        "rel_cdna_pos": 0.4
      },
      "feature_id": "ENST00000417324",
      "gene_id": "ENSG00000237613",

```

```
"genename": "FAM138A"
},
"gerp": {
  "n": 1.29,
  "s": -0.558
},
"isknownvariant": "FALSE",
"istv": "FALSE",
"length": 0,
"mapability": {
  "20bp": 0,
  "35bp": 0
},
"min_dist_tse": 122,
"min_dist_tss": 707,
"mutindex": 70,
"phast_cons": {
  "mammalian": 0.003,
  "primate": 0.013,
  "vertebrate": 0.003
},
"phred": 1.493,
"phylop": {
  "mammalian": -0.155,
  "primate": 0.151,
  "vertebrate": -0.128
},
"pos": 35367,
"rawscore": -0.160079,
"ref": "G",
"scoresegdup": 0.99,
"segway": "D",
"type": "SNV"
},
"dbnsfp": {
  "aa": {
    "aapos_sift": "ENSP00000409362:P44L",
    "alt": "L",
    "codonpos": 2,
    "pos": 44,
    "ref": "P",
    "refcodon": "CCG"
  },
  "alt": "A",
  "ancestral_allele": "G",
  "cadd": {
    "phred": 1.493,
    "raw": -0.160079,
    "raw_rankscore": 0.05963
  },
  "cds_strand": "-",
  "chrom": "1",
  "ensembl": {
    "geneid": "ENSG00000237613",
    "transcriptid": "ENST00000417324"
  },
  "fold-degenerate": 0,
  "genename": "FAM138A",
```

```

"gerp++": {
  "nr": 1.29,
  "rs": -0.558,
  "rs_rankscore": 0.11796
},
"hg18": {
  "end": 25230,
  "start": 25230
},
"hg19": {
  "end": 35367,
  "start": 35367
},
"hg38": {
  "end": 35367,
  "start": 35367
},
"lr": {
  "pred": "T",
  "rankscore": 0.32941,
  "score": 0.0846
},
"mutationtaster": {
  "converted_rankscore": 0.10124,
  "pred": "N",
  "score": 1.0
},
"phastcons": {
  "100way": {
    "vertebrate": 0.001,
    "vertebrate_rankscore": 0.1272
  },
  "46way": {
    "placental": 0.003,
    "placental_rankscore": 0.11579,
    "primate": 0.336,
    "primate_rankscore": 0.28762
  }
},
"phylop": {
  "100way": {
    "vertebrate": 0.055,
    "vertebrate_rankscore": 0.14229
  },
  "46way": {
    "placental": -0.143,
    "placental_rankscore": 0.11334,
    "primate": 0.175,
    "primate_rankscore": 0.17021
  }
},
"provean": {
  "converted_rankscore": 0.92415,
  "pred": "D",
  "score": -6.73
},
"radialsvm": {
  "pred": "T",

```

```
    "rankscore": 0.32188,
    "score": -0.9916
  },
  "ref": "G",
  "reliability_index": 5,
  "sift": {
    "converted_rankscore": 0.2575,
    "pred": "T",
    "score": 0.13
  },
  "siphy_29way": {
    "logodds": 3.5364,
    "logodds_rankscore": 0.07795,
    "pi": {
      "a": 0.5429,
      "c": 0.0,
      "g": 0.4571,
      "t": 0.0
    }
  },
  "vest3": {
    "rankscore": 0.16309,
    "score": 0.137
  }
}
```

Batch queries via POST

Although making simple GET requests above to our variant query service is sufficient in most use cases, there are some times you might find it's easier to batch query (e.g., retrieving variant annotations for multiple variants). Fortunately, you can also make batch queries via POST requests when you need:

```
URL: http://myvariant.info/v1/variant
HTTP method: POST
```

Query parameters

ids

Required. Accept multiple HGVS variant ids separated by comma, e.g., "ids=chr1:g.35367C>T,chr7:g.55241707G>T,chr16:g.28883241A>G". Note that currently we only take the input ids up to **1000** maximum, the rest will be omitted.

fields

Optional, can be a comma-separated fields to limit the fields returned from the matching hits. If "fields=all", all available fields will be returned. Note that it supports dot notation as well, e.g., you can pass "dbnsfp", "dbnsfp.genename", or "dbnsfp.aa.*". Default: "all".

email

Optional, if you are regular users of our services, we encourage you to provide us an email, so that we can better track the usage or follow up with you.

Example code

Unlike GET requests, you can easily test them from browser, make a POST request is often done via a piece of code, still trivial of course. Here is a sample python snippet:

```
import httplib2
h = httplib2.Http()
headers = {'content-type': 'application/x-www-form-urlencoded'}
params = 'ids=chr16:g.28883241A>G,chr1:g.35367G>A&fields=dbnsfp.genename,cadd.gene'
res, con = h.request('http://myvariant.info/v1/variant', 'POST', params,
↳headers=headers)
```

Returned object

Returned result (the value of “con” variable above) from above example code should look like this:

```
[
  {
    "_id": "chr16:g.28883241A>G",
    "cadd": {
      "gene": {
        "ccds_id": "CCDS53996.1",
        "cds": {
          "cdna_pos": 1889,
          "cds_pos": 1450,
          "rel_cdna_pos": 0.61,
          "rel_cds_pos": 0.64
        },
        "feature_id": "ENST00000322610",
        "gene_id": "ENSG00000178188",
        "genename": "SH2B1",
        "prot": {
          "protpos": 484, "rel_prot_pos": 0.64
        }
      }
    },
    "dbnsfp": {
      "genename": "SH2B1"
    },
    "query": "chr16:g.28883241A>G"
  },
  {
    "_id": "chr1:g.35367G>A",
    "cadd": {
      "gene": {
        "cds": {
          "cdna_pos": 476,
          "rel_cdna_pos": 0.4
        },
        "feature_id": "ENST00000417324",
```

```
    "gene_id": "ENSG00000237613",
    "genename": "FAM138A"
  },
  "dbnsfp": {
    "genename": "FAM138A"
  },
  "query": "chr1:g.35367G>A"
}
]
```

Server response

The MyVariant.info server returns a variety of query responses, and response status codes. They are listed here.

Note: These examples show query responses using the python [requests](#) package.

Status code 200

A **200** status code indicates a successful query, and is accompanied by the query response payload.

```
In [1]: import requests

In [2]: r = requests.get('http://myvariant.info/v1/query?q=_exists_:clinvar')

In [3]: r.status_code
Out[3]: 200

In [4]: data = r.json()

In [5]: data.keys()
Out[5]: dict_keys(['total', 'max_score', 'took', 'hits'])
```

Status code 400

A **400** status code indicates an improperly formed query, and is accompanied by a response payload describing the source of the error.

```
In [6]: r = requests.get('http://myvariant.info/v1/query?q=_exists_:clinvar&size=u')

In [7]: r.status_code
Out[7]: 400

In [8]: data = r.json()

In [9]: data
Out[9]:
{'error': "Expected 'size' parameter to have integer type.  Couldn't convert 'u' to_
↪integer",
'success': False}
```

Status code 404

A **404** status code indicates either an unrecognized URL, as in (*/query* is misspelled */quer* resulting in an unrecognized URL):

```
In [10]: r = requests.get('http://myvariant.info/v1/quer?q=_exists_:clinvar')
In [11]: r.status_code
Out[11]: 404
```

or, for the */variant* endpoint, a **404** status code could be from querying for a nonexistent HGVS ID, as in:

```
In [12]: r = requests.get('http://myvariant.info/v1/variant/5')
In [13]: r.status_code
Out[13]: 404

In [14]: data = r.json()

In [15]: data
Out[15]:
{'error': "ID '5' not found",
 'success': False}
```

Status code 5xx

Any **5xx** status codes are the result of uncaught query errors. Ideally, these should never occur. We routinely check our logs for these types of errors and add code to catch them, but if you see any status **5xx** responses, please submit a bug report to help@myvariant.info.

Third-party packages

This page lists the third-party packages/modules built upon the [MyVariant.info](#) services.

MyVariant python module

“myvariant” is an easy-to-use Python wrapper to access [MyVariant.info](#) services.

You can install it easily using either `pip` or `easy_install`:

```
pip install myvariant #this is preferred
```

or:

```
easy_install myvariant
```

This is a brief example:

```
In [1]: import myvariant
In [2]: mv = myvariant.MyVariantInfo()
In [3]: mv.getvariant('chr1:g.35367G>A')
```

```
Out[3]:
{'_id': 'chr1:g.35367G>A',
 '_version': 1,
 'cadd': {'alt': 'A',
 'annotype': 'NonCodingTranscript',
 'bstatistic': 994,
 'chmm': {'bivflnk': 0.0,
 'enh': 0.0,
 'enhbiv': 0.0,
 'het': 0.0,
 'quies': 1.0,
 'reprpc': 0.0,
 'reprpcwk': 0.0,
 'tssa': 0.0,
 'tssaflnk': 0.0,
 'tssbiv': 0.0,
 'tx': 0.0,
 'txflnk': 0.0,
 'txwk': 0.0,
 'znfrpts': 0.0},
 'chrom': 1,
 'consdetail': 'non_coding_exon,nc',
 'consequence': 'NONCODING_CHANGE',
 'consscore': 5,
 'cpg': 0.03,
 'dna': {'helt': -2.04, 'mgw': 0.01, 'prot': 1.54, 'roll': -0.63},
 'encode': {'exp': 31.46, 'h3k27ac': 23.44, 'h3k4me1': 23.8, 'h3k4me3': 8.6},
 'exon': '2/3',
 'fitcons': 0.577621,
 'gc': 0.48,
 'gene': {'cds': {'cdna_pos': 476, 'rel_cdna_pos': 0.4},
 'feature_id': 'ENST00000417324',
 'gene_id': 'ENSG00000237613',
 'genename': 'FAM138A'},
 'gerp': {'n': 1.29, 's': -0.558},
 'isknownvariant': 'FALSE',
 'istv': 'FALSE',
 'length': 0,
 'mapability': {'20bp': 0, '35bp': 0},
 'min_dist_tse': 122,
 'min_dist_tss': 707,
 'mutindex': 70,
 'phast_cons': {'mammalian': 0.003, 'primate': 0.013, 'vertebrate': 0.003},
 'phred': 1.493,
 'phylop': {'mammalian': -0.155, 'primate': 0.151, 'vertebrate': -0.128},
 'pos': 35367,
 'rawscore': -0.160079,
 'ref': 'G',
 'scoresegdup': 0.99,
 'segway': 'D',
 'type': 'SNV'},
 'snpeff': {'ann': [{'effect': 'non_coding_exon_variant',
 'feature_id': 'NR_026818.1',
 'feature_type': 'transcript',
 'gene_id': 'FAM138A',
 'gene_name': 'FAM138A',
 'hgvs_c': 'n.476C>T',
 'putative_impact': 'MODIFIER',
```

```
'rank': '2',
'total': '3',
'transcript_biotype': 'Noncoding'},
{'effect': 'non_coding_exon_variant',
'feature_id': 'NR_026820.1.2',
'feature_type': 'transcript',
'gene_id': 'FAM138F.2',
'gene_name': 'FAM138F',
'hgvs_c': 'n.476C>T',
'putative_impact': 'MODIFIER',
'rank': '2',
'total': '3',
'transcript_biotype': 'Noncoding'}}},
'vcf': {'alt': 'A', 'position': '35367', 'ref': 'G'}}
```

See <https://pypi.python.org/pypi/myvariant> for more details.

MyVariant R package

An R wrapper for the MyVariant.info API is available in Bioconductor since v3.2. To install:

```
source("https://bioconductor.org/biocLite.R")
biocLite("myvariant")
```

To view documentation for your installation, enter R and type:

```
browseVignettes("myvariant")
```

For more information, visit the [Bioconductor myvariant](#) page.

MyVariant Node.js package

`myvariantjs` is a Node.js wrapper for the MyVariant.info API, developed and maintained by [Larry Hengl](#). To install:

```
npm install myvariantjs --save
```

Some brief usage examples:

```
var mv = require('myvariantjs');
mv.getvariant('chr9:g.107620835G>A');
mv.getvariant('chr9:g.107620835G>A', ['dbnsfp.genename', 'cadd.phred']);

mv.getvariants("chr1:g.866422C>T,chr1:g.876664G>A,chr1:g.69635G>C"); // string of ↵
↵delimited ids
mv.getvariants(["chr1:g.866422C>T", "chr1:g.876664G>A","chr1:g.69635G>C"]);

mv.query("chr1:69000-70000", {fields:'dbnsfp.genename'});
mv.query("dbnp.rsid:rs58991260", {fields:'dbnsfp'});

mv.querymany(['rs58991260', 'rs2500'], 'dbnp.rsid');
mv.querymany(['RCV000083620', 'RCV000083611', 'RCV000083584'], 'clinvar.rcv_accession
↵');
```

For more information, visit its [API and usage docs](#), and its [github code repository](#).

You can also check out this [neat demo application](#) developed by Larry using this `myvariantjs` package.

Another MyVariant.info python module

This is another python wrapper of MyVariant.info services created by Brian Schrader. The repository is available here. You can install this package with PyPI like this:

```
pip install myvariant-api
```

A JBrowse plugin for MyVariant.info and MyGene.info

JBrowse provides a fast, embeddable genome browser built completely with JavaScript and HTML5.

Colin from the JBrowse team made a very nice plugin to visualize the gene and variant annotations in JBrowse Genome Browser, using the data served from both MyGene.info and MyVariant.info APIs.

- Live demo

To see it live, here is [the demo site](#). It has been tested with hg38, hg19, and zebrafish and has mygene.info and myvariant.info integrations

- Source code

<https://github.com/elsiklab/myvariantviewer>

The screenshot displays the MyVariant.info viewer interface. On the left, there is a sidebar with 'Available Tracks' including MyGene.info v2, MyGene.info v3, and MyVariant.info. The main window shows a genomic track for chromosome 19 (hg19) with a zoomed-in view of a variant. The variant details are shown in a central panel:

Primary Data

- Name: chr1:g.19568918C>T
- Position: chr1:19568918..19568918
- Length: 1 bp

Attributes

clinvar_attr (9)

Name	Value
allele_id	Z86635
alt	T
chrom	1
cytogenetic	1p36.13
omim	616846.0001
ref	C
rsid	rs869320623
type	single nucleotide variant

clinvar_hg19

end	start
19568918	19568918

clinvar_hg38

end	start
19242424	19242424

clinvar_hgvs_attr coding NM_015047.2:c.430G>A

clinvar_hgvs_genomic

NC_000001.10:g.19568918C>T	NC_000001.11:g.19242424C>T	NG_002948.1:g.14136G>A

clinvar_rcv_attr (7)

Name	Value
accession	RCV000210590
clinical_significance	Uncertain significance
last_evaluated	2013-02-01
number_submitters	1
origin	germline
preferred_name	NM_015047.2(EMC1):c.430G>A (p.Ala144Thr)
review_status	no assertion criteria provided

clinvar_rcv_conditions

identifiers	medgen
	CN221809

identifiers

name
not provided

id chr1:g.19568918C>T

Region sequence reference sequence not available

The right side of the screenshot shows a genomic track with various annotations, including gene models for CAPZB and AKR1F2, and variant annotations represented by colored dots.

- A screenshot

CHAPTER 4

Related links

- [github repository](#)