Using RESTful Web Services URL

Overview

The best way to use of the RESTful Web Services is through the client libraries implemented for different programming languages. Nevertheless, under certain circumstances it may be required to directly access the RESTful API. In order to provide a solution for those occasions we are here describing the RESTful API. We will focus on those end points of the RESTful API which are of more interest for HGVA users, giving examples of their use and pin pointing certain peculiarities of the parameters for HGVA. Data is hierarchically organised in Projects and Studies. Please, to ensure a proper understanding of the API have a look at Datasets and Studies in order to understand how data is organised: *Projects, Studies* and *Cohorts*. For details on the query parameters, please refer to the Swagger documentation at:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices

Further details on the API specification can be found at the RESTful Web Services and Clients section.

Getting information about genomic variants

Getting variant data from a given study:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/analysis/variant/query?studies={project}:{study}

An extensive list of filtering parameters allow great flexibility on the queries (check Swagger documentation link above). For example, get TTN variants from the Genome of the Netherlands study, which is framed within the *reference_grch37* project. We will also limit the number of returned results to 3:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/analysis/variant/query?gene=TTN&studies=GONL&limit=3

Getting information about projects

Getting all metadata from a particular project:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/projects/{projects}/info

For example, getting all metadata for the *reference_grch37* project:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/projects/reference_grch37/info

Getting all metadata from all studies associated to a particular project:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/projects/{projects}/studies

For example, getting all studies and their metadata for the *cancer_grch37* project:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/projects/cancer_grch37/studies

Getting information about studies

Get all available studies and their metadata. **Please note, of special interest will be here the field *alias* which contains the study identifier to be used as an input whenever a study must be passed as a parameter:**

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/search

For example, getting all metadata for all available studies:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/search

Getting summary data from a particular study:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/{project}:{study}/summary

For example, getting summary data for study *1kG_phase3* which is framed within project *reference_grch37*:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/reference_grch37:1kG_phase3/summary

Getting all available metadata for a particular study:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/{project}:{study}/info

For example, getting all metadata for study *GONL* which is framed within the project *reference_grch37*:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/reference_grch37:GONL/info

Getting all samples metadata for a given study:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/{project}:{study}/samples

For example, getting all samples metadata for study *1kG_phase3* which is framed within project *reference_grch37*. Please, note that not all studies contain samples data, e.g. GONL, ExAC, among others, only provide variant lists and aggregated frequencies, i.e. no sample genotypes.

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/studies/reference_grch37:1kG_phase3/samples

Getting information about samples

Get all metadata for a particular sample:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/samples/{sample}/info?study={project}:{study}

For example, get all metadata for sample *HG00096* of the *1kG_phase3* study which is framed within the *reference_grch37* project:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/samples/HG00096/info?study=reference_grch37:1kG_phase3

Getting information about cohorts

Getting all samples metadata in a given cohort:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/cohorts/{cohort}/samples?study={project}:{study}

For example, get all samples metadata for cohort *GBR* from study *1kG_phase3* which is framed within project *reference_grch37*:

http://bioinfo.hpc.cam.ac.uk/hgva/webservices/rest/v1/cohorts/GBR/samples?study=reference_grch37:1kG_phase3