### **Datasets and Studies**

## Data organisation

OpenCGA uses a two-level structure to organise datasets, these are *Projects* and *Studies* and are used to organise HGVA data and metadata:

- **Projects** is the top-level and can contain one or more *studies*. *Projects* are specific for one species and assembly, all *studies* in a *project* are stored and indexed together in the same database and, therefore, they share the variant annotation.
- Study, in turn, represents a particular dataset which can contain samples metadata and cohorts, and obviously all the genomic variants. For example, the 1000 Genomes Project is defined as a study in OpenCGA and belongs to Reference GRCh37 project. You can also define cohorts in the studies, they are just a set of samples defined within a study. For example, populations and super-populations within The 1000 Genomes Project are defined as cohorts, so EUR, AMR or GBR are examples of cohorts.

You can get more information about data organisation at OpenCGA Catalog Data Management. *Projects* and *Studies* have a unique *alias* to ease their usage from the command-line and REST API, you can find more information about how to query data programmatically at RESTful Web Services and Clients. Please, see next section the full list and organisation of the currently available *Projects* and *Studies* (datasets) in HVGVA.

### **Datasets**

In this sections you can find all datasets loaded in HGVA and how they are organised in *Projects* and *Stu dies* (see previous section).

	Studies		HGVA Version (date)	
Project name <i>(alias)</i>	Name	Alias	v1 ( <i>Dec.</i> 2016)	v2 ( <i>Jan.</i> 2018)
Reference GRCh37 (reference_grch 37)	1000 Genomes Project GRCh37	1kG_phase3	Phase 3 2016-05	Phase 3 2016-05
	Exome Sequencing Project (ESP6500)	ESP6500	2016-05	2016-05
	Exome Aggregation Consortium (ExAC)	EXAC	0.3.1 2016- 05	0.3.1 2016- 05
	Genome of the Netherlands (GoNL)	GONL	Release 5 2016-05	Release 5 2016-05
	UK10K Project	UK10k	2016-05	2016-05
	DiscovEHR	DISCOVEHR	-	
	Genome Aggregation Database (gnomAD Exomes)	GNOMAD_EXOM ES	-	
	Genome Aggregation Database (gnomAD Genomes)	GNOMAD_GENO MES	-	
	Spanish Medical Genome Project (MGP)	MGP	2016-12	2016-12
Reference GRCh38	1000 Genomes Project GRCh38	1kG_phase3	Phase 3 2016-10	Phase 3 2016-10
(reference_grch 38)	ESP6500	ESP6500	-	
	UK10K Project (*)	UK10K	-	
	DiscovEHR (*)	DISCOVEHR	-	
	Genome Aggregation Database (gnomAD Exomes) (*)	GNOMAD_EXOM ES	-	
	Genome Aggregation Database (gnomAD Genomes) (*)	GNOMAD_GENO MES	-	
Cancer GRCh37	QIMR Berghofer Melanoma	QIMR_Berghofer_ Melanoma	2016-12	2016-12
(cancer_grch37)	Chronic Myeloid Leukemia - Russian Academy of Medical Sciences	RAMS_CML	2016-12	2016-12
Platinum	Illumina Platinum	illumina_platinum	2015-08	2015-08
(platinum)				

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(\*) Liftover carried out by Genomics England (GEL)

# **Variant Anotation**

Variant annotation was carried out by the CellBase project. Please, check CellBase documentation for details on additional data sources: Data sources and species