Beacon v2 - Feature-rich Implementation of the Genomic Data Discovery Protocol

Michael Baudis, Rahel Paloots, Hangjia Zhao, Ziying Yang

... supported by Members of the Beacon Developer Community

Department of Molecular Life Sciences and Swiss Institute of Bioinformatics, University of Zurich, Switzerland

V2 of the GA4GH Beacon protocol provides a framework for extended, metadata-rich query and response options

- The Global Alliance for Genomics and Health together with ELIXIR provides the Beacon protocol, for federated discovery and sharing of genomic variation data.
- Beacon v2 was designed with support for rich biomedical data queries and data delivery, in distributed and standalone environments.
- Since 2016, the Beacon+ implementation on top of the Progenetix resource has served as testbed for Beacon protocol extensions on a live dataset.

Progenetix is a Reference Resource for oncogenomic profiling data with a focus on copy number variations (CNVs) built on a Stack of GA4GH Beacon v2 Code

- Somatic genomic Copy Number Variations are the largest quantitative contributors to the cancer mutation landscape.
- Progenetix is an open oncogenomic reference resource with > 140'000 CNV profiles from cancers and reference samples.
- With lead involvement in the Beacon development, the Progenetix utilizes the Beacon v2 protocol for communication and empowers external data integration through its openly accessible Beacon v2 API.
- The current Beacon+ implements the Beacon v2 default model with extensions, e.g. supporting the GA4GH Phenopackets format as well as various aggregated exports over handover objects.

Regional CNV Frequencies for Most Cancer Types provided over Beacon handover objects

Progenetix is a, well, Progenetix R API using handover objects

Visualization of CNV features using the pgxRpi R package. Aggregated CNV data for cancer types displayed using Circos or frequency plots in a local R environment. The R package relies on the Beacon v2 API to communicate with Progenetix.

Links and Info
progenetix.org | docs.GenomeBeacons.org | github.com/progenetix/pgxRpi | github.com/ga4gh-beacon/beacon-v2