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

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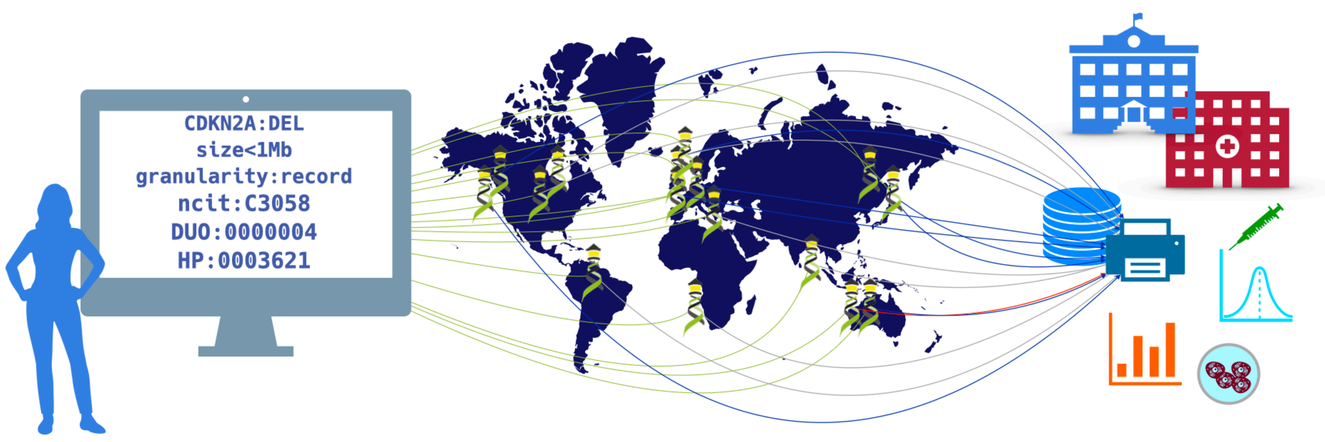
V2 of the GA4GH Beacon protocol provides a framework for extended, metadata-rich query and response options

- 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.



Can you provide data about focal deletions in CDKN2A in Glioblastomas from juvenile patients with unrestricted access?

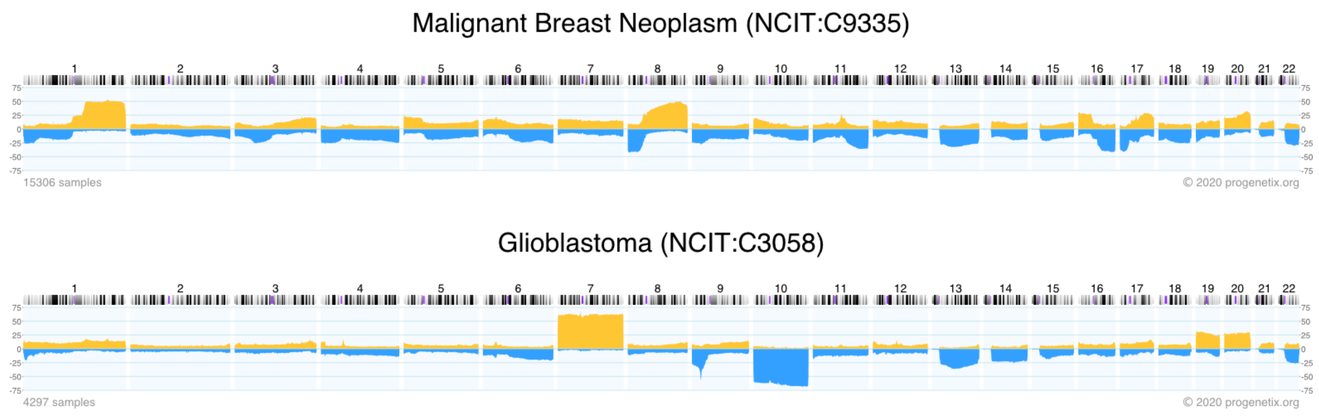
Beacon *v2* API

The Beacon API v2 represents a simple but powerful **genomics API** for ***federated*** data discovery and retrieval

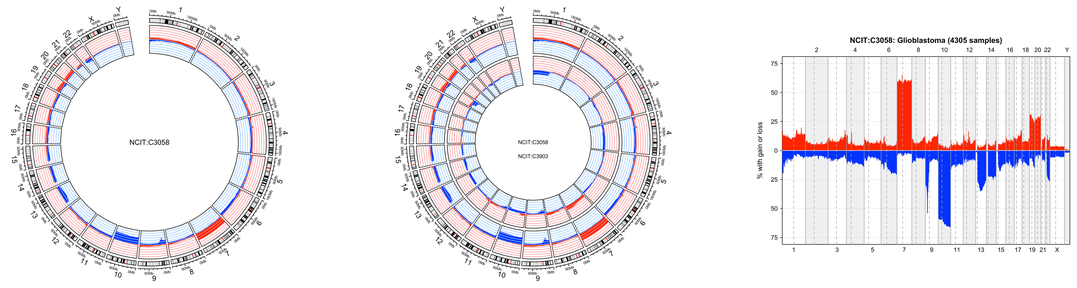
Progenetix is a Reference Resource for oncogenomic profiling data with a focus on copy number variations (CNVs) built on a Stack of GA4GH Beacon v2 Code

- 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



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



Visualization of CNV features using the pgxRpi R package. Aggregated CNV data for cancer typesis 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