Implementation of the GA4GH *Beacon v2* Protocol for Discovery and Sharing of genomic copy number variation data

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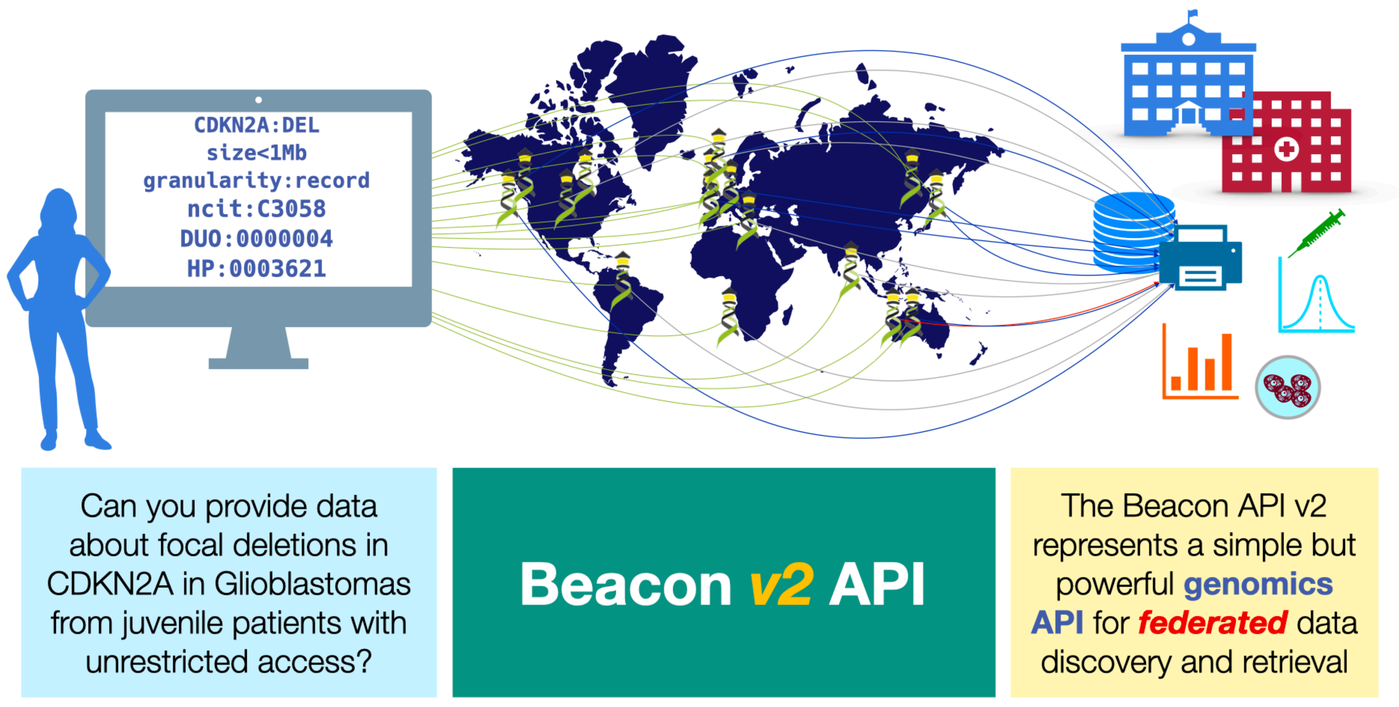
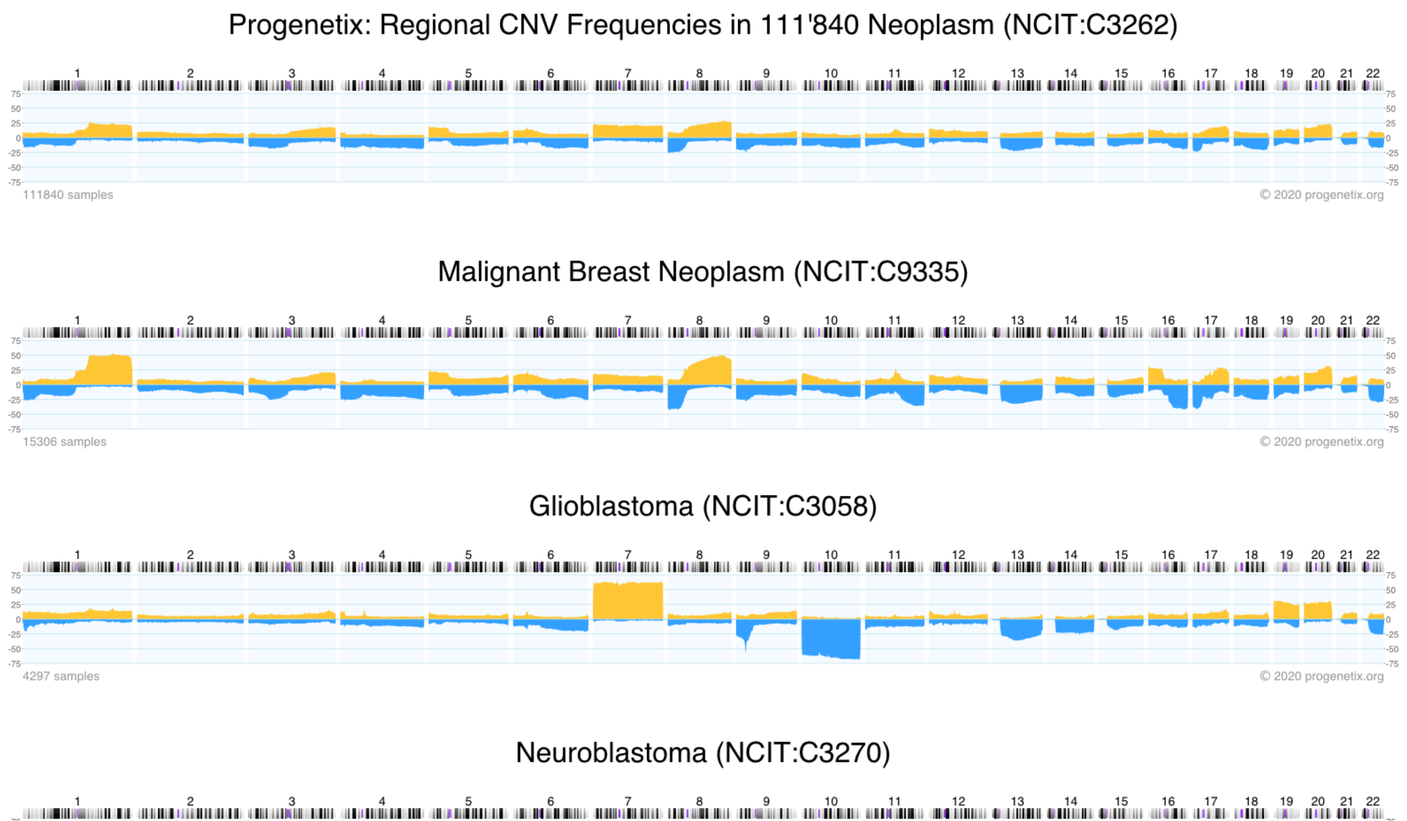
Progenetix is a Reference Resource for CNVs built on a Stack of GA4GH Beacon v2 Code

- Somatic genomic Copy Number Variations are the largest quantitative contributors to the cancer mutation landscape.
- The Progenetix database is a freely accessible reference resource, containing more than 140'000 CNV profiles from cancers and related reference samples.
- Since 2014, the Global Alliance for Genomics and Health together with ELIXIR has worked 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.
- 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



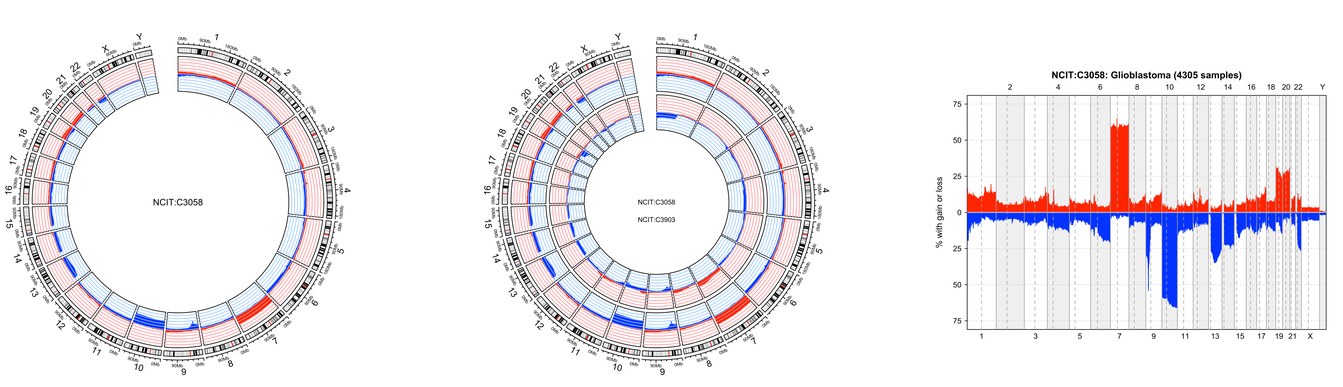
Regional CNV Frequencies for Most Cancer Types



Links and Info

- progenetix.org
- docs.progenetix.org
- docs.genomebeacons.org
- cnvar.org
- github.com/progenetix/pgxRpi
- github.com/ga4gh-beacon/beacon-v2
- The extended abstract can be accessed through the "News" category at docs.progenetix.org

Example API Use - pgxRpi



Visualization of CNV features using the pgxRpi R package. Here, aggregated CNV data for single or two (center) cancer types using Circos or frequency plots in a local R environment.
The R package relies on the Beacon v2 API to communicate with the Progenetix resource for direct data access and retrieval.