

Beacon Network: A System for Global Genomic Data Sharing

Motivation

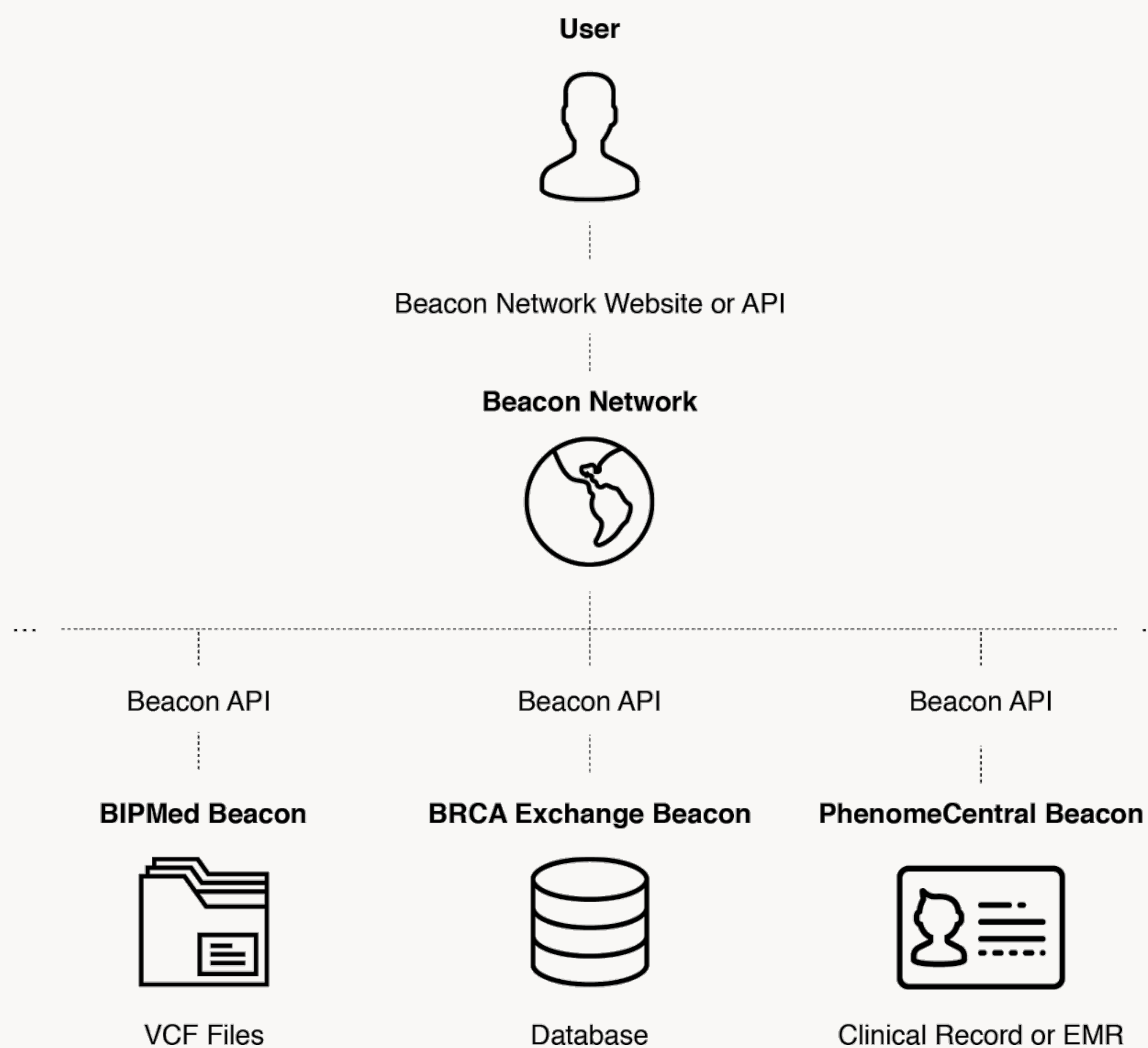
- ▶ Analysis of large volumes of genomic data has the potential to drive discoveries and applications in medicine.
- ▶ No institution has enough resources to capture all human variation.
- ▶ A global federated network of shared genomic data is needed.

Beacon

- ▶ Standard defining a **web service** for **discovery** and **sharing** of information about alleles.
- ▶ API:
 - Request: *Do you have information about this mutation?*
 - Response: *Yes/No (and optionally additional information).*
- ▶ Initiative of the Global Alliance for Genomics & Health.
- ▶ More information: <https://ga4gh.org/#/beacon>.

Beacon Network

- ▶ **Search engine** across the world's beacons.
- ▶ Translates genomic queries for each beacon, intelligently distributes the queries through the network, and aggregates the results.
- ▶ API:
 - Request: *Who has information about this mutation?*
 - Response: *List of beacons and their responses.*



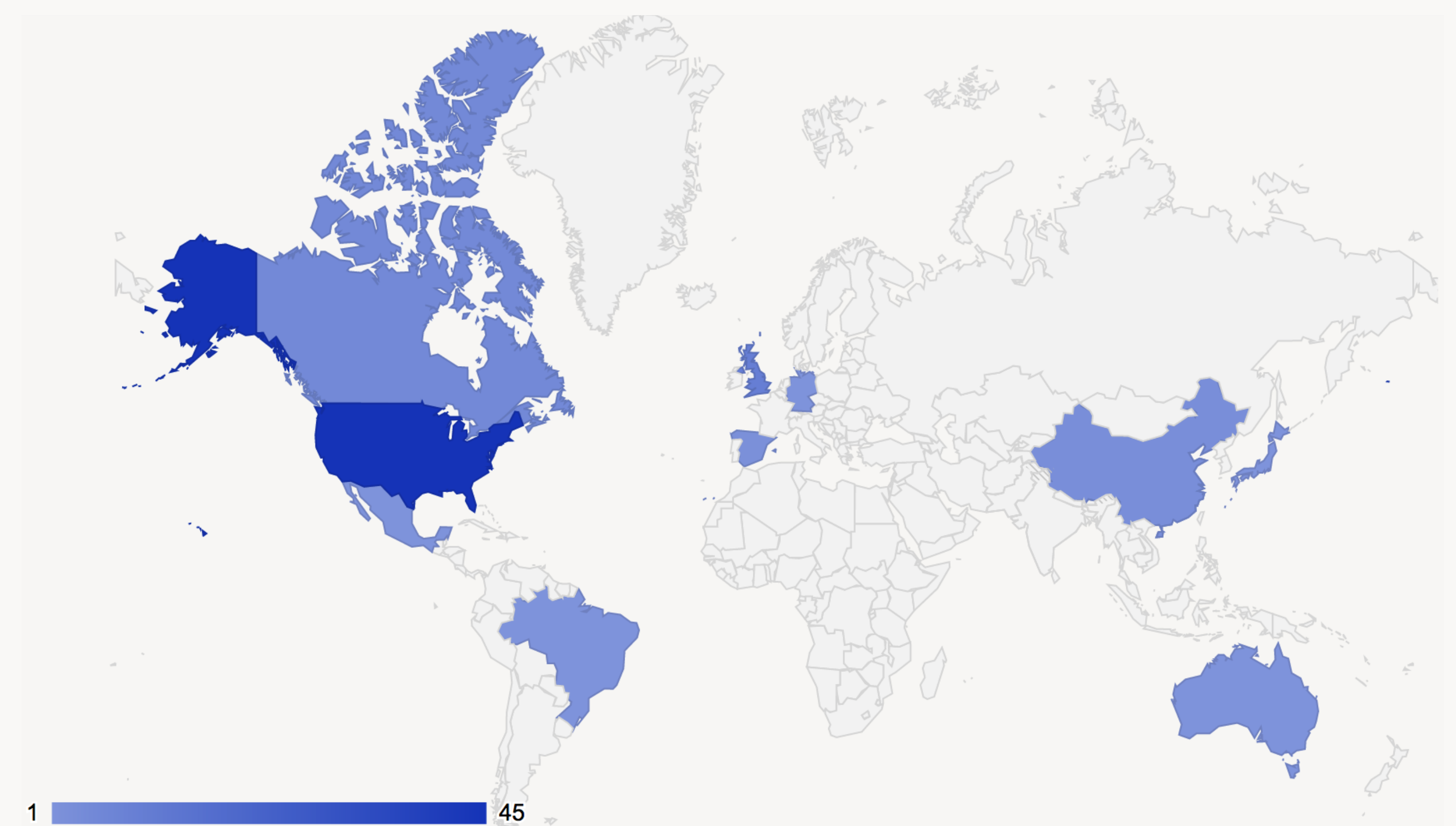
- ▶ Provides REST API, web client, command line client, query library, compatible beacon implementations in multiple programming languages, and adapters for various sources of genomic data.
- ▶ More information: <http://beacon-network.org>.

User interface

The screenshot shows the Beacon Network user interface. At the top, there is a search bar with the text "Search all beacons for allele". Below the search bar, there is a dropdown menu for "GRCh37" and a text input field containing "13 : 32936732 G > C". A "Search" button is located to the right of the input field. Below the search bar, there is a table of results. The table has columns for "Response", "All", and "None". The "Response" column shows "Found", "Not Found", and "Not Applicable". The "All" column shows "14", "29", and "22". The "None" column is empty. Below the table, there is a list of organizations. The organizations listed are "Altruist", "AMPLab - 1000 Genomes Project", "Biobase - HGMD", and "BRCA Exchange". Each organization has a status indicator: "Not Found", "Not Found", "Found", and "Found" respectively. Below the list of organizations, there is a table of clinical significance citations. The table has columns for "Clinical significance citations", "HGVS cDNA", "Condition ID type", "Date last evaluated", "HGVS protein", "Assertion method citation", "ClinVarAccession", "alt", "Condition category", and "Comment on clinical". The table contains one row of data.

Adoption

- ▶ 25+ of the world's top genomic organizations, 60+ beacons.



- ▶ Served 400K+ queries from 6K+ users in 100+ countries, resulting in 2M+ queries dispatched to the participants of the network.

