

Beacon: A Protocol for Federated Discovery and Sharing of Genomic Data

Motivation



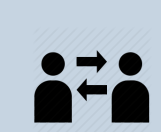
- ▶ Analysis of large volumes of genomic data has the potential to drive discoveries and applications in medicine.
- ▶ No institution has enough resources to capture all human variation.
- ▶ Standards for interoperability enabling sharing across a federated network of genomic data resources are needed.

Beacon



- ▶ Standard defining a web service for discovery and sharing of information about alleles.
- ▶ Demonstration of willingness of international organizations to share data.
- ▶ Initiative of the Global Alliance for Genomics & Health.
- ▶ More information: <https://ga4gh.org/#/beacon>.

API



- ▶ Request: *"Do you have information about this mutation?"*.

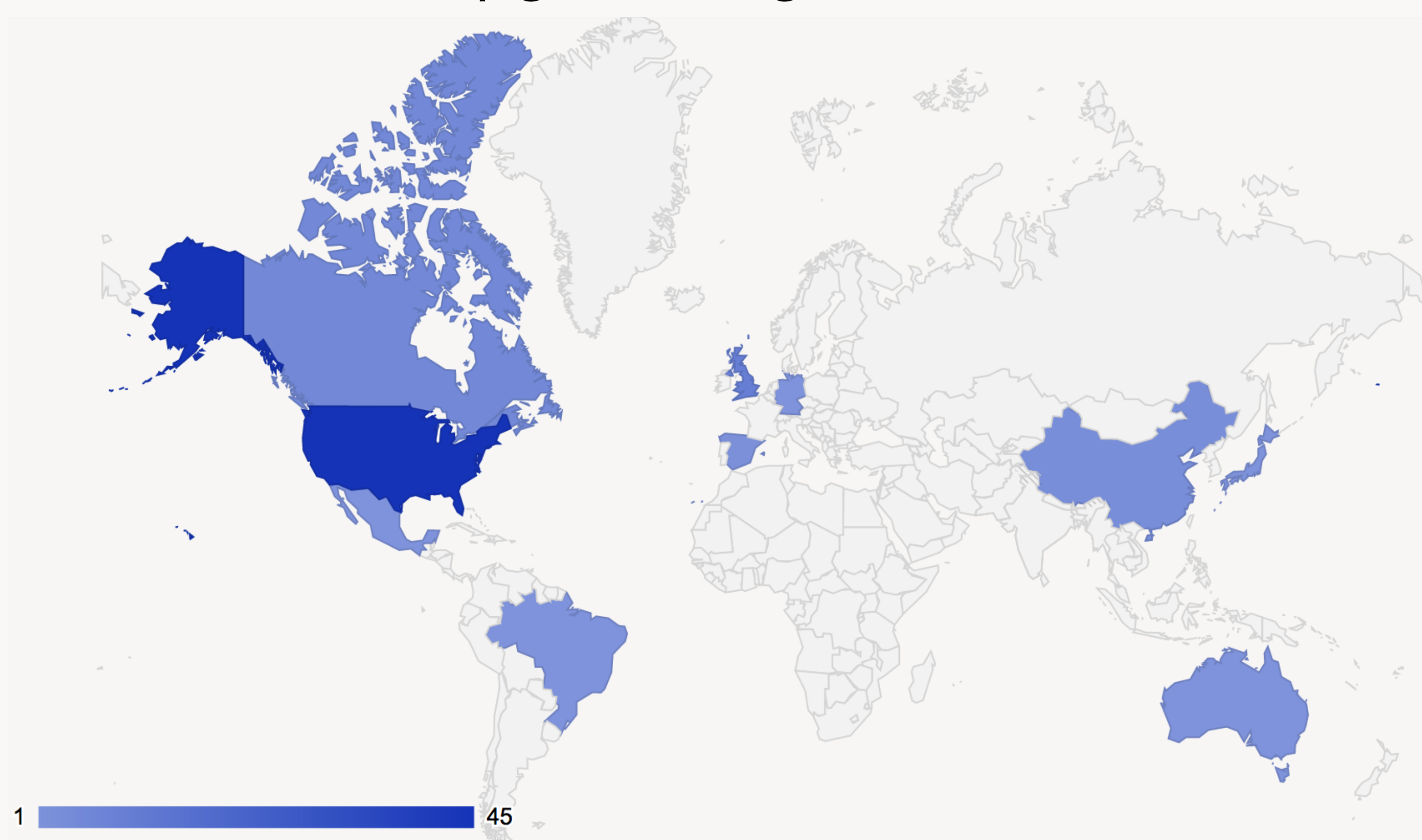
```
{  "referenceName": "13",  "start": 32936731,  "referenceBases": "G",  "alternateBases": "C",  "assemblyId": "GRCh37"}
```
- ▶ Response: *"yes"/"no"* (optionally additional information about the mutation).

```
{  "exists": true,  "datasetAlleleResponses": [    {      "exists": true,      "info": {        ...      }    }  ]}
```
- ▶ Supported by a variety of tools, e.g. reference implementation, command line client, query library, sample beacons in multiple programming languages, adapters for various sources of genomic data.
- ▶ More information: <https://github.com/ga4gh/beacon-team>.

Participants



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

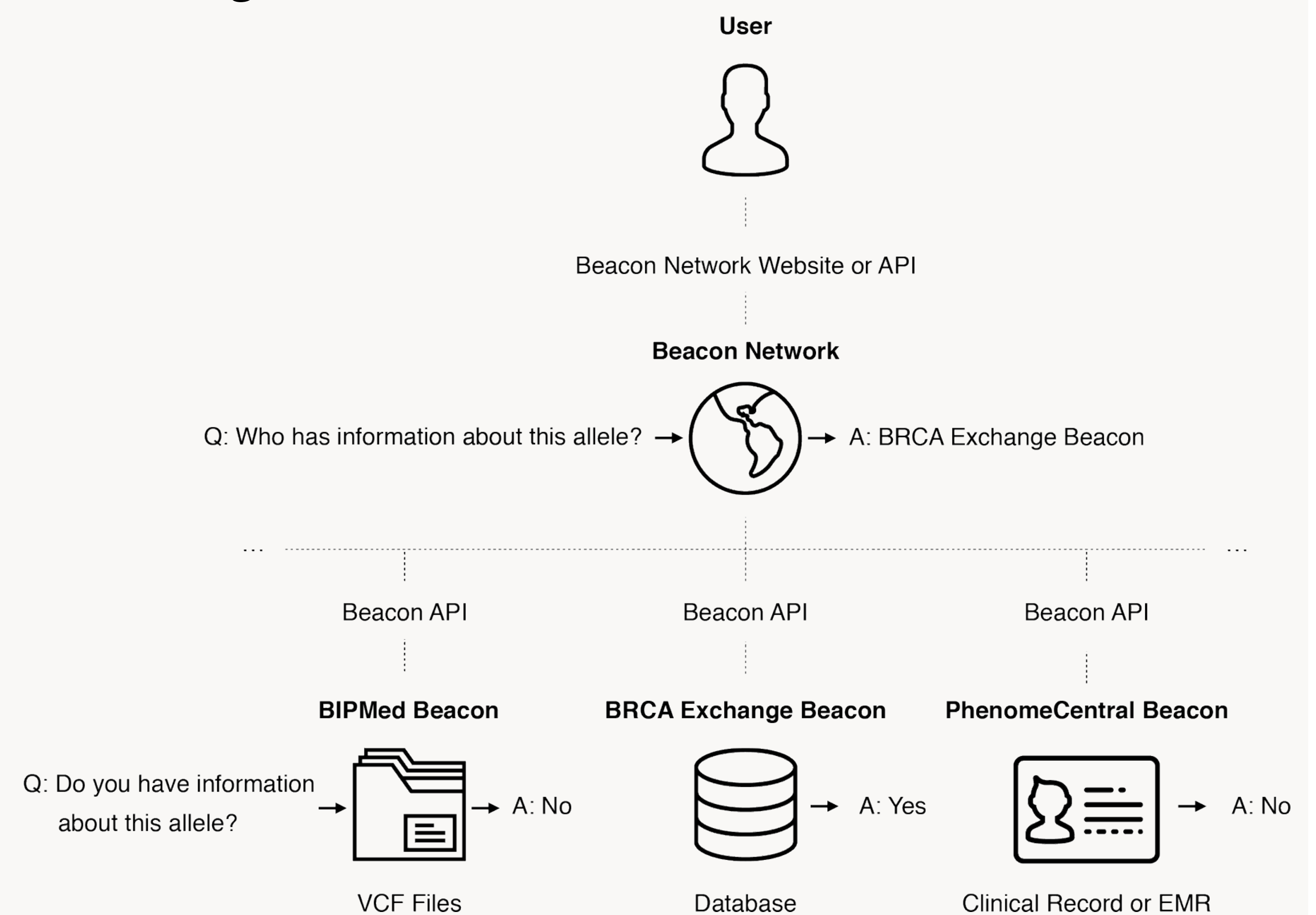


- ▶ 160+ datasets of various origins, e.g. sequence observations from patients, population studies, in-silico predictions, curated/crowd-sourced databases, scientific literature.

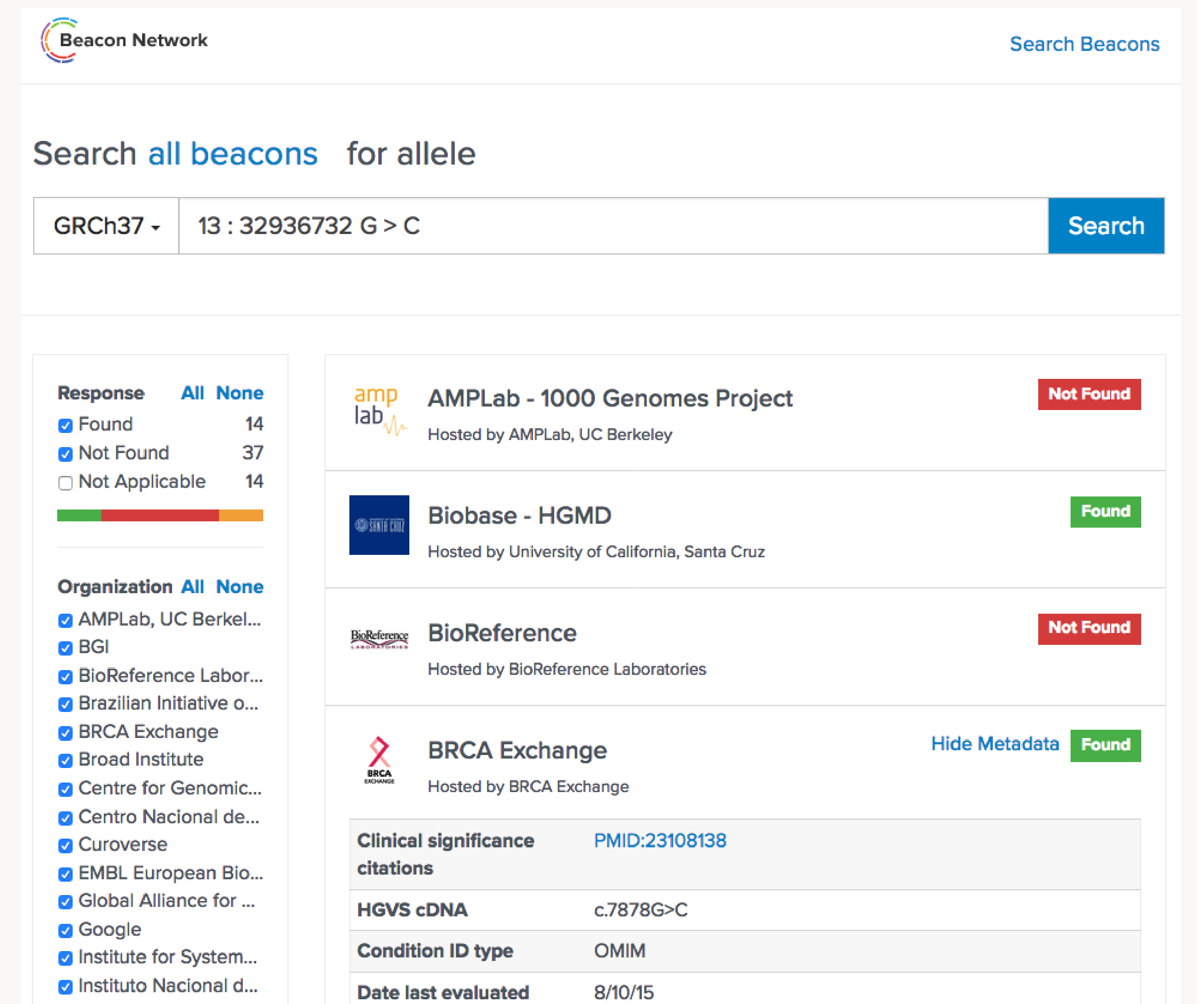
Beacon Network



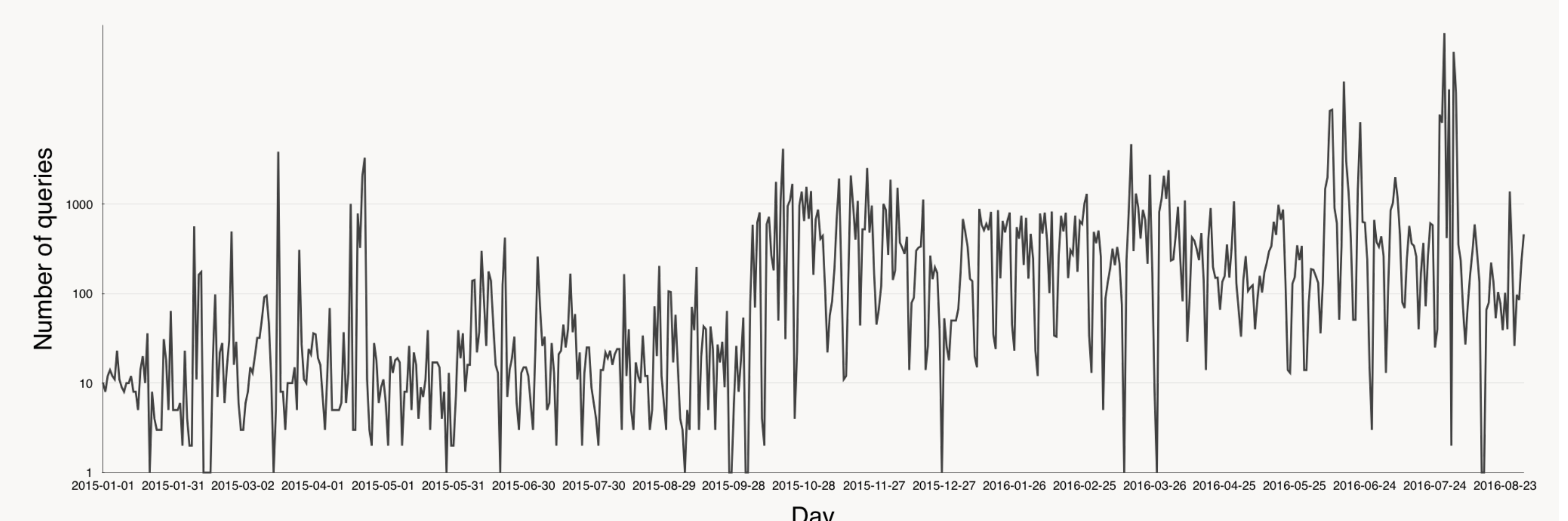
- ▶ Search engine across the world's beacons.



- ▶ Translates genomic queries for each beacon, intelligently distributes the queries through the network, and aggregates the results.



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



- ▶ More information: <http://beacon-network.org>.

