Beacon Network

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17/10/2016

2 years ago

Global Alliance for Genomics and Health
Beacon of Beacons

| Beacon: all | Chromosome: 15 | Position: 41087869 | Alt: A |

Beacon	Result
Wellcome Trust Sanger Institute	No
EMBL-EBI	No
UniProt	No
NCBI ClinVar	No
NCBI	No
Leiden Open Variation	No
AMPLab	Yes

Today

Adoption

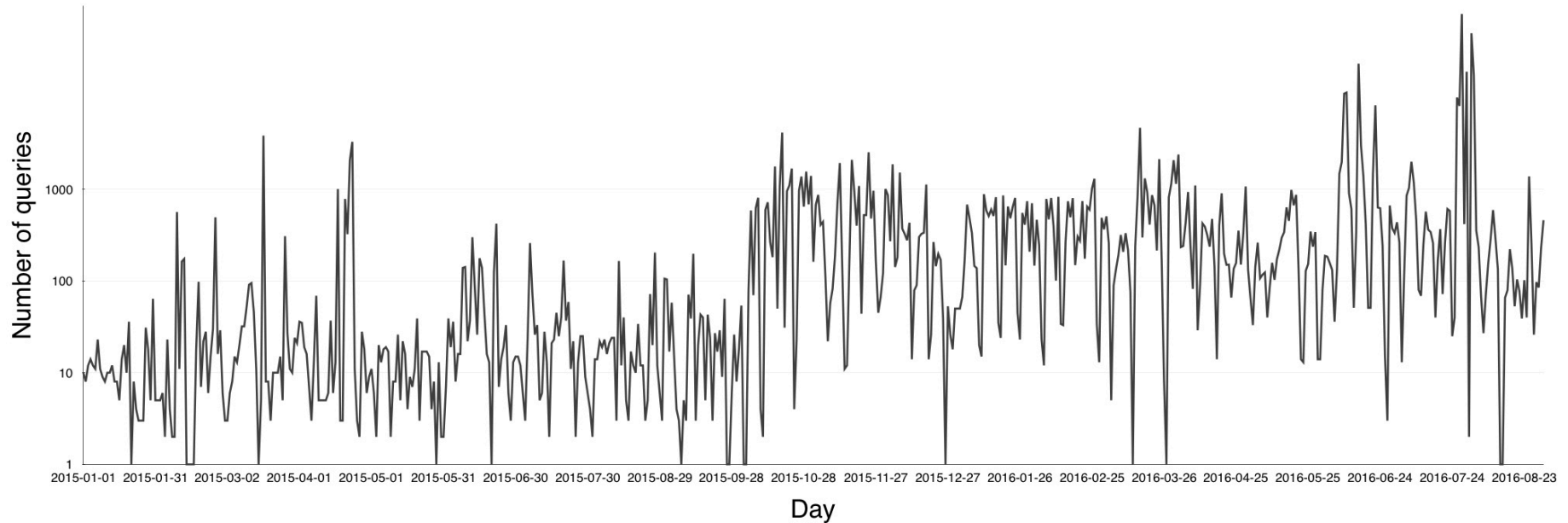- **25+** of the world's top genomic organizations, **60+** beacons.
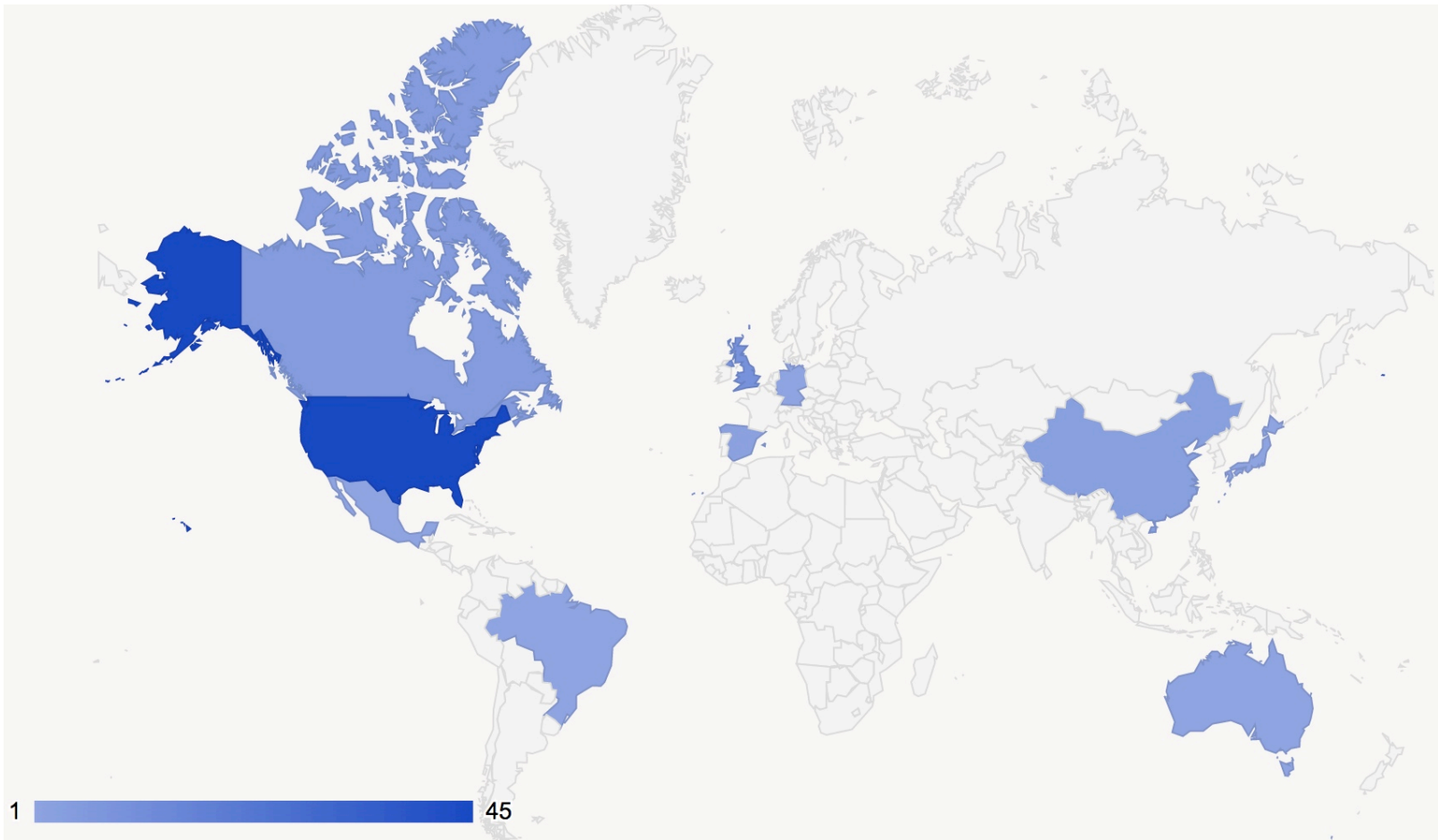
Usage

- **400K+** queries received ⟶ **2M+** queries dispatched.

Usage

- **6K+** users in **100+** countries.

https://beacon-network.org
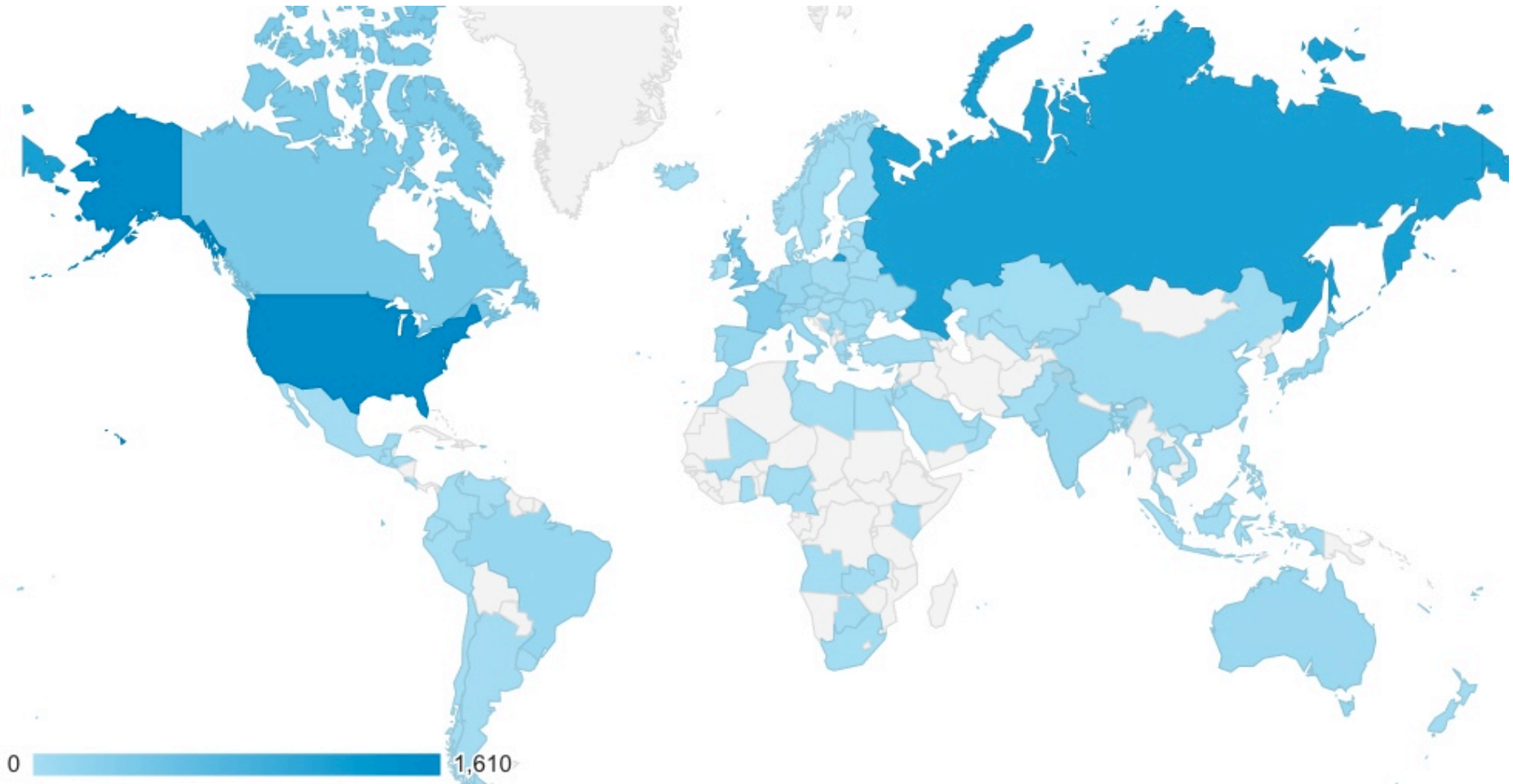
Usage

- Other metrics:
  - Size/growth.

  - Requests.

  - User visits.

  - Assemblies, chromosomes, variants.

  - Harmfulness.

  - Rarity.

  - Genes.

  - Disorders, clinical abnormalities.

Features

- Federation of queries across beacons.

- Registry of public beacons.

- Unified beacon API.

- Input/output conversion.

- Distribution of queries through the network.

- Aggregation.

Features

- Metadata support.

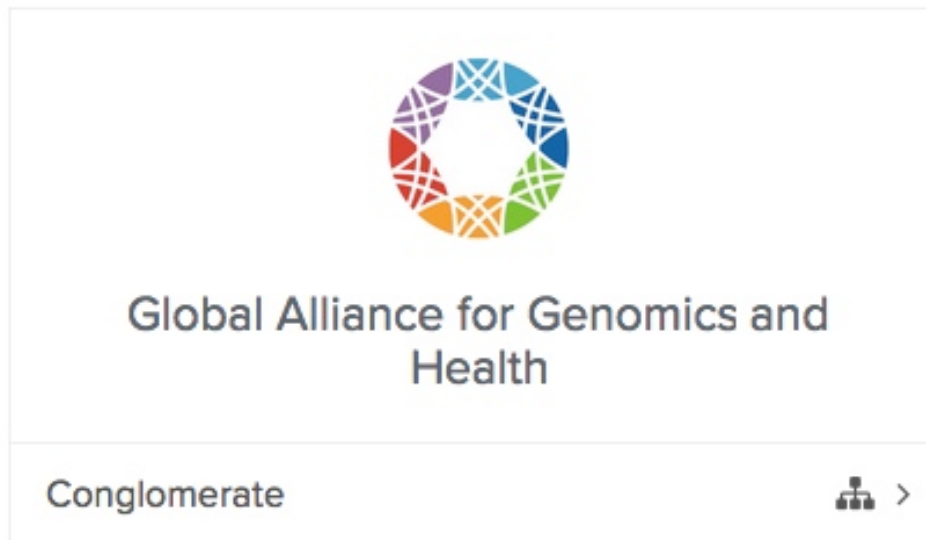| BRCA Exchange	
Hosted by BRCA Exchange	**Hide Metadata** **Found**
Clinical significance citations	PMID:23108138
HGVS cDNA	c.7878G>C
Condition ID type	OMIM
Date last evaluated	8/10/15
HGVS protein	p.(Trp2626Cys)
Assertion method citation	http://enigmaconsortium.org/documents/ENIGMA_Rules_2015-03-26.pdf
ClinVarAccession	SCV000244475
alt	C
Condition category	Disease
Comment on clinical significance	IARC class based on posterior probability from multifactorial likelihood analysis, thresholds for class as per Plon et al. 2008 (PMID: 18951446). Class 5 based on posterior probability = 1
Reference sequence	NM_000059.3
Gene symbol	BRCA2
URL	http://hci-exlovd.hci.utah.edu/variants.php?select_db=BRCA2&action=search_all&search_Variant%2FDNA=c.7878G>C
chrom	13
Abbrev AA change	W2626C
ref	G
Assertion method	ENIGMA BRCA1/2 Classification Criteria (2015)
BIC Nomenclature	8106G>C
Clinical significance	Pathogenic

Features

- External system integration (e.g. secured beacons).

Search 1000 Genomes Project - Yoruban Trio for allele

| GRCh37 ▾ | 1 : 695744 G > T | | Search |

Response	All	None
☑ Found		1
☐ Not Found		0

1000 Genomes Project - Yoruban Trio
Hosted by DNAstack

Found

ℹ More info...

Features

• External system integration (e.g. secured beacons).

Search 1000 Genomes Project - Yoruban Trio for allele

| GRCh37 ▾ | 1 : 695744 G > T | Search |

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☑ Found   1
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1000 Genomes Project - Yoruban Trio
Hosted by DNAstack

Found

ℹ More info...

username or email

password

○ Remember me   Log in
Forgot password?
Register for an account

Features

![Global Alliance for Genomics & Health]

- External system integration (e.g. secured beacons).

Tools

- REST API
    - https://beacon-network.org/api/

Endpoint	Description	Example
/beacons	Lists beacons.	Go to example
/beacons/{beacon}	Shows a beacon.	Go to example
/organizations	Lists organizations.	Go to example
/organizations/{organization}	Shows an organization.	Go to example
/responses	Queries beacons.	Go to example
/responses/{beacon}	Queries a beacon.	Go to example
/chromosomes	Lists supported chromosome.	Go to example
/alleles	Lists supported alleles.	Go to example
/references	Lists supported reference genomes.	Go to example

https://beacon-network.org/#/developers/api/beacon-network

Tools

- Open-source client library (Java)
  - https://github.com/mcupak/beacon-network-client
  - https://mcupak.github.io/beacon-network-client/

```
BeaconNetworkClient bnc = new BeaconNetworkClientImpl();
BeaconResponseDto response = bnc.getResponse(ChromosomeDto.CHR13, 32936732L,
                            AlleleDto.C, ReferenceDto.HG19, "amplab");
```

public interface **BeaconNetworkClient**

Beacon Network API. All methods throw either operation specific exceptions or `InternalException`.

Method Summary

| All Methods | Instance Methods | Abstract Methods |

Modifier and Type	Method and Description
com.dnastack.bob.service.dto.BeaconDto	**getBeacon**(java.lang.String beaconId) Shows information on the beacon specified by its id.
java.util.List<com.dnastack.bob.service.dto.BeaconDto>	**getBeacons**() Lists available beacons.

Tools

- Open-source CLI
    - https://github.com/mcupak/beacon-network-cli

```
$ java -jar cli.jar beacon —help

Beacon Network CLI Usage:
 [beacon | organization | response] : Use one of these commands. To get help on
                                       each command, Type -h with the
                                       corresponding command.
 -u (--url) VAL                      : Beacon Network URL (default =
                                       https://beacon-network.org/api/)
```

Tools

- Embeddable widget
    - https://beacon-network.org/#/developers/embed

```
<iframe src="https://beacon-network.org:443/#/widget?rs=GRCh37&chrom=13&pos=32936732&ref=G&allele=C"
        style="width: 500px; height: 700px; overflow: auto;"
        marginwidth="0" marginheight="0" frameborder="0" vspace="0" hspace="0">
</iframe>
```

Future

- Support for the latest version of the Beacon API.
- User accounts.
- Secured beacon support.
- Programmatic beacon registration, beacon management.
- Workflow for contacting beacon operators.
- Continued support for tooling.
- **Beacon Network specification.**

Questions?

💬 goo.gl/PsMLhr

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