How to light a beacon

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Options

Global Alliance for Genomics & Health
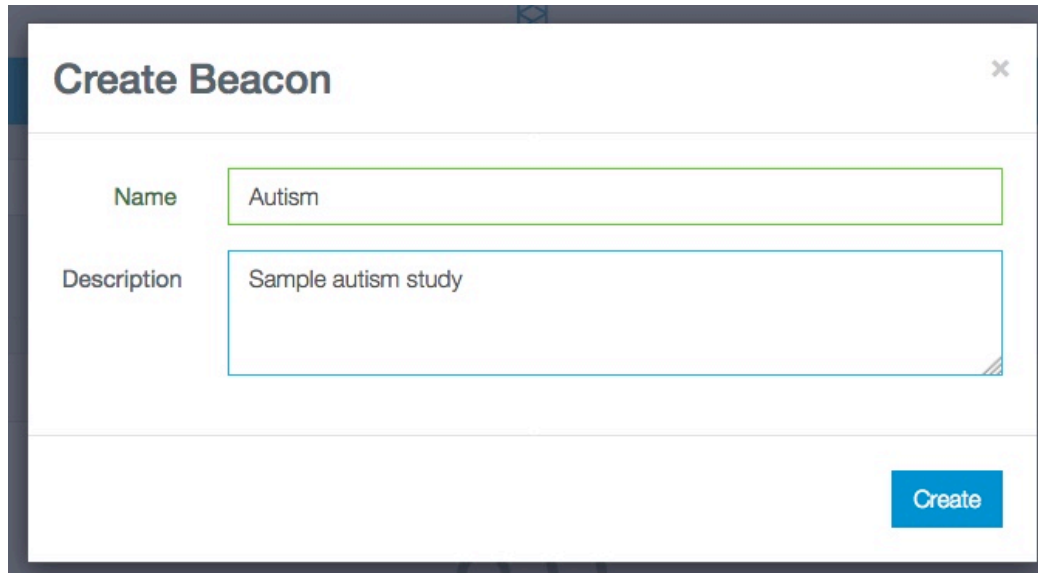
	Ease of implementation	Flexibility
#1 **Managed solutions**	***	*
#2 **Wrapping existing APIs**	**	**
#3 **Implementing beacon from scratch**	*	***

https://mirocupak.com/files/beacon-sme-2017.pdf

#1 Managed solutions

- Light a beacon by clicking a button.
- Easiest, fully automated, no development effort needed.
- Data hosted by a 3rd party.

- E.g. DNAstack (https://dnastack.com/).

#1 DNAstack

#1 DNAstack

#2 API wrappers

- Beacon on top of your existing system (API).
- E.g. GA4GH Core API (https://github.com/ga4gh/schemas/).

- There are open-source projects to help you get started.
  - Apache 2.0 license.
- Host yourself or contact us.

#2 Beacon Development Kits (BDK)

- Open-source quickstarts.
- Compliant with the latest beacon specification.
- Proven to work well.
- Available in:
    - Java
        - https://github.com/mcupak/beacon-java
    - Python
        - https://github.com/mcupak/beacon-python
    - JavaScript
        - https://github.com/mcupak/beacon-javascript

```
117    # TODO: override with the details of your beacon
118    @app.route('/beacon-python/', methods=['GET'])
119    def info():
120        return jsonify(Beacon)
```

#2 Java BDK

- Most advanced of the BDKs.

- Provides:
  - Compatible beacon API.
  - Sample beacon implementation.
  - Integration of Beacon data models.
  - Various utilities: output serialization, input normalization, error handling and status code mapping…
  - Test suite.
  - Security (prototype).

#2 BDK adapter system

- Adapter API:
    - https://github.com/mcupak/beacon-adapter-api
- Adapter implementations for common data stores/APIs:
    - Variants API
        - https://github.com/mcupak/beacon-adapter-variants
        - variant set ⟶ beacon dataset
    - Annotation API
        - https://github.com/mcupak/beacon-adapter-annotations
        - annotation set ⟶ beacon dataset
    - VCF
        - https://github.com/mcupak/beacon-adapter-vcf
        - file ⟶ beacon dataset
    - Easy to implement your own and plug into Java BDK.

#2 Beaconizer

- Extension of Java BDK for more complex *beaconizing*.
- Support for multiple adapters and multiple beacons.
- Open-source: https://github.com/mcupak/beaconizer

```json
{
    "name": "beacon_id",
    "adapterClass": "com.dnastack.beacon.core.adapter.impl.SampleBeaconAdapterImpl",
    "configValues": [
        {
            "name":"sampleName",
            "value":"33-03-03"
        }
    ]
}
```

```
GET /beacons
GET /beacons/:id
GET /beacons/:id/query
```

https://mirocupak.com/files/beacon-sme-2017.pdf

#3 Implementing beacon

- DIY.
- Most flexibility, control and software development effort.
- Not hard.
  - Beacon API is simple.
  - Many institutions have done this before.
  - Many open-source implementations to help you.

#3 How to start?

- Check out the API specification.
  - https://github.com/ga4gh/beacon-team/
  - Very simple and easy to understand.
  - Data model with support for various programming languages.

- Implement 2 REST endpoints returning JSON.
  - Information about the beacon and its datasets.
  - Query for an allele.

#3 Query API

Request:

"Do you have information about this mutation?".

```
{
  "referenceName": "13",
  "start": 32936731,
  "referenceBases": "G",
  "alternateBases": "C",
  "assemblyId": "GRCh37"
}
```

Response:

"yes"/"no"

(+additional info about the mutation)

```
{
  "exists": true,
  "datasetAlleleResponses": [
    {
      "exists": true,
      "info": {
...
```

#3 Status of the Beacon API

- Latest: 0.3
  - https://github.com/ga4gh/beacon-team/releases/tag/v0.3.0
  - Improved support for datasets and cross-dataset queries.
  - Modular and extensible.
  - Data versioning.
  - Improved data model.
  - Tooling.

- WIP: 0.4
  - Complex variants.
  - Improved data use conditions.
  - Documentation.
  - Developer experience.
  - Various minor fixes.

  - Future: 1.0
    - Help us define what's needed.
    - https://github.com/ga4gh/beacon-team/issues

#3 Beacon implementations

- Full list: https://github.com/ga4gh/beacon-team/wiki/Resources
- Java
  - https://github.com/mcupak/beacon-java
  - https://github.com/elixirhub/human-data-beacon
  - https://github.com/prota/cnag-beacon
- Python
  - https://github.com/Genecloud/simplebeacon
  - https://github.com/maximilianh/ucscBeacon
  - https://github.com/kozbo/Beacon-on-GA4GH-API
- JavaScript
  - https://github.com/ElementoLab/beacon
- Go
  - https://github.com/googlegenomics/beacon-go

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#3 Other tools: Compliance suite

- Test beacon compatibility with 0.3 specification.
- Groovy-based prototype.
- Open source: https://github.com/mcupak/beacon-compliance

https://mirocupak.com/files/beacon-sme-2017.pdf

#3 Other tools: Utilities

- Coordinate conversion library.
    - Wraps Liftover and HGVS.
    - Extensible, with built-in converters for standard assemblies.
    - Open-source: https://github.com/mcupak/genome-coordinates-converter

```
LiftOver intervalLiftOver = UCSCLiftOver.hg19ToHg38();
Interval newInterval = intervalLiftOver.liftOver("chr1",743267,743268);
```

```
String hgvs = "NM_182763.2:c.688+403C>T"
GenomeInterval interval = HGVSConverter.hgvsToGenomic(hgvs);
```

#3 Other tools: Client and CLI

- Client library.
  - Query beacons from Java.
  - Integration with Beacon 0.3 schema.
  - Open-source: https://github.com/mcupak/beacon-client

```
BeaconNetworkClient bnc = new BeaconNetworkClientImpl();
bnc.getResponse(CHR1, 1L, A, HG19, "brca-exchange");
```

- CLI.
  - Query beacons from your shell.
  - Open-source: https://github.com/mcupak/beacon-cli

```
$ java -jar beacon-cli.jar -u http://localhost:5000/test/ response \
-a GRCh37 -ab A -r 1 -rb T -s 1000

{
  "beaconId": "test",
  "exists": true,
  "alleleRequest": …
}
```

Beacon Network

https://beacon-network.org/

Beacon Network

- Tooling:
    - REST API: https://beacon-network.org/api/
    - Client library: https://github.com/mcupak/beacon-network-client
    - CLI: https://github.com/mcupak/beacon-network-cli
    - Widget: https://beacon-network.org/#/developers/embed

- Registration:
    - Automated for managed solutions.
    - Manual if you implement yourself.
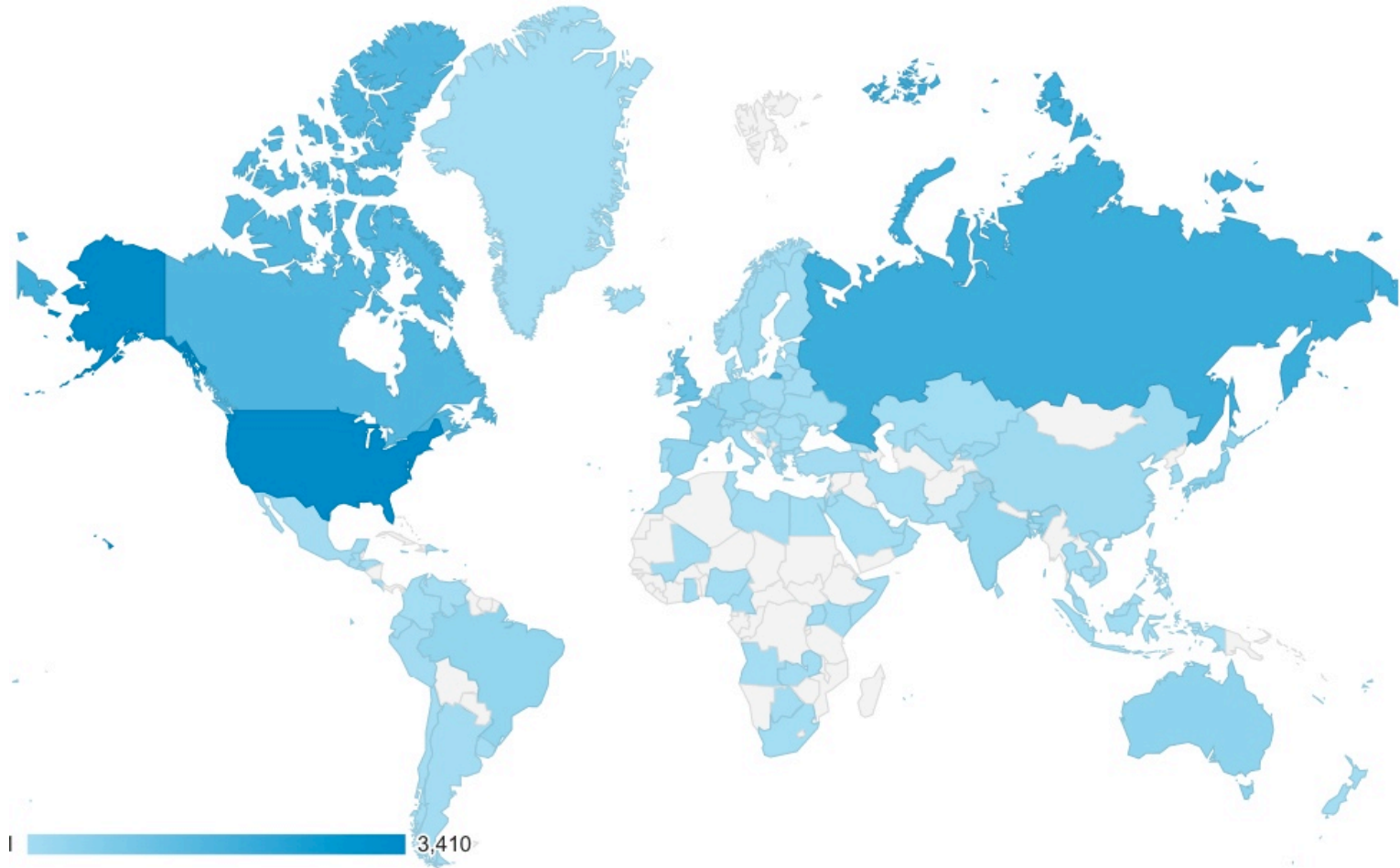   - Email ⟶ API.

Adoption

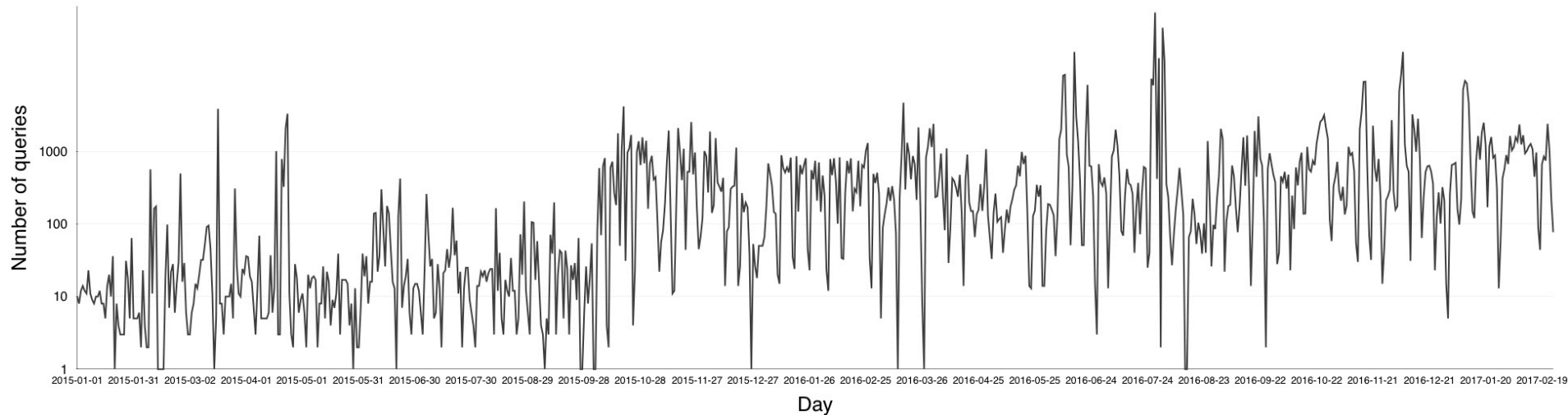- **38** organizations, **70** beacons, **17** countries, **6** continents.
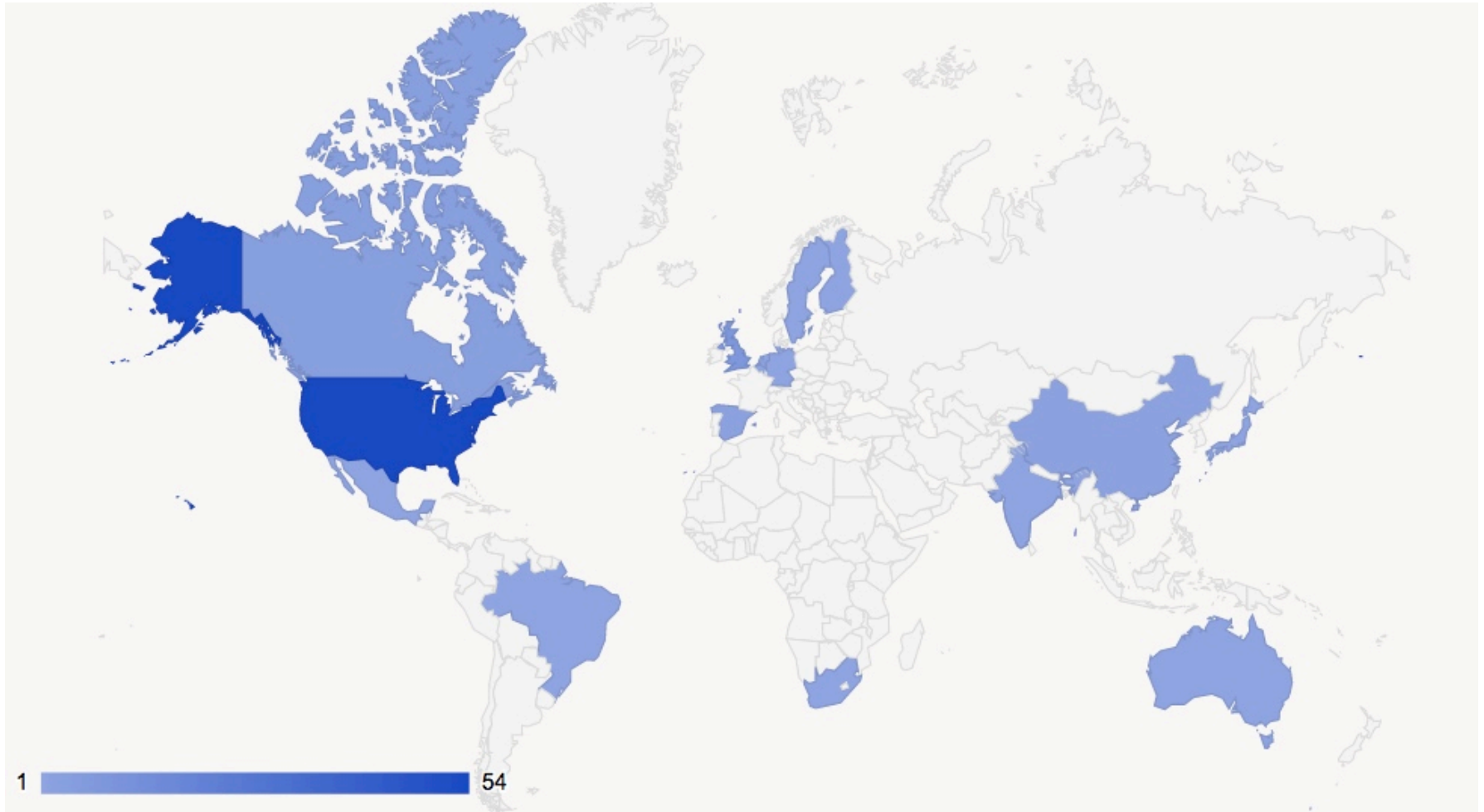
Usage

- **8.5K+** users in **118** countries.

https://mirocupak.com/files/beacon-sme-2017.pdf

Usage

- **600K+** queries received ⟶ **4M+** queries dispatched.

Contact

Beacon

- https://github.com/ga4gh/beacon-team/issues/
- https://beacon-team-slackin.herokuapp.com/
- Mailing list and bi-weekly conference calls.

Beacon Network

- goo.gl/PsMLhr
- info@beacon-network.org
- @BeaconOfBeacons

Me

- miro@dnastack.com
- @mirocupak

Questions?

https://mirocupak.com/files/beacon-sme-2017.pdf