

# ELIXIR Beacon Strategic Calls Minutes and Actions Q4. 2018

A GA4GH Discovery Work Stream Subgroup

These are the minutes for the Beacon meetings, a driver project of the GA4GH Discovery Work Stream. For further information, please visit the GA4GH Work Stream page at <https://ga4gh.org>. (See also the [Vision Statement](#) and recent [Activities](#))

Other minutes documents are available:

[Current](#)

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## 2018-12-18: Technical Call

Attending: Juha, Jordi, Sabela, Gary, Johan, Malin, Miro, Teemu

Sprint [document](#)

Beacon Network [document](#)

Updated [PR #230](#): Add handover mechanism. Miro to review. (Move forward, avoid this becoming a blocker.)

## 2018-12-11: Last of 2018

**Chair:** David Lloyd

**Attendees:** Michael Baudis, Melissa Cline, Tim Beck, Tom Shorter, Tony Brookes, Sabela de la Torre, Miro Cupak, Dylan Spalding, Dietlind Gerloff,

**Regrets:** Gary (OOO) Ilkka, Jordi,

	Actions Arising	Assigned To	Deadline	Update
1.				

	Agenda Item	Speaker	Time
1.	Approve <a href="#">previous minutes</a> and confirm Agenda	Chair	1 min
2.	Review of Actions (See below)	Chair	1 min
3.	Technical Update <a href="#">slides</a>	Juha	10 min
4.	Feedback from GDPR meeting (10th Dec, Brussels)	David	10 min

5.	BRCA Exchange ( <a href="#">slides</a> )	Melissa Cline	20 min
6.	University of Leicester infrastructure (Beacon and use of OIDC) and HDRUK (slides)	Tim Beck	15 min
7.	Forthcoming activity: 18th Dec Technical Call 25th Dec cancelled		

	Previous Actions	Assigned	Deadline	Update
1.				

Notes:

### GDPR workshop (10th Dec, Brussels) [Agenda & Notes](#)

Outcome: to establish an ELIXIR webpage containing overview of:

- best practice
- Use Cases
- wider activities
- Training opportunities
- Materials (policy and other documents)
- Tools
- Issues

Coordinated by a working group

Aim to have this in place ahead of AH (June 2019)

Also the issue of the Beacon Network and ELIXIR AAI wrt GDPR was discussed. This is the most pressing part for the Beacon. Other Beacon issues that were raised at the GDPR workshop are not really Beacon specific.

### Melissa Cline BRCA Exchange

BRCA1 and 2 have strong medical implications

High number of VUS

BRCA Exchange run monthly pipeline to aggregate ClinVar, LOVD, BIO, ExAC, and others

Primary users: Variant curators and clinicians

Integrated views for variants on the BRCA gene

- Also has a hyperlink for Beacon query link

Would like to use an evidence Beacon; no evidence that needs to be protected - potentially with a handover mechanism

Also potential for protected evidence Beacon

MB: this is great. Would like to use this as a prototype for evidence Beacon - can use this to identify which query parameters are needed, what coordinates, etc.

The next thing is connect evidences from one Beacon to another. This is a step for down the road.

Thirdly, BRCA could be a data consumer of data through the Beacon

### Tim Beck UoL

HDR UK - six sites across 22 universities and research institutes

Make healthcare and biobank data discoverable

>40 fellowships awarded

Each of the 6 sites responsible for a set of deliverables

Cafe Variome works in a federated network

Searches can be performed across the network

>30 CV installations - mostly for closed networks, or to share siloed data from one resource

CV has a Beacon. Enables discovery across the two public CVs. Using v0.3 of the API

Want to extend to include restricted CV installations

EDS CV - the data owner consented to allow restricted access

Have used OpenID Connect to share data from a restricted access (EDS) version

Demod at the GA4GH 4th Plenary

Since then have extended the OpenID provider at UoL using KeyCloak

Based on verifying bone fide researcher claim which is associated with the KeyCloak OIDC account

Can they use ELIXIR AAI as ID provider?

Nearly through the registration process

Plan to upgrade Beacon to v1.0.1

Restricted Beacon to work with ELIXIR AAI

Examine ELIXIR AAI for CV users

2018-11-27:

**Chair:** Michael

**Attendees:**

**Regrets:** Gary (EAPM), David (BioData),

	Actions Arising	Assigned To	Deadline	Update
1.				

	Agenda Item	Speaker	Time
1.	Approve <a href="#">previous minutes</a> and confirm Agenda	Chair	1 min
2.	Review of Actions (See below)	Chair	2 min
3.	Technical Update <a href="#">slides</a>	Juha	10 min
4.	Continuation of handover... and more discussion	Jordi	15 min
5.	Belgian Beacon update	Amin	20 min
6.	Wildcard Range Matches <ul style="list-style-type: none"><li>• <a href="#">proposal/introduction</a> and previous <a href="#">notes</a></li><li>• Positioning in dev timeline</li><li>• Alignment w/ handover for resolving responses to wildcard requests (related discussions <a href="#">here</a> and <a href="#">there</a>)</li></ul>	Michael	10min
7.	Forthcoming activity: 4th Dec Technical Call 11th Dec Strategic Call 18th Dec Technical Call 25th Dec cancelled		

	Previous Actions	Assigned	Deadline	Update
1.	Continue discussion of Jordi's presentation from last time	Gary	27/11	In agenda
2.	Follow up on 2018 IS <a href="#">Deliverables</a>	Gary	27/11	All Nodes on target 2018 IS may be extended

- Main:
- reporting from the dev team about things making their way / having been patched (e.g. wildcard queries supported by protocol, although no disambiguation in delivery)
  - re-emphasis that we'll go ahead w/ a component model as presented by Jordi (in spirit, if not necessary in detail)
  - clarification by me to use & drive the development to "outward looking" documentation/proposals, and not only through technical timelines (link them?!), to engage interesting parties, deliver something to look forward to/inquire about
    - \* asking Juha et al to align / link
  - short demo about the current Beacon+ state (wildcard range queries, handover object use), as basis for "clinical" etc

## 2018-11-19: Technical Call [slides](#)

**Attending:** Juha, Sabela, Jordi, Johan, David

Notes & discussion captured in the slides.

Johan: Swedish beacon is in python based on UCSC implementation. (No controlled access or handover)  
Looking to develop in discussion with other tech. developers.

## 2018-11-13: Updated handover concept... and more!

**Chair:** Gary

**Attendees:** Dietlind, Ilkka, Jordi, Marc, Michael, Juha, Malin, Sabela, Jella, David, Stephanie

**Regrets:** Dylan

(Details for regular attendees affiliation is available at the [foot of this document](#))

	Actions Arising	Assigned To	Deadline	Update
1.	Imp. Study Milestones & Deliverables status	Gary		

	Agenda Item	Speaker	Time
1.	Approve <a href="#">previous minutes</a> and confirm Agenda	Chair	1 min
2.	Review of Actions (See below)	David	2 min
3.	Technical Update <a href="#">slides</a>	Juha	10 min
4.	Gary Update: - Beacon internship - have conducted 4 interviews - Forthcoming opportunities: - ELIXIR SME - Plants - Proteomics - BRCA Exchange - GeL - Beacon Development Team in 2019 - <a href="#">ELIXIR 2018 Implementation Study Deliverables</a>	Gary	10 min
5.	Beacon Handover... and more <a href="#">GitHub</a>	Jordi	30 min
6.	A.O.B? <a href="#">Beacon Webinar</a> Thursday 15th November.		
7.	Forthcoming activity: 19th Nov Technical Call 27th Nov Strategic Call		

	Previous Actions	Assigned	Deadline	Update
1.	Further handover examples	Jordi / Sabela	Presentation today	
2.	Website edits, starting with <a href="#">team pages</a> <a href="#">beacon webpage</a>	everybody	(Gary done)	

## Notes:

Introduction: Jella (Netherlands Cancer Inst.)

Website: Encourage all to add content, personal information etc.

Tech. Update:

Opportunities:

- SME event, 4th March
  - Beacon Workshop to demonstrate a Beacon with Controlled Access
  - Genomics England and others interested
  - Ilkka: New ELIXIR Node coordinator, will put in contact, establish best route to put this in place.

Plants:

- ELIXIR have funded project 4pm, led by Bordeaux (Apricot genome data, proprietary data, cannot add to EGA etc.)

- Will require changing hard coding of Human Chromosome, aim to make something applicable to other plants
- Planning a hackathon Q1 2019.

Gary presenting at PAG (Plant & Animal Genome) looking to pick up potential users for Beacon

Proteomics:

- Greater complexity (AA code not DNA code)
- Planning a workshop to establish a use case for sensitive proteomic data (March 2019?)
- Will then look to update reference

Hub to identify someone able to present Gary to find slot in strategic TC

Looking to have presentations on regular TC for Use Cases

(eg. Tim Beck, sharing variant data in Transmart environment)

Genomics England

Looking to light a public Beacon within the GeL Environment

Aim to encourage researchers to use GeL data within their environment, demonstrate

Jordi: Good to have a demonstration to understand the Use Cases they are planning

Gary: A presentation will need the project plan to be approved by GeL board (early 2019)

Development Team

Moving to WP1 in Barcelona, led by Jordi.

Juha moving to new role within CSC

Imp Study

Review of [Milestones & Deliverables](#)

Jordi: [slides](#) different flavours of Beacon

Esp from new communities (hCNV, Plants).

Most searches are looking for rare variants. (The concept is growing up!)

Outline of evolving beacon [slide 7](#)

Handover: more details of the discussion to date on [GitHub](#)

“Custom” where no meaningful ontology term (Beacon, RD-connect) with note to explain

Two mandatory terms: URL, ID

pull/230

Working with RD-connect, into EGA and

Looking to develop this with other use cases

Juha: Like the Use Case, the openAPI looks useful

AOB: Stephanie: Recent NIH data [sharing policy updates](#) to allow more sharing of summary data.

2018-11-06: Technical Update [slides](#)

2018-10-30: Updated website

**Chair:** David

**Attendees:** Michael, Jordi, Deitlind, Dylan, Mike Qu, Sabela,

**Regrets:** Gary

	Actions Arising	Assigned To	Deadline
1.	Further handover examples	Jordi / Sabela	
2.	Website edits, starting with <a href="#">team pages</a>	everybody	

	Agenda Item	Speaker	Time
1.	Approve <a href="#">previous minutes</a> and confirm Agenda	Chair	5 min
2.	Review of Actions (None)	David	0 min

3.	Technical Update ( <i>Sprint extended to 6th Nov.</i> <a href="#">link</a> )	Juha	0 min
4.	Beacon <a href="#">Website</a> - scope and maintenance of site? - location? I like the idea of integrating with the ELIXIR Europe site. - timelines and directions of work?	Michael	20 min
5.	Beacon Intern update (close Wed)		5 min
6.	Schema examples (ClinPhen group, Phenopackets) ( <i>rolled over from last call</i> ) • First draft of schema elements on <a href="#">Github</a>	Michael	15 min
7.	A.O.B? - Handover specification example ( <a href="#">slide</a> )	Jordi	5 min
8.	Forthcoming activity: 23rd Oct Technical Call 30th Oct Strategic Call		

Minutes:

**Beacon Website:** an overview of recent activities on the [Website](#).

- Using Ruby-based Jekyll platform, update through [Github](#), changes visible, fully formatted, in minutes.
- Encouragement that this site become a living record of activities & discussions. [Link](#)
- Either direct edits (apply for membership in group) or through PRs, issues
- Outline and details of how to update the pages
- managing/creating posts are in the README
- local installation [instructions](#) for testing/editing

**Beacon Intern:** 2 applicants, close on Wednesday (tomorrow)

**Handover example** ([roadmap page](#), [draft example slide](#))

- Initial work, more development in hand.
- Talking with RDconnect, to jump to RDconnect visualization? (URL in development)
- Looks promising as a model for many things.
- MB: Resembles current implementation on SIB [Beacon+](#) with the addition of “handover” objects at different levels (e.g. Beacon response, allele response) => go ahead
  - link to handover discussion on Beacon [webpage](#)
- Good to have some well formed implementations with this working.
- More examples on next call

**Schema Examples:**

- Building on work in GA4GH Metadata team, now [Clinical Phenotype](#) (esp Phenopackets) and [Genomic Knowledge Standards](#) WS.
- Schema laid out in the [README](#)
- Schema definitions in [YAML file](#)
  - [Variant](#) (currently extension of VCF-derived GA4GH schema)
  - [Individual](#) (Generally human)
  - [Common](#) (Schema blocks / shared objects)
  - [Biosample](#) (e.g. DNA / proteins from genomic array / sequencing)
- [Classes](#) (for each of the above)

## 2018-10-16: Follow up from GA4GH Plenary

**Chair:** Gary

**Attendees:** Michael, Tony, Sabela, Niclas, Verkata, Marko, David, Lee Peng.

**Regrets:**

	Actions Arising	Assigned To	Deadline
1.	Beacon inventory		
2.	Beacon “branding”		

	Agenda Item	Speaker	Time
1.	Approve <a href="#">previous minutes</a> and confirm Agenda	Chair	5 min
2.	ELIXIR-SE Beacon project plans ( <a href="#">slides</a> )	Niclas	15 min
3.	Review of Actions (None)	David	0 min
4.	GA4GH Beacon workshop review ( <a href="#">Agenda</a> )	Gary	5 min
5.	2019-21 Project Plan approved by HoN; contracts being generated at Hub	Gary	5 min
6.	Technical <a href="#">Update</a> ( <i>Current sprint in this <a href="#">link</a></i> )	Juha	10 min
7.	Schema examples (ClinPhen group, Phenopackets) ( <i>rolled over from last call</i> ) <ul style="list-style-type: none"> <li>• First draft of schema elements on <a href="#">Github</a></li> </ul>	Michael	15 min
8.	A.O.B?		5 min
9.	Forthcoming activity: 6th Nov Technical Call 13th Nov Strategic Call		

	Previous Actions	Assigned	Deadline	Update
1.				

Minutes:

NJ:

- 20 Healthcare universities across SE - some have hospitals
- Also have SciLife lab - national hub for health and environmental research:
  - Funded by 4 universities and the national government
  - Includes NBIS (the Swedish ELIXIR Node), which represents 11 institutes
- Data Centre has a Beacon; Clinical hospitals have a Beacon
- Research Beacon: 1000 individuals
- Clinical Beacon: Rare Diseases WGS data
- Want closed registered access Beacon Network - Genomics Medicine Network Sweden; then expand into other countries
- Willing to be a test case for Clinical Beacon

Q: research Beacon using latest version of the specification?

Current Beacon is homebrewed. Switching over to the FI ref python implementation

Q: research Beacon - is this a public Beacon?

Q: handover? We are building this during this sprint (hopefully) within this sprint

Q: role in the clinical beacon?

A: clinical Beacon not part of the SE Node; but Node collaborates with them; both SciLife lab

Don't expect ELIXIR funding; national funding

Q: TB: EJP-RD - 24 ERN (European Reference Networks): software, standards, networks, etc.

GA4GH Workshop summary

A good chance to meet face to face, a significant turn out.

Presentation included only one ref. implementation - several across Europe

- Barcelona
- Finland

Which beacons exist, which are using these Implementations or an aggregate.

Gary - to contact each Beacon, (add details on the webpage)

GA4GH developments

Search API, whether complimenting / aligned with Beacon, fine.

Range Query; Match any variables in a range

(Neutral issue for Clinical data)

AI: branding for Beacon; and scope out the Beacons across ELIXIR

E.g.

**Standard Beacon**

- sample or aggregate

**Clinical Beacon**

- single sample (non-aggregate) BAM/phenotypic data
- allows the backend to connect the found variant to samples or individuals, including metadata. (Controlled access dataset)

**Evidence Beacon**

- On data collection of knowledge resources (for example ClinVar: VICC)
- Search delivers variants and the biological impact

Handover specification e.g. BRCAexchange Beacon - use the handover plus range query to have all variants in e.g. exon 1. Can download a VCF with the lines, or (better) use htsgget to stream into a genome browser

Establish these definitions, then ask across Beacons as to who has which.

## 2018-10-09 Tech. Call.

Agreed to list outstanding PR on the Sprint Slides ahead of the TC - encourage people to review these PR.

Current and next sprint: [document](#)

General agreement that technical development could be more active and effective.