The ELIXIR Beacon in 2018: A driver project of GA4GH

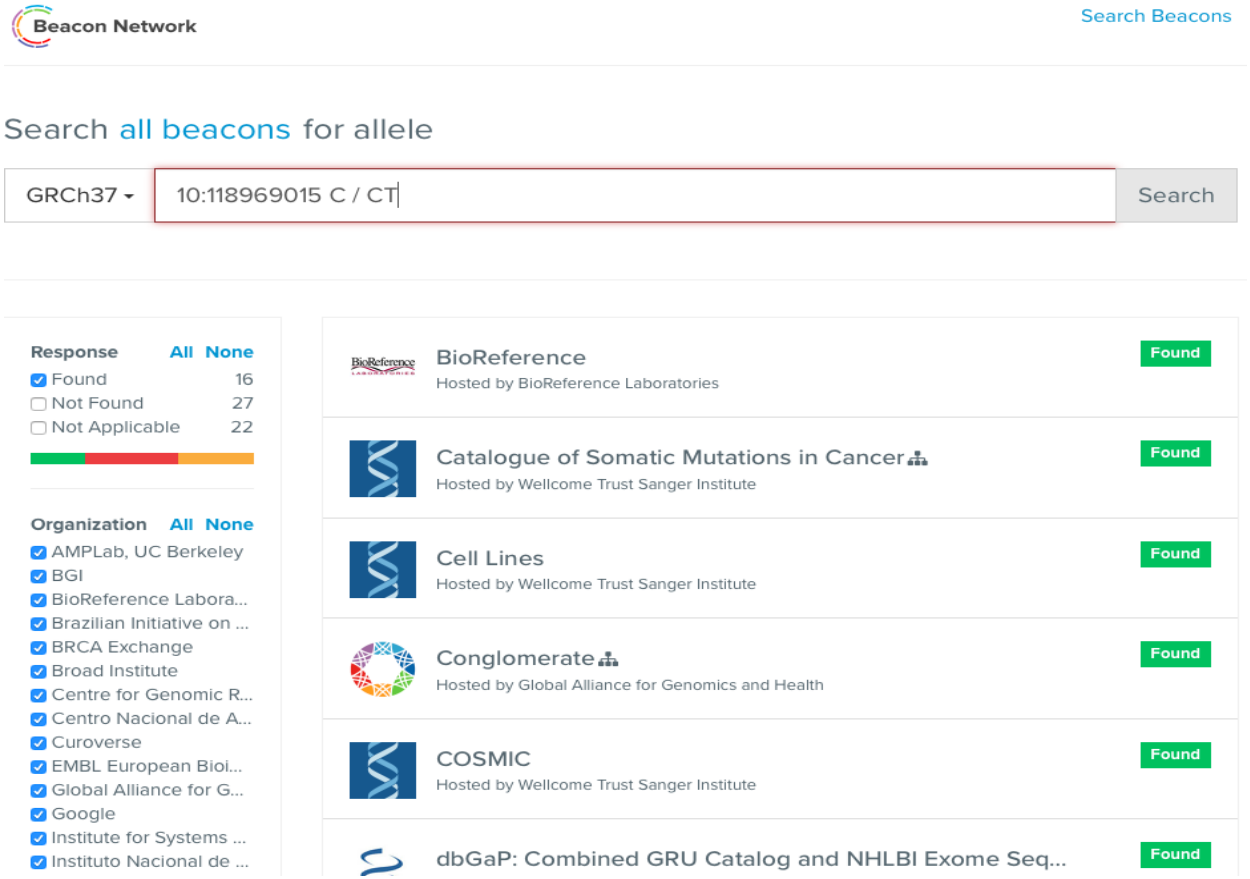
Michael Baudis, University of Zurich | **SIB**

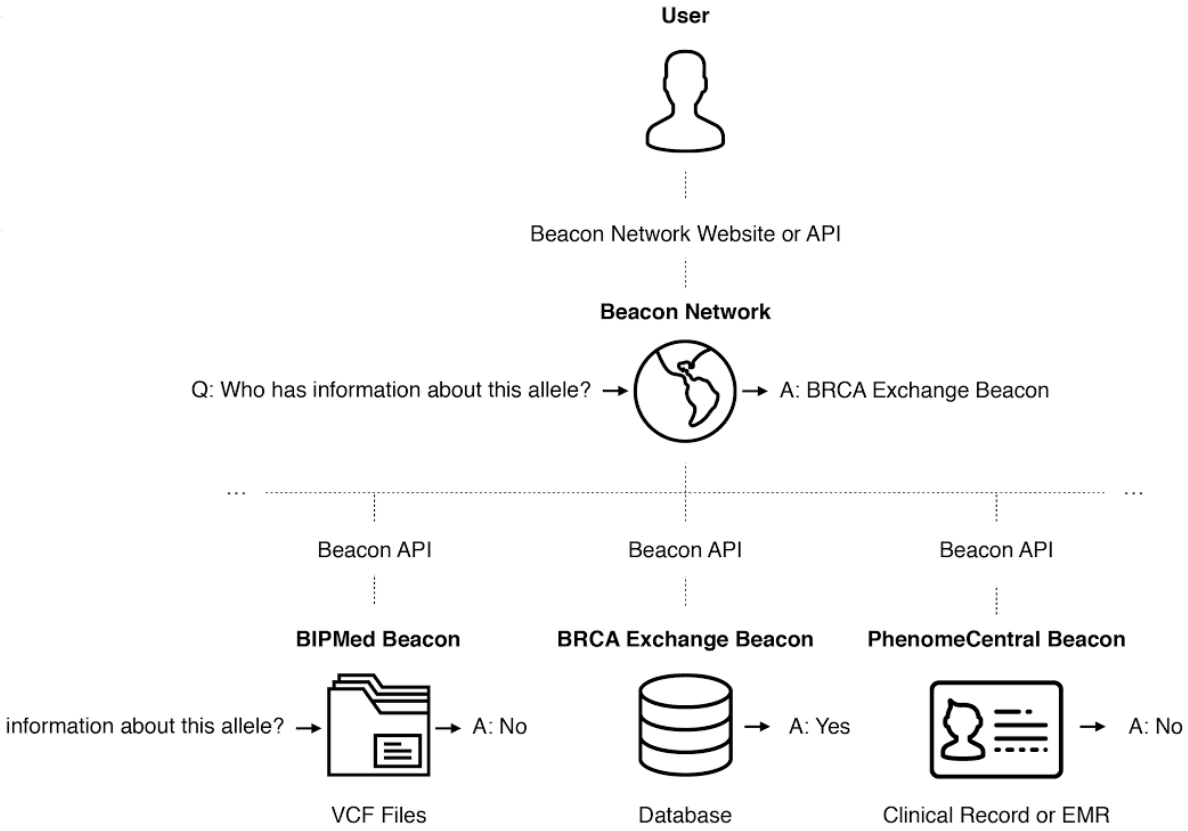
ELIXIR All Hands 2018, 4-7 June 2018, Berlin, Germany

Global Alliance
for Genomics & Health

Beacon Project

An open web service that tests the willingness of international sites to share genetic data.





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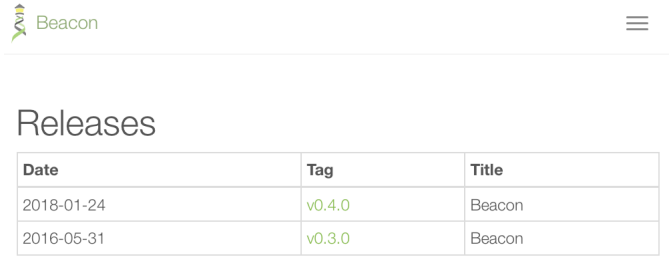
35+ Organizations

90+ Beacons

200+ Datasets

100K+ Individuals



ELIXIR Beacon Project 2018

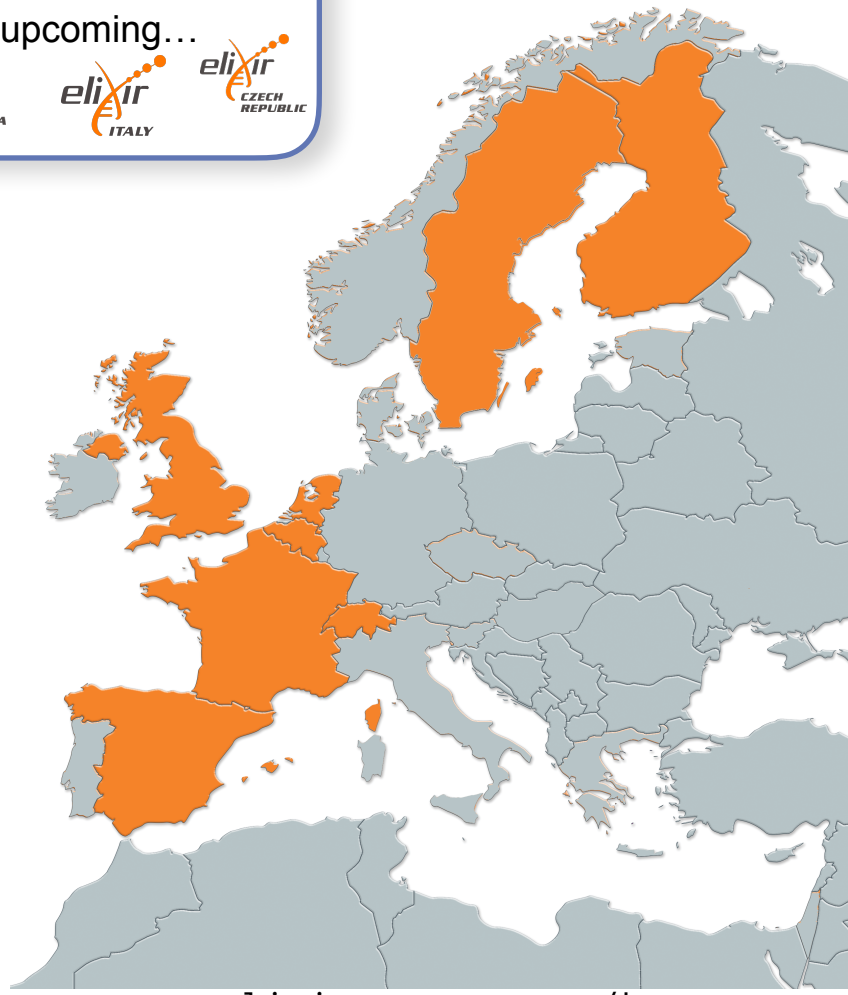
- WP1 - Development of Beacon API specification with the GA4GH
- WP2 - Supporting new ELIXIR Beacon development and deployment across Europe
- WP3 - Implementation of an ELIXIR Beacon Network & Registry
- WP4 - Security of ELIXIR Beacon and Beacon Network
- WP5 - Developing Indicators to establish Registry and Beacon as an Emerging ELIXIR Service
- WP6 - Strategic partnerships with national data cohorts and biobanks
- WP7 - Project Management and Communication

Node	Name of PI	Role	PMs	Other Contrib	Work Packages						
					1	2	3	4	5	6	7
FI	Ilkka Lappalainen	co-lead	5		X	X	X	X	X	X	X
ES	Jordi Rambla	lead	5		X	X	X				X
CH	Michael Baudis, (Heinz Stockinger)	co-lead	5		X					X	X
EBI	Dylan Spalding	co-lead	5		X	X	X	X			
SE	Niclas Jareborg	member	1			X					
BE	Yves Moreau	member	1			X					
FR	Macha Nikolski	member	1			X					
NL	Morris Swertz	member	1			X					
IT	David Horner	member	-								
FI	ELIXIR Compute Platform - Tommi Nyrönen			2							
TBD[6]	ELIXIR Training Platform			1		X					
HUB	Serena Scollen, Susheel Varma	member coordinator	0.5		X				X	X	X

Node work package participation, as in 2018 project plan

2018 Progress

- Release of v.0.4 specification
  - basis for **GA4GH Beacon v.1.0**
  - undergoing GA4GH certification (first for procedure)
- Building flexible **Beacon Network**(s)
- Beacon paper under review in *Nature Biotechnology*
- Post-2018 planning, documented in **5-year plan**
- Widening Node participation & additional use cases:
  - ELIXIR Norway joined Implementation Study, to light Beacon(s) against **Marine data**
  - Widen this out to more use cases? Plants, other…



www.elixir-europe.org/beacons

Security and GDPR

- Requirements analysis document, outlining **re-identification mitigation**
  - Individual Beacon and network of Beacons
- GA4GH Data **Security questionnaire** completed
- GDPR workshop in Brussels:
  - Raw data subject to recitals 33 (scientific research) and 34 (genetic data)
  - GDPR not applicable to anonymous data (recital 26)
- Genetic data kept anonymous by re-identification mitigation techniques
- Access restrictions (**tiered Beacon access**) can be adapted for each node
  - Accounts for local legal interpretations

Extending the protocol ...

Beacon+

beacon.progenetix.org

This forward looking Beacon interface implements additional, planned features beyond the current GA4GH specifications. Info

Query

SNV Example | DGV Example | CNV Example

Dataset	tcga
Reference name*	9
Genome Assembly*	GRCh38 / hg
Start *min* Position*	19,500,000
Start *max* Position	21,975,098
End *min* Position	21,967,753
End *max* Position	24,500,000
Alt. Base(s)*	DEL
Bio-ontology	icdot:c50.9: (4065)

Beacon Implementations

- implementing existing resources with Beacon protocol
- e.g. TCGA cancer variants (structural and SNV)

Beacon Response

- quantitative (counts for variants, callsets and samples)
- *Handover* to authentication system for data retrieval
- **no exposure** of data beyond standard Beacon response and additional pointer to matched data

Prototyping Query Extensions

- testing e.g. bio-metadata queries using ontology terms

Dataset	Assembly	Chro	Start Range	End Range	Pos	Ref Alt	Bio Query	Variants Calls Samples	$f_{alleles}$	Response Context
tcga	hg38	9	19,500,000 21,975,098	21,967,753 24,500,000		DEL	icdot:c50.9	54 54 54	0.0243	JSON UCSC Handover

arrayMap progenetix This Beacon implementation is developed by the Computational Oncogenomics Group at the University of Zurich, with support from the SIB Technology group and ELIXIR.

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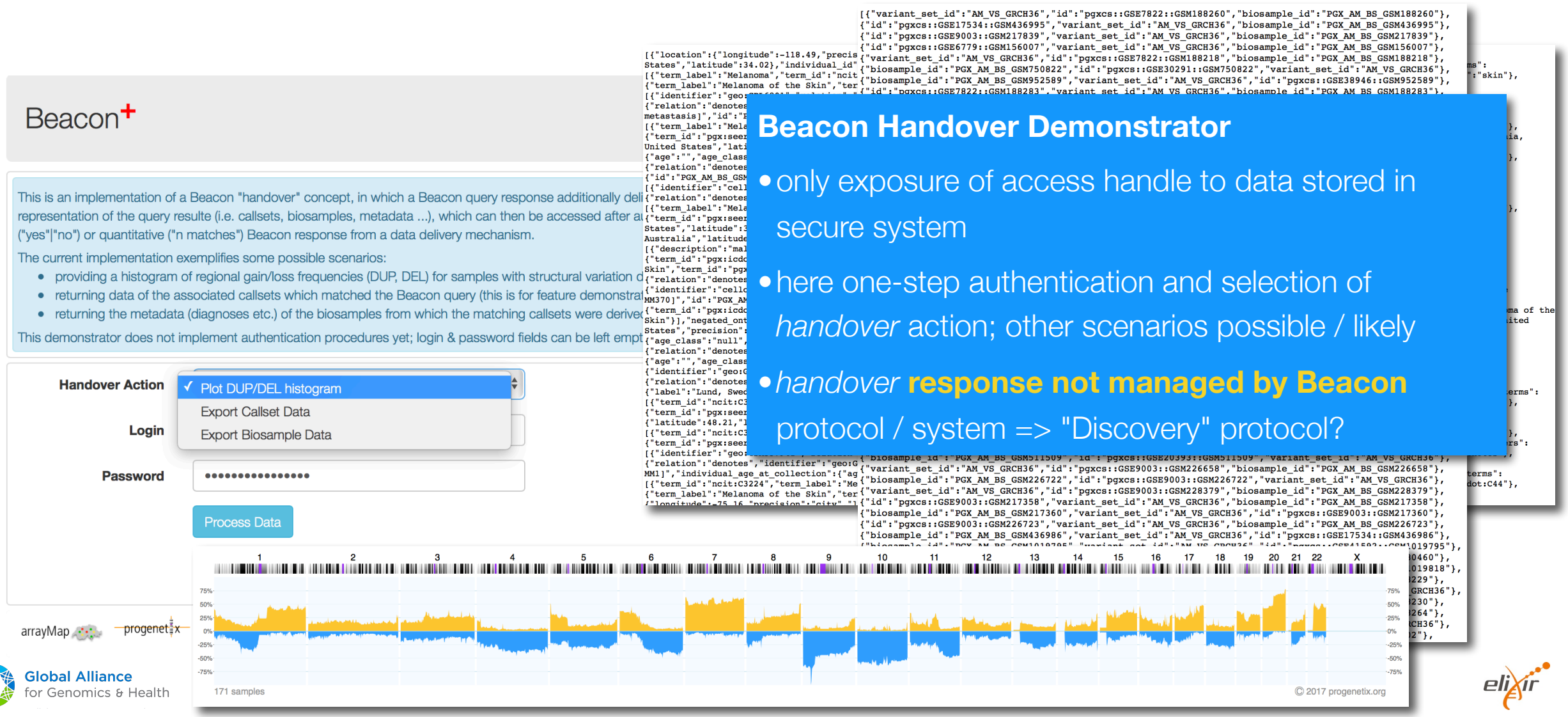
Beacon query => Handover Handle => Authentication => Data Retrieval

Beacon+ example implementation using public somatic variation data



Beacon Handover Demonstrator

- only exposure of access handle to data stored in secure system
- here one-step authentication and selection of *handover* action; other scenarios possible / likely
- *handover* **response not managed by Beacon** protocol / system => "Discovery" protocol?

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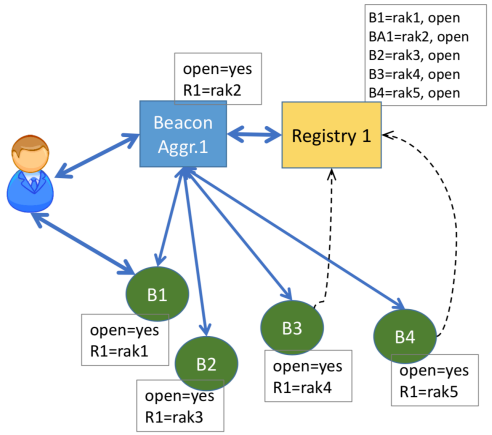
Towards a shining future...

- extensive/complete representation of genome **variant types** in query
  - close coordination with **GA4GH::GKS** and **GA4GH::Discovery** work streams
- providing a tested model for layered **registered access**
  - ELIXIR AAI
- implementing **Beacon network**(s) throughout ELIXIR
  - open protocols for extension and external implementations
- extending Beacon query protocol (**metadata**...)
  - keeping "aggregate response" model
- Beacon queries as entry points for data delivery, using "**handoff**" scenarios



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Towards a shining future... **"The" Genome Query API**

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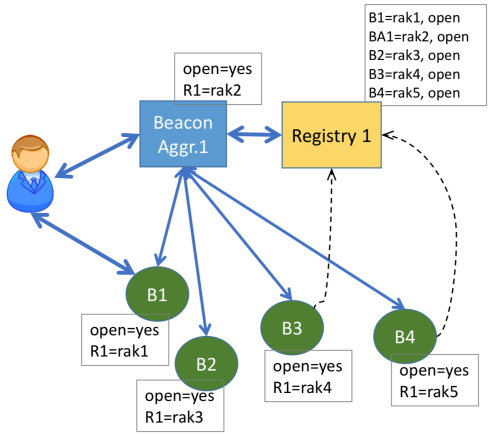

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Reminder: Beacon WorkShop Thursday a.m.

Thank You!

... and many more

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