Beacon Beacon Interface implements additional, planned features beyond the current GA4GH specifications.														
Query								SNV Ex	kample DG\	V Example	CNV Example			
	Dataset	arraymap			\$									
Reference name*		9												
Genome Assembly*		GRCh36 / hg18 \$				Beacon Response								
Variant type*		DEL (Deletion) \$				 quantitative (counts for variants, callsets and samples) 								
Start min Position* Start max Position End min Position End max Position Bio-ontology		19,500,000 21,964,826 21,958,228 24,500,000 ncit:c3224: Melanoma (1098)				 Handover to authentication system for data retrieval no exposure of data beyond standard Beacon 								
						resp	UHSE	and at	JUILION		ter to m	lateriec	Uala	-5555
								Beacon Qu	ery					
Response														
Dataset	Assembly	Chro	Var Type	Start Range	End Range	Pos	Ref Alt	Bio Query	Variants Calls Samples	f _{alleles}	Response Context			
arraymap	hg18	9	DEL	19,500,000 21,964,826	21,958,228 24,500,000			ncit:c3224	157 171 171	0.1557	JSON UCSC Handover			
arrayMap 🐽	−progenet ‡x			is developed by the logy group and ELIX		Oncogenomic	s Group at	the University of	f Zurich, with	Univers				

```
"dataset allele responses" : [
         "dataset id" : "arraymap",
         "call count" : "171",
         "sample count" : "171",
         "variant count" : "157",
         "error" : null,
         "exists" : true,
         "external url": "http://beacon.arraymap.org",
         "frequency": "0.1557",
         "info": {
            "callset access handle" : "d5850347-d411-11e7-8c89-ec436516cb41",
            "description" : "The query was against database \"arraymap ga4gh\",
variant collection \"variants cnv grch36\". 171 matched calls for 157 distinct
variants. Out of 62033 biosamples in the database, 1098 matched the biosample
query; of those, 171 had the variant.",
         "note" : ""
```

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 outside of Beacon protocol / system



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

Beacon+ example implementation using public somatic variation data

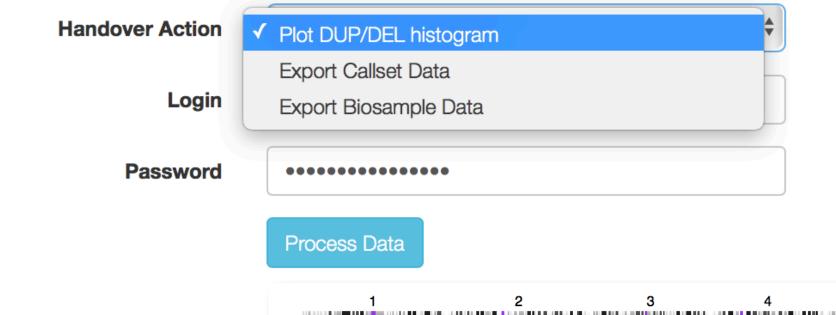


This is an implementation of a Beacon "handover" concept, in which a Beacon guery response additionally de representation of the guery resulte (i.e. callsets, biosamples, metadata ...), which can then be accessed after ("yes"|"no") or quantitative ("n matches") Beacon response from a data delivery mechanism.

The current implementation exemplifies some possible scenarios:

- providing a histogram of regional gain/loss frequencies (DUP, DEL) for samples with structural variation
- returning data of the associated callsets which matched the Beacon guery (this is for feature demonstrated)
- returning the metadata (diagnoses etc.) of the biosamples from which the matching callsets were derived {"term_id":"pgx:icdd

This demonstrator does not implement authentication procedures yet; login & password fields can be left empt | States , precision | Precis



[{"identifier": "geo: {"relation": "denotes {"term_id":"pgx:seen United States", "lat. {"age":"","age_class [{"identifier":"cel {"relation": "denotes [{"term_label":"Mela l {"term_id":"pgx:seer States","latitude":3 Australia","latitude [{"description": "mal {"term_id":"pgx:icdo Skin","term_id":"pgx [{ "relation": "denotes at {"identifier":"celle MM370]","id":"PGX_AN Skin" }] , "negated_ont States", "precision" {"relation": "denotes {"age":"","age_class {"identifier": "geo: {"relation":"denotes [{"term_id":"ncit:C3 {"term_id":"pgx:see {"latitude":48.21, [{"term_id":"ncit:C3 {"term_id":"pgx:see:

[{"identifier": "geo:

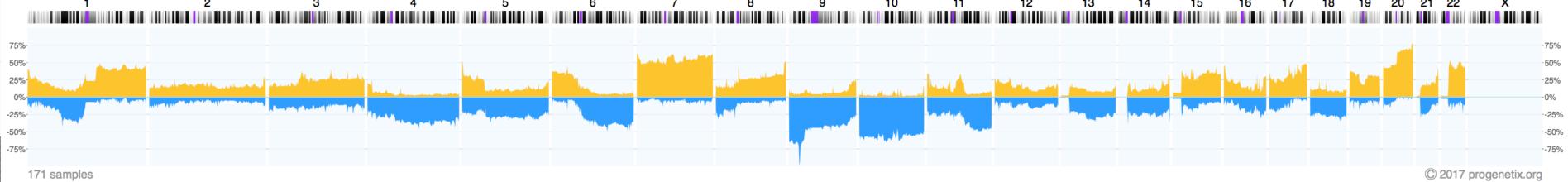
["id":"pgxcs::GSE17534::GSM436995","variant set id":"AM VS GRCH36","biosample id":"PGX AM BS GSM436995"} ["id":"pgxcs::GSE9003::GSM217839","variant set id":"AM VS GRCH36","biosample id":"PGX AM BS GSM217839"}, ["id":"pgxcs::GSE6779::GSM156007","variant set id":"AM VS GRCH36","biosample id":"PGX AM BS GSM156007"}, {"location":{"longitude":-118.49, "precis { "variant_set_id":"AM_VS_GRCH36", "id":"pgxcs::GSE7822::GSM188218", "biosample_id":"PGX_AM_BS_GSM188218"}, States", "latitude":34.02}, "individual_id" {"variant_set_id":"AM_VS_GRCH36", "id":"pgxcs::GSE7822::GSM188218", "biosample_id":"PGX_AM_BS_GSM188218"},
[{"term_label":"Melanoma", "term_id":"ncit {"biosample_id":"PGX_AM_BS_GSM750822", "id":"pgxcs::GSE30291::GSM750822", "variant_set_id":"AM_VS_GRCH36"},
{"term_label":"Melanoma of the Skin" "ter {"biosample_id":"PGX_AM_BS_GSM952589", "variant_set_id":"AM_VS_GRCH36", "id":"pgxcs::GSE38946::GSM952589"},

{"variant set id":"AM VS GRCH36","id":"pgxcs::GSE7822::GSM188260","biosample id":"PGX AM BS GSM188260"}

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?

[{"term_id": "ncit:C3224", "term_label": "Me {"biosample_id": "PGX_AM_BS_GSM226722", "id": "pgxcs::GSE9003::GSM226722", "variant_set_id": "AM_VS_GRCH36"}, {"term_label": "Melanoma of the Skin", "ter{"variant_set_id": "AM_VS_GRCH36", "id": "pgxcs::GSE9003::GSM228379", "biosample_id": "PGX_AM_BS_GSM228379"}, ogitude".-75 16 "precision"."city" "1 {"id":"pgxcs::GSE9003::GSM217358","variant set id":"AM VS GRCH36","biosample id":"PGX AM BS GSM217358"}, {"biosample id": "PGX AM BS GSM217360", "variant set id": "AM VS GRCH36", "id": "pgxcs::GSE9003::GSM217360"}, {"id":"pgxcs::GSE9003::GSM226723","variant_set_id":"AM_VS_GRCH36","biosample_id":"PGX_AM_BS_GSM226723"}, {"biosample id":"PGX AM BS GSM436986","variant set id":"AM VS GRCH36","id":"pgxcs::GSE17534::GSM436986"}



dot:C44"},

CH36"},