



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

Info

Query

SNV Example

DGV Example

CNV Example

Dataset

arraymap

Reference name*

9

Genome Assembly*

GRCh36 / hg18

Variant type*

DEL (Deletion)

Start *min* Position*

19,500,000

Start *max* Position

21,964,826

End *min* Position

21,958,228

End *max* Position

24,500,000

Bio-ontology

ncit:c3224: Melanoma (1098)

Beacon Query

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

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

beacon.progenetix.org



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**

```
"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" : "",  
  },  
],
```



