A Linked Data Representation for Summary Statistics and Grouping Criteria

RPI IDEA/Tetherless World Constellation

James P. McCusker, Michel Dumontier, Shruthi Chari, Joanne S. Luciano, and Deborah L. McGuinness

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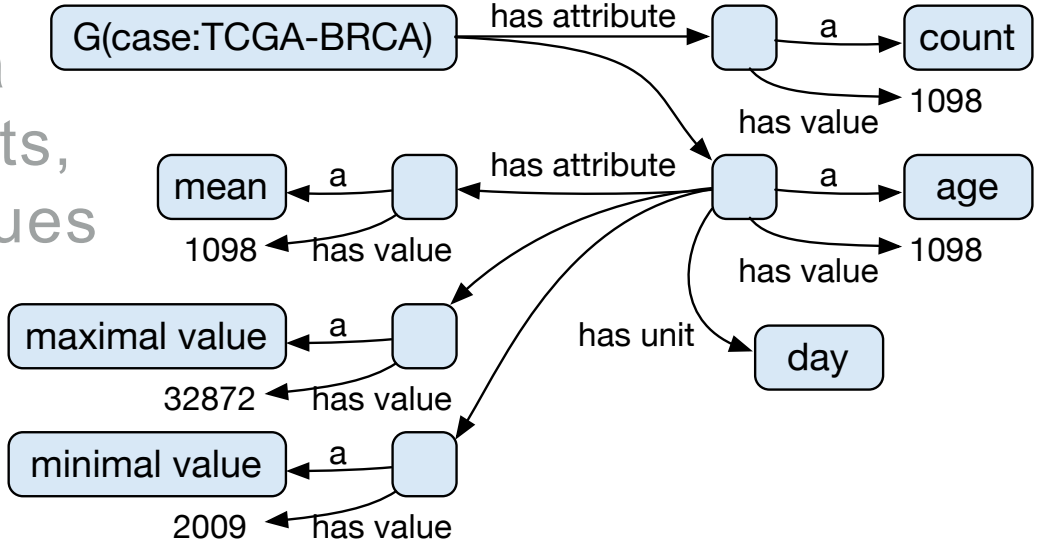
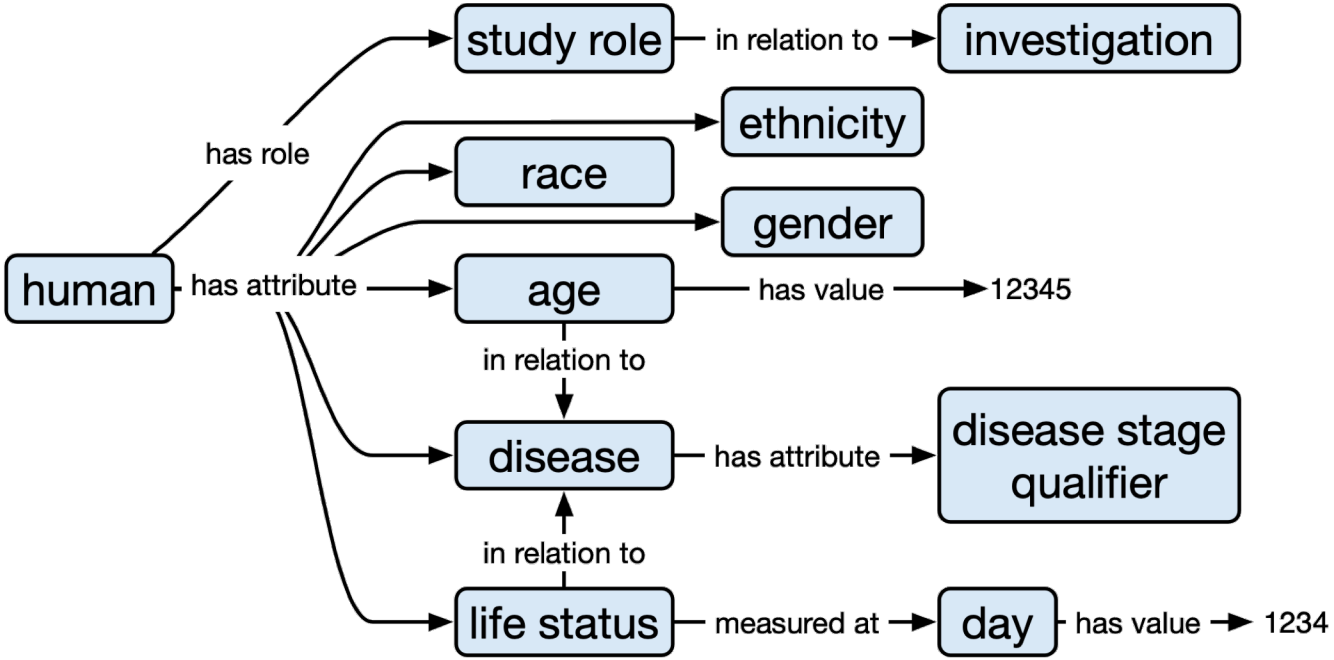
Summary statistics across groups can be formalized as **linked data** using *owl:Class*-based sets, expressing aggregate values as attributes of those classes.

Class: G(case:TCGA-BRCA)
SubClassOf: sio:human and
sio:'has role' some (sio:'subject role'
and sio:'in relation to' value case:TCGA-BRCA)



Example Data Schema – Genomic Data Commons Clinical Annotations



Defining Grouping Criteria (starting with Calvanese *et al.* 2008)

OWL

```
Class: GDC_Subject
EquivalentTo: sio:human
  and sio:'has role' some (sio:'subject role'
    and sio:'in relation to' some sio:investigation)
```

SPARQL

```
select ?GDC_Subject WHERE {
  ?GDC_Subject a sio:SIO_000485; # human
   sio:SIO_000228 [ # has role
      a sio:SIO_000883; # study subject
      sio:SIO_000668 [ # in relation to
        a sio:SIO_000747 # investigation
      ]
    ].
}
```

Defining Grouping Criteria (starting with Calvanese *et al.* 2008)

$$q\left(\bar{x}, \alpha\left(\bar{y}\right)\right) \leftarrow \phi$$

where

$$\texttt{Class:}\ \bar{x}$$
$$\texttt{SubClassOf:}\ \phi$$

We will reserve $\alpha(\bar{y})$ for later.

Class: $\bar{x}$
SubClassOf: ϕ

$$\bar{x} = G(g_1, \ldots, g_n)$$

Class: $G(\ g_1, \ldots, g_n)$
SubClassOf: ϕ

```
Class: G(?x)
SubClassOf: sio:human
  and sio:'has role' some (sio:'subject role'
    and sio:'in relation to' value ?x)
```

Grouping Criteria as a SPARQL query

```
select ?GDC_Subject ?x where {
   ?GDC_Subject a sio:SIO_000485; # human
   sio:SIO_000228 [ # has role
      a sio:SIO_000883; # study subject
      sio:SIO_000668 ?x # in relation to
   ].
   ?x a sio:SIO_000747 # investigation
}
```

```
Class: G(?x)
SubClassOf: sio:human
   and sio:'has role' some (sio:'subject role'
      and sio:'in relation to' value ?x)
```

Grouped Criteria as expanded classes

```
Class: G(?x)
SubClassOf: sio:human
  and sio:'has role' some (sio:'subject role'
    and sio:'in relation to' value ?x)
```

Class: G(case:FM-AD)
SubClassOf: sio:human
and sio:'has role' some (sio:'subject role'
and sio:'in relation to' value case:FM-AD)

Class: G(case:TARGET-NBL)
SubClassOf: sio:human and
sio:'has role' some (sio:'subject role'
and sio:'in relation to' value case:TARGET-NBL)

…

owl:Classes with **property restriction** definitions can be **assigned URIs automatically** based on the **graph digest** of that property restriction using RGDA1 or similar graph digest algorithms.

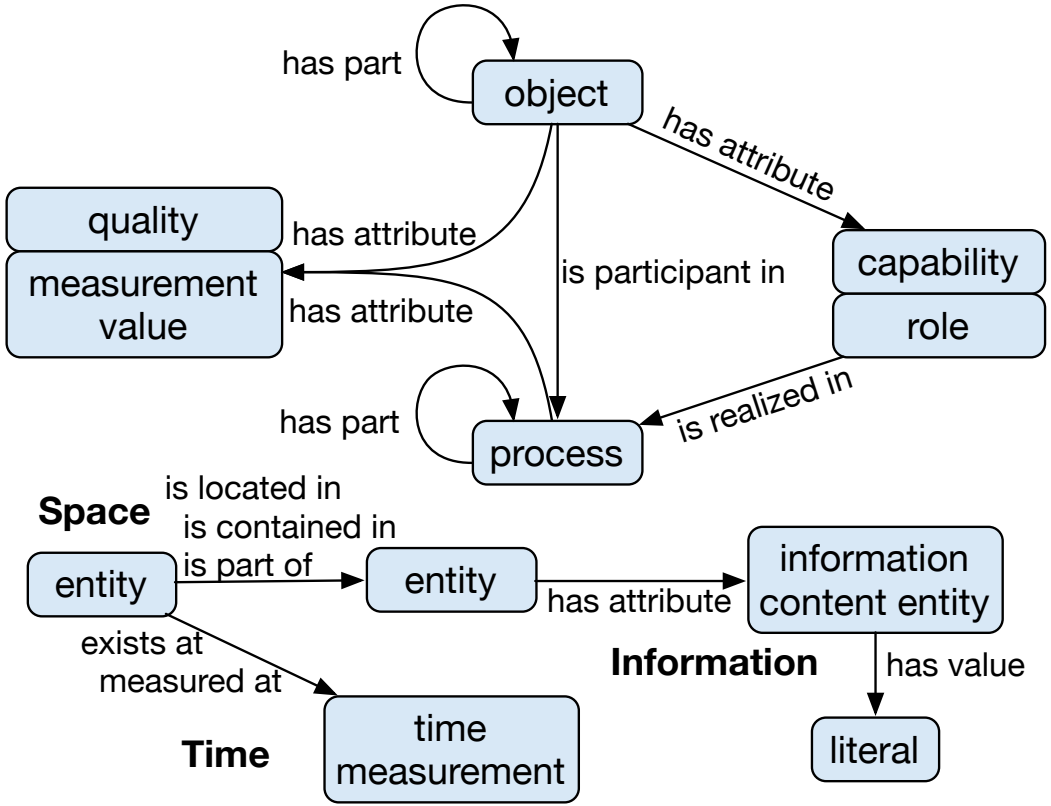
```
graph = IsomorphicGraph()

graph = source_graph.query("""
describe ?restr where {
  ?G owl:equivalentClass|rdfs:subClassOf ?restr.
}""", initBindings={"G":my.Class} )

digest = graph.graph_digest()

source_graph.add((
  my.Class,
  owl:equivalentClass,
  digest_prefix[digest]
))
```

WARNING! We will be discussing the use of OWL 2 puns.

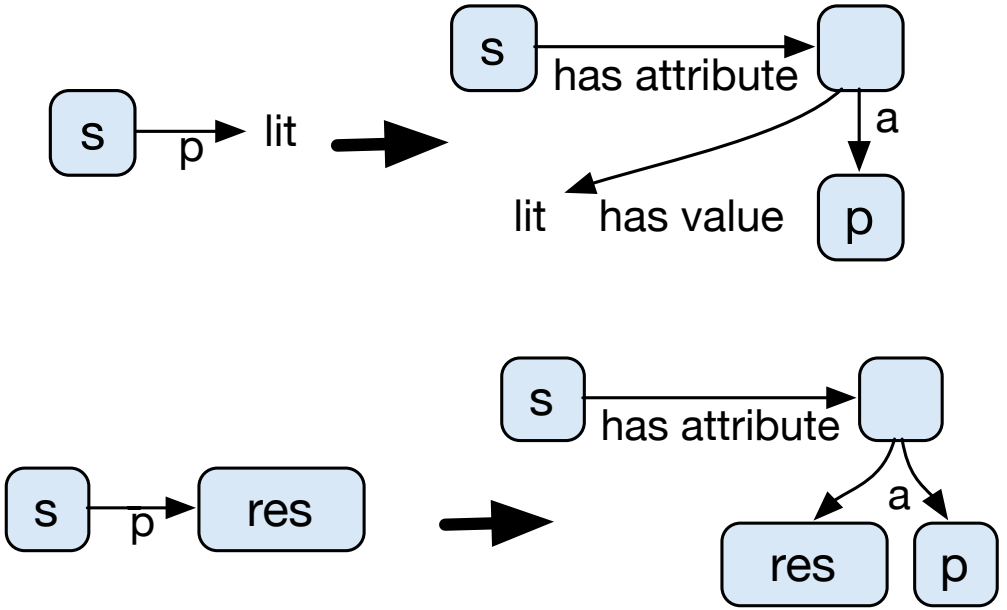
TL;DR for **OWL 2 Punning**:

Statements asserted about a resource as an OWL Class **cannot be used to draw inferences** about the resource as an OWL Individual or vice-versa.

Expressing aggregate values relies on the Semanticscience Integated Ontology, or an expressive equivalent.



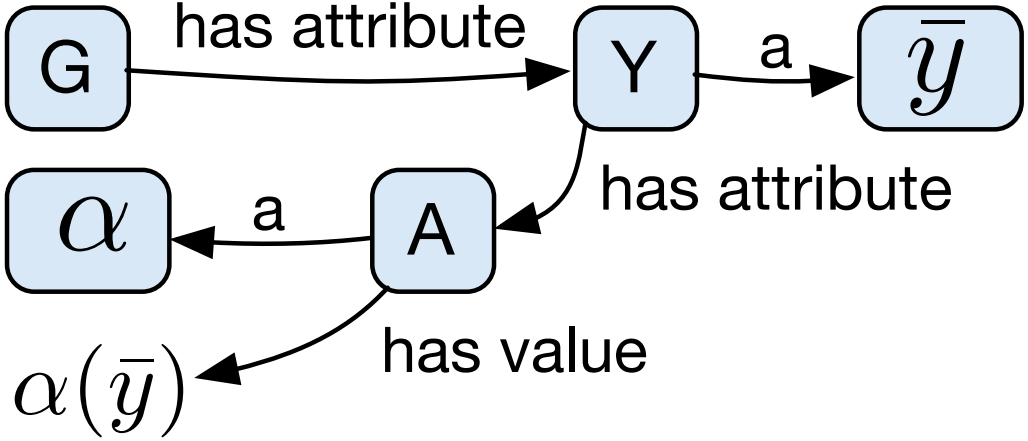
First, if needed we reify non-SIO statements as attributes.



Finally, here's what we do with $\alpha(\bar{y})$.

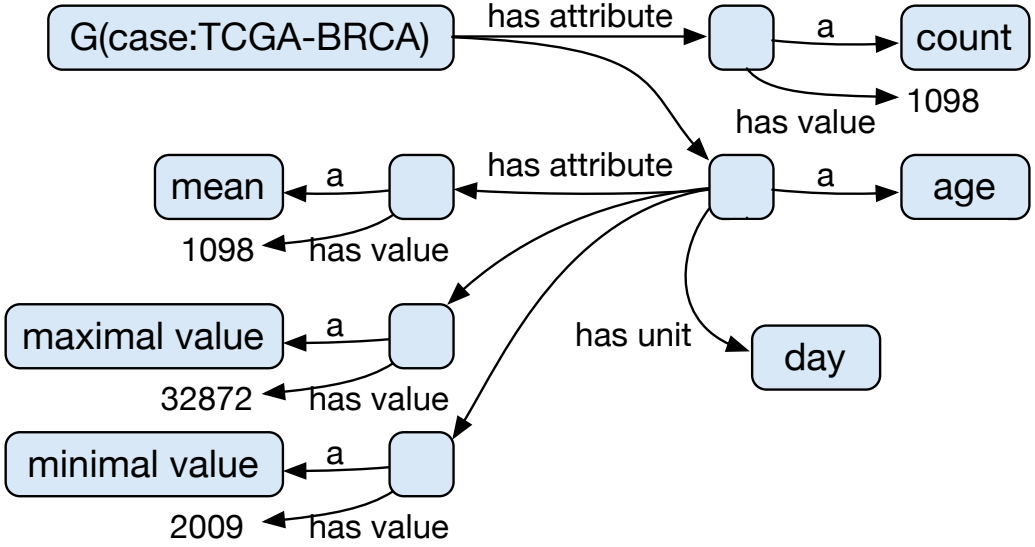
$$\forall G, \alpha(\bar{y}) \exists A \in \alpha, Y \in \bar{y}$$

$$attr\,(G, Y) \wedge attr\,(Y, A) \wedge val\,(A, \alpha(\bar{y}))$$



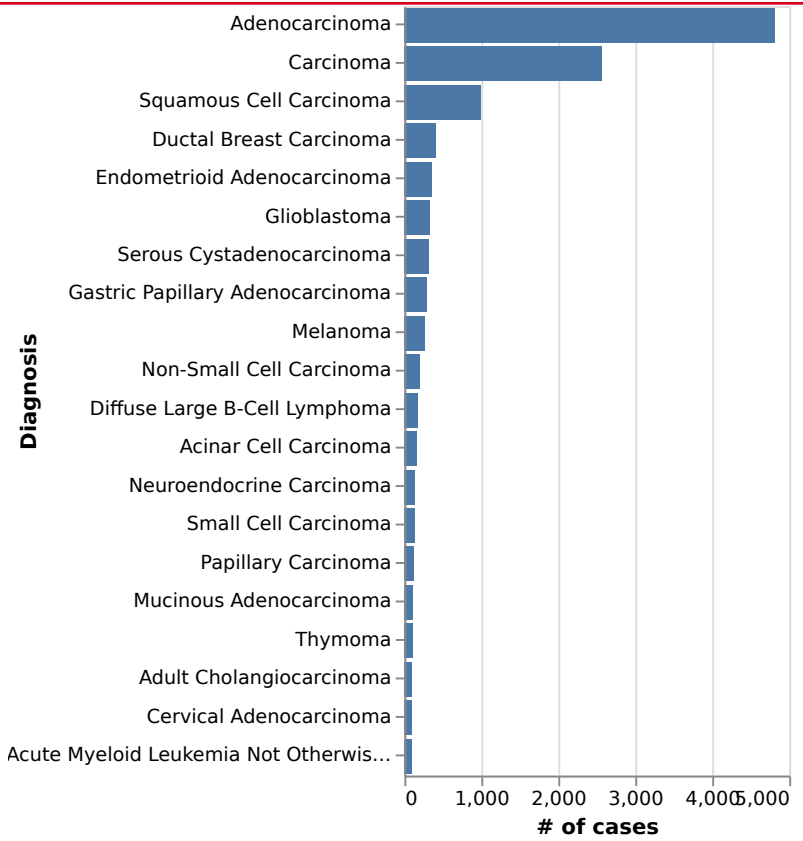
Here's what it looks like in practice.

Class: G(case:TCGA-BRCA)
SubClassOf: sio:human and
sio:'has role' some (sio:'subject role'
and sio:'in relation to' value case:TCGA-BRCA)



Implementation in Jupyter Notebook

- We can query summary statistics from an RDF graph and put the results into it's own graph.
- We query the statistics out and display them using Vega-Lite.



Many thanks to:

Coauthors: Deborah, Michel, Joanne, and Shruthi
Others whom I've bothered about this:
John Erickson, Patrice Seyed, and James Michaelis.

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