

The COVID-19 epidemiology and monitoring ontology

CEMO

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Precision intervention



Precision epidemiology

PERSPECTIVE

<https://doi.org/10.1038/s41591-019-0345-2>

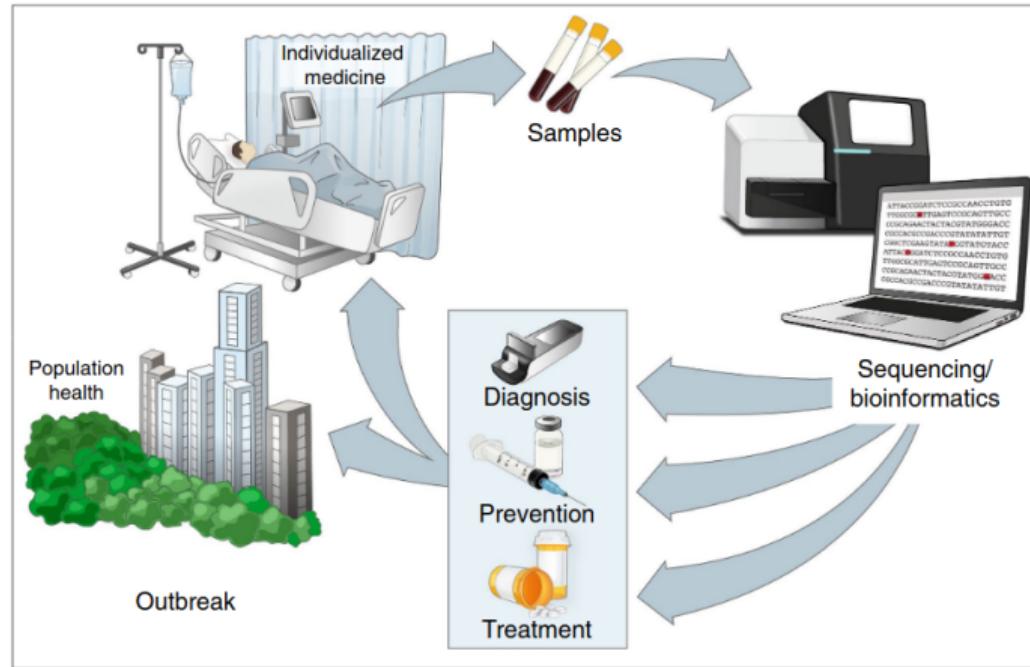
nature
medicine

Precision epidemiology for infectious disease control

Jason T. Ladner¹, Nathan D. Grubaugh², Oliver G. Pybus³ and Kristian G. Andersen^{4,5*}

Advances in genomics and computing are transforming the capacity for the characterization of biological systems, and researchers are now poised for a precision-focused transformation in the way they prepare for, and respond to, infectious diseases. This includes the use of genome-based approaches to inform molecular diagnosis and individual-level treatment regimens. In addition, advances in the speed and granularity of pathogen genome generation have improved the capability to track and understand pathogen transmission, leading to potential improvements in the design and implementation of population-level public health interventions. In this Perspective, we outline several trends that are driving the development of precision epidemiology of infectious disease and their implications for scientists' ability to respond to outbreaks.

Precision epidemiology



source: <https://doi.org/10.1038/s41591-019-0345-2>

Measurements for outbreak monitoring



source: <https://www.nature.com/collections/bedbejdbij>

Measurements for outbreak monitoring

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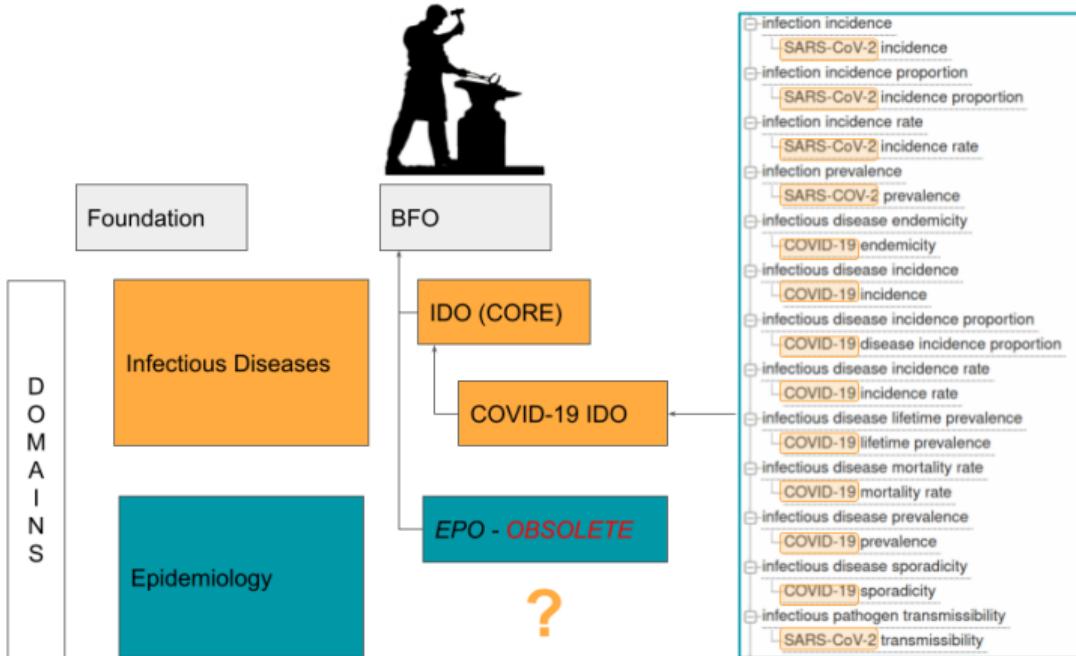
EDITORIAL | 27 January 2021

How epidemiology has shaped the COVID pandemic

Nature's third progress report, coming at the end of the pandemic's first year, highlights key findings from epidemiology – from sounding the early alarm to following the impact of new variants.

source: <https://media.nature.com/original/magazine-assets/d41586-021-00183-z/d41586-021-00183-z.pdf>

Ontologies for efficient epidemiology



source: <http://www.obofoundry.org/>

Hypothesis

Research question

- Semantic status for COVID epidemiology in OBO?

Objective

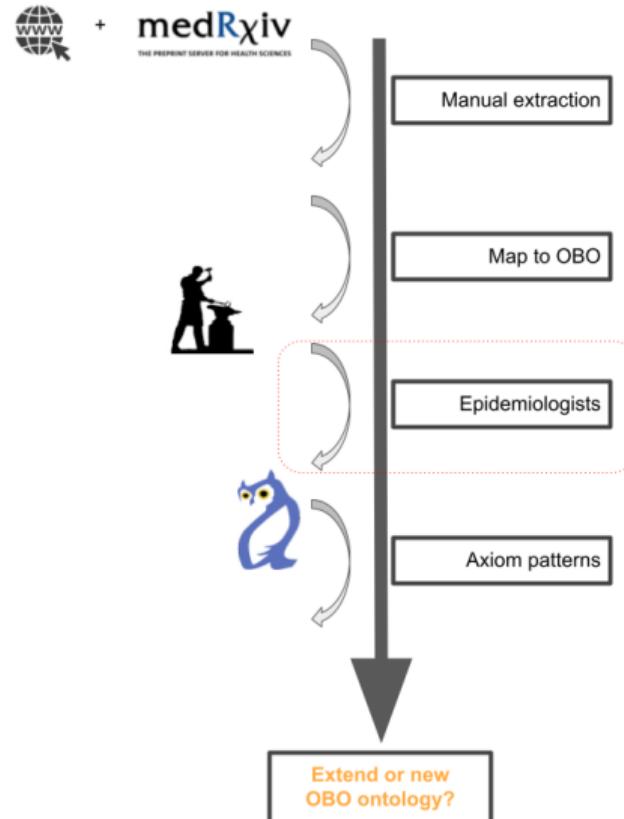
Improve epidemiological measurements ontological representation

Method

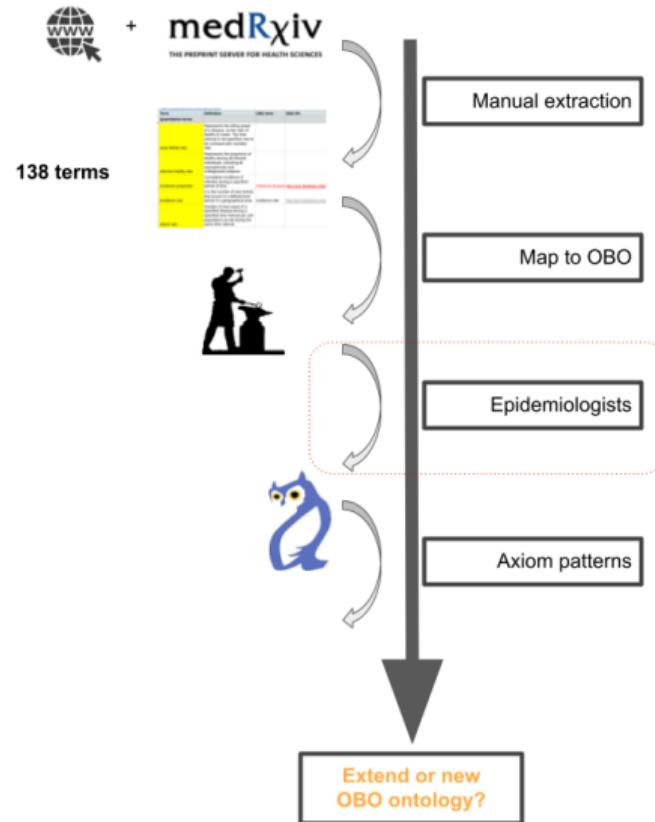
- Determine ontological status of epidemic measures terms
- Develop a minimal formal model
- Hackathons: **COVID-19 vBH20**, **BH20-EU**, **SWATATHON 21**



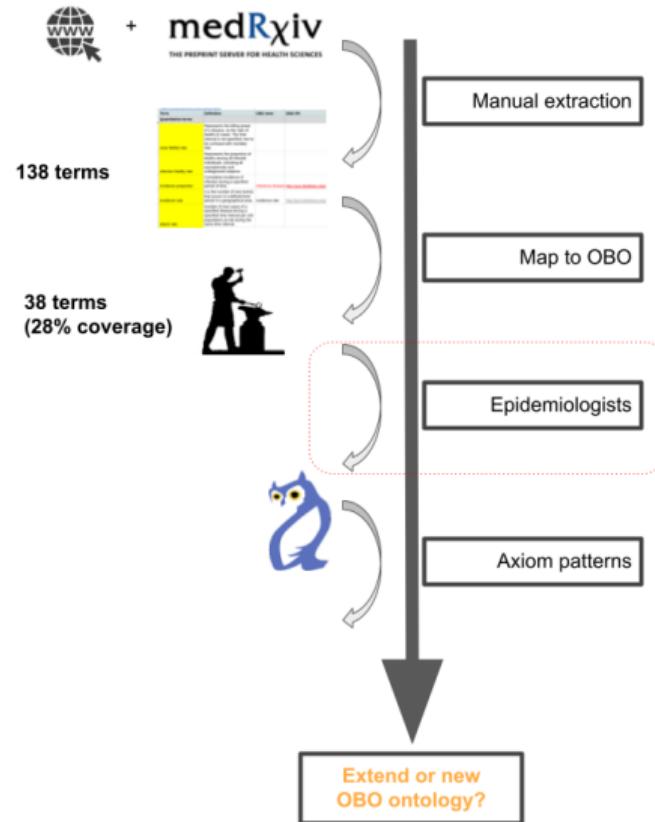
Workflow



Results

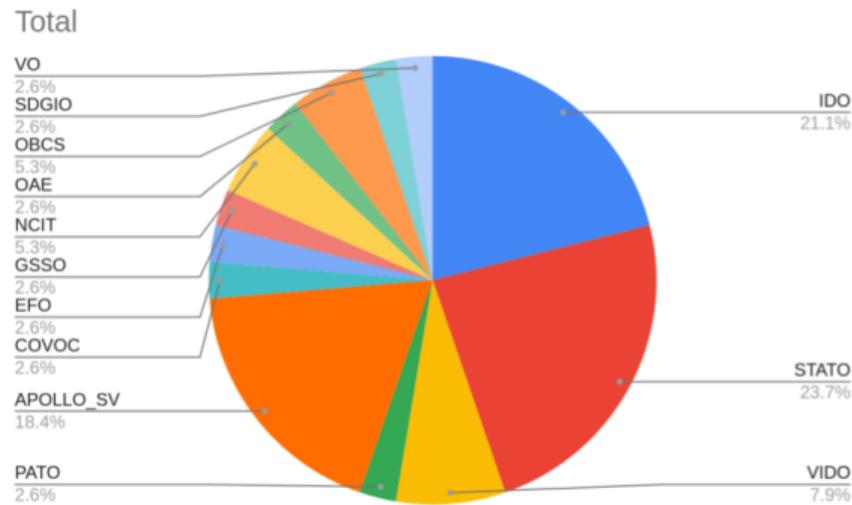


Results



Results

Low coverage and distributed in different ontologies



curated terms: <https://docs.google.com/spreadsheets/d/1kSOnSc6lVjrRj1ZoP8i2ZJI3UIwEzeHZ6BayxZXarSE/edit?usp=sharing>

Results

Described as quality

Infectious Disease Ontology

A set of interoperable ontologies that will together provide coverage of the infectious disease domain. IDO core is the upper-level ontology that hosts terms of general relevance across the domain, while extension ontologies host terms to specific to a particular part of the domain.

Search IDO Search

Terms Download Ontology Homepage Request a Term

Browse Terms Browse Properties Ontology history

Obsolete Class entity

- continuant
 - generically dependent continuant
 - independent continuant
 - specifically dependent continuant
 - quality
 - infection incidence
 - infection incidence proportion
 - infection incidence rate
 - infection prevalence
 - infectious agent seroprevalence
 - infectious agent transmissibility
 - infectious disease endemicity
 - infectious disease incidence
 - infectious disease incidence proportion
 - infectious disease incidence rate
 - infectious disease lifetime prevalence
 - infectious disease mortality rate
 - infectious disease prevalence
 - infectious disease sporadicity
 - infectivity
 - relational quality
 - susceptibility
 - virulence
 - realizable entity

Ontology information

Ontology IRI: <http://purl.obolibrary.org/obo/ido.owl>
Version IRI: <http://purl.obolibrary.org/obo/ido/2017-11-03/ido.owl>
Ontology ID: ido
Version: 2017-11-03
Number of terms: 518
Last loaded: Sun Aug 16 13:23:07 BST 2020

date 2017-11-03T15:13:52

license

creator Albert Goldstein
Barry Smith
Lindsay Cowell
Alan Rutenberg
Björn Peters
Alexander Diehl

defaultLanguage en

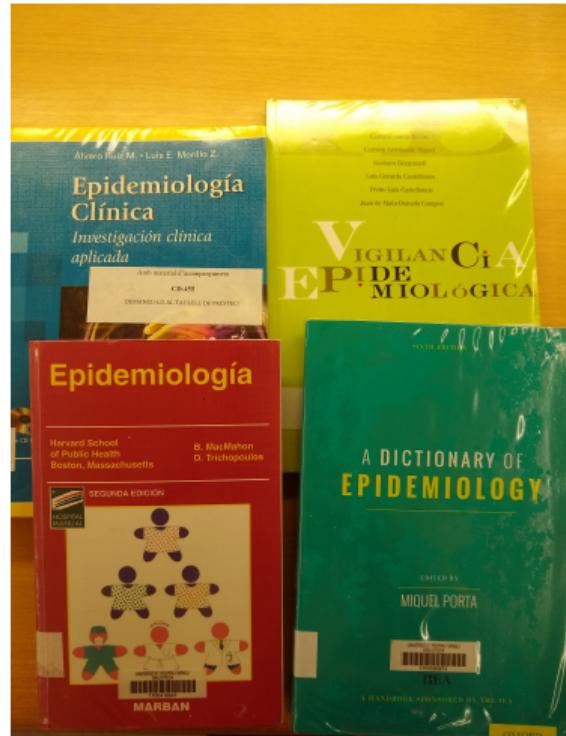
source: <https://www.ebi.ac.uk/ols/ontologies/ido>

Results

- **CEMO ontology (OWL)**
- Best practices: reuse and modular
- OBO ontology (BFO)
- Describes domain concepts and epidemic measures
- Axiom patterns
- Ontological model (SIO)

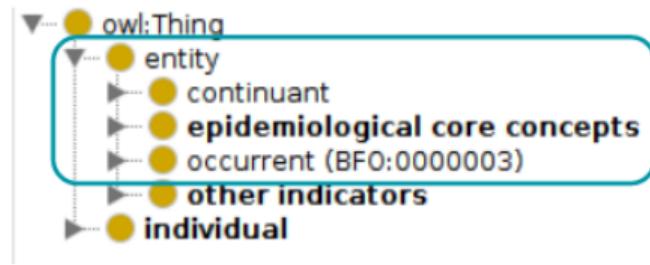
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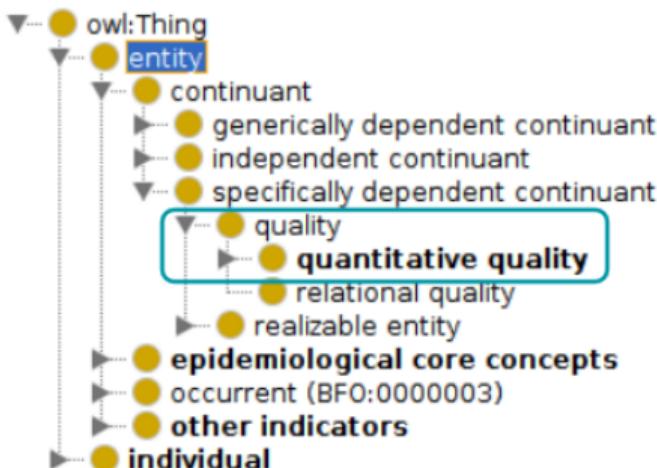
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Epidemic measures

:quantitative-quality SubClassOf
[(**:is-attribute-of** some *:descriptive-epi*)
and (**:is-about** some *:interpretation*)]

Lines of time (GFO)

:timeline SubClassOf
:process
and (**:caused-by** only *:process*)
and (**:has-participant** only *:person*)
and (**:projects-to** exactly 1 *gfo:chronoid*)]

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- **Ontological model (SIO)**

SIO Design Pattern

```
'quantity'
subClassOf
'has value' some Literal # consider owl:real (xsd:float, xsd:double, xsd:int, xsd:long)
and 'has unit' only 'measurement unit'
and 'is output of' only 'measurement process'
and 'is attribute of' some 'entity'
and 'measured at' some ('time instant' or 'time interval' and 'has value' some xsd:dateTime) #optional
```

Thus, entities can be described in terms of their quantified attributes as follows:

```
'entity'
subClassOf
'has attribute' some ( 'quantity' that 'has value' some Literal and 'has unit' some 'measurement unit' )
```

Sometimes, it is desirable to express that the value of a part of an entity:

```
'measurement of x'
subClassOf
'quantity'
and 'has value' some Literal
and 'has unit' some 'measurement unit'
and 'is attribute of' some ( 'object' and 'is part of' some 'object' )
```

Finally, we can represent the generation of a quantity as follows:

```
'measuring process'
subClassOf
'process'
and 'has agent' some 'object' # the agent (person, software, etc) initiating the measurement
and 'has target' some ('has capability' some 'to be measured') # the entity being measured
and 'has output' some 'quantity' # the measurement (see above)
and 'has attribute' some 'time interval' # the time at which the measuring took place
and 'has participant' some (object and 'has function' some 'to measure') # the device used to undertake the measurement
and 'conforms to' some 'effective specification' # a the protocol followed, if any
```

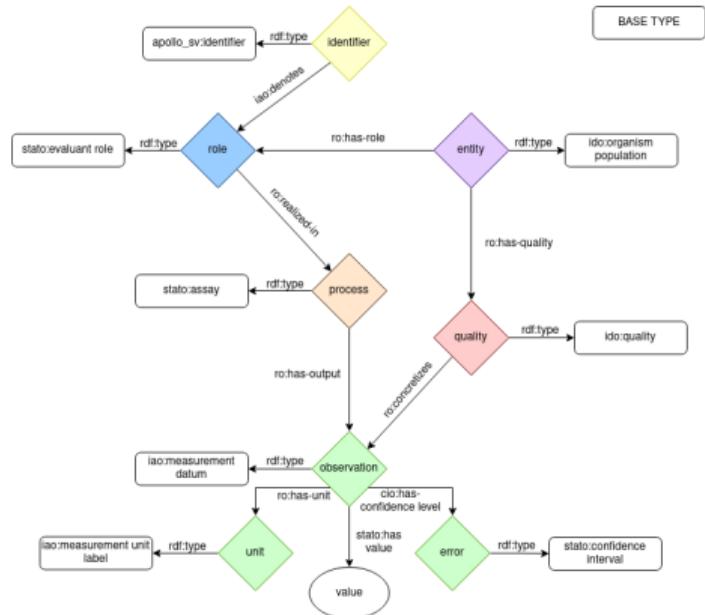
Results

EJP RD core model based on SIO (representation of health data **at patient level**)

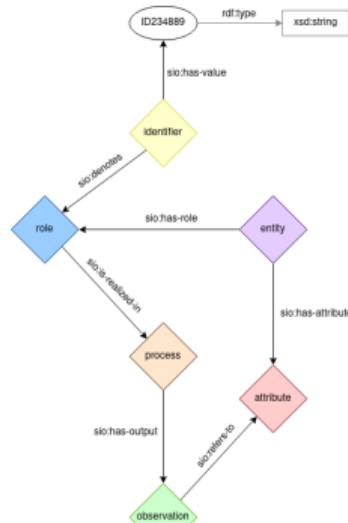


Results

CEMO ontological model (representation of statistical profiles at population level)



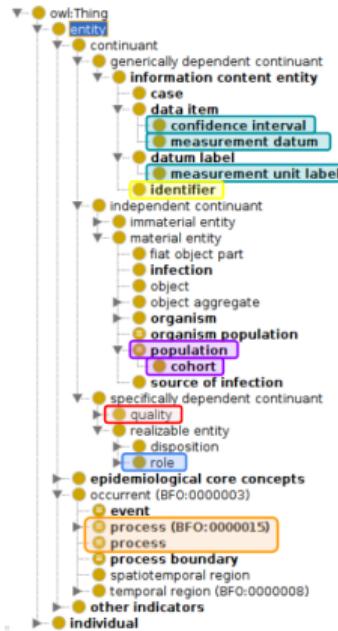
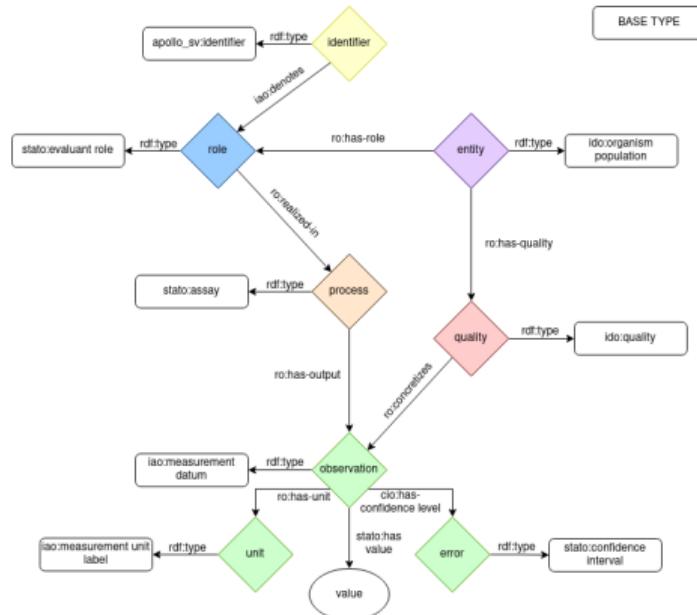
EJP RD core model based on SIO (representation of health data at patient level)



EJP RD Core model: <https://github.com/ejp-rd-vp/CDE-semantic-model/wiki/Core-model-SIO>

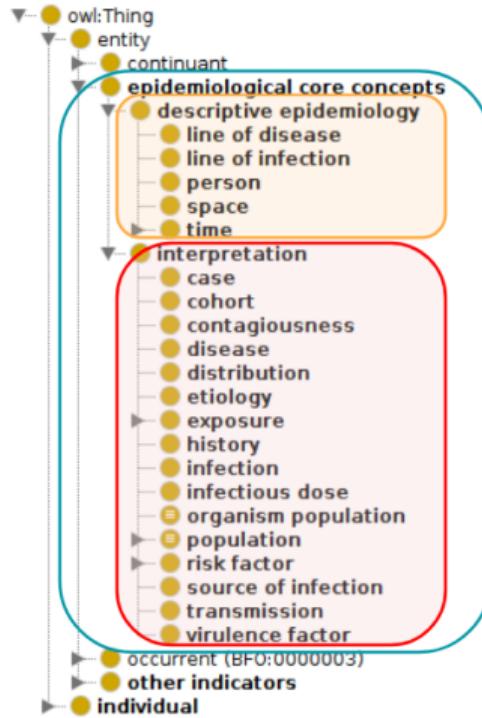
Results

CEMO ontological model and mapped to IDO



Results

Described in the **scope of domain** core concepts (axiom pattern)



Results

- EJP RD core model based on SIO
- CEMO measurement model and mapped to IDO
- Described in the scope of domain core concepts
- **Patient-population link FAIR recommendation**
based on **GA4GH Phenopackets::cohort block**



Discussion

Discussion

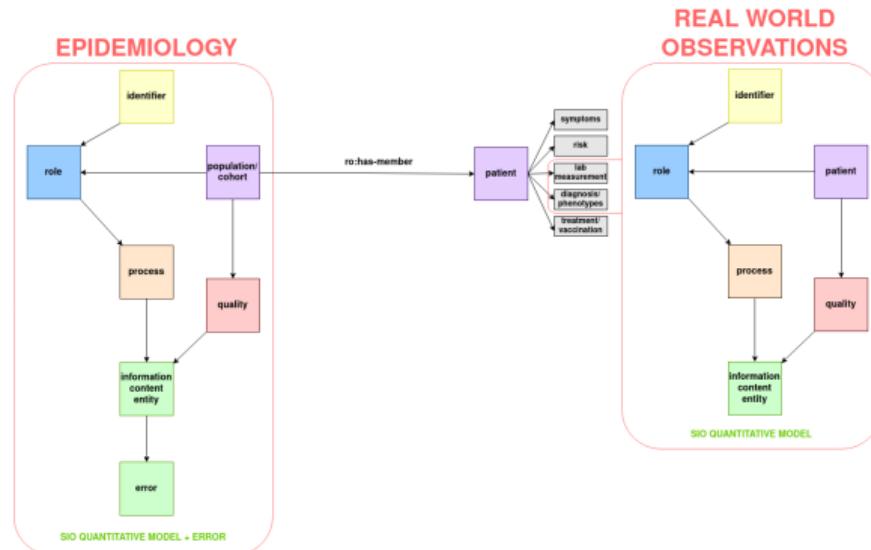
- Fill the gap
- New ontology
- To enable precision intervention
- OBO for biomedical research
- Reuse of measurement design pattern

Limitation

- Evaluation (BFO-GFO)

Future

- More expressive formal population-patient link



Conclusions

We provide a new ontology, CEMO, to enable efficient precision epidemiology

The screenshot shows the OBOLibrary interface for the CEMO ontology. The top navigation bar includes tabs for Active ontology, Entities, Class matrix, Property matrix, and DL Query. Below the navigation is an 'Ontology header' section containing the Ontology IRI (<http://purl.obolibrary.org/obo/cemo.owl>) and Ontology Version IRI (<http://purl.obolibrary.org/releases/16-05-2021/cemo.owl>). The main content area is divided into sections: Annotations, Metrics, Class axioms, Object property axioms, and Direct imports.

Annotations:

- dcterms:license: <https://creativecommons.org/licenses/by/4.0/>
- dc:creator: <http://orcid.org/0000-0003-0169-8149>
- dc:contributor: Elena de la Calle
- dc:contributor: <https://orcid.org/0000-0001-8149-5890>
- dc:contributor: <https://orcid.org/0000-0001-8888-635X>
- dc:contributor: <https://orcid.org/0002-5111-7263>
- dc:contributor: <https://orcid.org/0000-0002-8691-772X>
- dc:contributor: <https://orcid.org/0000-0003-0351-6523>
- dc:contributor: <https://orcid.org/0003-1795-5930>

Metrics:

Metric	Count
Axiom	1345
Logical axiom count	331
Declaration axioms count	275
Class count	227
Object property count	21
Data property count	1
Individual count	0
Annotation Property count	30

Class axioms:

Axiom Type	Count
SubClassOf	260
EquivalentClasses	7
DisjointClasses	29
GCI count	0
Hidden GCI Count	9

Object property axioms:

Axiom Type	Count
SubObjectPropertyOf	4
EquivalentObjectProperties	0
InverseObjectProperties	2
DisjointObjectProperties	0
FunctionalObjectProperty	6
InverseFunctionalObjectProperty	4
TransitiveObjectProperty	0

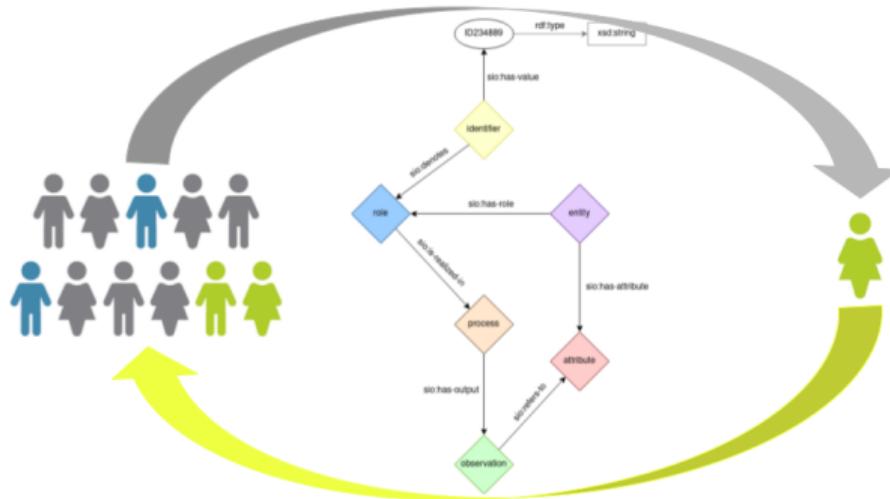
Direct Imports:

- bfo: <http://purl.obolibrary.org/obo/bfo/2019-08-26/bfo.owl>

Ontology IRI: <<http://purl.obolibrary.org/obo/cemo.owl>>
Version IRI: <<http://purl.obolibrary.org/obo/bfo/2019-08-26/bfo.owl>>
Location: <http://purl.obolibrary.org/obo/bfo/2019-08-26/bfo.owl>

source: <https://github.com/NuriaQueralt/covid19-epidemiology-ontology>

Precision intervention



Acknowledgements

- Co-authors

- Paul N. Schofield
- Robert Hoehndorf
- Claus Weiland
- Erik Schultes
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Time for Q/A

Thank you!

