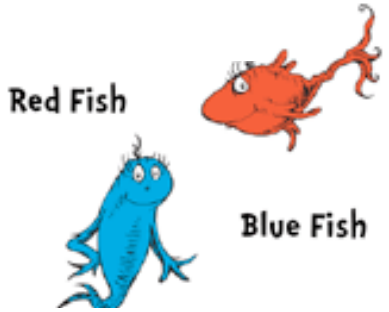
Software and database resources for curation and management of evolutionary phenotypes



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US National Evolutionary Synthesis Center

Durham NC

PHENOSCAPE

National Evolutionary Synthesis Center

NESCent

Questions for a comparative anatomical knowledgebase

- What do we know about phenotypic variation for a given anatomical feature?
- What do we know about the phenotype of a given taxon?
- What sister taxa vary from each other in same way that a given genetic mutant varies from wild type?

Toward *computable* phenotypes

- Hard-code detailed semantics into a database application
- Capture expert knowledge in ontologies and let the computer do generic logical reasoning

THE NATIONAL CENTER FOR BIOMEDICAL ONTOLOGY

BioPortal

Browse | Search

Teleost taxonomy

Search Results

Tree View

Tree view constructed based on *is_a* hierarchy

- Chordata
  - Craniata
    - Myxinomorphi
    - Vertebrata
      - Gnathostomata
        - Chondrichthyes
        - Osteichthyes
          - Actinopterygii
            - Chondrostei
            - Cladistia
            - Neopterygii
              - Halecostomi
                - Amiiformes
                - Teleostei
                  - Elopomorpha
                  - Euteleostei
                  - Ostarioclupeomorpha
                  - Osteoglossomorpha

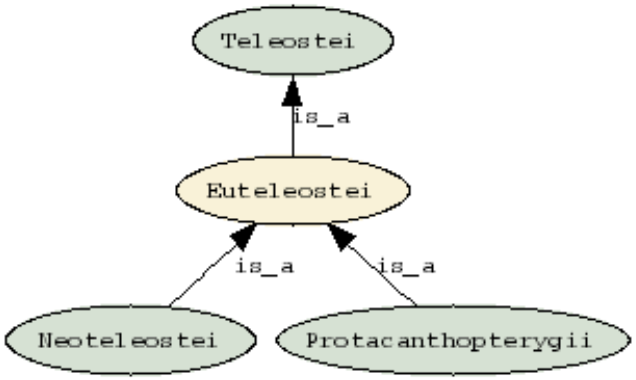
Class/Type Details

General

Class/Type Name	**Euteleostei**
Id	**TTO:254**

Graph View

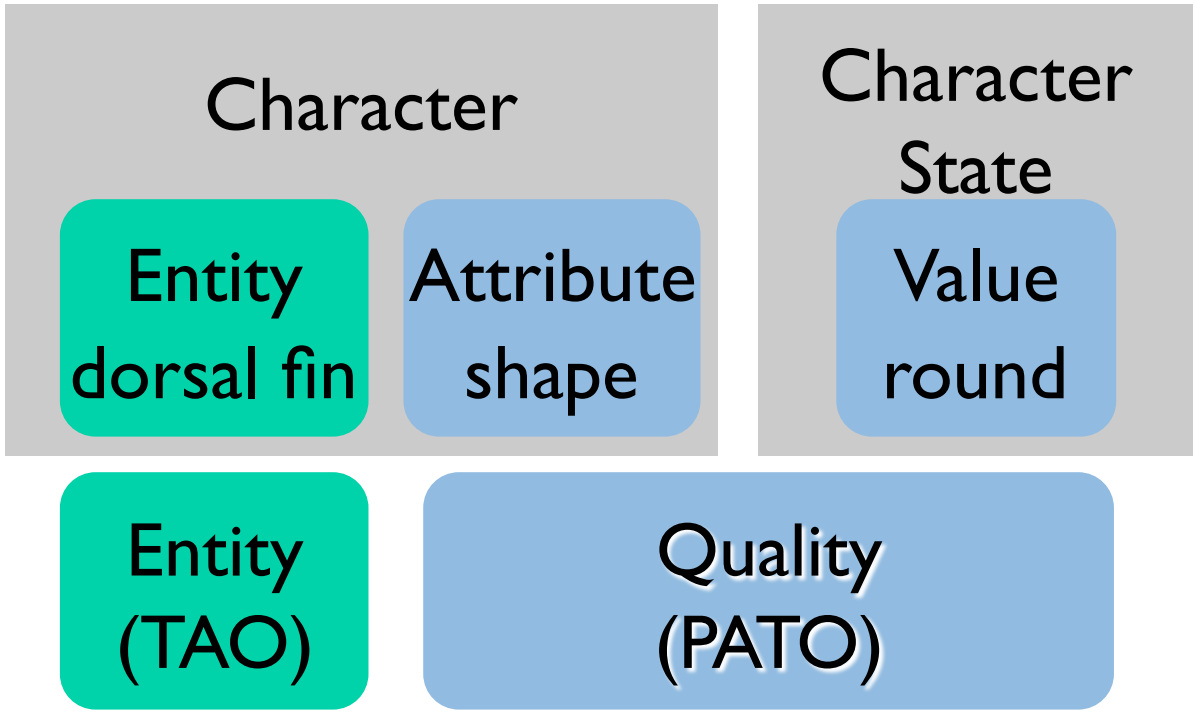
Graph Type: Local Neighborhood



Informatics requirements for application to clade ***X***

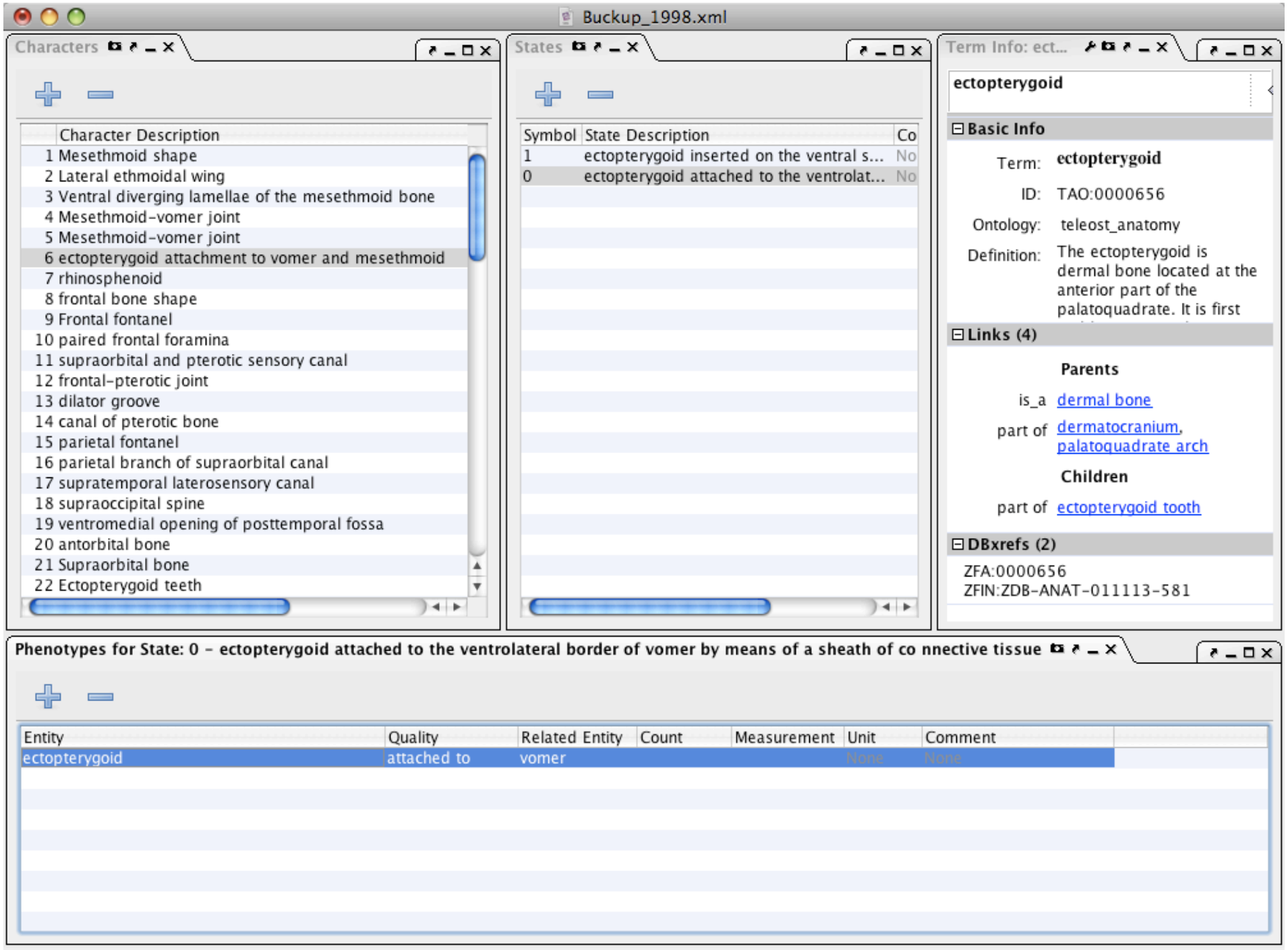
- Ontologies
  - Entities: **X Anatomy Ontology**
  - Qualities: **Phenotype and Trait Ontology (PATO)**
  - Taxonomy: **X taxonomy ontology**
- Curation tool: **Phenex**
  - Mutant phenotypes from zebrafish (14K in ZFIN)
  - Cypriniform "evolutionary phenotypes" from the literature
- Ontology-driven database: **OBD**
  - With a user interface tailored for evolutionary queries

Character Matrix and Entity-Quality syntax



Phenex data curation software

- Separates ontology curation from data curation
- User interface
  - Displays original character matrix-style assignments
  - Guides user to select appropriate ontology terms for EQ annotations
  - Incorporates references to literature and specimens
- Technical features
  - Loads current ontologies at startup
  - Generates NeXML files that can be easily loaded into the database
  - Platform independent
  - Open-source code base


Buckup_1998.xml
Characters
Character Description
1 Mesethmoid shape
2 Lateral ethmoidal wing
3 Ventral diverging lamellae of the mesethmoid bone
4 Mesethmoid-vomer joint
5 Mesethmoid-vomer joint
6 ectopterygoid attachment to vomer and mesethmoid
7 rhinosphenoid
8 frontal bone shape
9 Frontal fontanel
10 paired frontal foramina
11 supraorbital and pterotic sensory canal
12 frontal-pterotic joint
13 dilator groove
14 canal of pterotic bone
15 parietal fontanel
16 parietal branch of supraorbital canal
17 supratemporal laterosensory canal
18 supraoccipital spine
19 ventromedial opening of posttemporal fossa
20 antorbital bone
21 Supraorbital bone
22 Ectopterygoid teeth
States
Symbol
State Description
Co
1
ectopterygoid inserted on the ventral s...
No
0
ectopterygoid attached to the ventrolat...
No
Term Info: ect...
ectopterygoid
Basic Info
Term:
ectopterygoid
ID:
TAO:0000656
Ontology:
teleost_anatomy
Definition:
The ectopterygoid is dermal bone located at the anterior part of the palatoquadrate. It is first
Links (4)
Parents
is_a
dermal bone
part of
dermatocranium,
palatoquadrate arch
Children
part of
ectopterygoid tooth
DBxrefs (2)
ZFA:0000656
ZFIN:ZDB-ANAT-011113-581
Phenotypes for State: 0 - ectopterygoid attached to the ventrolateral border of vomer by means of a sheath of co nnective tissue
Entity
Quality
Related Entity
Count
Measurement
Unit
Comment
ectopterygoid
attached to
vomer
None
None

```
<state id="da8b85c2-e88e-4777-bfcb-e5daab15d829"
  label="ectopterygoid attached to the ventrolateral border of
  vomer by means of a sheath of connective tissue" symbol="0">
    <dict>
      <key>OBO_phenotype</key>
      <any>
        <phen:phenotype>
          <phen:phenotype_character>
            <phen:description/>
            <phen:bearer>
              <phen:typeref about="TAO:0000656"/>
            </phen:bearer>
            <phen:quality>
              <phen:typeref about="PATO:0001667"/>
              <phen:related_entity>
                <phen:typeref about="TAO:0000308"/>
              </phen:related_entity>
            </phen:quality>
          </phen:phenotype_character>
        </phen:phenotype>
      </any>
    </dict>
  </state>
```

The Phenoscape database

- Based on the Open Biomedical Database (OBD) model
  - Developed by Chris Mungall
- Generic (ie organism-agnostic)
- Represents both the ontologies and data annotations
- Exploits logical structure of the ontologies
  - Querying annotations of “dermal bone” will return annotations of “ectopterygoid”

Nodes

node_id	ontology term	label
548	PATO:0000462	"absent from organism"
10373	TTO:1001979	"Danio rerio"
16249	TTO:101040	"Danio"
53	OBO_REL:part_of	"part_of"
77	OBO_REL:is_a	"is_a"
48988	PHENOSCAPE:exhibits	"exhibits"

Links

node_id	predicate	node_id
10373	77	16249

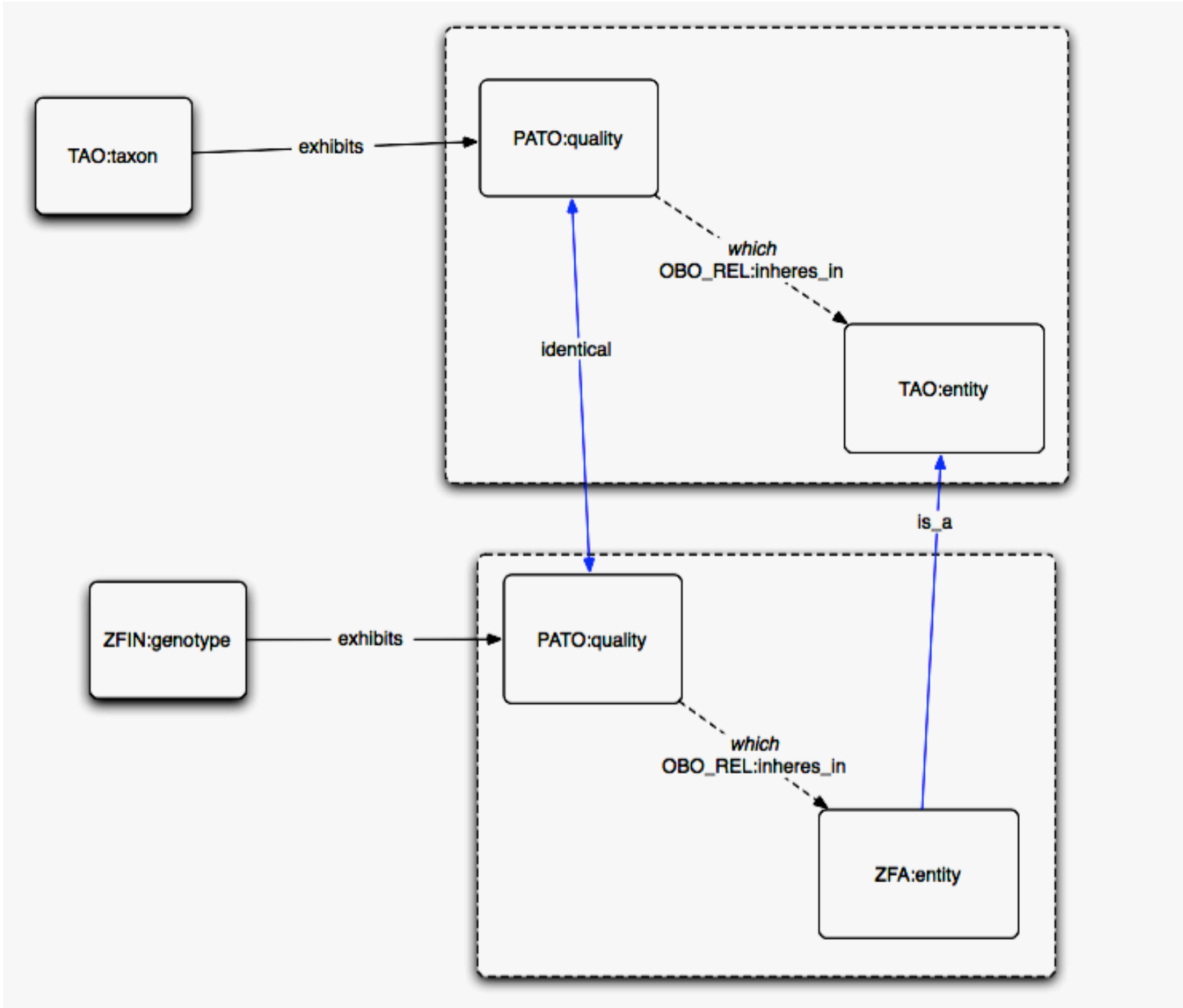
--> "Danio rerio is_a Danio"

10373	48988	109224

--> "Danio rerio exhibits absence in a process which is part of infraorbital 1"

User interface

- Search for anatomical entity
  - What do we know about phenotypic variation for a given anatomical feature?
- Search for taxon
  - What do we know about the phenotype of a given taxon?
- Search for gene
  - What sister taxa vary from each other in same way that a given genetic mutant varies from wild type?


TAO:taxon
exhibits
PATO:quality
which
OBO_REL:inheres_in
TAO:entity
identical
is_a
ZFIN:genotype
exhibits
PATO:quality
which
OBO_REL:inheres_in
ZFA:entity

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Acknowledgements

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