Challenges in Developing Multi-species Anatomy Ontologies

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and

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PHENOSCAPE

Single species ontologies

- Represent the generalized structures of a species
  - Zebrafish Anatomical Ontology
  - Foundational Model of Anatomy

Multi-species ontologies

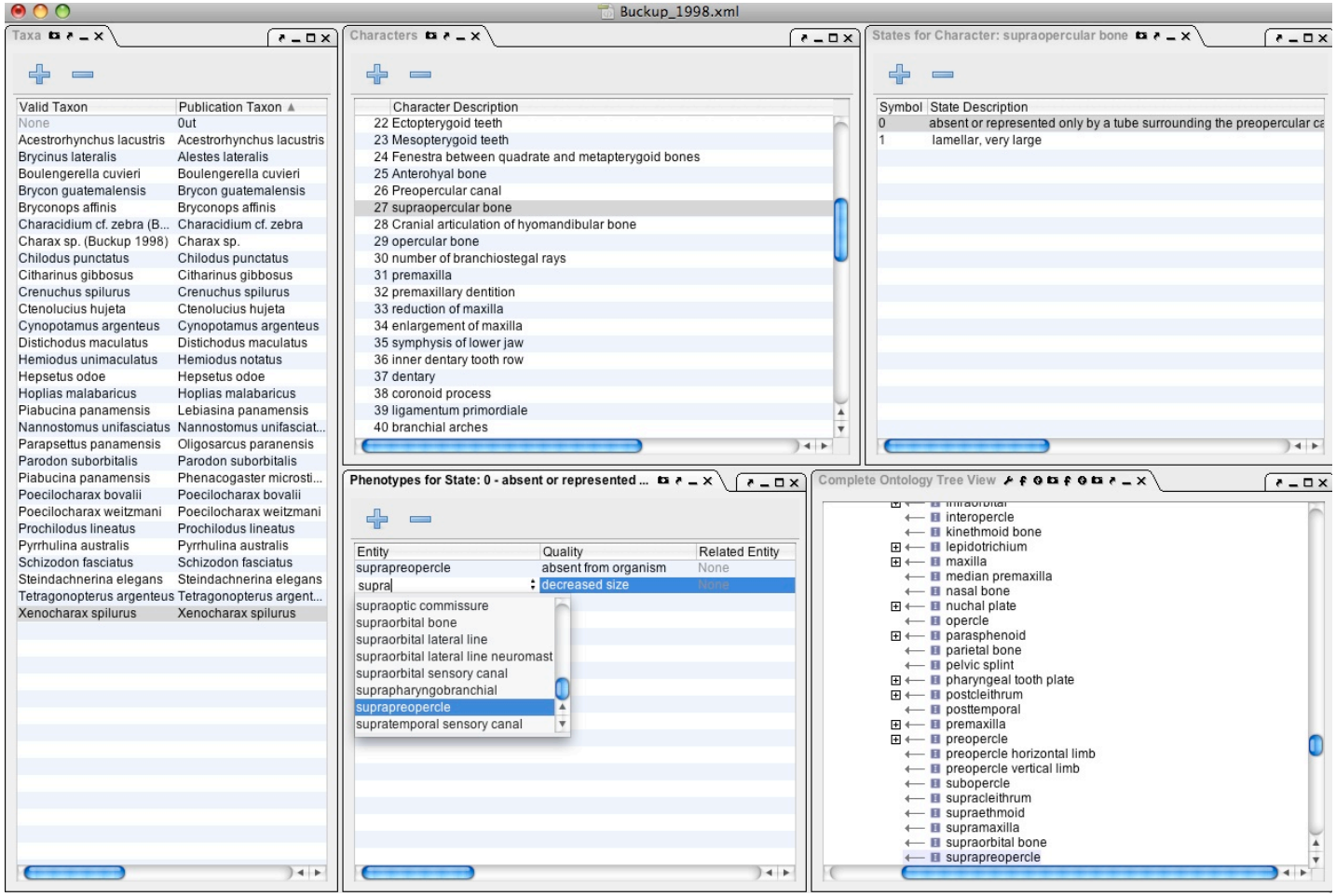
- Represent the variety of structures observed in all species in a clade
  - Teleost Anatomy Ontology (TAO)
  - Plant Structure Ontology
  - Amphibian Anatomy Ontology

Why use ontologies?

- Standardization of vocabulary among communities
- Free text not computable
- Integration across databases
- Comparison of structures across monophyletic groups of organisms
- Query across large amount of data

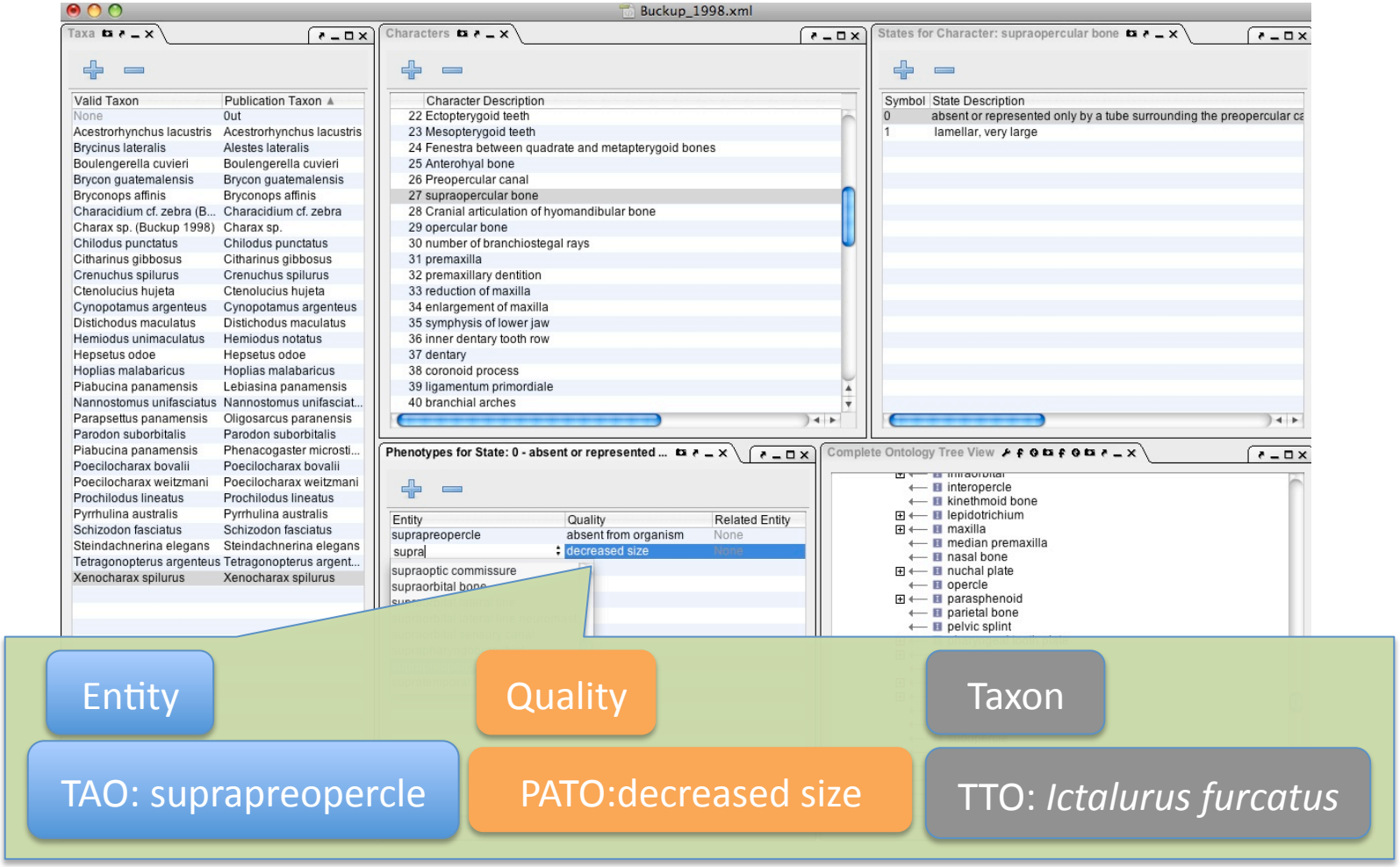
Uses of multi-species anatomy ontologies

Phenoscape: Annotation of evolutionary phenotypes



Uses of multi-species anatomy ontologies

Phenoscape: Annotation of evolutionary phenotypes



Uses of multi-species anatomy ontologies

Image annotation

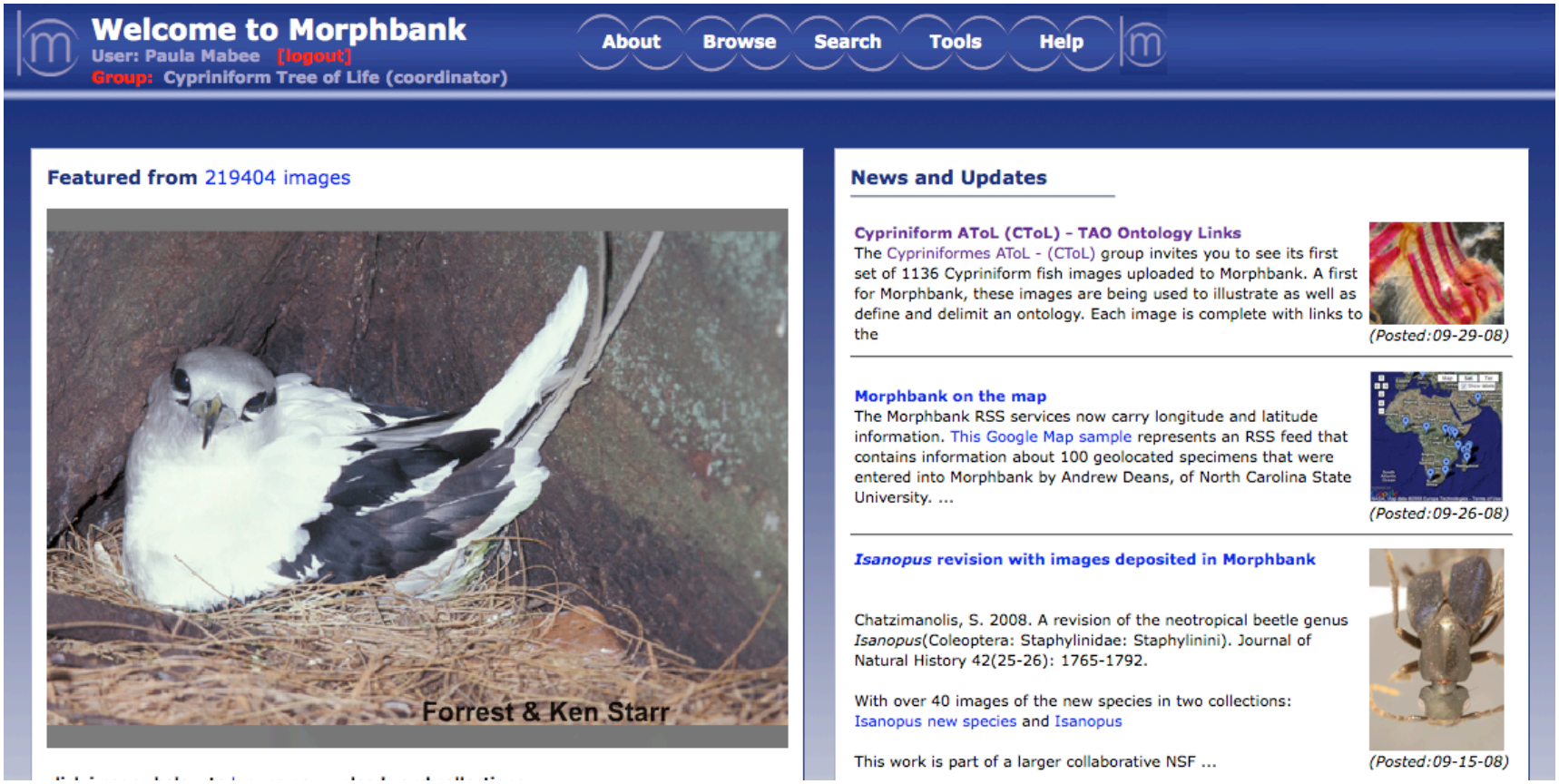


Image Record: [460945] Chromobotia macracanthus

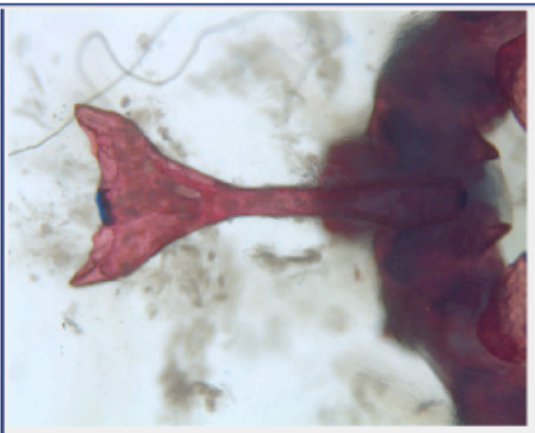
Contributor: Cypriniform Tree of Life
Submitter: Paula Mabee
Date Submitted: 2008-09-08
Last Modified: 2008-09-08
Publish Date: 2008-09-07

Description: basihyal From spreadsheet line 417
CToLDateSubmitted: 14-Jun-11
CToLSubmittedBy: Ericka Grey

Magnification: NULL
Dimension (px): 1280x1024
Resolution (PPI): 100
Submitted as: jpg
Original File Name: Botia_macracanthus_1165_25x_EG_Basihyal_501.jpg

View id: 459122
Specimen part: basihyal
Angle: Dorsal
Technique: Digital Camera
Preparation: Cleared and counterstained for bone (Alizarin red) and cartilage (Alcian blue)

Download: tiff (1.21 MB)
jpeg (131.37 KB)
Copyright: Ericka Grey and Paula Mabee
License:



FSI Viewer

Specimen

Specimen id: 460920
Basis of record: [S] - Specimen
Sex: unknown
Form: unknown
Stage: Juvenile/Adult
Catalog number: 199848
Collector:
Date collected:

Locality

Locality Id:
Continent ocean:
Country:
Locality:
Latitude:
Longitude:
Elevation (m):

Determination

Class: Actinopterygii
Order: Cypriniformes
Family: Cobitidae
Genus: Chromobotia
Species: Chromobotia macracanthus

Determination annotations

(Add Annotation...)

External links/identifiers

External identification: CToL-S:0001165
External identification: CToL-I:0000501

Ontology:

Teleost Anatomy Ontology TAO:0000316

Other Annotations

(Add Annotation...)

THE NATIONAL CENTER FOR BIOMEDICAL ONTOLOGY

BioPortal 2.0

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Home | Browse | Search | Projects | All Mappings | Teleost anatomy and development

- pharyngeal arch 1
- pharyngeal vasculature
- splanchnocranium
- pharyngeal pouch
- pharyngeal musculature
- pharyngeal arch 2
  - hyoid muscle
  - pharyngeal arch 2 skele
    - dorsal hyoid arch
    - ventral hyoid arch
      - basihyal tooth pla
      - ceratohyal bone
      - epihyal
      - interhyal cartilage
      - basihyal cartilage
      - hypohyal
      - ceratohyal cartila
      - interhyal bone
      - urohyal
      - branchiostegal ray
      - basihyal

basihyal (Link To Concept)

Details | Visualization | Marginal Notes | Mappings | Resources

Class Name: basihyal

ID: TAO:0000316

Definition: The basihyal is the 'tongue' of the a fish, the median and anteriorly projecting element of the ventral hyoid arch.Ossification of the cartilaginous basihyal is first visible at the posterior end of the club-shaped basihyal cartilage (6.0 mm). Ossification spreads throughout the cartilage, except for the anterior tip, which remains cartilaginous. The posteroventral tip extends between the left and right dorsal hypohyals.

Database References: ZFIN

Is A: endochondral bone

Develops From: basihyal cartilage

Part Of: ventral hyoid arch

Related Synonym: "basihyoid" , "glossohyal"

Why multi-species?

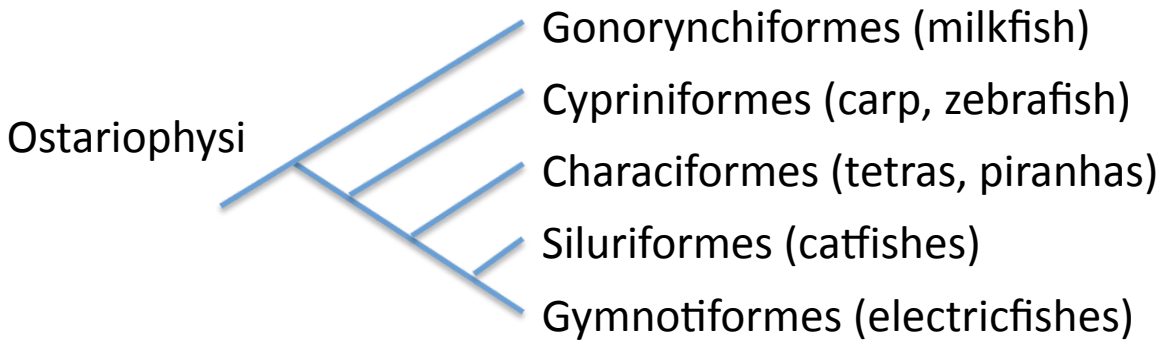
- Practicality: too many species to make a separate ontology for each

- Domain knowledge: represention of the knowledge of comparative biologists for groups of species rather than single species

Developing the Teleost Anatomy Ontology (TAO)

- Multi-species anatomy ontology for teleost fishes
- Represent all of the variation observed in all species in a clade
- Requirements and challenges

Scope of Teleost Anatomy Ontology



(Fink and Fink, 1981)

Teleost anatomy ontology (TAO) cloned on Zebrafish Anatomical Ontology

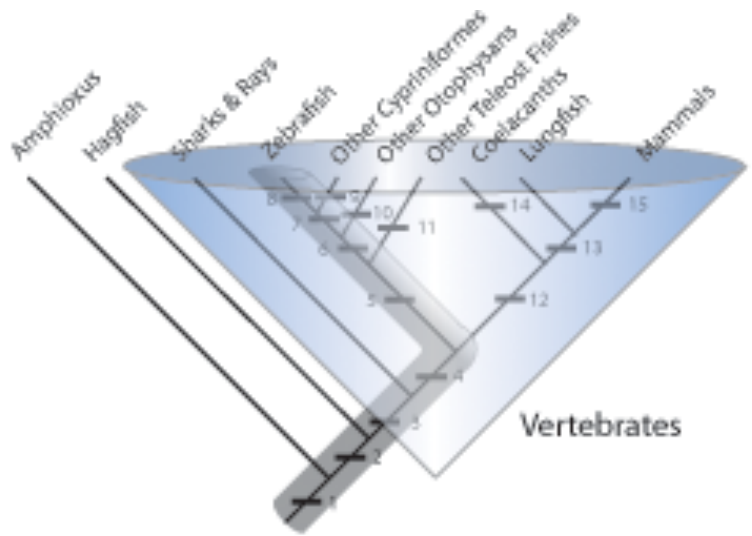


Fig. 1. Expansion of zebrafish core anatomical ontology (grey inner cylinder) to include all vertebrates (blue cone). The zebrafish anatomical ontology (~1,500 terms) includes features that evolved at various times along the common vertebrate lineage (characters 1—8). In order to make evolutionary comparisons across vertebrates, this ontology must be extended to cover the characteristics of the other teleost fishes (characters 9—11) and other vertebrates (characters 12—15).

Zebrafish terms are subtypes of Teleost terms

Zebrafish Anatomical Ontology

- zebrafish anatomical entity
  - anatomical space
  - anatomical structure
    - acellular anatomical structure
    - anatomical group
    - cell
    - compound organ
    - embryonic structure
    - extraembryonic structure
    - multi-tissue structure
    - organism subdivision
    - portion of tissue
      - actinotrichium
      - chordo neural hinge
      - cranial nerve root
      - dental papilla
      - endocardial ring
      - endocardium
      - epithelium
      - external cellular layer
      - ganglion

is_a

is_a

Teleost Anatomy Ontology

- teleost anatomical entity
  - anatomical space
  - anatomical structure
    - acellular anatomical structure
    - anatomical group
    - cell
    - compound organ
    - embryonic structure
    - extraembryonic structure
    - multi-tissue structure
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ZFIN

Upper-level nodes of TAO aligned with Common Anatomy Reference Ontology

- Classes
  - anatomical entity
    - immaterial anatomical entity
      - anatomical line
      - anatomical point
      - anatomical space
      - anatomical surface
    - material anatomical entity
      - anatomical structure
        - acellular anatomical structure
        - anatomical group
        - cell
        - cell component
        - compound organ
        - extraembryonic structure
        - multi-cellular organism
        - multi-tissue structure
        - organism subdivision
        - portion of tissue
      - portion of organism substance
- Relations
- Obsolete

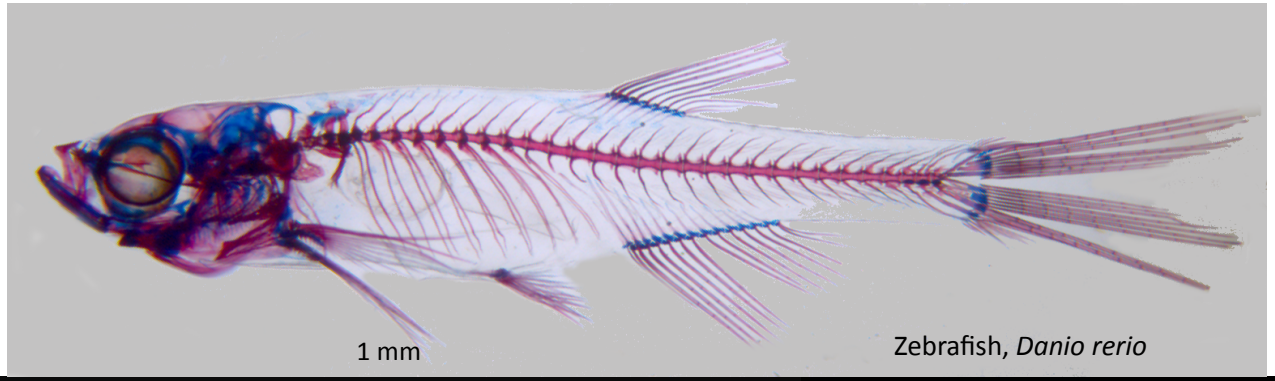
CARO

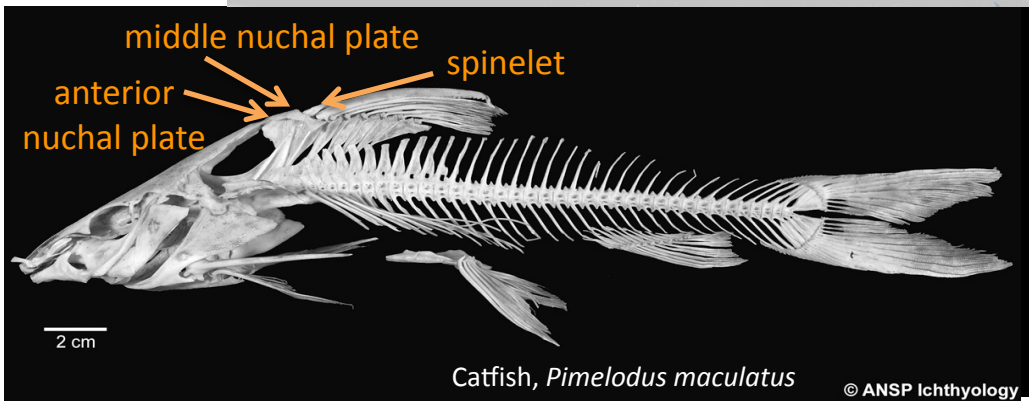
- Template for top-level nodes of anatomical ontologies

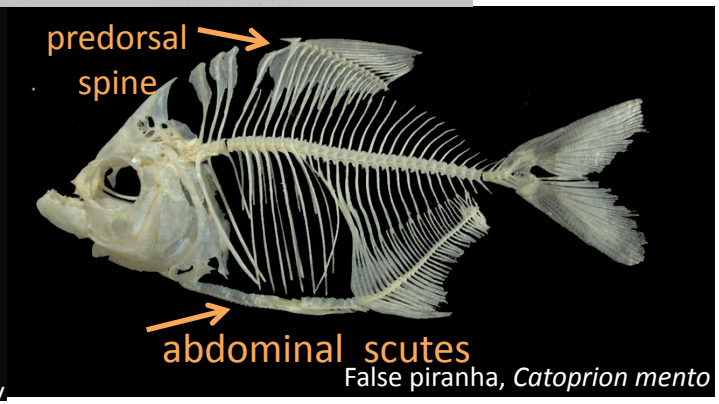
- Promotes interoperability among anatomical ontologies

Growth of Teleost anatomy ontology

- Since cloning from Zebrafish anatomical ontology in Sept 2007,
  395 (207 skeletal) new terms added
  Total of 2,371 terms as of Dec 2008







Teleost anatomy terms

```
term name: basihyal bone
id: TAO:0000316
def: ”Endochondral bone that is the median and anteriorly
   projecting element of the ventral hyoid arch."
synonym: "basihyoid"
synonym: "glossohyal”
is_a: TAO:0001591 ! endochondral bone
relationship: develops_from TAO:0001510 ! basihyal cartilage
relationship: part_of TAO:0001402 ! ventral hyoid arch
```

unique identifier

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definition

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Logical relationships to other terms

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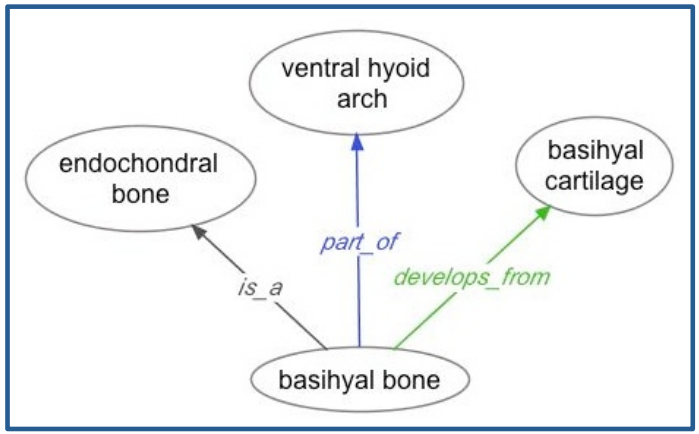
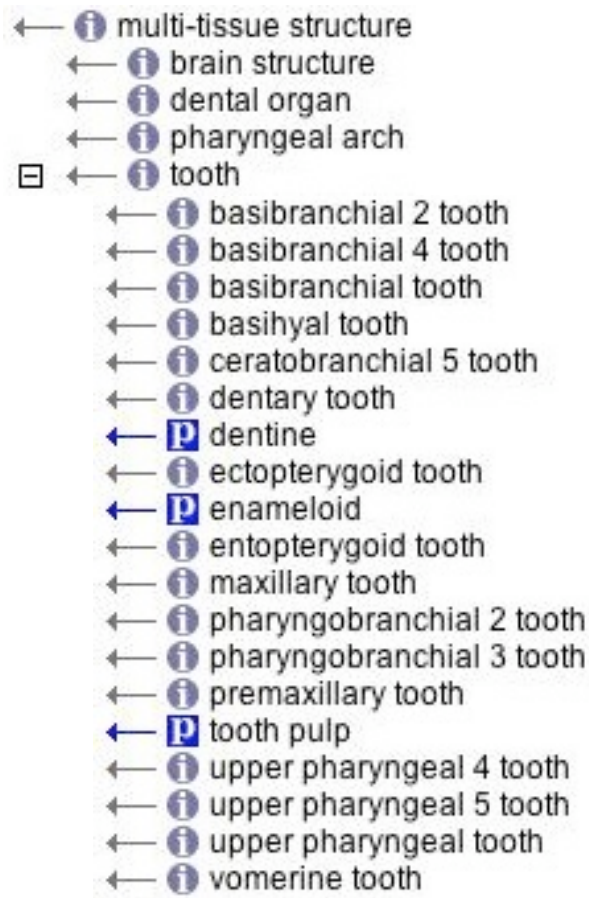


TAO development workflow



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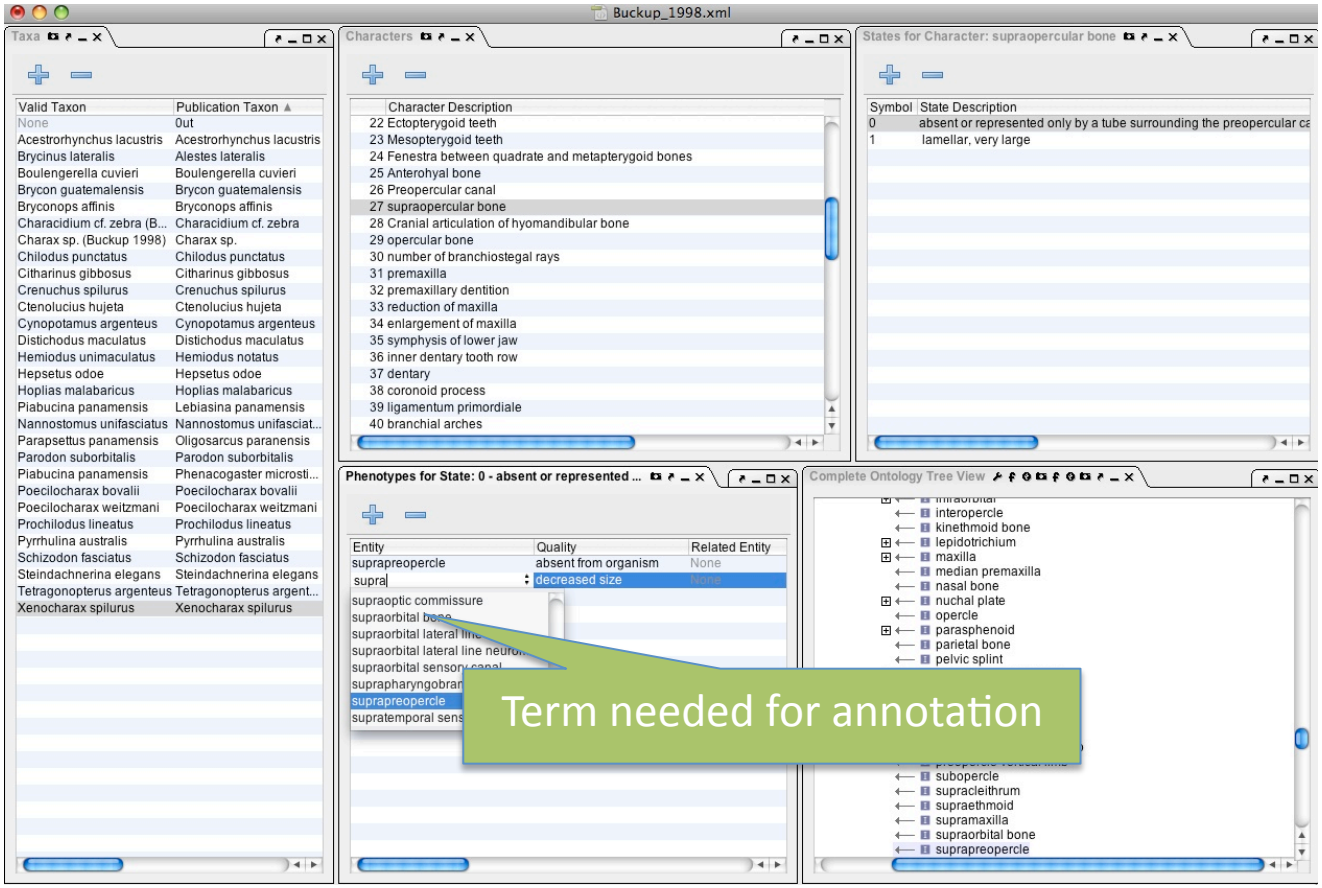

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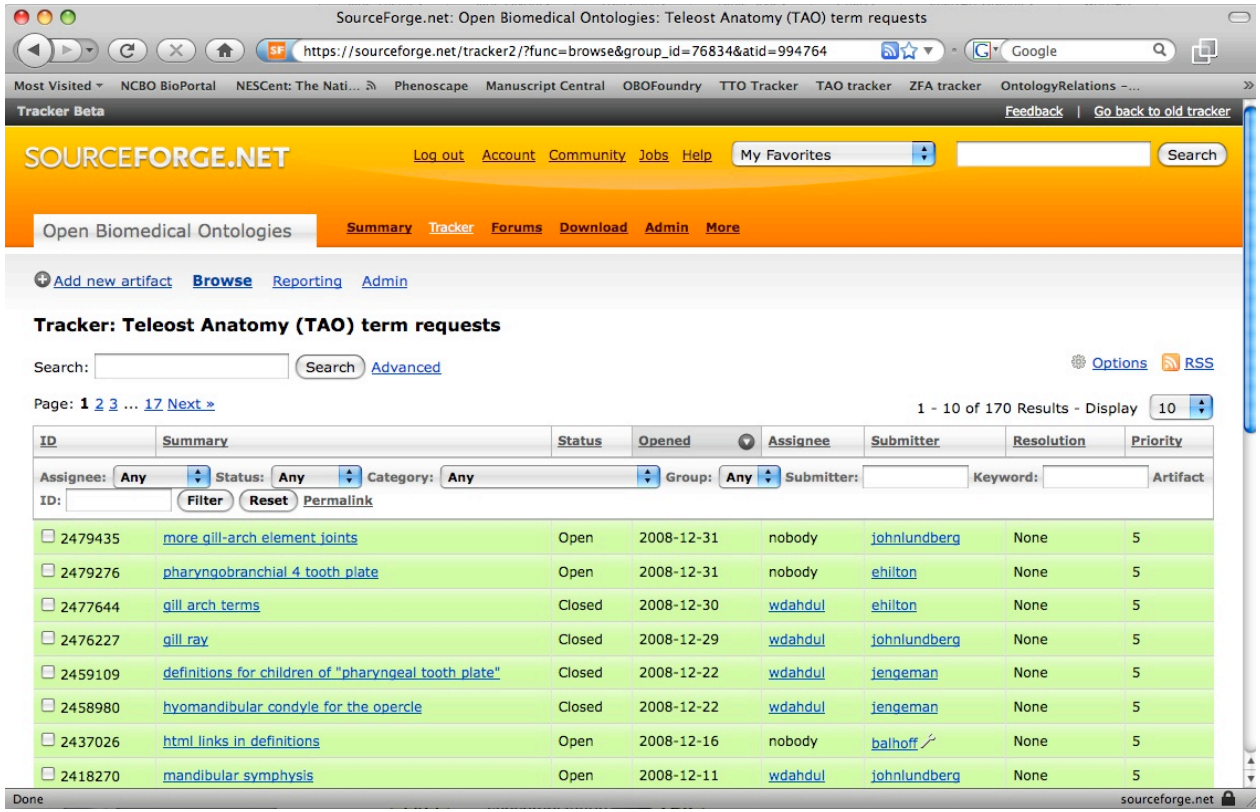

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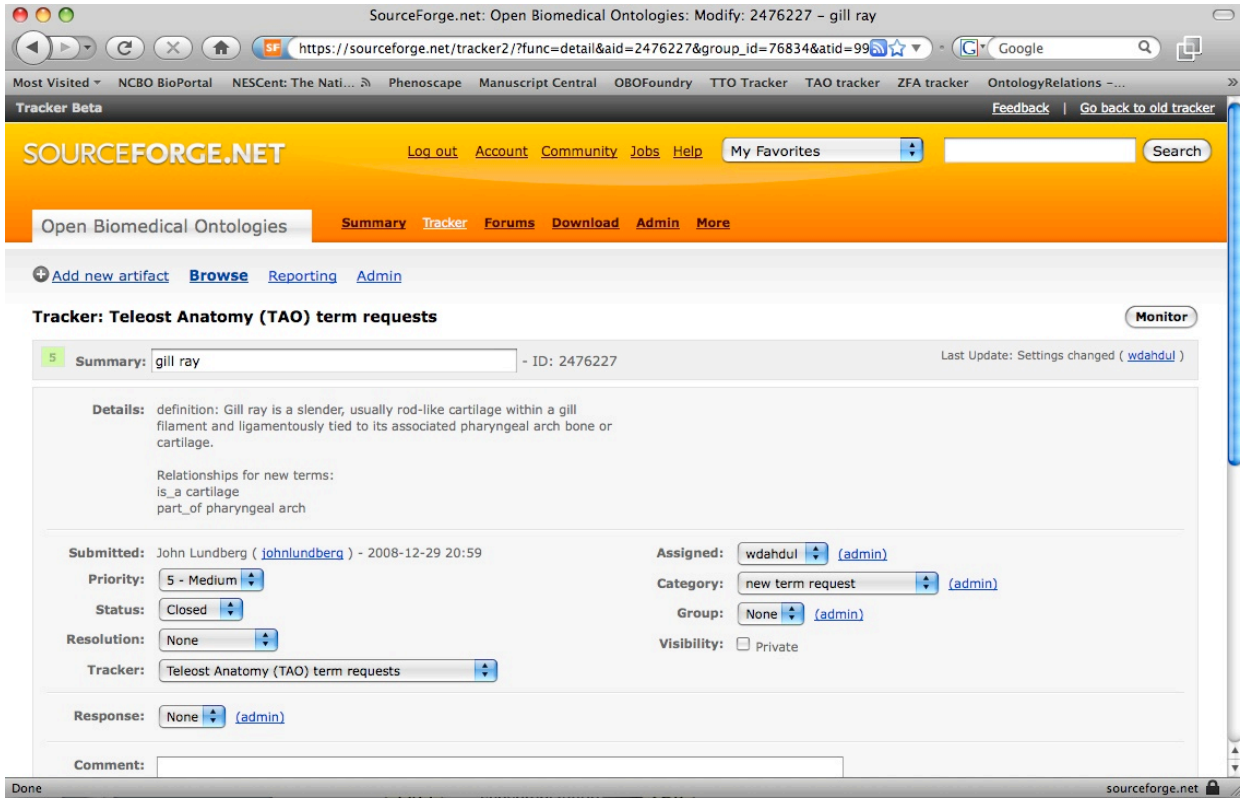

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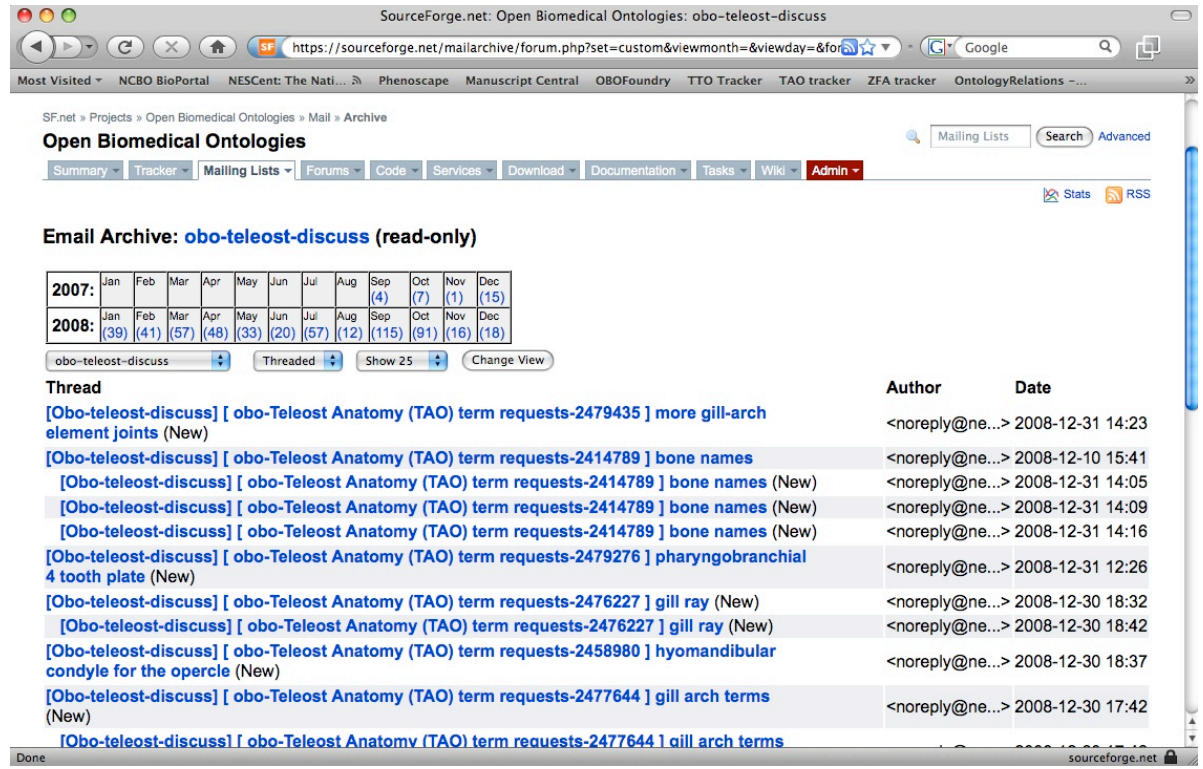

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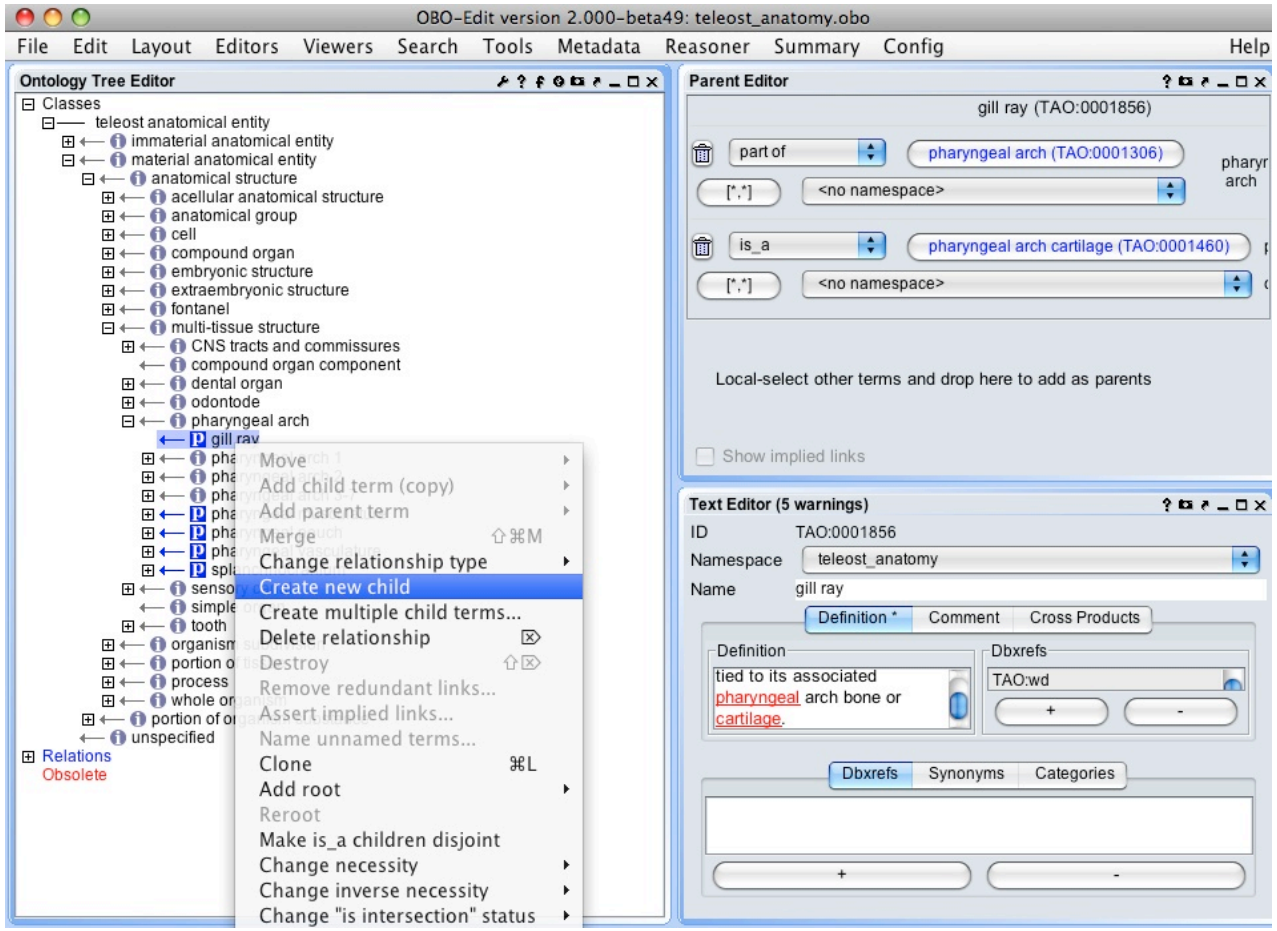

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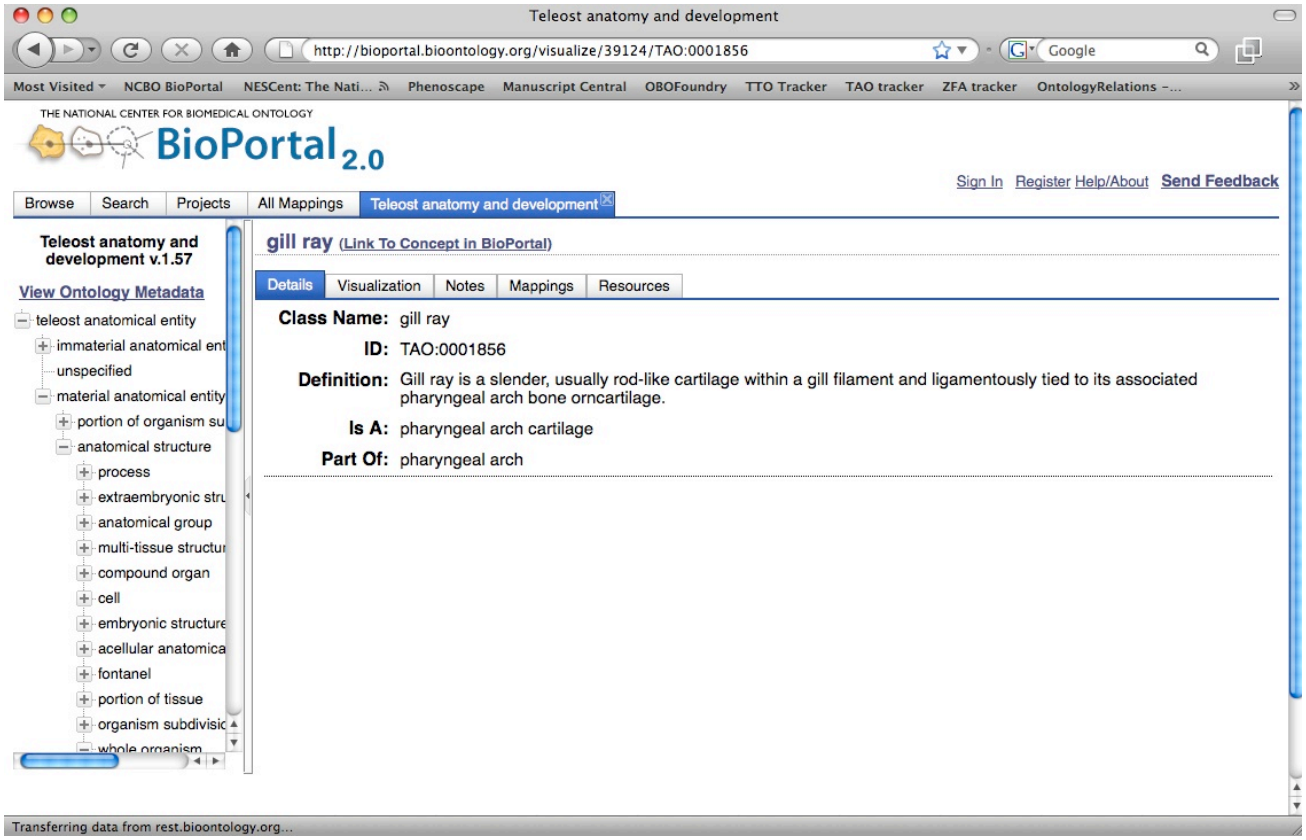

Requirements and challenges in TAO development

- Requirements:
  - Refinement of definitions
  - Additional classes
  - Classes become qualities
- Challenges
  - Add new terms in ontology or created on the fly by combining existing terms?
  - Representation of taxonomically variable relationships
  - Homology

Refinement of definitions

Infraorbital 1

- *Old definition:*
  - Dermal bone that covers the region between the anterior margin of the eye and the upper jaw. Infraorbital 1 (the lachrymal) is the first and largest bone in the infraorbital series. Bears the anterior most part of the infraorbital canal.

- *New definition*:
  - Infraorbital that is the first (anteriormost) bone of the infraorbital series.

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Multi-species ontologies may need classes to reflect more detailed hierarchy of terms

Zebrafish Anatomical Ontology

- multi-tissue structure
  - brain structure
  - ceratobranchial 5 tooth
  - dental organ
  - pharyngeal arch

Teleost Anatomy Ontology

- multi-tissue structure
  - brain structure
  - dental organ
  - pharyngeal arch
  - tooth
    - basibranchial 2 tooth
    - basibranchial 4 tooth
    - basibranchial tooth
    - basihyal tooth
    - ceratobranchial 5 tooth
    - dentary tooth
    - dentine
    - ectopterygoid tooth
    - enameloid
    - entopterygoid tooth
    - maxillary tooth
    - pharyngobranchial 2 tooth
    - pharyngobranchial 3 tooth
    - premaxillary tooth
    - tooth pulp
    - upper pharyngeal 4 tooth
    - upper pharyngeal 5 tooth
    - upper pharyngeal tooth
    - vomerine tooth

Some classes from single-species ontologies move to quality ontology

Foundational Model of Anatomy

- Joint
  - Joint of principal body part
  - Nonsynovial joint
    - Cartilaginous joint
      - Symphysis
        - Intervertebral symphysis
        - Mandibular symphysis
        - Manubriosternal joint
        - Pubic symphysis
        - Xiphisternal joint
      - Synchondrosis
    - Fibrous joint
      - Dento-alveolar joint
      - Suture
      - Syndesmosis
    - Synostosis
  - Synovial joint

Teleost Anatomy Ontology

- joint
  - autopalatine-lateral ethmoid joint
  - autopalatine-vomer joint
  - ceratohyal-branchiostegal ray joint
  - ceratohyal-dorsal hypohyal joint
  - ceratohyal-ventral hypohyal joint
  - dentary-anguloarticular joint
  - dorsal hypohyal-urohyal joint
  - dorsal hypohyal-ventral hypohyal joint
  - epihyal-branchiostegal ray joint
  - epihyal-ceratohyal joint
  - frontal-pterotic joint
  - inter-ventral hypohyal joint
  - interhyal-epihyal joint
  - mandibular symphysis

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Solution 1: Joint terms in TAO have multiple parents

Some classes from single-species ontologies move to quality ontology

Foundational Model of Anatomy

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  - epihyal-ceratohyal joint
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  - inter-ventral hypohyal joint
  - interhyal-epihyal joint
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Solution 2: anatomy classes become quality terms in a different ontology:

Entity	Quality	Taxon
TAO: prootic-exoccipital joint	PATO:cartilaginous	TTO: *Ictalurus furcatus*

Managing ontology growth

- New terms can be added to the ontology
  - Possess unique identifier
  - *premaxilla ascending process* TAO:0000699
    - *is_a* TAO:process
    - *part_of* TAO:premaxilla
- Alternatively, terms can be “post-composed” at time of annotation by combining existing terms to create a new one:

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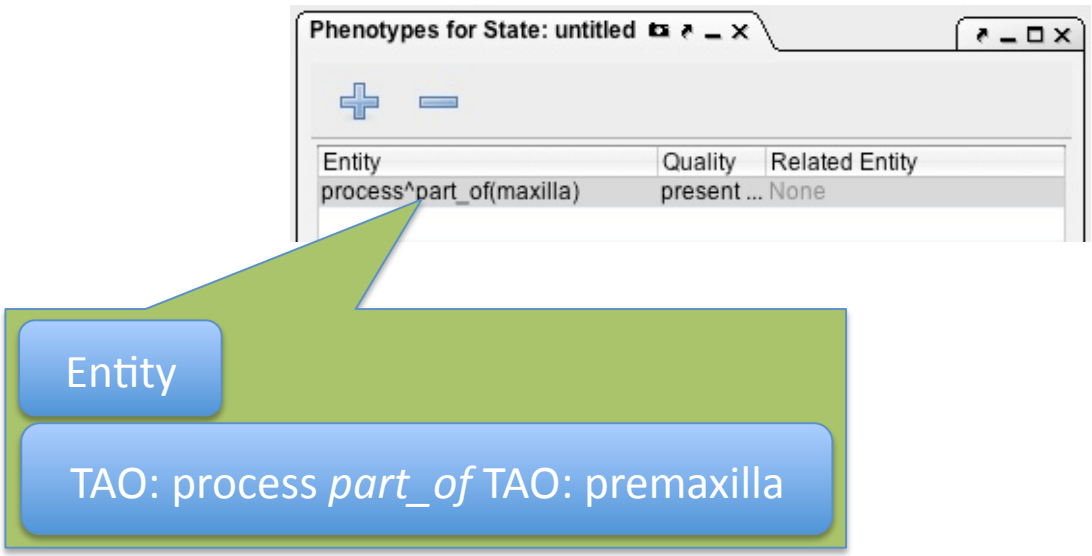

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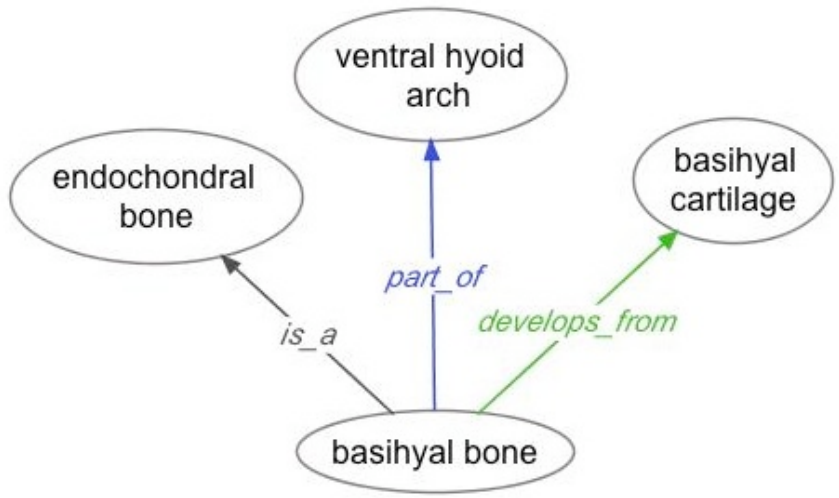
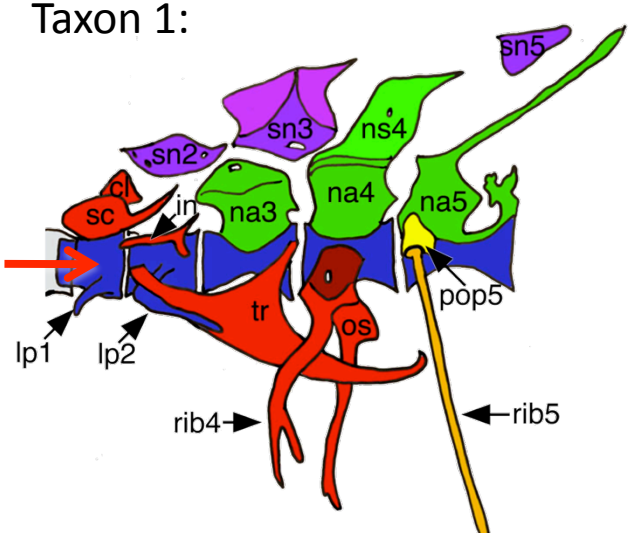
    - *part_of* TAO:premaxilla
- Alternatively, terms can be "post-composed" at time of annotation by combining existing terms to create a new one:
- Guideline: add new term to ontology if it will be used repeatedly

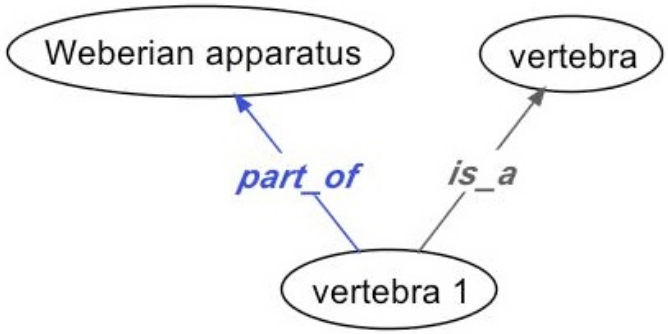
Different relationships for different taxa



Different relationships for different taxa

Taxon 1:

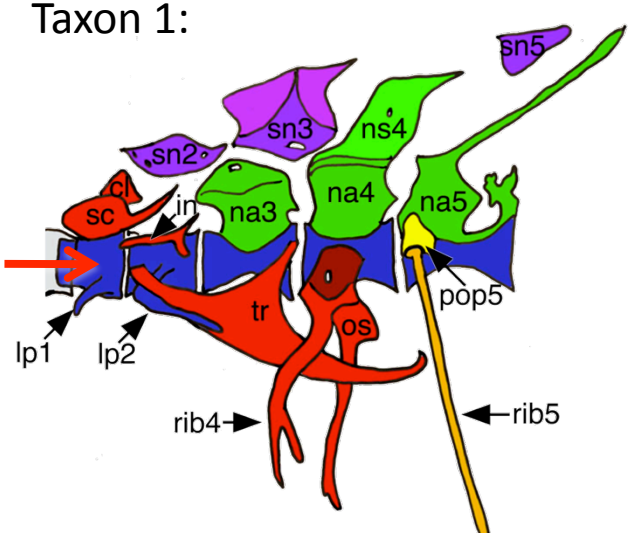


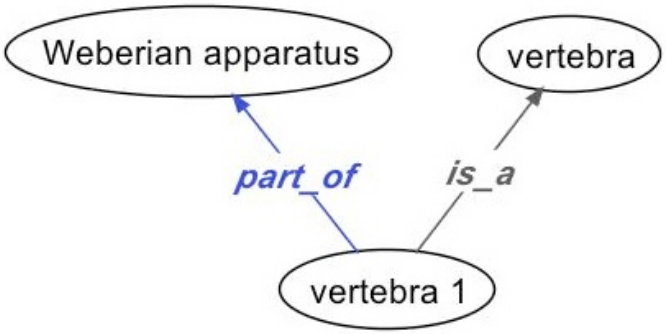


Weberian apparatus consists of the modified anteriormost four vertebrae and associated structures of otophysans which transduce sound or pressure waves from the tunica externa of the swimbladder to the inner ear (Alexander, 1962).

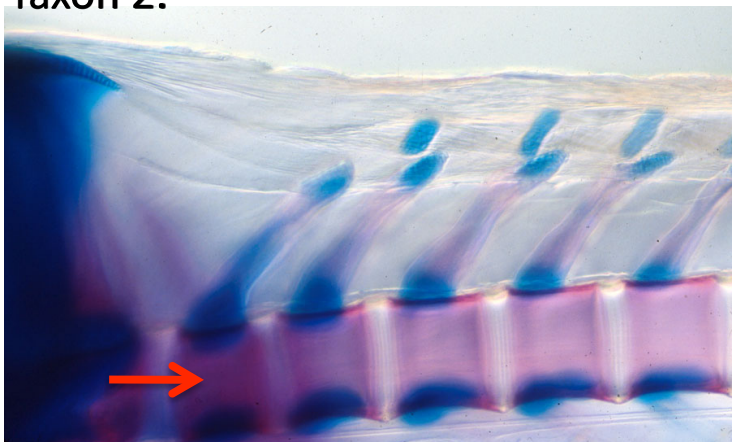
Different relationships for different taxa

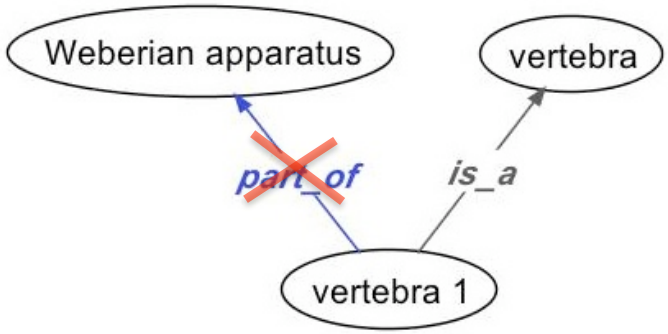
Taxon 1:





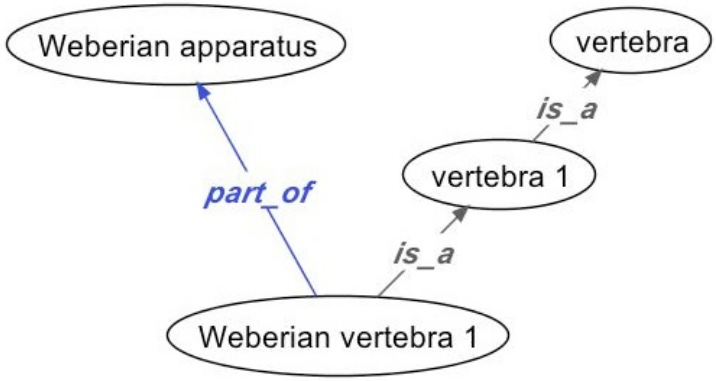
Taxon 2:





Different relationships for different taxa

Solution 1: create child of vertebra 1 that is part of the Weberian apparatus



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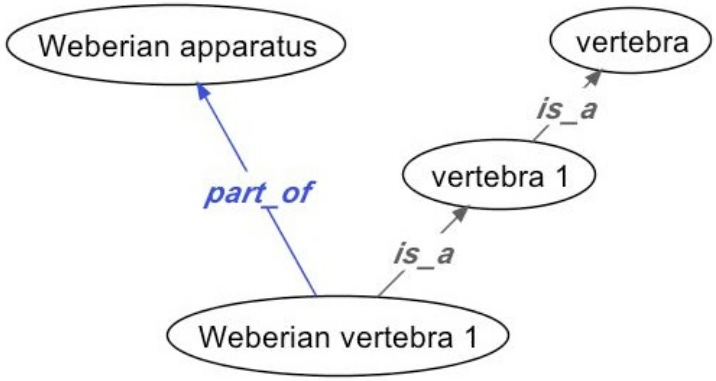
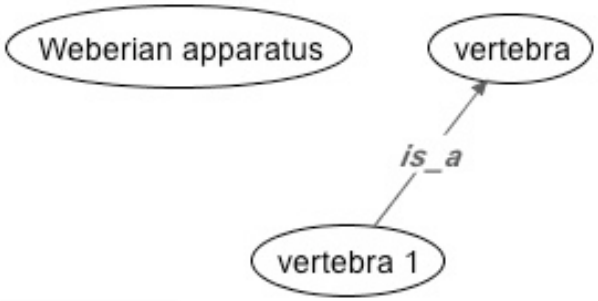


Solution 2: describe variation in annotation statement



Entity	Quality	Taxon
TAO: vertebra 1 *part_of* Weberian apparatus	PATO:present	TTO: *Ictalurus furcatus*

Homology

- TAO represents all possible anatomical structures in teleosts, but does not imply homology between terms

- Homology is a hypothesis and its designation requires evidence and attribution

Homology evidence codes

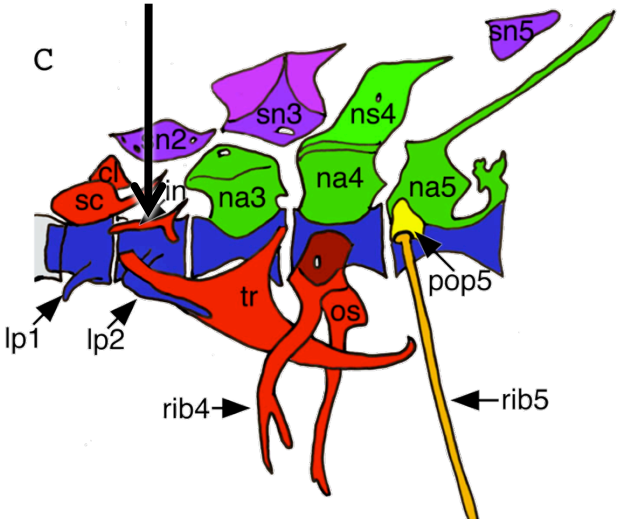
Available from Evidence Code Ontology

- Inferred from positional similarity (IPS), ECO:0000060
- Inferred from compositional similarity (ICS), ECO:0000063
- Inferred from developmental similarity (IDS), ECO: 0000067
- Inferred from morphological similarity (IMS), ECO:0000071
- Inferred from gene expression similarity (IGES), ECO:0000075
- Inferred from phylogeny (IP), ECO:0000080

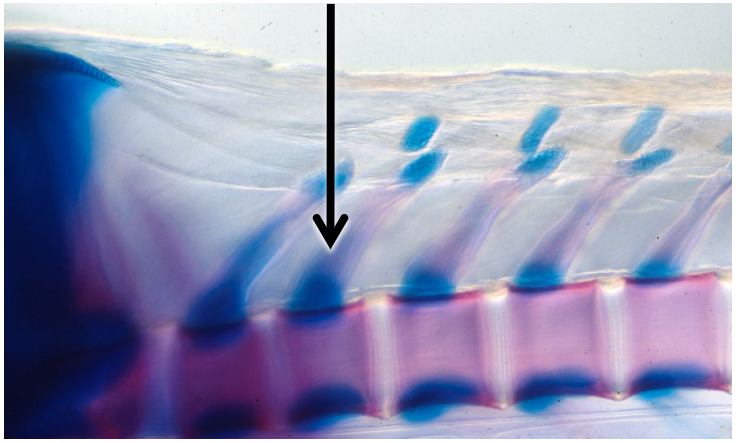
Publication	Entity 1	Taxon 1	Entity 2	Taxon 2	Evidence
Fink and Fink 1981	neural arch 2	Teleostei	intercalarium	Otophysi	inferred from positional similarity

Homology

intercalarium



neural arch 2

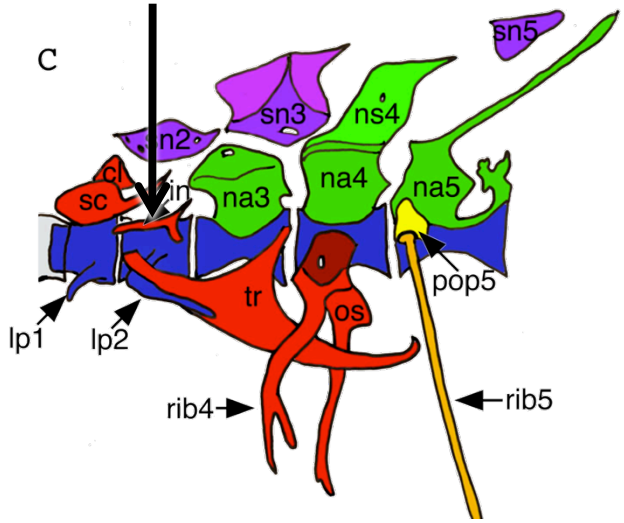


Representation in TAO:

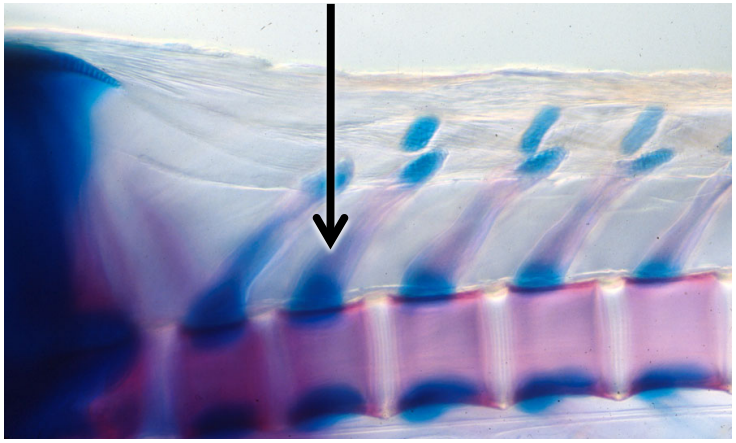
- ⊟ neural arch
  - ⊞ intercalarium
  - neural arch 2
  - ⊞ neural arch 3
  - neural arch 4

Homology

intercalarium



neural arch 2



Representation in TAO:

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Homology annotation:

Publication	Entity 1	Taxon 1	Entity 2	Taxon 2	Evidence
Fink and Fink 1981	neural arch 2	Teleostei	intercalarium	Otophysi	inferred from positional similarity

Multi-species Ontologies

- Required to represent evolutionary variability
- Logical challenges in representating variability across a clade in an ontology
- Homology can be treated explicitly with a formalized relation between terms

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