

Challenges in Developing Multi-species Anatomy Ontologies

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and

National Evolutionary Synthesis Center



Single species ontologies

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

Multi-species ontologies

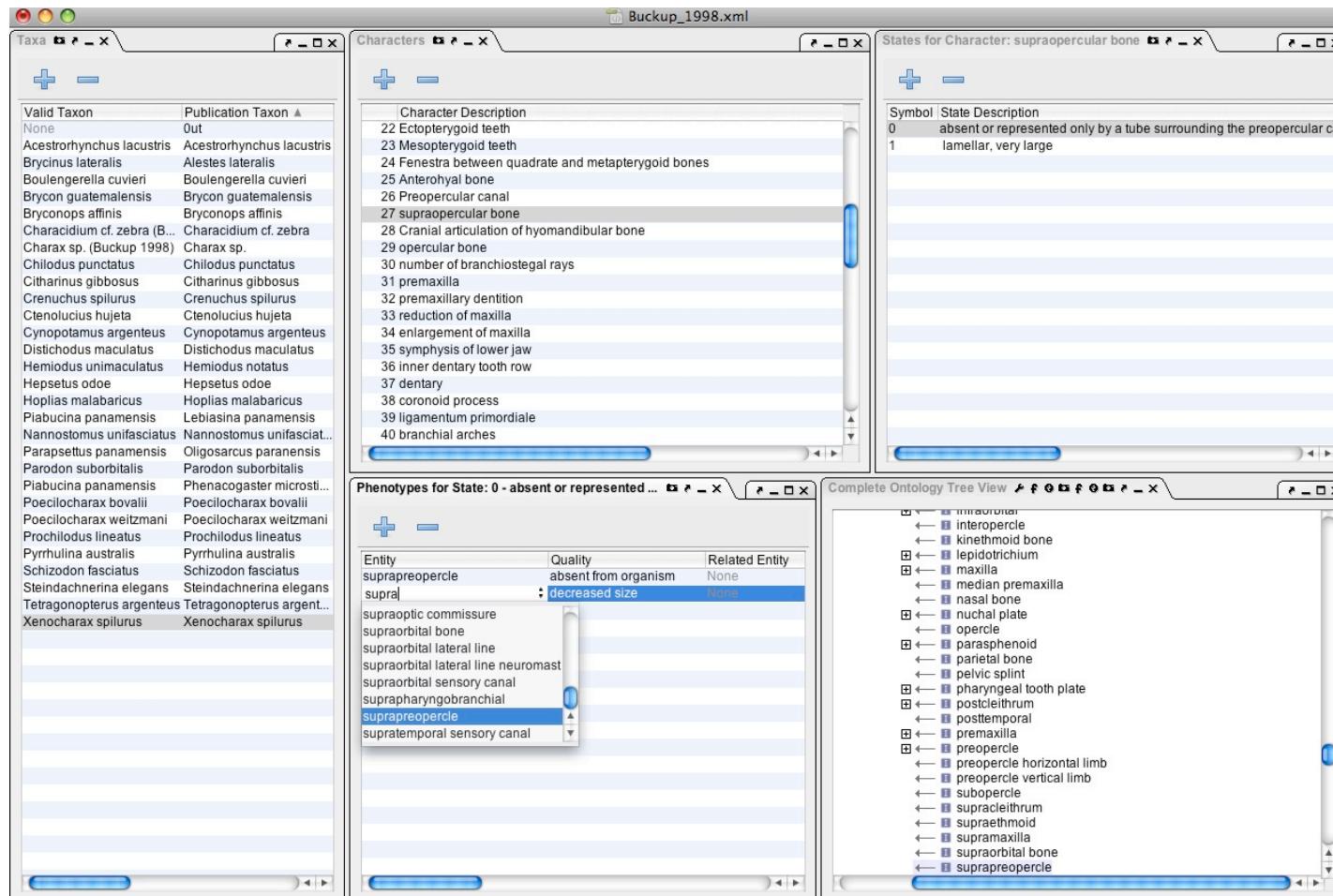
- Represent the variety of structures observed in all species in a clade
 - Teleost Anatomy Ontology (TAO)
 - Plant Structure Ontology
 - Amphibian Anatomy Ontology

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

The screenshot shows the Phenoscape annotation interface with four main windows:

- Taxa**: A list of valid and publication taxons.
- Characters**: A list of 40 anatomical characters, with character 27 (supraopercular bone) selected.
- States for Character: supraopercular bone**: A list of states for character 27, with state 0 (absent or represented only by a tube surrounding the preopercular canal) selected.
- Complete Ontology Tree View**: A tree view of the ontology, with the node for supraopercle expanded.

Below the interface is a green box containing four colored boxes:

- Entity** (blue)
- Quality** (orange)
- Taxon** (grey)
- TTO: *Ictalurus furcatus*** (grey)

Annotations below the interface:

- Entity**: TAO: suprapreopercle
- Quality**: PATO:decreased size
- Taxon**: TTO: *Ictalurus furcatus*

Uses of multi-species anatomy ontologies

Image annotation

Welcome to Morphbank
User: Paula Mabee [[logout](#)]
Group: Cypriniform Tree of Life (coordinator)

About Browse Search Tools Help

Featured from 219404 images



Forrest & Ken Starr

News and Updates

Cypriniform AToL (CToL) - TAO Ontology Links
The Cypriniformes AToL - (CToL) group invites you to see its first set of 1136 Cypriniform fish images uploaded to Morphbank. A first for Morphbank, these images are being used to illustrate as well as define and delimit an ontology. Each image is complete with links to the



(Posted:09-29-08)

Morphbank on the map
The Morphbank RSS services now carry longitude and latitude information. This Google Map sample represents an RSS feed that contains information about 100 geolocated specimens that were entered into Morphbank by Andrew Deans, of North Carolina State University. ...



(Posted:09-26-08)

***Isanopus* revision with images deposited in Morphbank**

Chatzimanolis, S. 2008. A revision of the neotropical beetle genus *Isanopus* (Coleoptera: Staphylinidae: Staphylinini). Journal of Natural History 42(25-26): 1765-1792.



With over 40 images of the new species in two collections: [Isanopus new species](#) and [Isanopus](#)

This work is part of a larger collaborative NSF ...

(Posted:09-15-08)

Image Record: [460945] Chromobota macracanthus

(m)

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 (Alician blue)

[Download: tiff \(1.21 MB\)](#)

[jpeg \(131.37 KB\)](#)

Copyright: Ericka Grey and Paula Mabee



[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 [\[i\]](#)

Order: Cypriniformes [\[i\]](#)

Family: Cobitidae [\[i\]](#)

Genus: Chromobota [\[i\]](#)

Species: Chromobota macracanthus [\[i\]](#)

[Determination annotations](#) [\[i\]](#) [\(Add Annotation...\)](#)

External links/identifiers

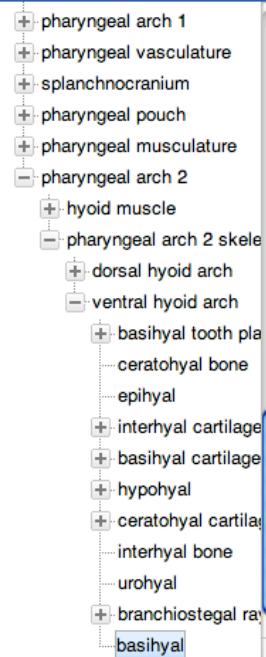
External identification: CToL-S:0001165

External identification: CToL-I:0000501

Ontology:

Teleost Anatomy Ontology TAO:0000316

[Other Annotations](#) [\[i\]](#) [\(Add Annotation...\)](#)

**basihyal** ([Link To Concept](#))[Details](#) [Visualization](#) [Marginal Notes](#) [Mappings](#) [Resources](#)**Class Name:** basihyal**ID:** TAO:0000316

Definition: The basihyal is the 'tongue' of the 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"

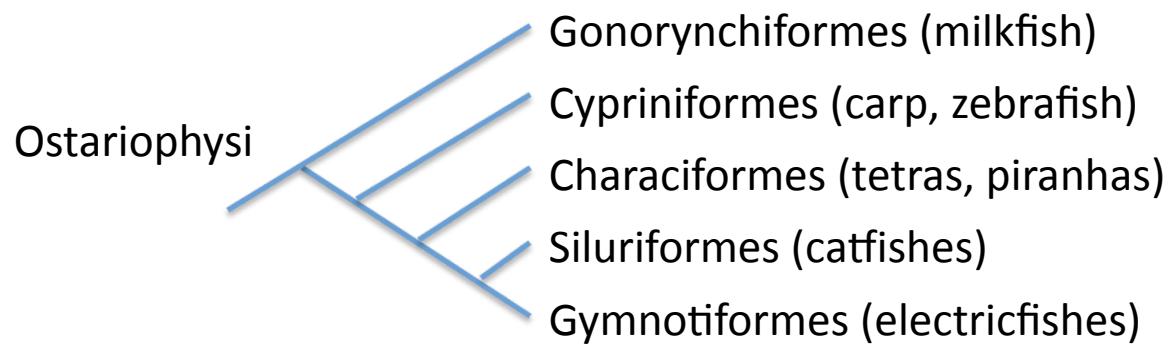
Why multi-species?

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

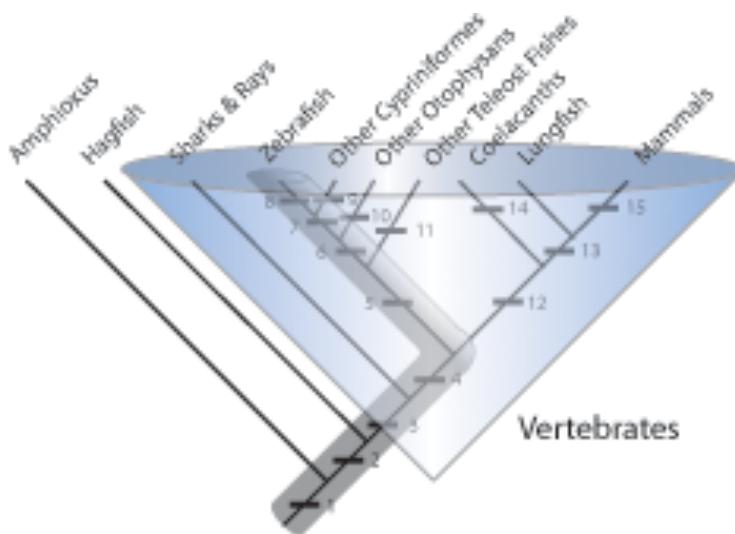
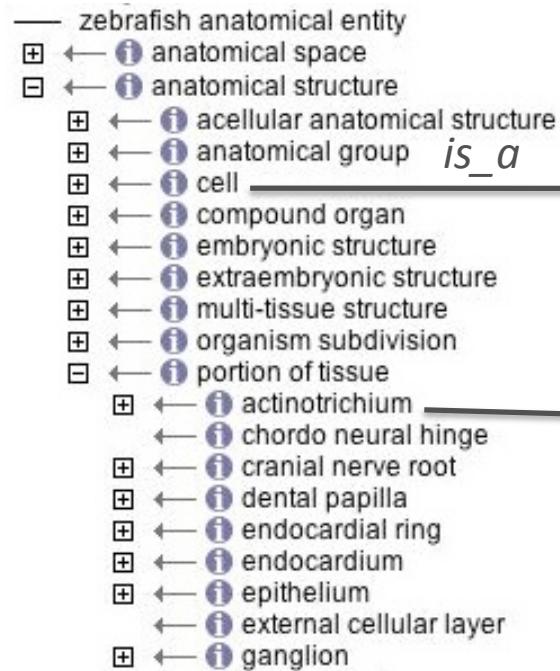


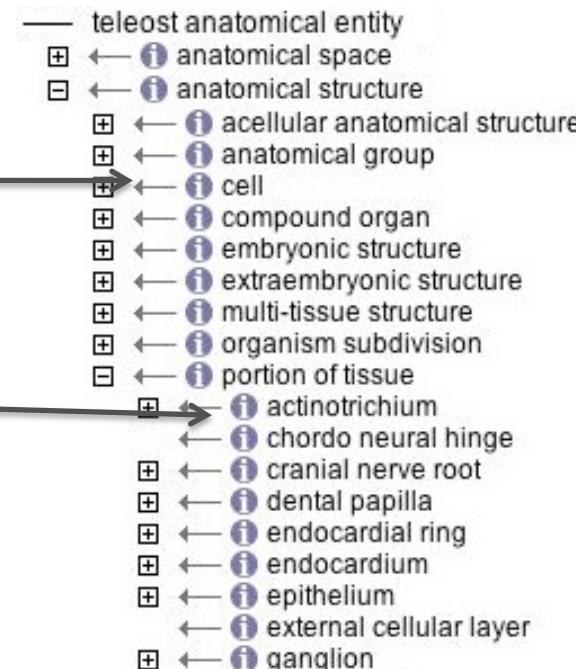
Fig. 1. Expansion of zebrafish core anatomical ontology (grey inner cylinder) to include all vertebrates (blue cone). The zebrafish anatomical ontology (~1,500 terms) includes features that evolved at various times along the common vertebrate lineage (characters 1–8). In order to make evolutionary comparisons 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



Teleost Anatomy Ontology



ZFIN

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

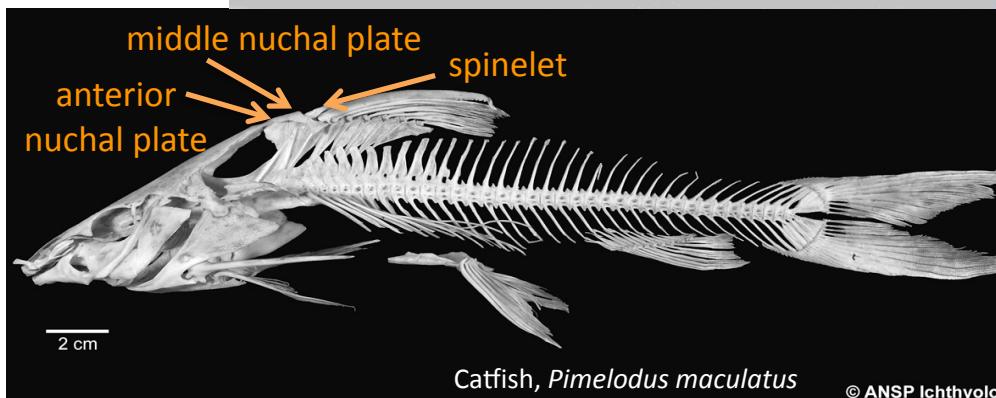
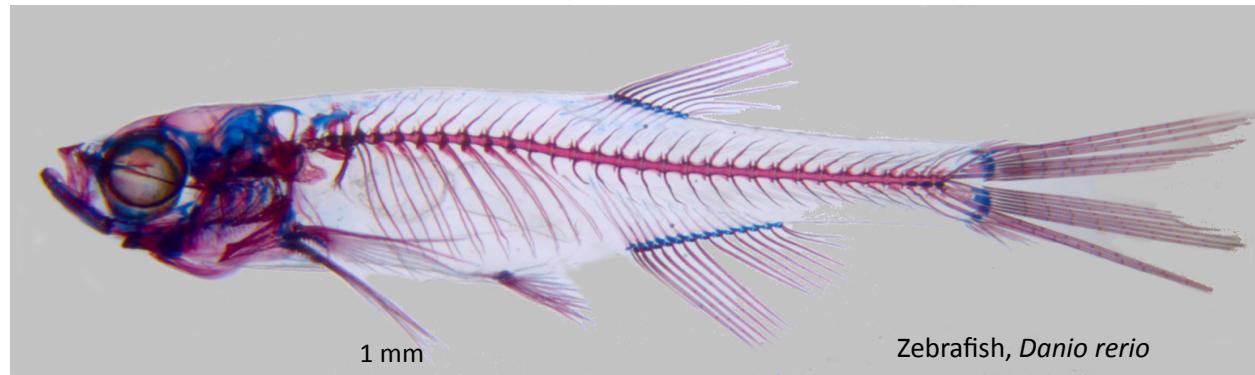
- Classes
 - — anatomical entity
 - ⊖ ← ⓘ immaterial anatomical entity
 - ← ⓘ anatomical line
 - ← ⓘ anatomical point
 - ⊕ ← ⓘ anatomical space
 - ← ⓘ anatomical surface
 - ← ⓘ material anatomical entity
 - ⊖ ← ⓘ anatomical structure
 - ⊖ ← ⓘ acellular anatomical structure
 - ⊖ ← ⓘ anatomical group
 - ⊕ ← ⓘ 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

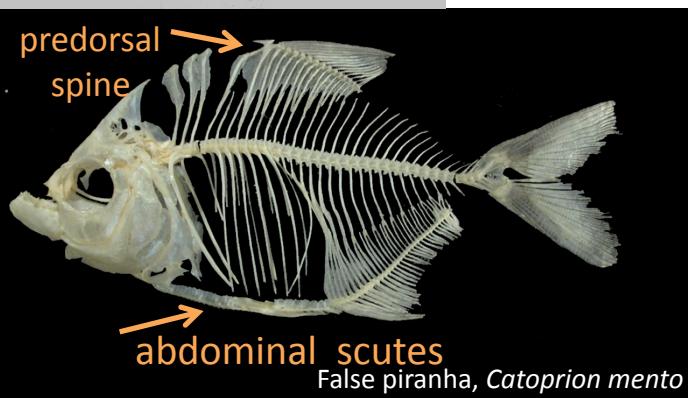
Growth of Teleost anatomy ontology

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



Catfish, *Pimelodus maculatus*

© ANSP Ichthyology



False piranha, *Catoprion mento*

Teleost anatomy terms

term name: **basihyal bone**

id: TAO:0000316  unique identifier

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

Teleost anatomy terms

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definition

Teleost anatomy terms

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Logical relationships to other
terms

Teleost anatomy terms

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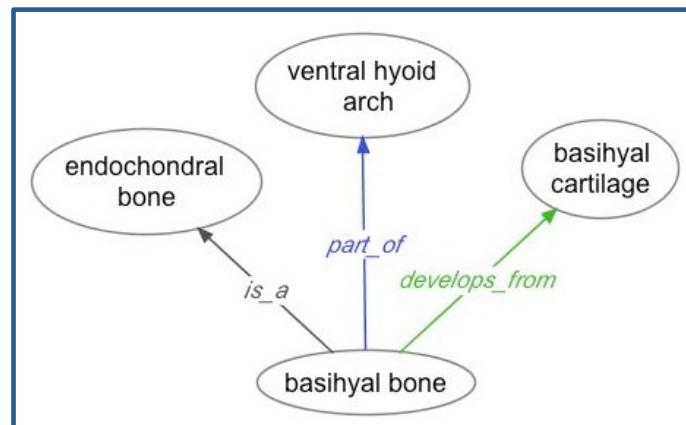
def: "Endochondral bone that is the median and anteriorly projecting element of the ventral hyoid arch."

synonym: "basihyoid"

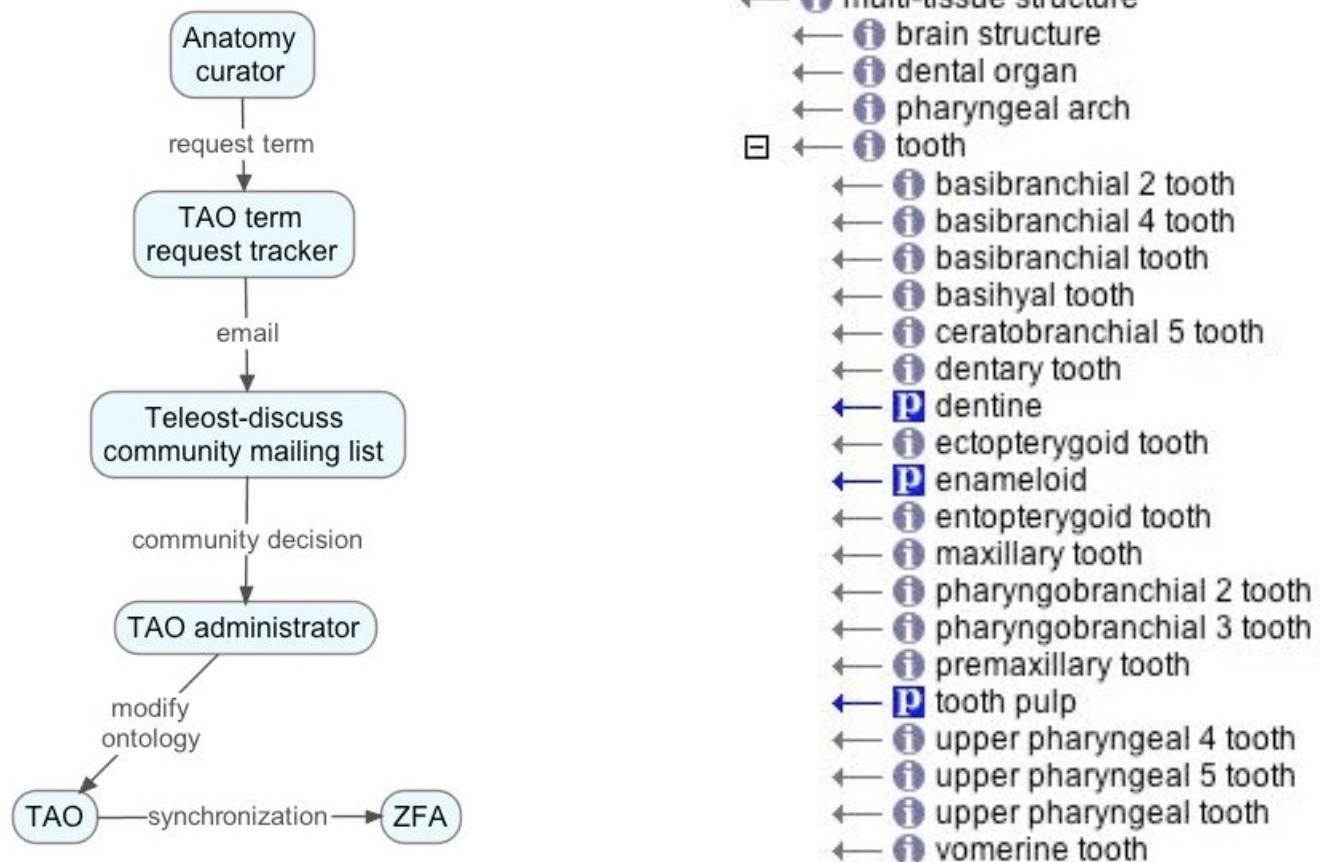
synonym: "glossohyal"

{ is_a: TAO:0001591 ! endochondral bone
relationship: develops_from TAO:0001510 ! basihyal cartilage
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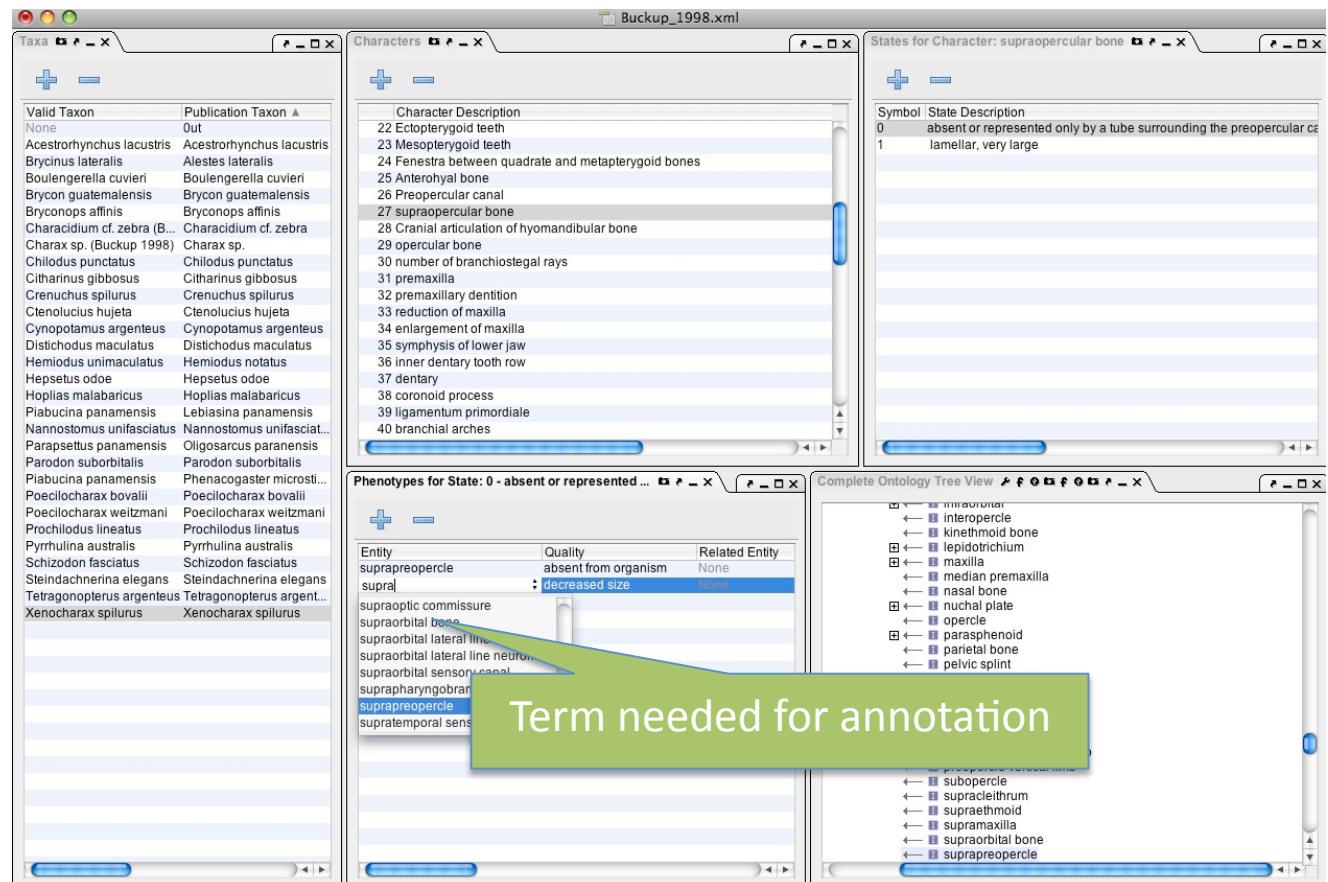
Logical relationships to other terms



TAO development workflow



TAO development workflow



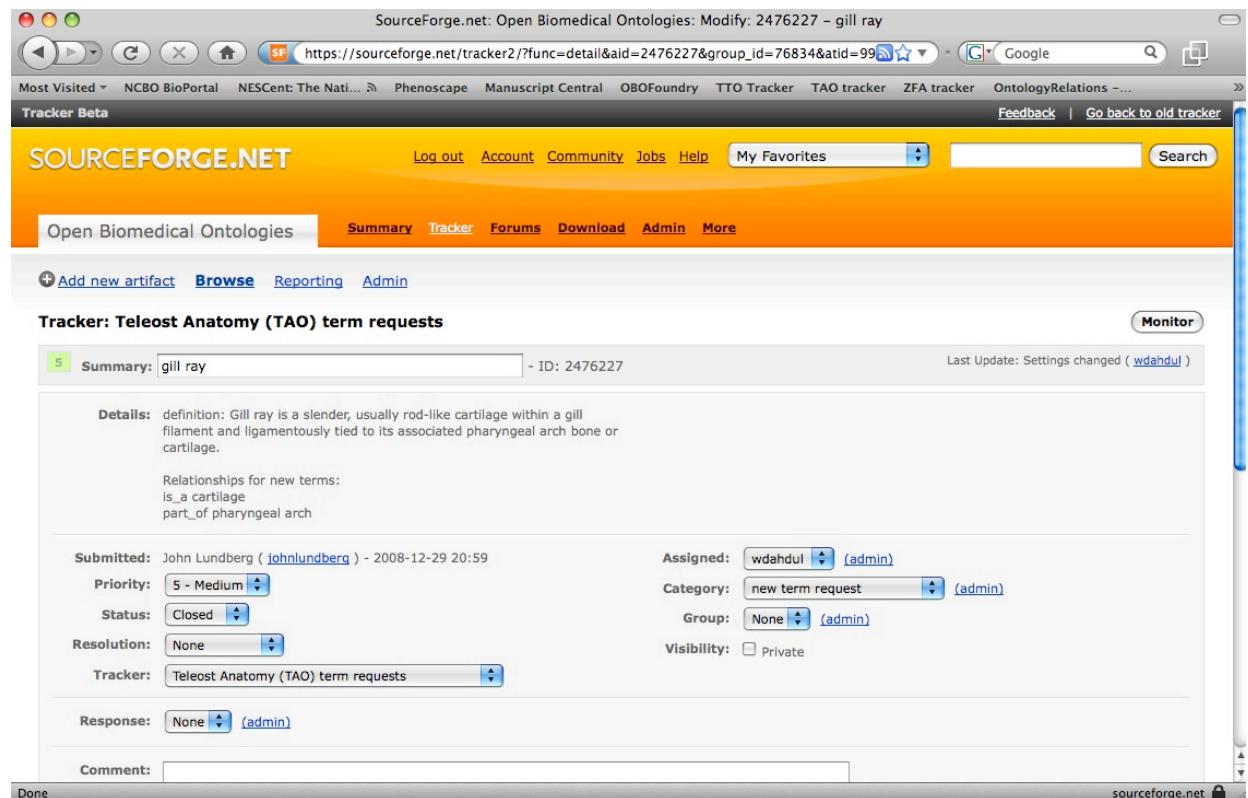
TAO development workflow



The screenshot shows the SourceForge.net Tracker Beta interface for 'Teleost Anatomy (TAO) term requests'. The page title is 'SourceForge.net: Open Biomedical Ontologies: Teleost Anatomy (TAO) term requests'. The main content area displays a table of term requests:

ID	Summary	Status	Opened	Assignee	Submitter	Resolution	Priority
2479435	more gill-arch element joints	Open	2008-12-31	nobody	johnlundberg	None	5
2479276	pharyngobranchial 4 tooth plate	Open	2008-12-31	nobody	ehilton	None	5
2477644	gill arch terms	Closed	2008-12-30	wdahdul	ehilton	None	5
2476227	gill ray	Closed	2008-12-29	wdahdul	johnlundberg	None	5
2459109	definitions for children of "pharyngeal tooth plate"	Closed	2008-12-22	wdahdul	iengeman	None	5
2458980	hyomandibular condyle for the opercle	Closed	2008-12-22	wdahdul	iengeman	None	5
2437026	html links in definitions	Open	2008-12-16	nobody	balhoff	None	5
2418270	mandibular symphysis	Open	2008-12-11	wdahdul	johnlundberg	None	5

TAO development workflow



TAO development workflow



The screenshot shows the SourceForge.net Email Archive for the **obo-teleost-discuss** mailing list. The interface includes a header with navigation links like SF.net, Most Visited, NCBO BioPortal, NESCent, Phenoscope, Manuscript Central, OBOFoundry, TTO Tracker, TAO tracker, ZFA tracker, and OntologyRelations. Below the header, there's a search bar and a link to the Mailing Lists page.

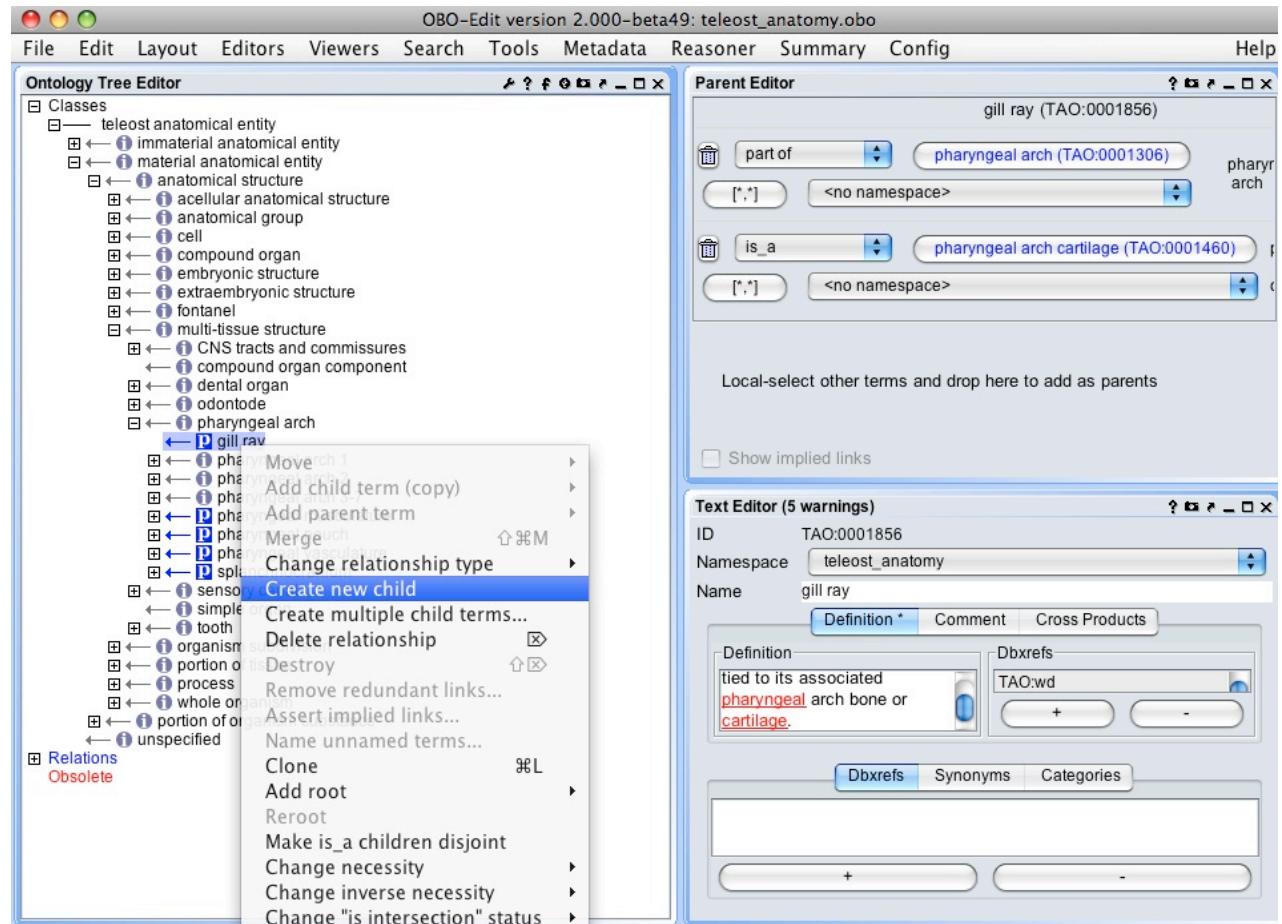
The main area displays the **Email Archive: obo-teleost-discuss (read-only)**. It features a monthly summary table for 2007 and 2008, showing the number of messages per month. Below the table are buttons for Threaded view, Show 25 messages, and Change View.

The message list starts with a thread titled "[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2479435] more gill-arch element joints (New)". The list continues with several other threads related to anatomy term requests, such as bone names, pharyngobranchial 4 tooth plate, and hyomandibular condyle for the opercle. Each message entry includes the author's email address and the date it was sent.

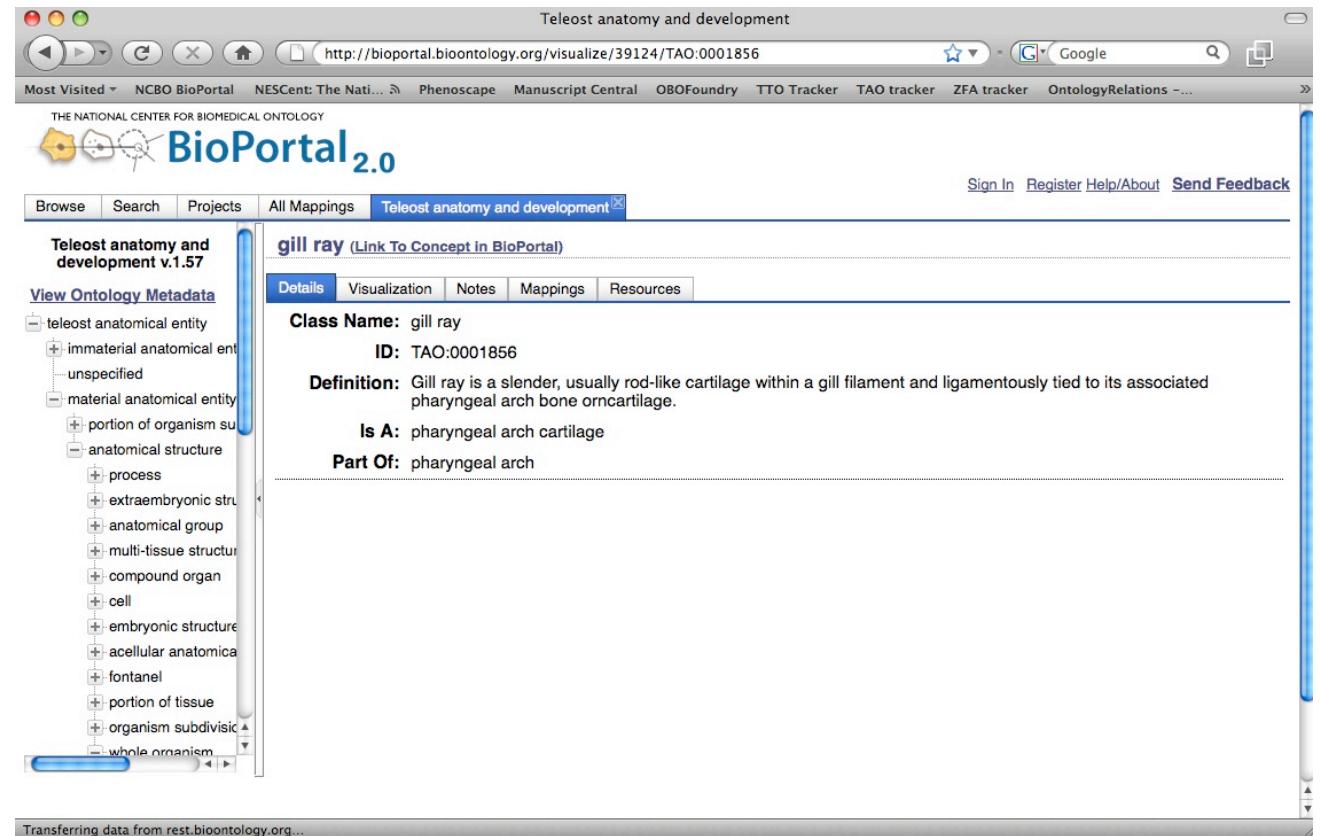
	Author	Date
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2479435] more gill-arch element joints (New)	<noreply@ne...>	2008-12-31 14:23
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2414789] bone names	<noreply@ne...>	2008-12-10 15:41
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2414789] bone names (New)	<noreply@ne...>	2008-12-31 14:05
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2414789] bone names (New)	<noreply@ne...>	2008-12-31 14:09
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2414789] bone names (New)	<noreply@ne...>	2008-12-31 14:16
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2479276] pharyngobranchial 4 tooth plate (New)	<noreply@ne...>	2008-12-31 12:26
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2476227] gill ray (New)	<noreply@ne...>	2008-12-30 18:32
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2476227] gill ray (New)	<noreply@ne...>	2008-12-30 18:42
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2458980] hyomandibular condyle for the opercle (New)	<noreply@ne...>	2008-12-30 18:37
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2477644] gill arch terms (New)	<noreply@ne...>	2008-12-30 17:42
[Obo-teleost-discuss] [obo-Teleost Anatomy (TAO) term requests-2477644] gill arch terms	<noreply@ne...>	

At the bottom of the archive page, there are links for Done, sourceforge.net, and a lock icon.

TAO development workflow



TAO development workflow



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.

Refinement of definitions

Infraorbital 1

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

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

Zebrafish Anatomical Ontology

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

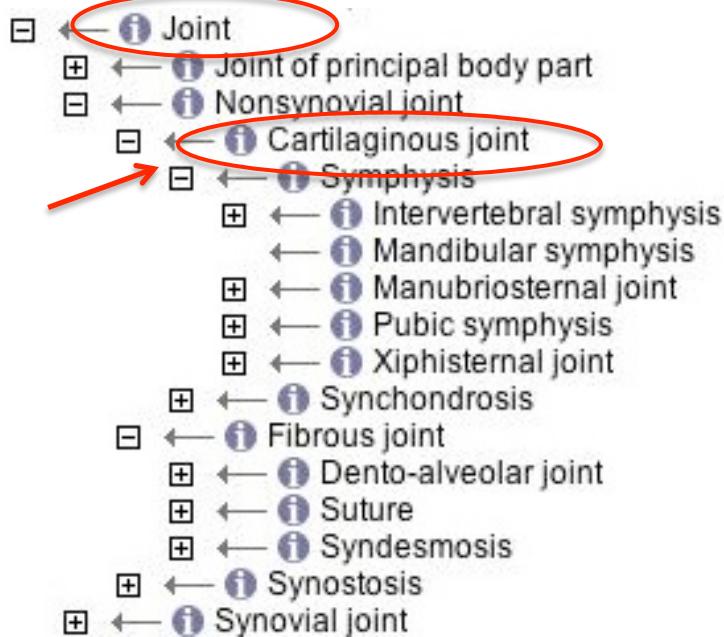
Teleost Anatomy Ontology

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

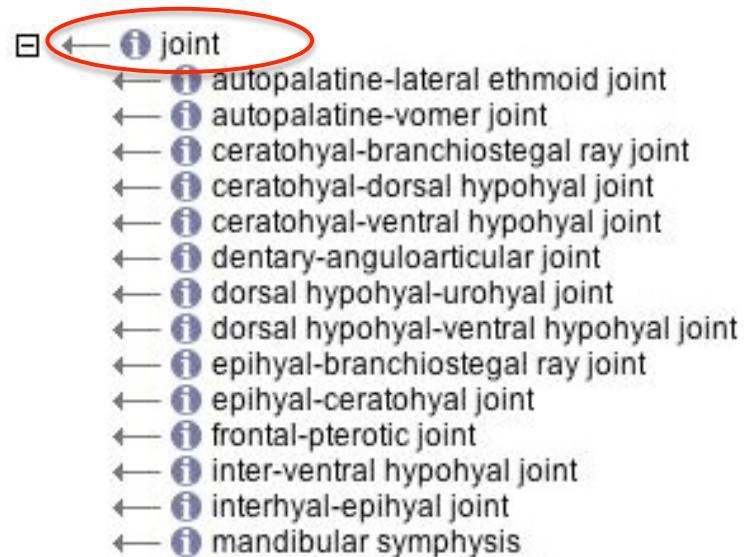


Some classes from single-species ontologies move to quality ontology

Foundational Model of Anatomy

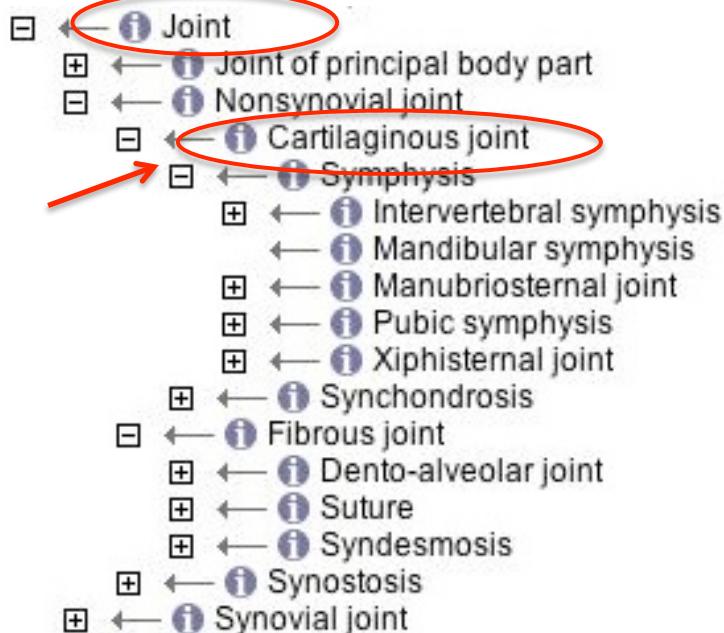


Teleost Anatomy Ontology

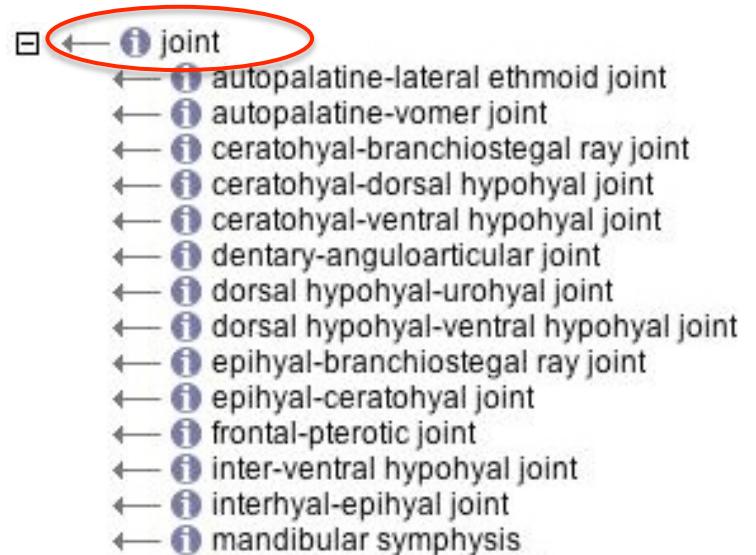


Some classes from single-species ontologies move to quality ontology

Foundational Model of Anatomy



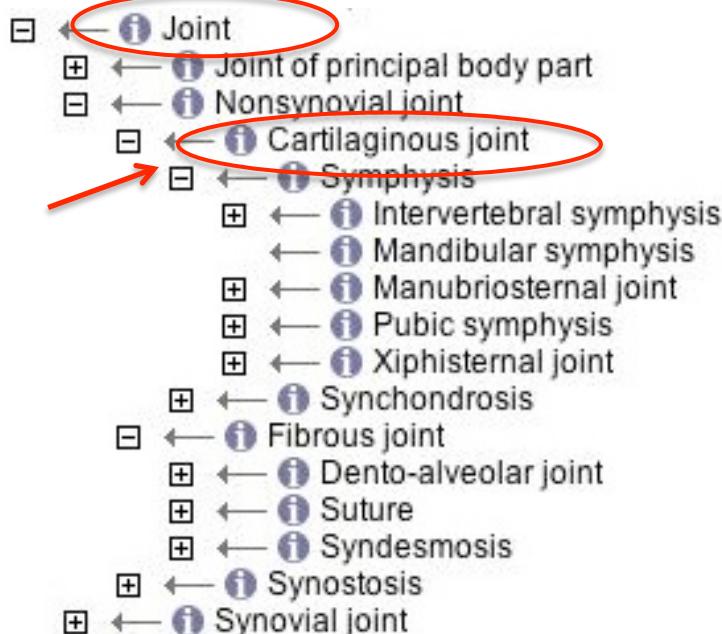
Teleost Anatomy Ontology



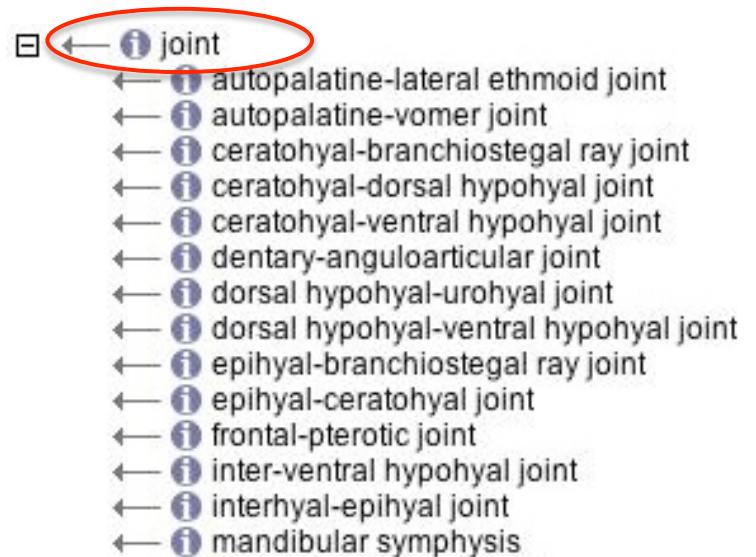
Solution 1: Joint terms in TAO have multiple parents

Some classes from single-species ontologies move to quality ontology

Foundational Model of Anatomy



Teleost Anatomy Ontology



Solution 2: anatomy classes become quality terms in a different ontology:

Entity

TAO: prootic-exoccipital joint

Quality

PATO:cartilaginous

Taxon

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:

Managing ontology growth

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

The screenshot shows a software window titled "Phenotypes for State: untitled". Inside the window, there is a table with three columns: "Entity", "Quality", and "Related Entity". The first row contains the value "process^part_of(maxilla)" in the "Entity" column, "present ..." in the "Quality" column, and "None" in the "Related Entity" column. A green callout box with a blue border and rounded corners points from the word "Entity" to the "Entity" column of the table. Below this callout box is another blue callout box containing the text "TAO: process part_of TAO: premaxilla".

Entity	Quality	Related Entity
process^part_of(maxilla)	present ...	None

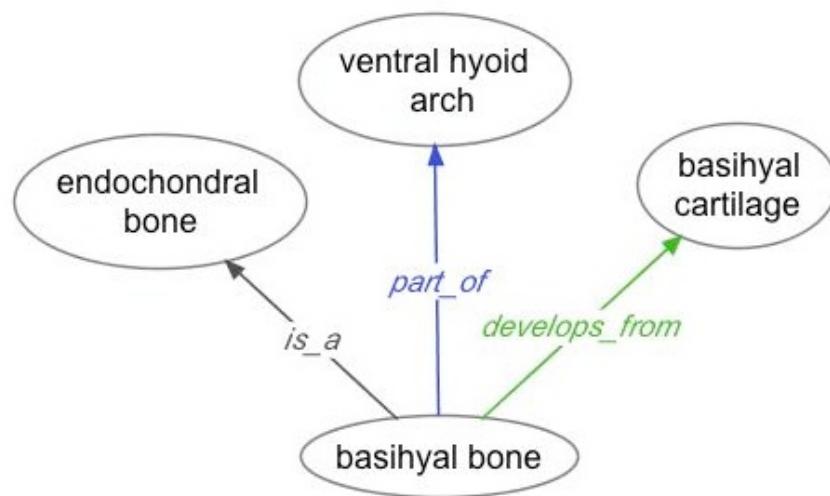
Entity

TAO: process *part_of* TAO: premaxilla

Managing ontology growth

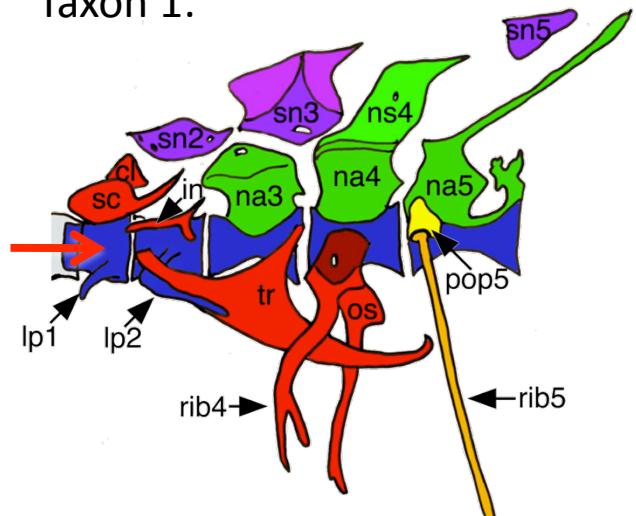
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- Alternatively, terms can be “post-composed” at time of annotation by combining existing terms to create a new one:
- Guideline: add new term to ontology if it will be used repeatedly

Different relationships for different taxa

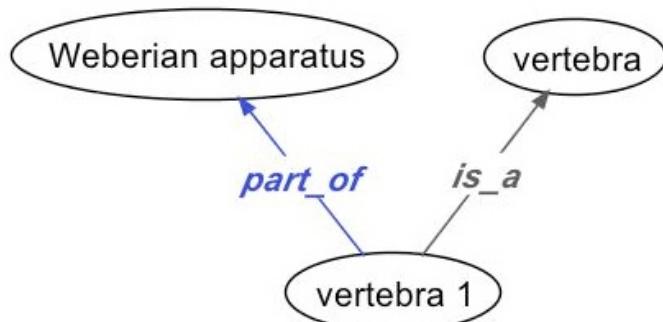


Different relationships for different taxa

Taxon 1:

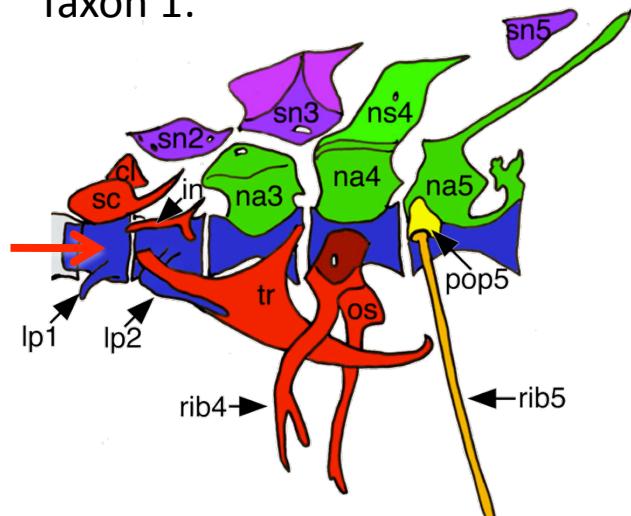


Weberian apparatus consists of the modified anteriormost four vertebrae and associated structures of otophysans which transduce sound or pressure waves from the tunica externa of the swimbladder to the inner ear (Alexander, 1962).

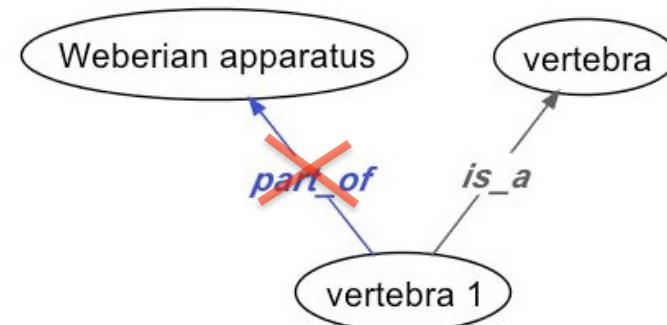
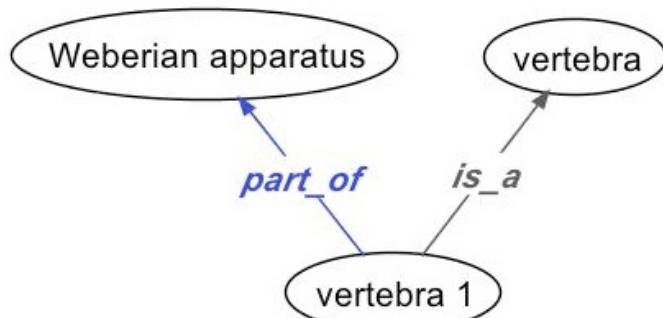
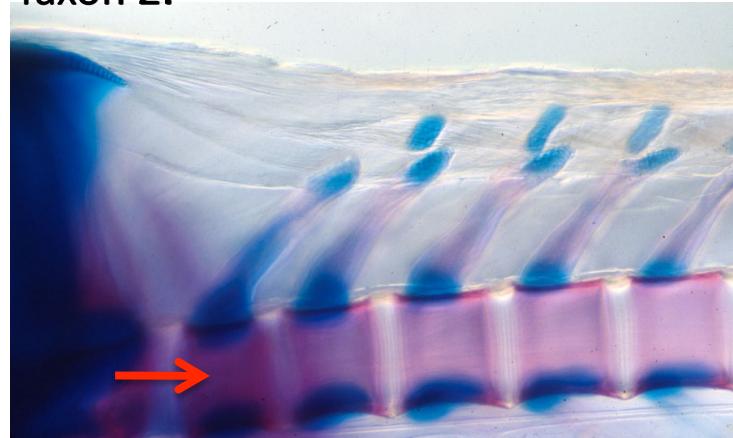


Different relationships for different taxa

Taxon 1:

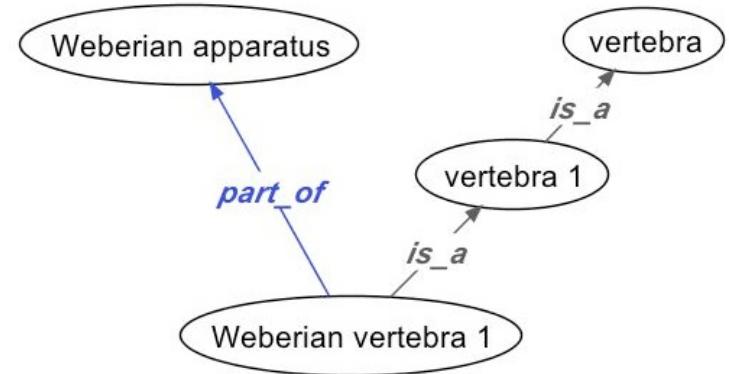


Taxon 2:



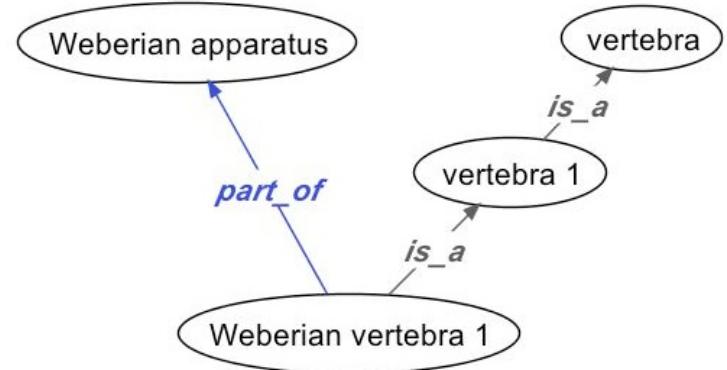
Different relationships for different taxa

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

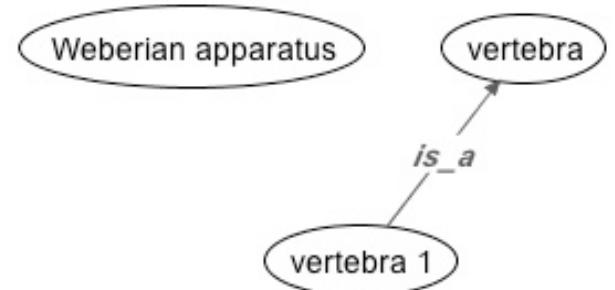


Different relationships for different taxa

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



Solution 2: describe variation in annotation statement



Entity

TAO: vertebra 1 *part_of*
Weberian apparatus

Quality

PATO:present

Taxon

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

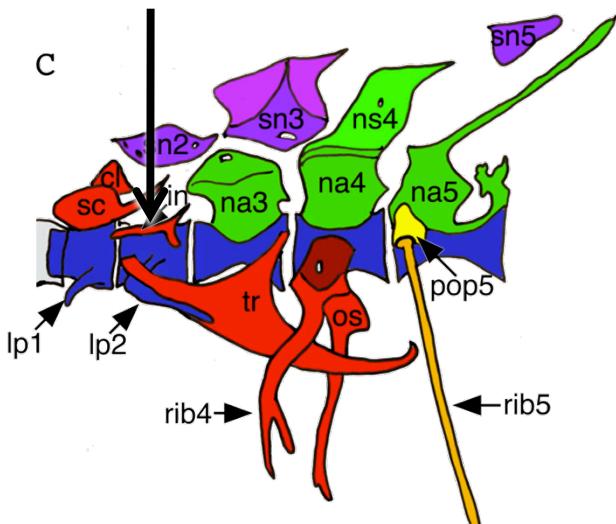
Available from Evidence Code Ontology

- 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

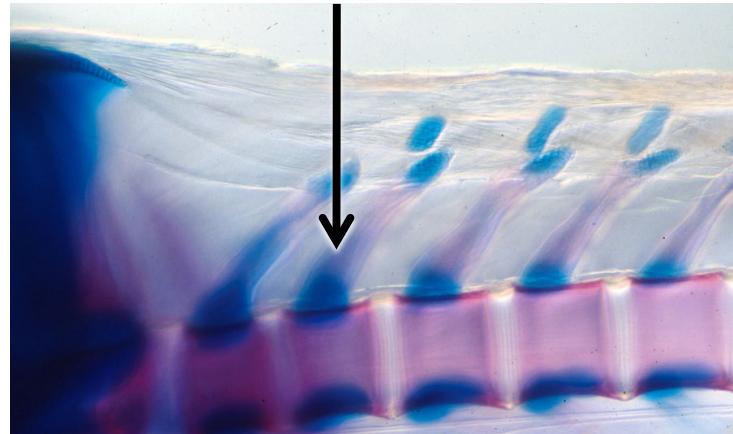
Publication	Entity 1	Taxon 1	Entity 2	Taxon 2	Evidence
Fink and Fink 1981	neural arch 2	Teleostei	intercalarium	Otophysi	inferred from positional similarity

Homology

intercalarium



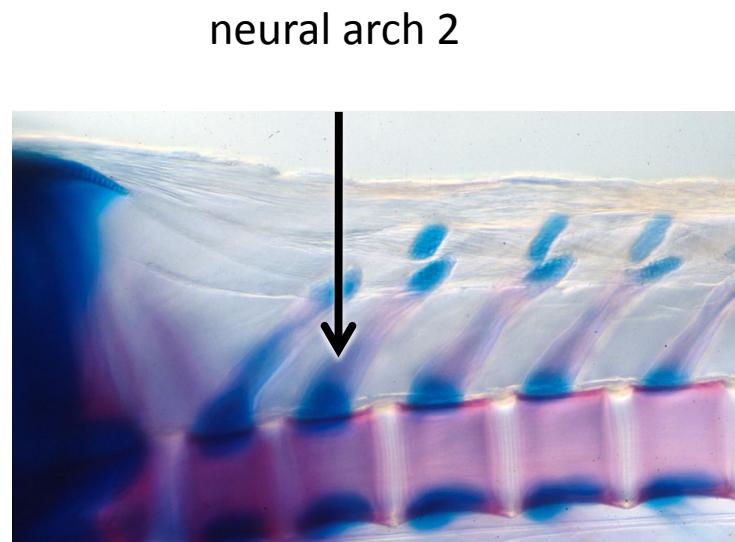
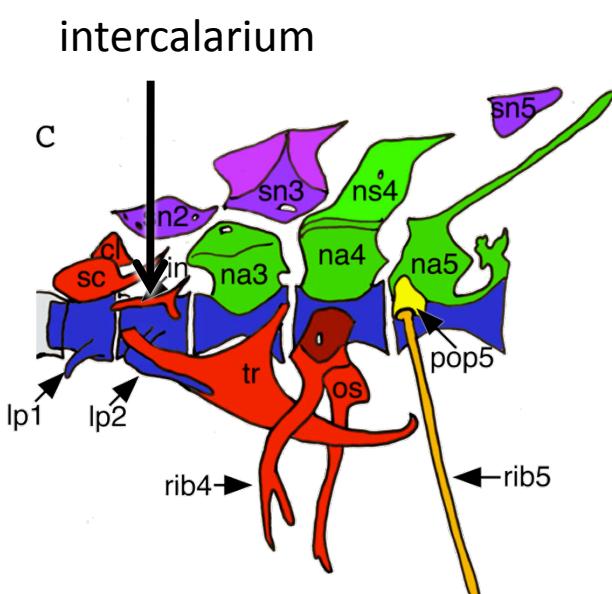
neural arch 2



Representation in TAO:

- ◻ ← ⓘ neural arch
- ◻ ← ⓘ intercalarium
- ← ⓘ neural arch 2
- ◻ ← ⓘ neural arch 3
- ← ⓘ neural arch 4

Homology



Representation in TAO:

- ← ⓘ neural arch
- ⊕ ← ⓘ intercalarium
- ← ⓘ neural arch 2
- ⊕ ← ⓘ neural arch 3
- ← ⓘ neural arch 4

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 representing variability across a clade in an ontology
- Homology can be treated explicitly with a formalized relation between terms

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