

Integrating anatomy and phenotype ontologies with taxonomic hierarchies

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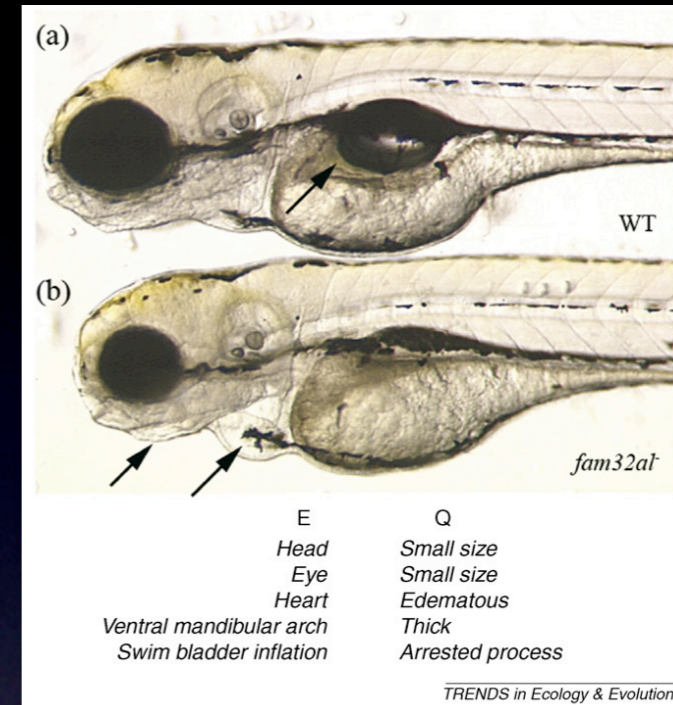
Comparative studies

Developmental genetics

TABLE 1. CHARACTER STATE MATRIX USED FOR PHYLOGENETIC ANALYSIS OF THE PLACEMENT OF *B. capapretum* WITHIN PIMELODIDAE AND *Brachyplatystoma*. Character states described in Appendix 1 and text.

	12345	1 67890	11111 12345	11112 67890	22222 12345	22223 67890	33333 12345
<i>Steindachnerdion</i>	11110	00000	00000	21000	00000	00000	00011
<i>Phractocephalus-Leiarius</i> group	11110	00000	00000	00000	00000	00000	00001
<i>Pimelodus</i> group	11111	11111	00000	00000	00000	00000	00000
<i>Calophysus</i> group	11111	11111	00000	00000	01110	00000	01201
<i>Zungaro</i>	11111	10000	00000	01001	20000	00000	00010
<i>Sorubim</i> group	11111	10000	00000	20001	20000	00000	00011
<i>Platynematichthys</i>	11111	10000	11000	00000	00000	00000	00000
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000	00000	00000	00000
<i>B. tigrinum</i>	11111	10000	11121	00111	11000	00000	10010
<i>B. platyneum</i>	11111	10000	11120	11111	11110	00000	11100
<i>B. filamentosum</i>	11111	10000	11111	11111	21101	11111	01101
<i>B. capapretum</i>	11111	10000	11111	11111	21101	11111	01101
<i>B. rousseauxii</i>	11111	10000	11111	11111	21101	11100	01101
Heptapteridae	00000	00000	00000	00001	00000	00000	00001
Pseudopimelodidae	00000	00000	00000	00000	01000	00000	10010
Bagridae	00100	00000	00000	01000	00000	00000	00000
Ictaluridae	00000	00000	00000	00001	00000	00000	00000

Lundberg & Akama 2005



Specimen-based descriptions



Length.—8 mm. Entirely black.
 Head shining, microscopically finely punctured, without furrows below antennae; malar area large, equal to half the length of eye; mandible with two teeth. Scape very long; in male a little longer than two following segments together, in female nearly as long as three following together; 3rd segment four times as long as 2nd, 4th three times as long as 2nd, in male; 3rd three times and 4th twice as long as 2nd in female.
 Prothorax very short; mesonotum shining, almost smooth, except for diffuse microscopic puncturation and one or two larger punctures; prescutum separated from scutum by deep lateral grooves; parapsidal furrows distinct; scutellum with scattered shallow punctures; mesopleura almost smooth; metapleura strongly and closely punctured. Propodeum with large shallow reticulate punctures with a tendency to lie in transverse rows. Boundary of propodeum with metapleuron marked by a regular horizontal row of shallow dorsoventrally elongated depressions. Fore wings with one recurrent nervure, one small closed submarginal cell, very large 1st discoidal cell, and 3rd discoidal cell open to wing margin; each wing with large pointed anal lobe; hind wings each with 10 hamuli. Hind coxae with a few scattered punctures, and hind legs nearly twice as long as the body. Abdomen shining, “club” sometimes reddish ventrally, strongly compressed laterally, triangular or almost ovate. Ovipositor not exerted.

<http://www.linnean-online.org/>

How to associate phenotypes with taxa and organisms?

Assumptions

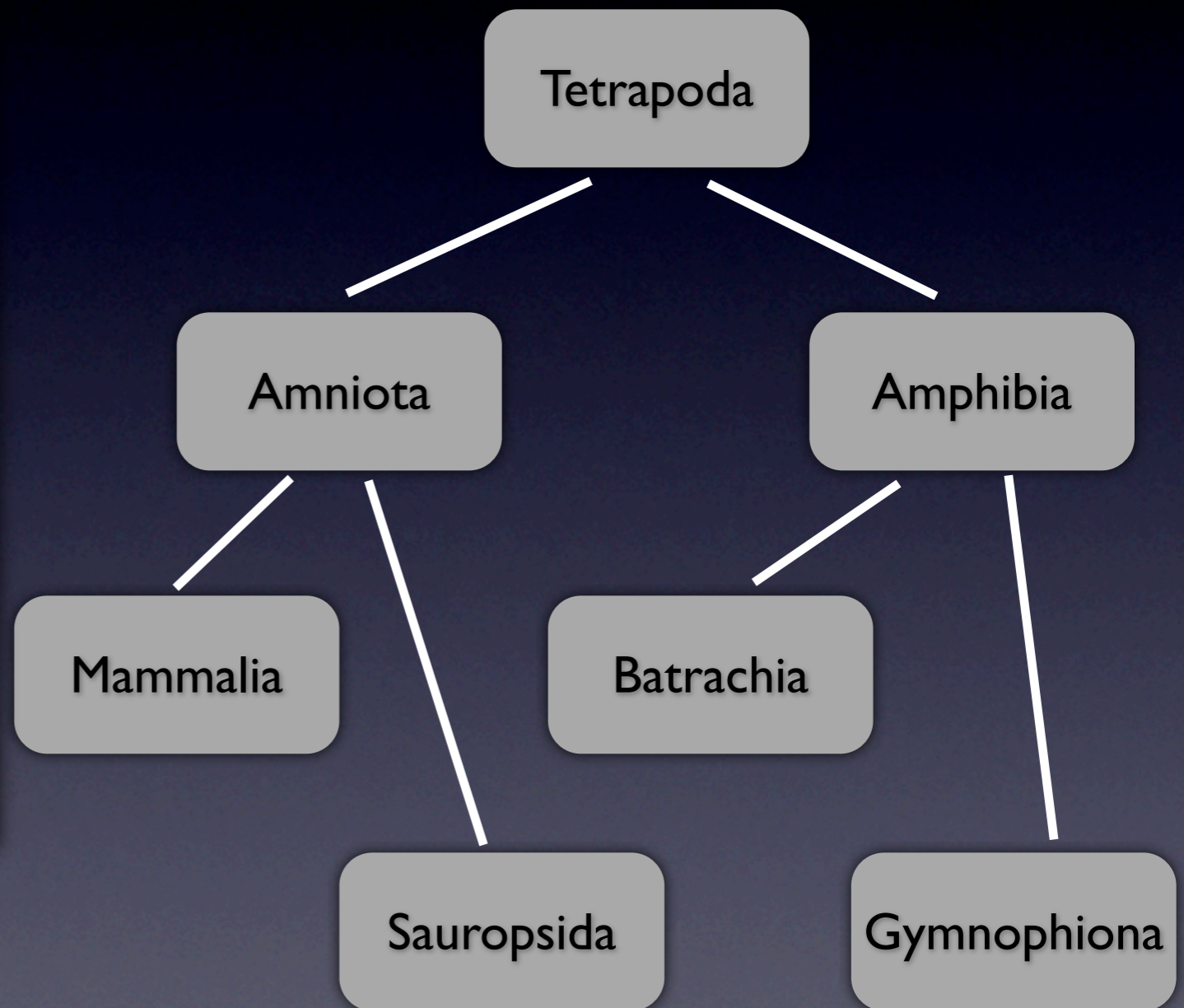
- EQ phenotype model: **opercle**, **triangular**
- OWL DL: classes vs. individuals
- *has_part* some (**opercle** and *bearer_of* some **triangular**)

Taxonomic hierarchy

◦ **Tetrapoda** (tetrapods) *Click on organism name to get more information*

- **Amniota** (amniotes)
 - **Mammalia** (mammals)
 - **Prototheria**
 - **Theria**
 - **Sauropsida** (sauropsids)
 - **Sauria** (diapsids)
 - **Testudines** (turtles)
- **Amphibia** (amphibians)
 - **Batrachia**
 - **Anura** (frogs and toads)
 - **Caudata**
 - **Gymnophiona**
 - **Caeciliidae** (true caecilians)
 - **Ichthyophiidae** (Asiatic tailed caecilians)
 - **Rhinatreumatidae** (neotropical tailed caecilians)
 - **Scolecophoridae** (African caecilians)
 - **Typhlonectidae** (aquatic caecilians)
 - **Uraeotyphlidae** (Indian caecilians)
 - **unclassified Gymnophiona**

<http://www.ncbi.nlm.nih.gov/>



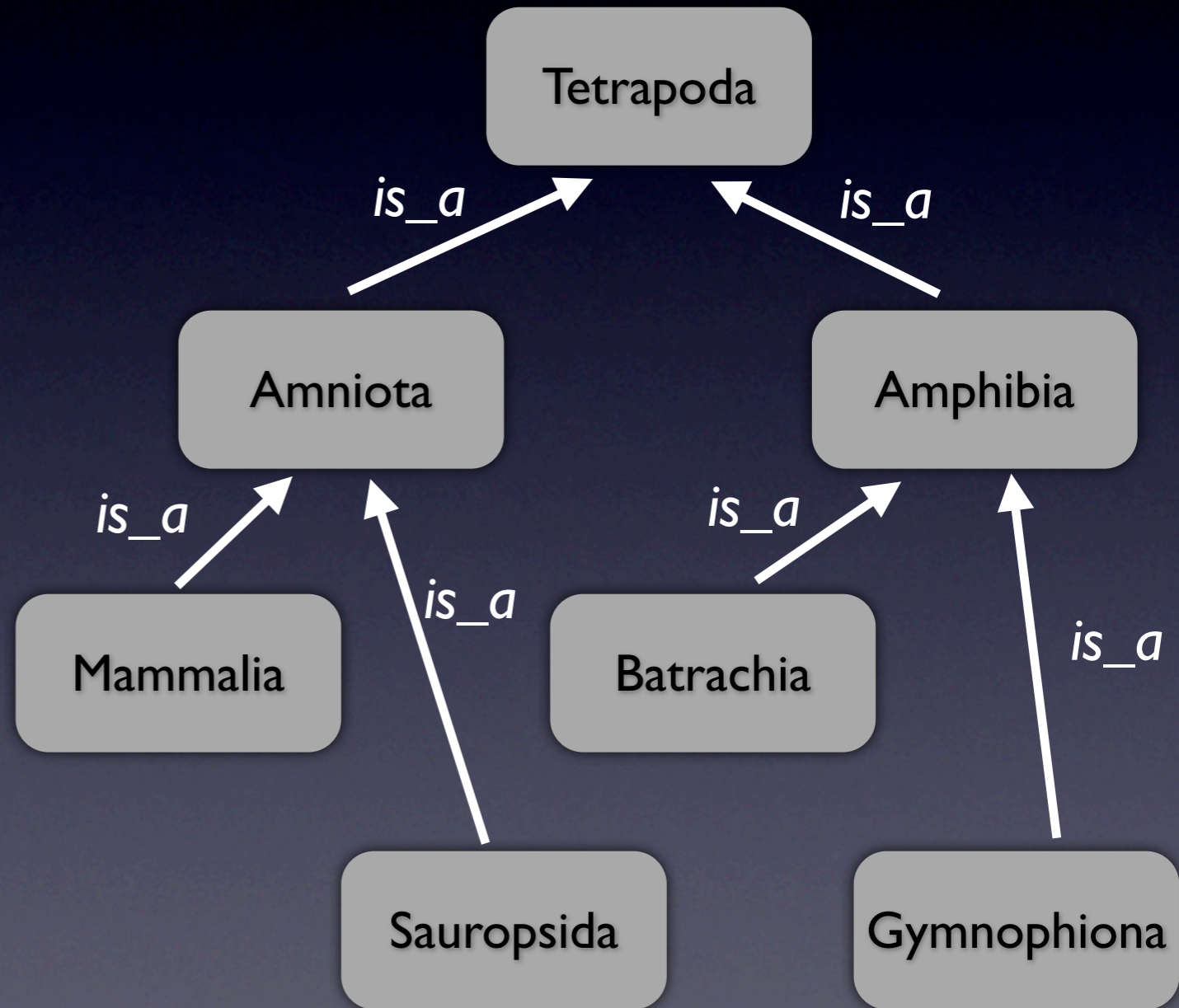
OBO format

```
[Term]
id: NCBITaxon:32523
name: Tetrapoda
is_a: NCBITaxon:8287
synonym: "tetrapods" EXACT genbank_common_name []
xref: GC_ID:1
```

```
[Term]
id: NCBITaxon:32524
name: Amniota
is_a: NCBITaxon:32523
synonym: "amniotes" EXACT genbank_common_name []
xref: GC_ID:1
```

```
[Term]
id: NCBITaxon:8457
name: Sauropsida
is_a: NCBITaxon:32524
synonym: "sauropsids" EXACT genbank_common_name []
xref: GC_ID:1
```

```
[Term]
id: NCBITaxon:40674
name: Mammalia
is_a: NCBITaxon:32524
synonym: "mammals" RELATED blast_name []
synonym: "mammals" EXACT genbank_common_name []
property_value: has_rank NCBITaxon:class
xref: GC_ID:1
```



```
χρητ: GC_ID:1
βιολογική_λάση: μαστοειδής NCBITaxon:32523
επινοήση: "μαστοειδής" EXACT γενεοκοινωνία []
επινοήση: "μαστοειδής" ΒΕΓΑΤΕD ρεσφ-μνοήση []
τξ-9: NCBITaxon:32524
```

Genus-differentia?

- Tetrapoda = (Vertebrata and *has_part* exactly 4 limb)
- Mammalia = (Amniota and *has_part* some hair and *has_part* some mammary_gland)

Are taxa really classes?

- “Organisms belong in a particular species because they are part of that genealogical nexus, not because they possess any essential traits” — David Hull
- Differentiating characteristics help us to identify groups, but groupings aim to represent shared evolutionary history

Practical requirements

- Which species exhibit a yellow fin?
- Which species exhibit both blue and yellow fins?
- In which families can we find organisms without scales?
- Which taxa possess hair as a direct ancestral condition?
- Which taxa had some ancestor with hair?
- Annotate phenotypes which differ from the ancestral condition

Class problems

- Tetrapoda = (Vertebrata and *has_part* exactly 4 limb)
- Snake: SubClassOf (Tetrapoda and *has_part* exactly 0 limb)
- Cumbersome to represent polymorphism, evolutionary reversals/losses without unintended inferences and logical inconsistencies

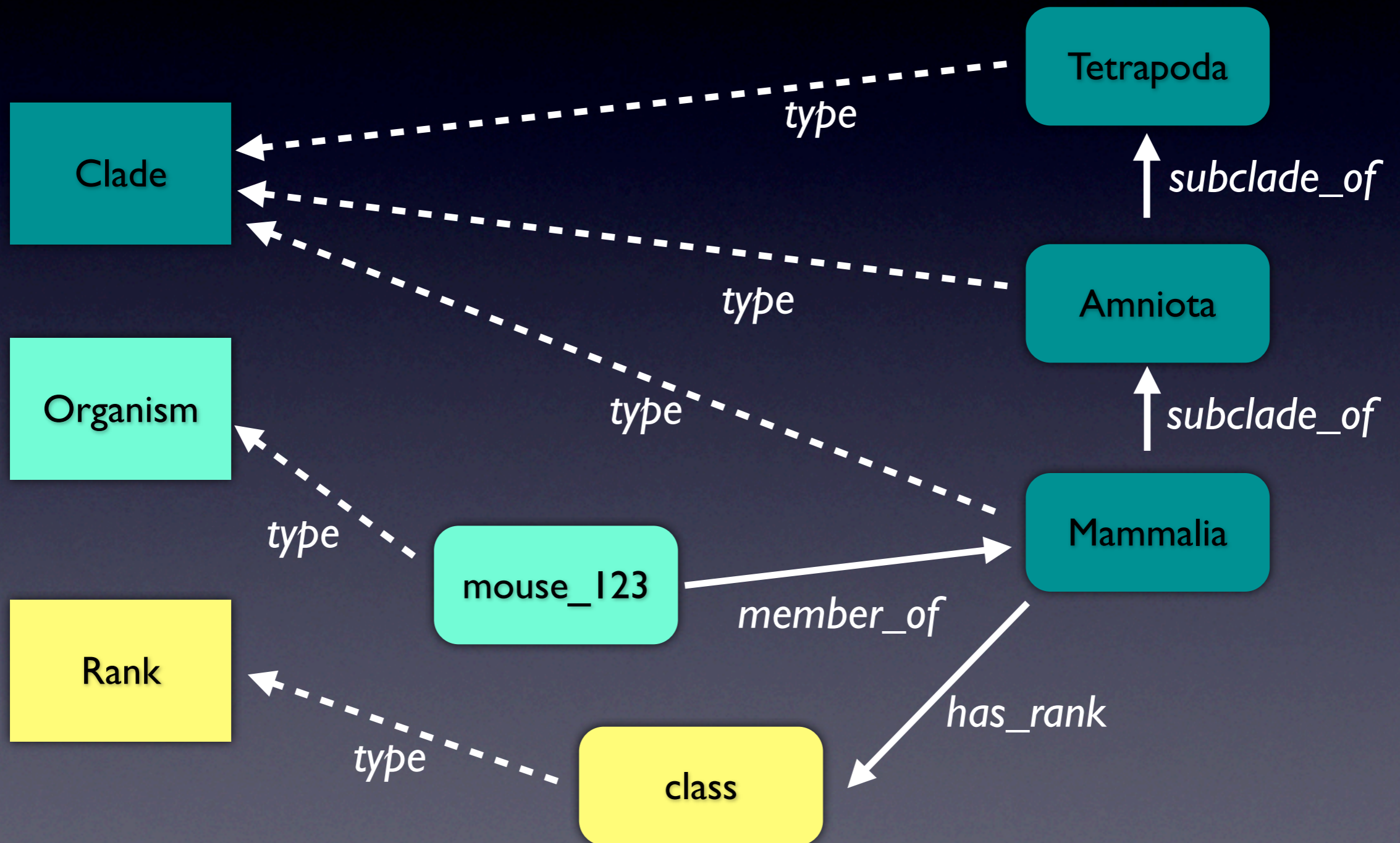
Class problems

- Tetrapoda = (Vertebrata and *has_part* exactly 4 limb)

- Snake: SubClassOf (Tetrapoda and *has_part* exactly 0 limb)

- Cumbersome to represent polymorphism, evolutionary reversals/losses without unintended inferences and logical inconsistencies

Individual model



Individual model

- *subclade_of/contains_clade* - taxon–taxon (transitive)
- *member_of/has_member* - organism–taxon
- *member_of* o *subclade_of* → *member_of*
- *has_ancestor/has_descendant* - organism–organism (transitive)
- *has_progenitor* - taxon–organism (ancestor of members)
- *member_of* o *has_progenitor* → *has_ancestor*
- *subclade_of* o *has_progenitor* → *has_ancestor_to_members*

Query for a basic phenotype, returning species only:

Description: *Ictalurus_punctatus*

Types +

- Clade
- has_member some (has_part some (caudal_fin and bearer_of some bifurcated))

Same individuals +

Different individuals +

Property assertions: *Ictalurus_punctatus*

Object property assertions +

- subclade_of *Ictalurus*
- has_rank species
- has_member catfish_1234
- subclade_of *Ictaluridae*
- subclade_of *Siluriformes*

Data property assertions +

Query:

Query (class expression)

(has_rank value species) and (has_member some (has_part some (fin and bearer_of some bifurcated)))

Execute Add to ontology

Query results

Instances (1)

- Ictalurus_punctatus

Legend:

- Super classes
- Ancestor classes
- Equivalent classes
- Subclasses
- Descendant classes
- Individuals

Polymorphism (maybe):

The screenshot displays two panels from a software application, likely a web ontology editor, showing information for the class *Ictalurus_furcatus*.

Description: Ictalurus_furcatus

Types +

- Clade
- has_member some (has_part some (scale and bearer_of some blue))
- has_member some (has_part some (scale and bearer_of some yellow))

Same individuals +

Property assertions: Ictalurus_furcatus

Object property assertions +

- has_rank species
- subclade_of Ictalurus
- subclade_of Ictaluridae
- subclade_of Siluriformes

Data property assertions +

Negative object property assertions +

A newly observed reversal of an ancestral condition:

Description: Siluriformes

Types +

- Clade
- has_progenitor some (has_part only (not scale))

Same individuals +

Different individuals +

Property assertions: Siluriformes

Object property assertions +

- has_rank order
- has_member catfish_1234
- has_member scaled_catfish_4321
- contains_clade Ictaluridae
- contains_clade Ictalurus
- contains_clade Ictalurus_punctatus

Data property assertions +

Description: scaled_catfish_4321

Types +

- has_part some scale
- Organism

Same individuals +

Different individuals +

Property assertions: scaled_catfish_4321

Object property assertions +

- member_of Ictaluridae
- member_of Siluriformes

Data property assertions +

Negative object property assertions +

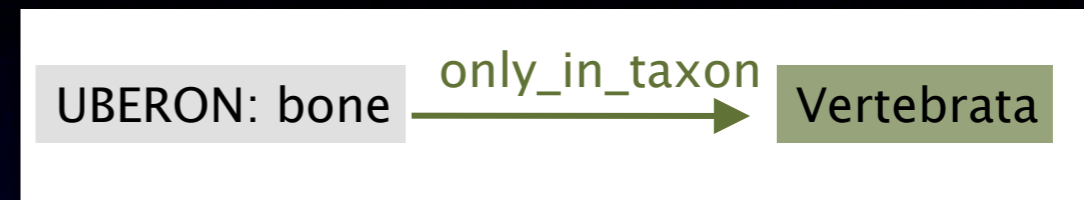
Negative data property assertions +

Class-based alternative?

- **Ictalurus_punctatus**: SubClassOf
has_ancestor some (**Ictalurus_punctatus**
and *has_descendant* some (*has_part* some
(**caudal_fin** and *bearer_of* some
bifurcated)))
- Not able to meet all the requirements, and
less understandable (to me)

Taxonomic restrictions

Uberon uses relations to constrain applicability of anatomical terms:



(Melissa Haendel)

UBERON:bone: SubClassOf
in_taxon only **Vertebrata**

Using our approach:

UBERON:bone: SubClassOf
in_taxon only (*member_of* value **Vertebrata**)

Summary

- In OWL-DL, at least, we have to choose class or individual for our model entities
- Individual representation...
 - perhaps closer fit to evolutionary reality
 - addresses query use-cases straightforwardly
- Can directly associate properties with taxa (e.g. ranks)

Feedback

- Use-cases which call for classes?
- Modifications or additions to the proposed taxon–organism relations?
- Example ontology:

➔ <http://tinyurl.com/ICBO-taxonomy>

Acknowledgments

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