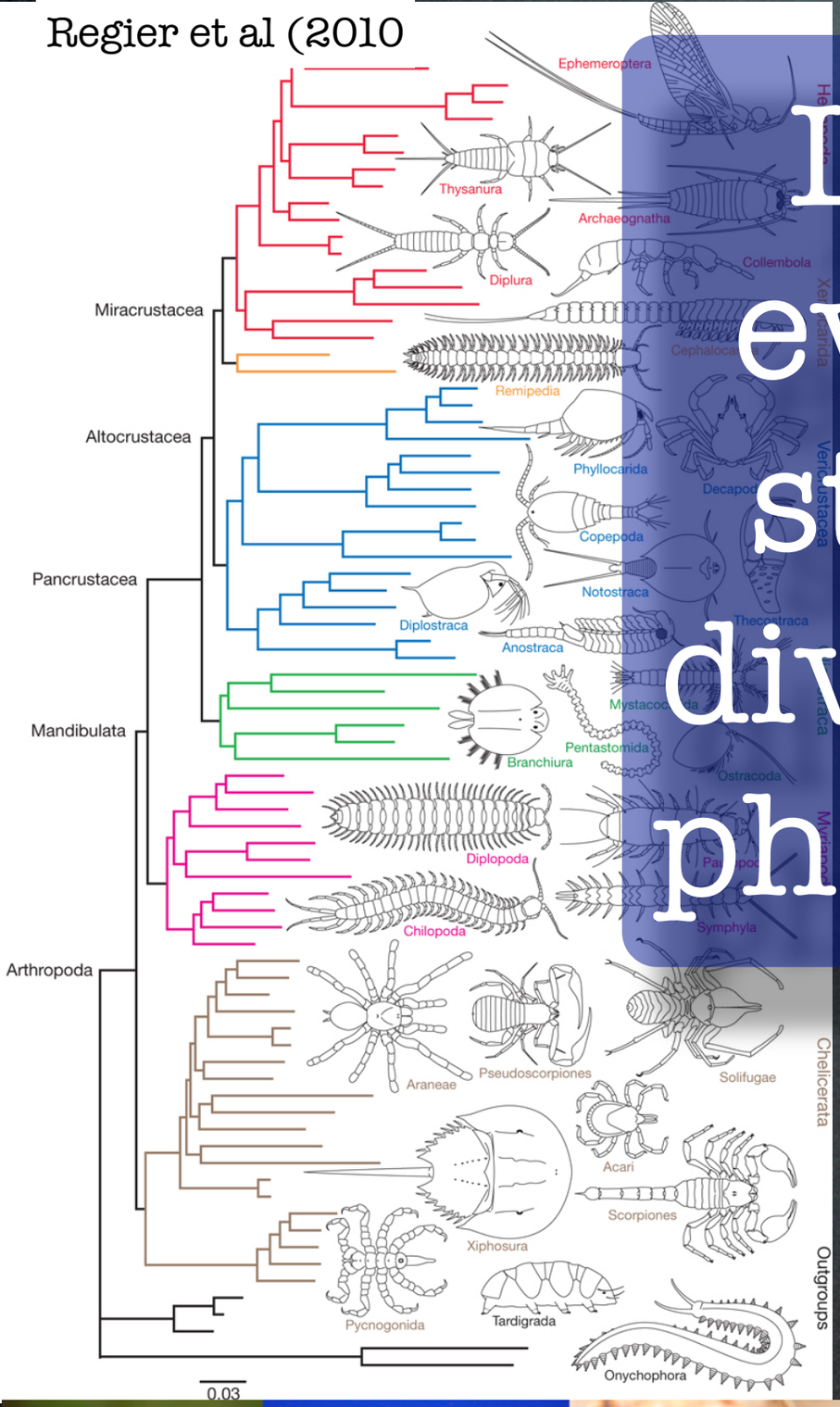
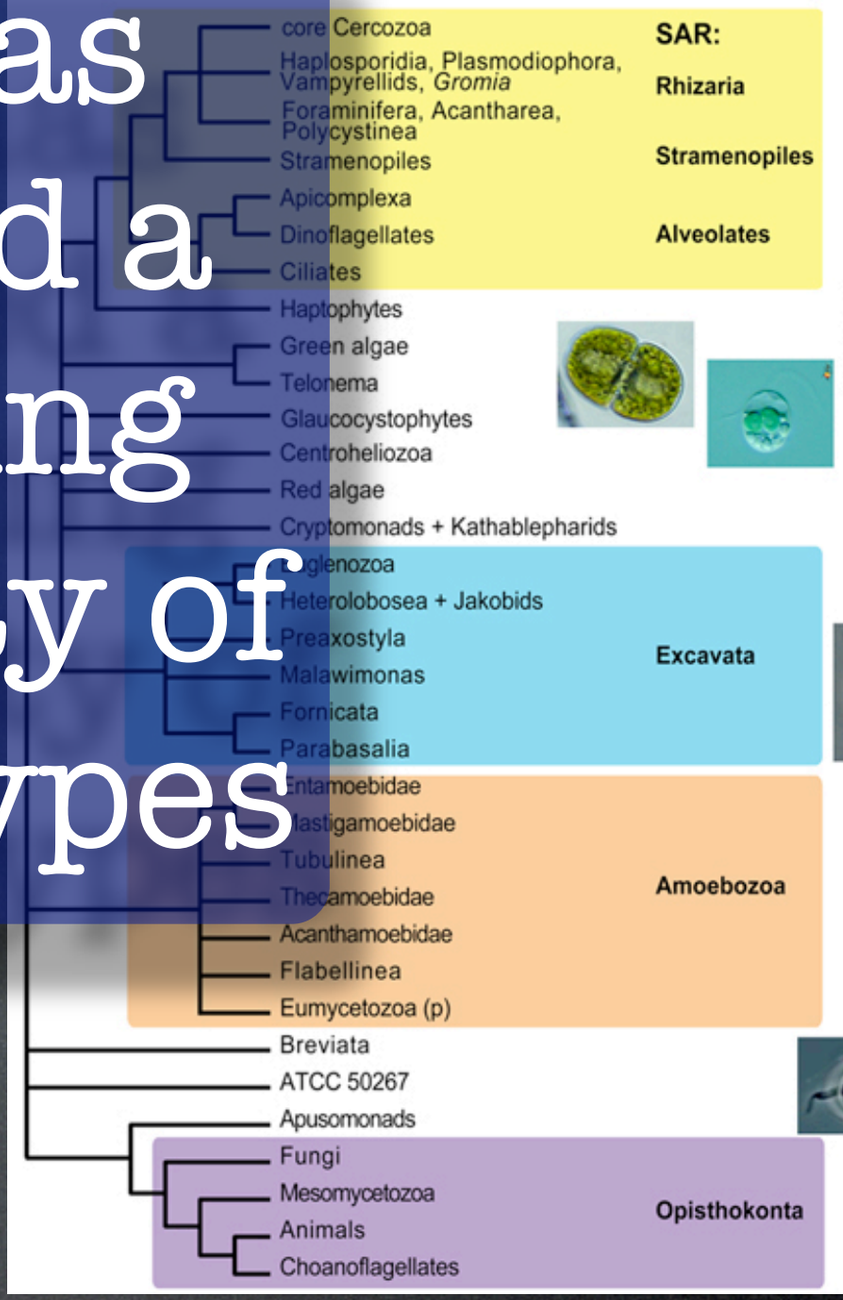
Reasoning over phenotype diversity, character change, and evolutionary descent

Hilmar Lapp
National Evolutionary Synthesis Center (NESCent)
Seminar at University of Florida, March 1, 2011

Regier et al (2010



Parfrey et al (2010, Parfrey & Katz



Life has evolved a stunning diversity of phenotypes

Images: Web Tree of Life (http://tolweb.org)

Large body of evolutionary phenotype documentation

PERGAMON

biochemical systematics and ecology

Biochemical Systematics and Ecology 28 (2000) 319–350

www.elsevier.com/locate/biochemsyseco

The interrelationships of Acanthomorph fishes: A total evidence approach using molecular and morphological data

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[b]*Division of Fishes, National Museum of Natural History, Washington, DC 20560, USA*

Received 30 N

Abstract

DNA sequence and morphologica
acanthomorph fishes and two speci
ctophiformes. A 572 base-pair (bp) s
from three regions of the 28S ribos
series were analyzed under the crite
resulted in a set of four most parsim
congruent with the hypothesis artic
554–626). © 2000 Elsevier Science

Keywords: Phylogenetic analysis; Molec
terygii; Euacanthopterygii; Acanthopter

1. Introduction

The Acanthomorpha (Rosen,
group of the major radiation of
and over 14,000 species, they
remarkable morphological and

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0305-1978/00/$ - see front matter © 20
PII: S0305-1978(99)00069-1

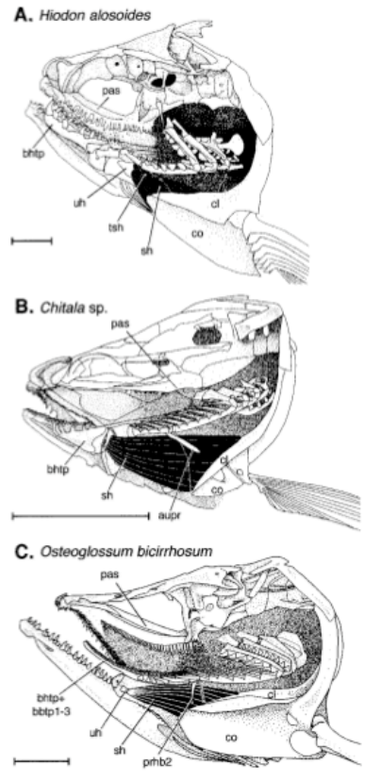

Fig. 1. The three osteoglossomorphs examined in this study showing the overall variation in the structure of the TBA. These specimens are seen in left lateral view with the left "cheek" skeleton removed; anterior is facing left. In addition to variation in the distribution and form of intraoral dentition, these taxa differ in orientation of the pectoral girdle. In Notopteroidei (represented by *Chitala* sp.), the pectoral girdle is more vertical, which brings it more posterior than in Hiodontiformes and Osteoglossoidei (represented by *Hiodon alosoides* and *Osteoglossum bicirrhosum*, respectively), in which the pectoral girdle has a distinct anterior horizontal arm. Osteoglossiformes (Notopteroidei + Osteoglossoidei) share the presence of autogenous bones (notopteroids) or processes (osteoglossoids) associated with the second gill arch, although the homology of these elements is questionable. The sternohyoideus muscle is shown in black and the orientation of the CBL is shown by a heavy gray line (the CBL actually lies between the left

1999) and between species (Sanford and Lauder, 1990). Such functional variation of the TBA suggests unappreciated complexity of the morphology of the TBA.

In this study, I examined the morphological variation in a character complex, the so-called tongue-bite apparatus (TBA), in three genera (*Hiodon*, *Osteoglossum*, and *Chitala*), which represent three living clades of Osteoglossomorpha (Fig. 1). The monophyly of Osteoglossomorpha, even w
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PROCEEDINGS OF THE ACADEMY OF NATURAL SCIENCES OF PHILADELPHIA 153: 119–15

Phylogeny of the subfamily Corydoradinae (Siluriformes: Callichthyidae), with a definiti

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ABSTRACT—A phylogenetic hypothesis of relationships within the callichthyi
based on 83 characters. The monophyly of the subfamily is well supported, as are
and *Brochis*. However, the monophyly of *Corydoras*, as traditionally defined, is n
included species being more closely related to *Aspidoras* or *Brochis* than to nomin
with a previous hypothesis, which considered *Brochis* and *Corydoras* as forming a
as its sister-group. A clade composed of *Aspidoras* and the species currently assi
cropterus, *C. prionotus* and *Corydoras* sp. A, is herein recognized based on the fol
portion of frontal bone long, hypobranchial 2 well ossified, free margin of oper
supraorbital canal long, and ossified portion of pectoral spine reduced. The ren
monophyletic assemblage with complex vertebra compact, posterior expansion o
triangular dorsal lamina on anguloarticular, and medial expansion of coracoid e
to accommodate the monophyletic groups defined in the present study. Discussi
clade are provided.

RESUMO—Uma hipótese sobre as relações filogenéticas da subfamília de Callich
base em 83 caracteres. O monofiletismo da subfamília é corroborado, assim c
Aspidoras e *Brochis*. Entretanto, o monofiletismo de *Corydoras*, como tradicionalm
algumas de suas espécies atuais sendo mais proximamente relacionadas a *Aspidor*
nominais. Estas conclusões contrastam com uma hipótese prévia que considera
pamento monofilético, com *Aspidoras* como seu grupo-irmão. Um clado compos
a *Corydoras*, *C. barbatus*, *C. macropterus*, *C. prionotus* e *Corydoras* sp. A, é diagnos
anterior do osso frontal alongado, hipobranquial 2 bem ossificado, margem livre
canal supra-orbital alongado, e porção ossificada do espinho peitoral reduzida. Os
um grupo monofilético definido pela vértebra complexa compacta, expansão p
entalhe, presença de lâmina dorsal triangular no ângulo-articular e expansão m
classificação é proposta para acomodar os grupos monofiléticos definidos no pres
de cada um dos principais clados são fornecidos.

INTRODUCTION

The subfamily Corydoradinae includes about 90% of the species in the Neotropical catfish family Callichthyidae and is one of the most diverse siluriform assemblages in the Neotropics, with approximately 170 valid species (Nelson, 1994; Reis, 1998a). Species of the Corydoradinae are of small

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Representatives
several freshwater environments, ranging from fast-flowing piedmont streams with sandy or rocky bottom to lowland pools with muddy bottom. They are well known among fishkeepers worldwide, with *Corydoras* species being particularly popular.

Zoological Journal of the Linnean Society, 2002, **135**, 529–546. With 14 figures

Comparative osteology of the *Danio* (Cyprinidae: Ostariophysi) axial skeleton with comments on *Danio* relationships based on molecules and morphology

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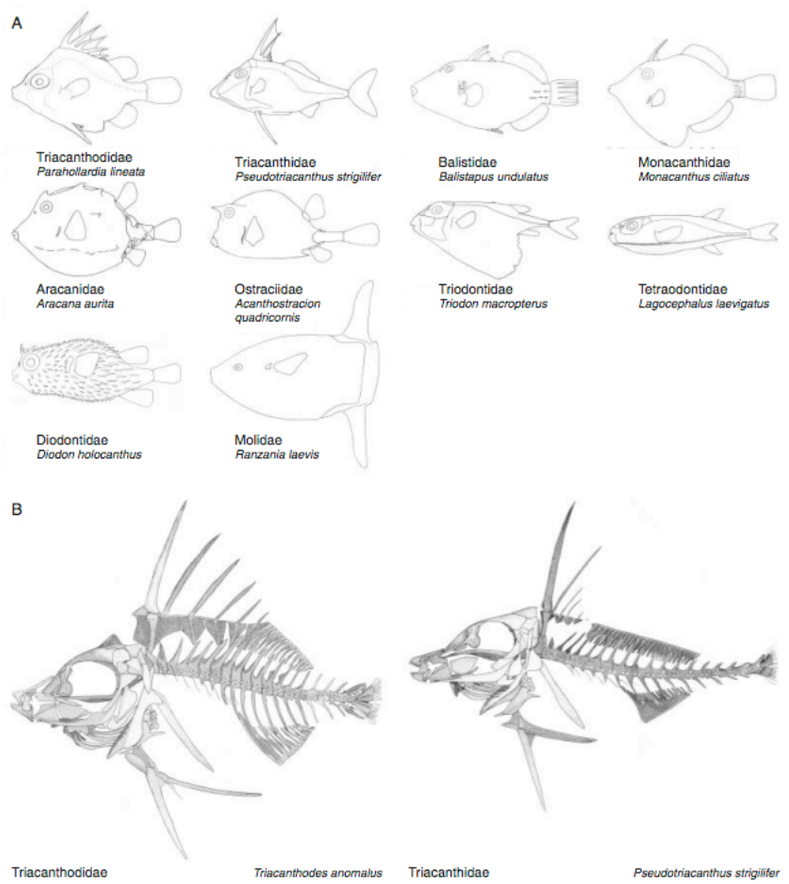


Figure 1. Illustrations of the skeletal anatomy and body shape of a generalized representative for each of the ten extant tetraodontiform families. Lateral views of (A) ten species, one from each extant family, and (B–F) of the skeletons of a representative species for each family, as follows: (B) triacanthodid *Triacanthodes anomalus* and triacanthid *Pseudotriacanthus strigilifer*; (C) balistid *Balistapus undulatus* and monacanthid *Monacanthus ciliatus*; (D) aracanid *Kentrocapros aculeatus* and ostraciid *Acanthostracion quadricornis*; (E) triodontid *Triodon macropterus* and tetraodontid *Lagocephalus laevigatus*; (F) diodontid *Diodon holocanthus* and molid *Ranzania laevis*.

water fishes native to Southern Asia. Biological
sh, *Danio rerio*, has become an important model
logy and genetics. Though several phylogenetic
sulting phylogenies conflict in detail. To examine
vel, we studied the axial skeleton for 11 species
ith DNA sequence data for five gene sequences
nio exhibits 14 characters that prove useful in
pport the monophyly of the danios included in
ades: a deep-bodied group and a slender-bodied
show that the molecular data resolves relation-
bodied subclade is determined by morphological
of the Linnean Society, 2002, **135**, 529–546.

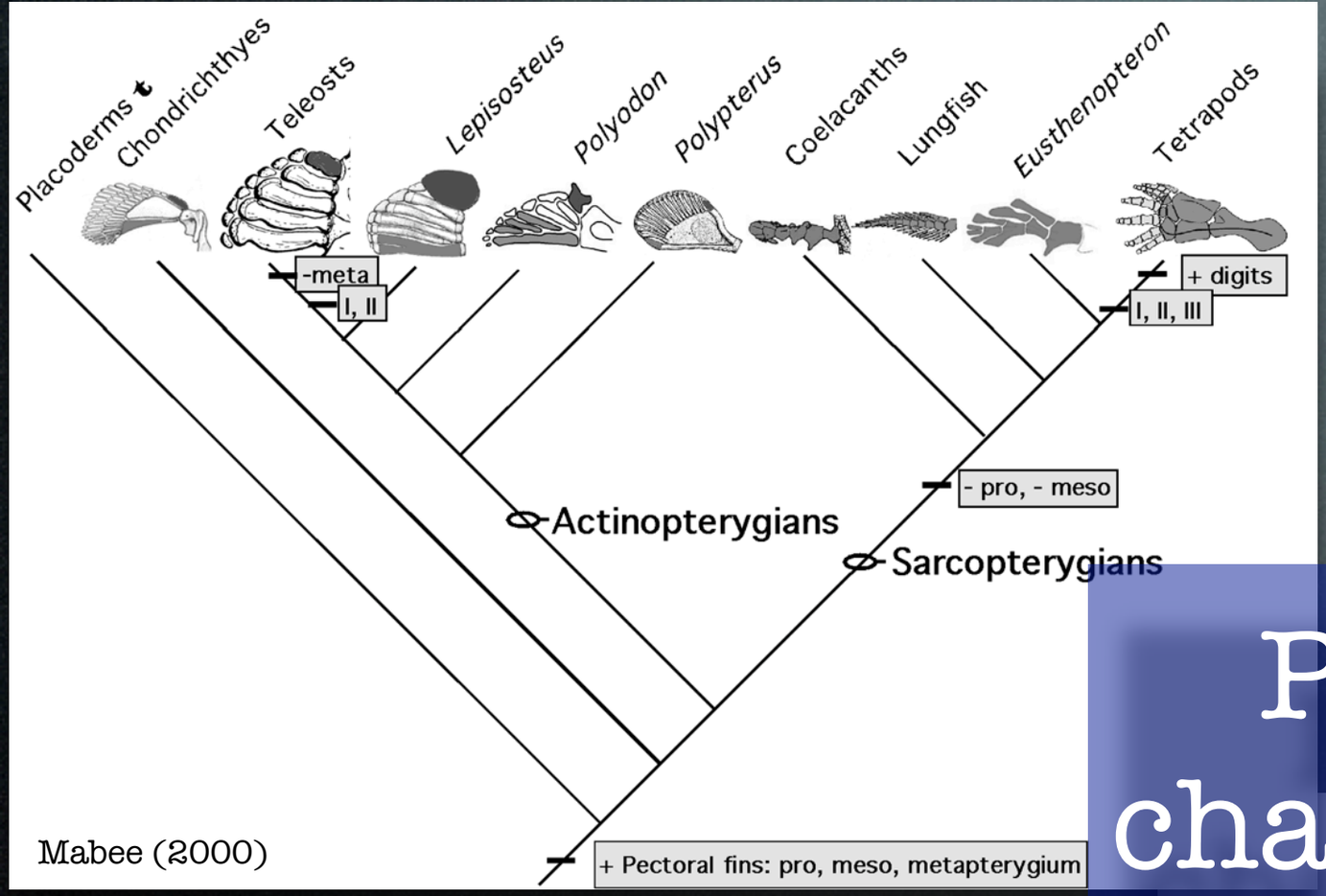
– caudal skeleton – *Danio rerio* – zebrafish –

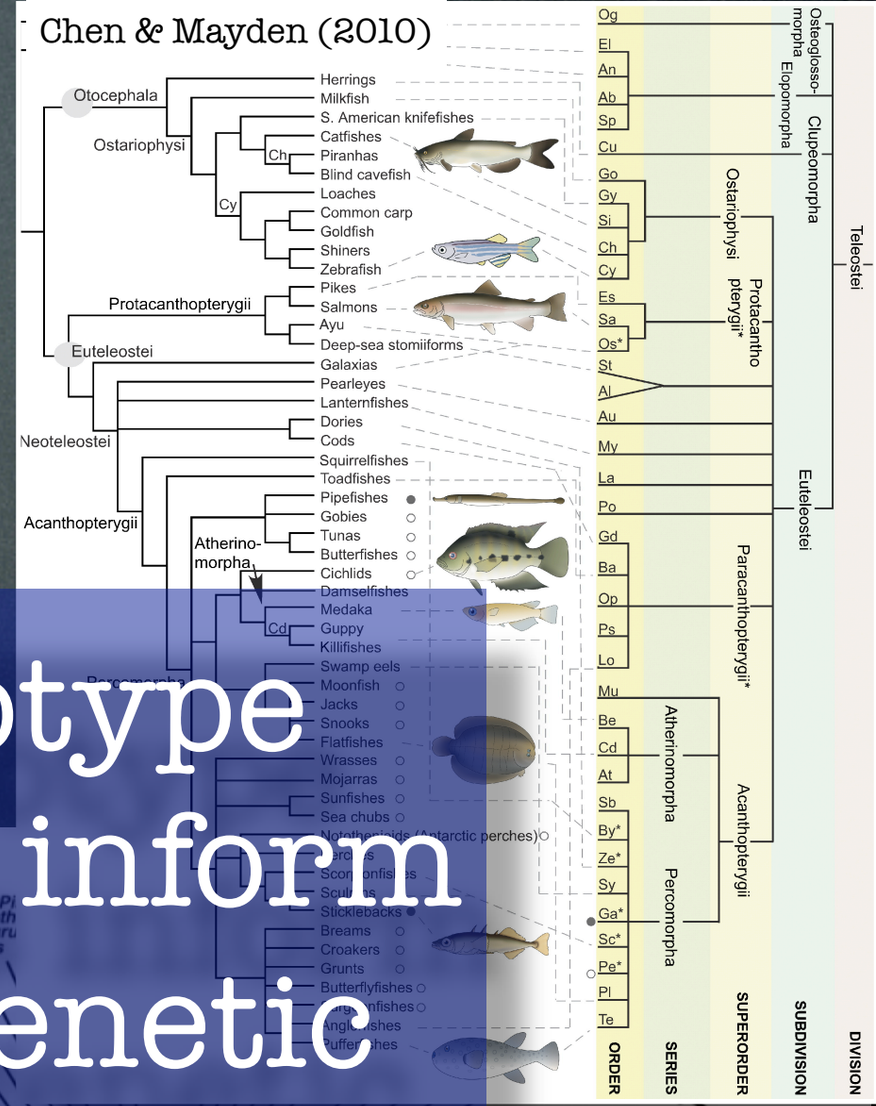
recedented opportunity to investigate patterns
chanisms of phenotypic transformation among
related vertebrate species. Such comparative
lutionary studies of danios require a species
ylogeny. While progress has been made in this
(e.g. Meyer *et al.* 1993, 1995; Zardoya *et al.*
he several available phylogenies, which rely
entirely on DNA sequence data, conflict in
nd include only a fraction of *Danio* species. In
dy we describe osteological variation found in
al skeleton of danios and assess the phyloge-
ility at the species level of these morphological
combination with molecular data, our mor-
al data provide a well-supported phylogeny for
pecies of *Danio* that we were able to obtain.
dy provides a foundation for a more compre-
phylogenetic analysis of osteological data for
pending completion of ongoing alpha taxonomic
by others (e.g. F. Fang, M. Kottelot).

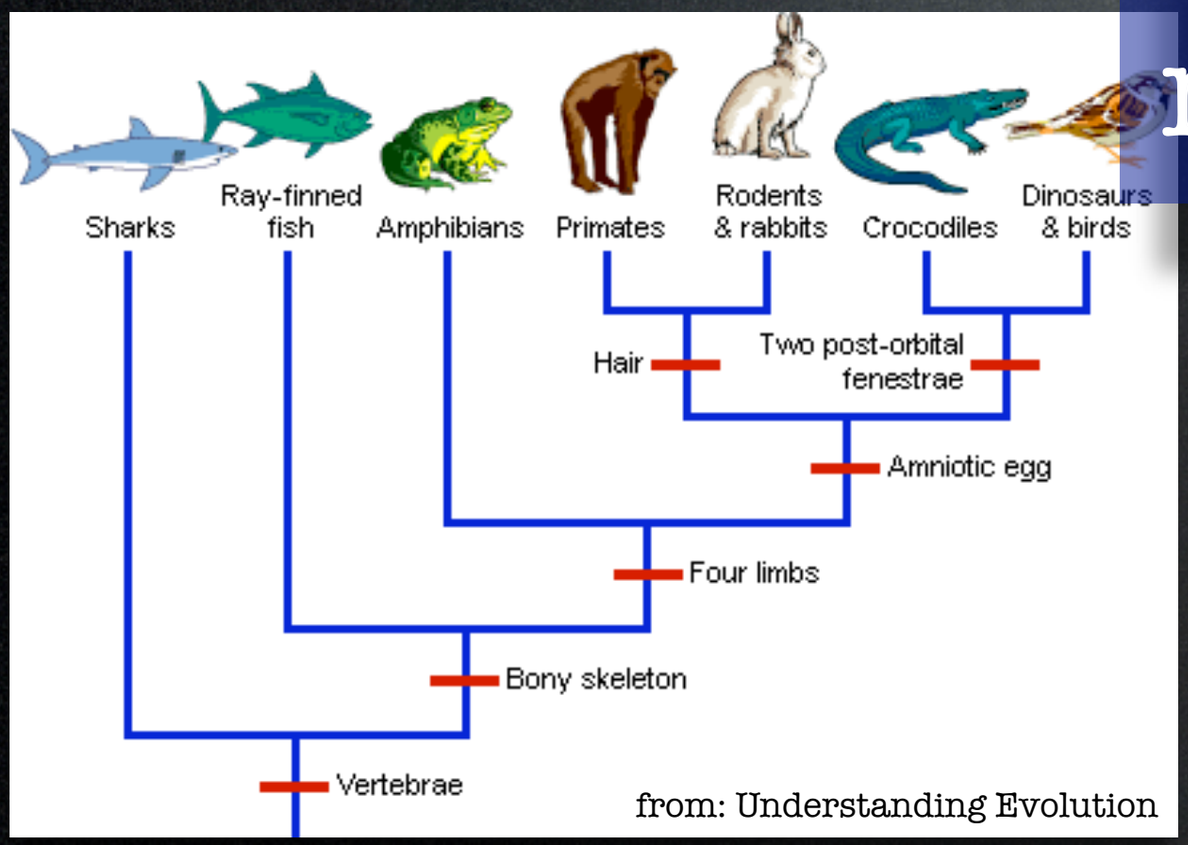
SYSTEMATIC BACKGROUND

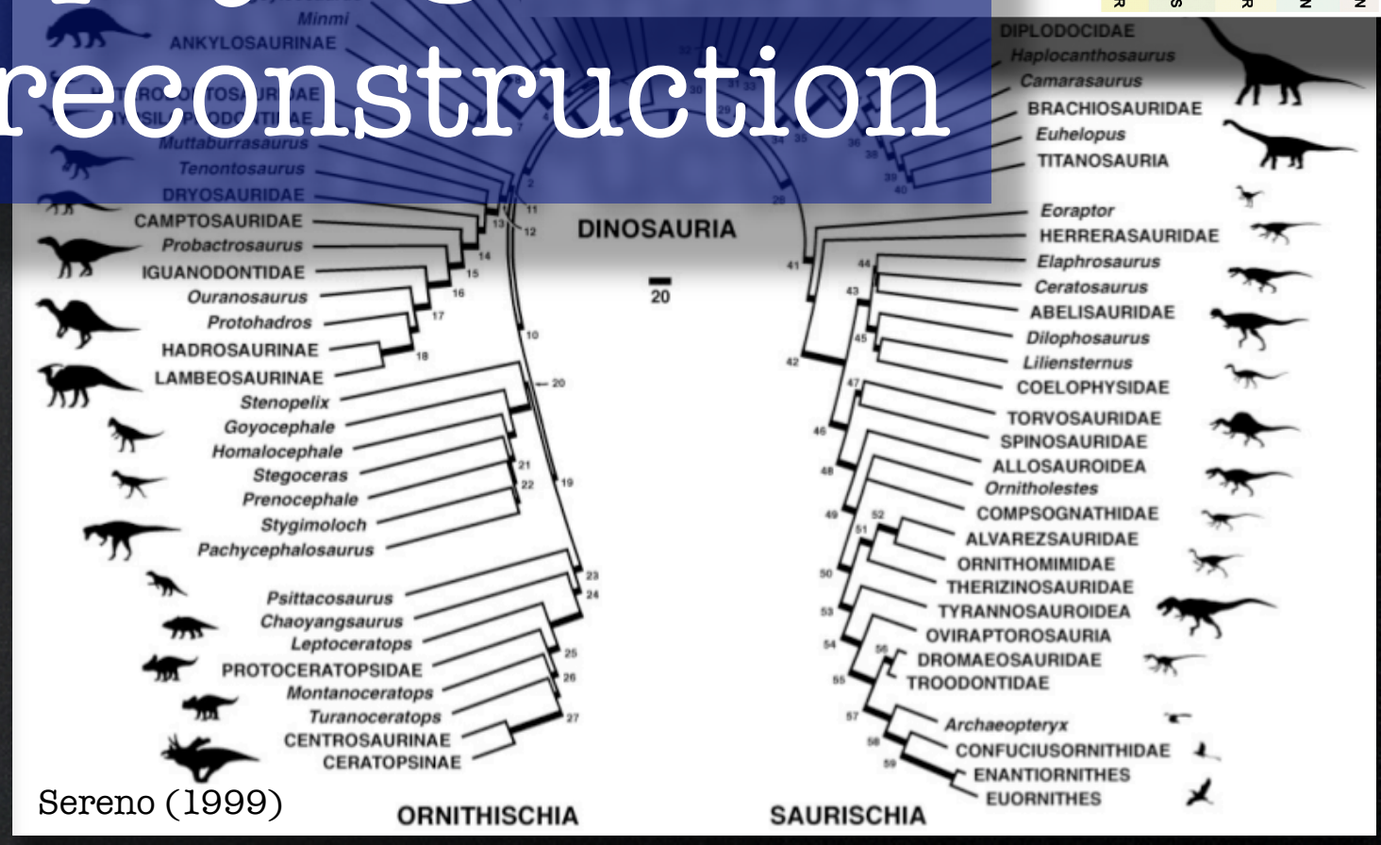
are ostariophysan fishes, which are the domi-
oup of freshwater fishes comprising approxi-

Phenotype changes inform phylogenetic reconstruction









Sereno (1999)

Phenotype observations important to many fields

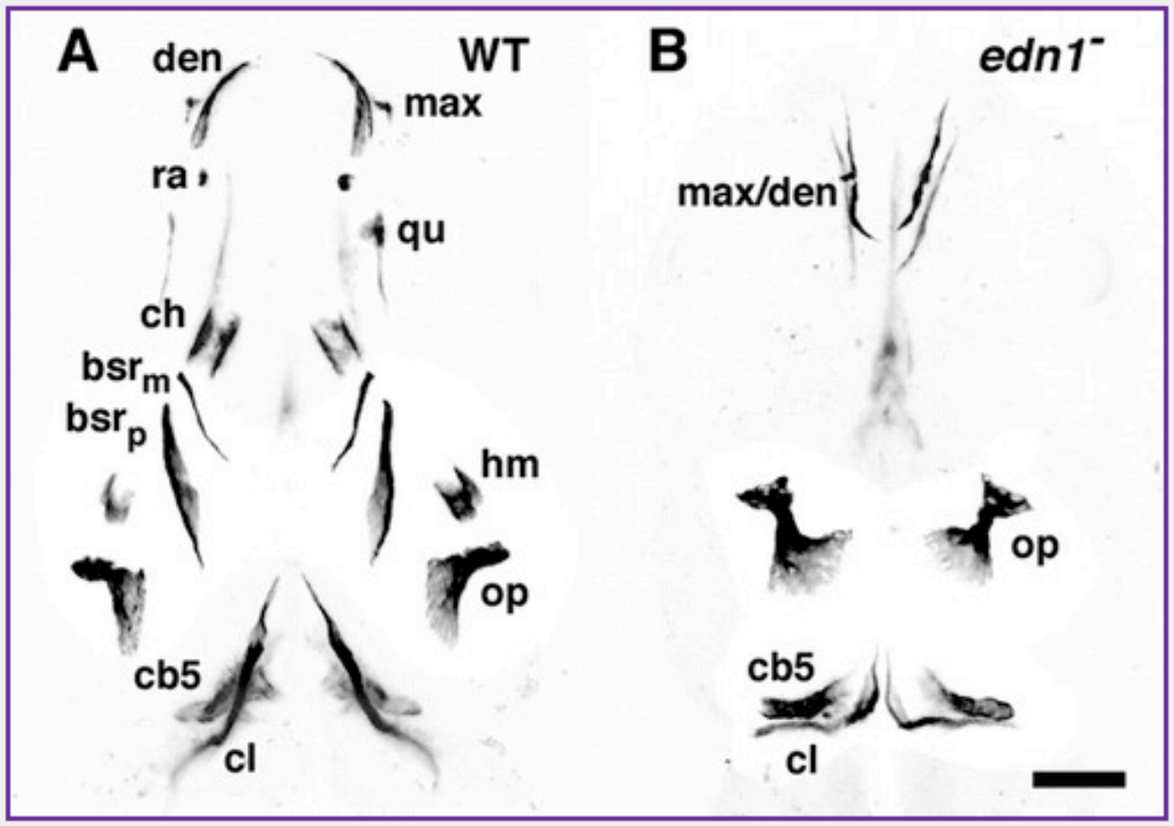
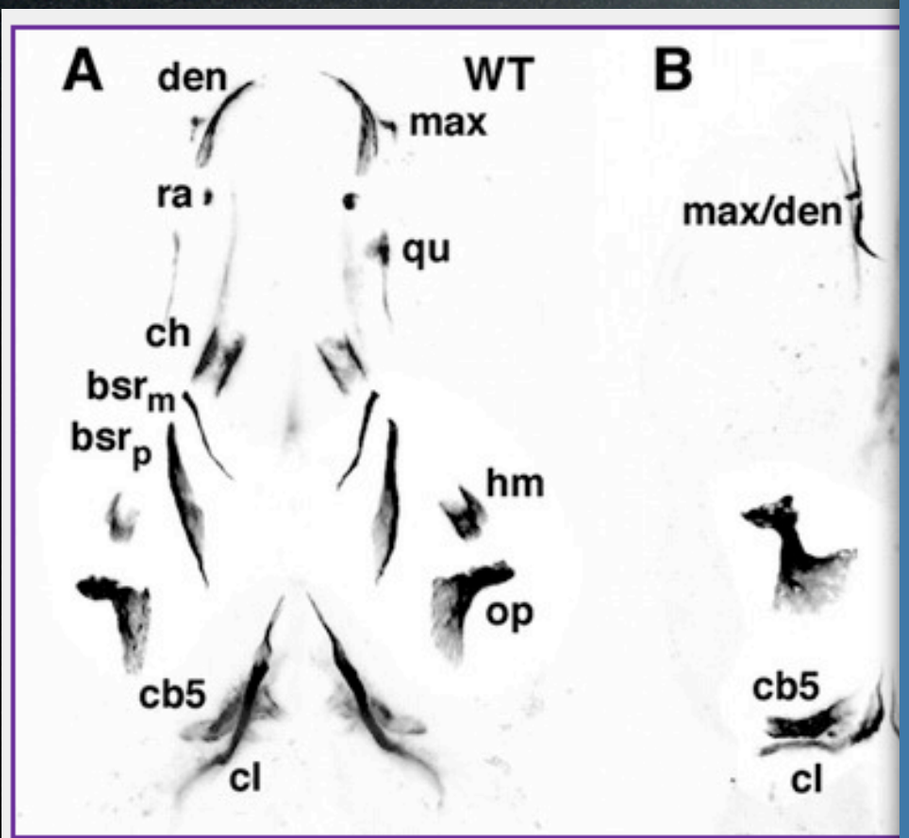


Fig. 2 Ossifications in the young wild-type (WT) zebrafish (A) and homozygous *edn1* mutant (B). Ventral views, anterior to the top, of negative images of bones fluorescently labeled with Calcein in larvae at 7-days postfertilization. Ventral bones of the pharyngeal arches are identified (Cubbage and Mabee, 1996) by their labels on the left side, and dorsal bones are labeled on the right side in both panels, all of them are present as bilateral pairs. (A) The wild type first or mandibular arch includes a dorsal and a ventral dermal bone, the maxilla (max) and dentary (den), and a dorsal and a ventral cartilage-replacement bone, the quadrate (qu) and retroarticular (ra). The second or hyoid arch includes a dorsal and two ventral dermal bones, opercle (op), and two branchiostegal rays (bsr_p and bsr_m), and dorsal and ventral cartilage replacement bones (very incompletely ossified at this stage), the hyomandibula (hm) and ceratohyal (ch). The most posterior arch includes a cartilage-replacement bone, ceratobranchial 5 (cb5). Overlaying ceratobranchial 5 is the cleithrum (cl), a long dermal bone connecting the posterior skull and the pectoral girdle. Two other craniofacial bones present at this stage lie deeper in the tissue and are not labeled, the parasphenoid and the endopterygoid. (B) Many of the anterior ossifications (in the first two arches) are missing in the *edn1* mutant. Ceratobranchial 5 and the cleithrum are present, shortened and somewhat malformed. In the mandibular arch dermal bones (max/den) are present but severely malformed, an example of the `wicket' phenotype discussed in the text (see also Fig. 3). In the hyoid arch the opercle is present and its joint region (upper part of the bone) is markedly expanded, a mild example of the `opercle-gain' phenotype described in the text and other Figures. Scale bar: 100 μm.

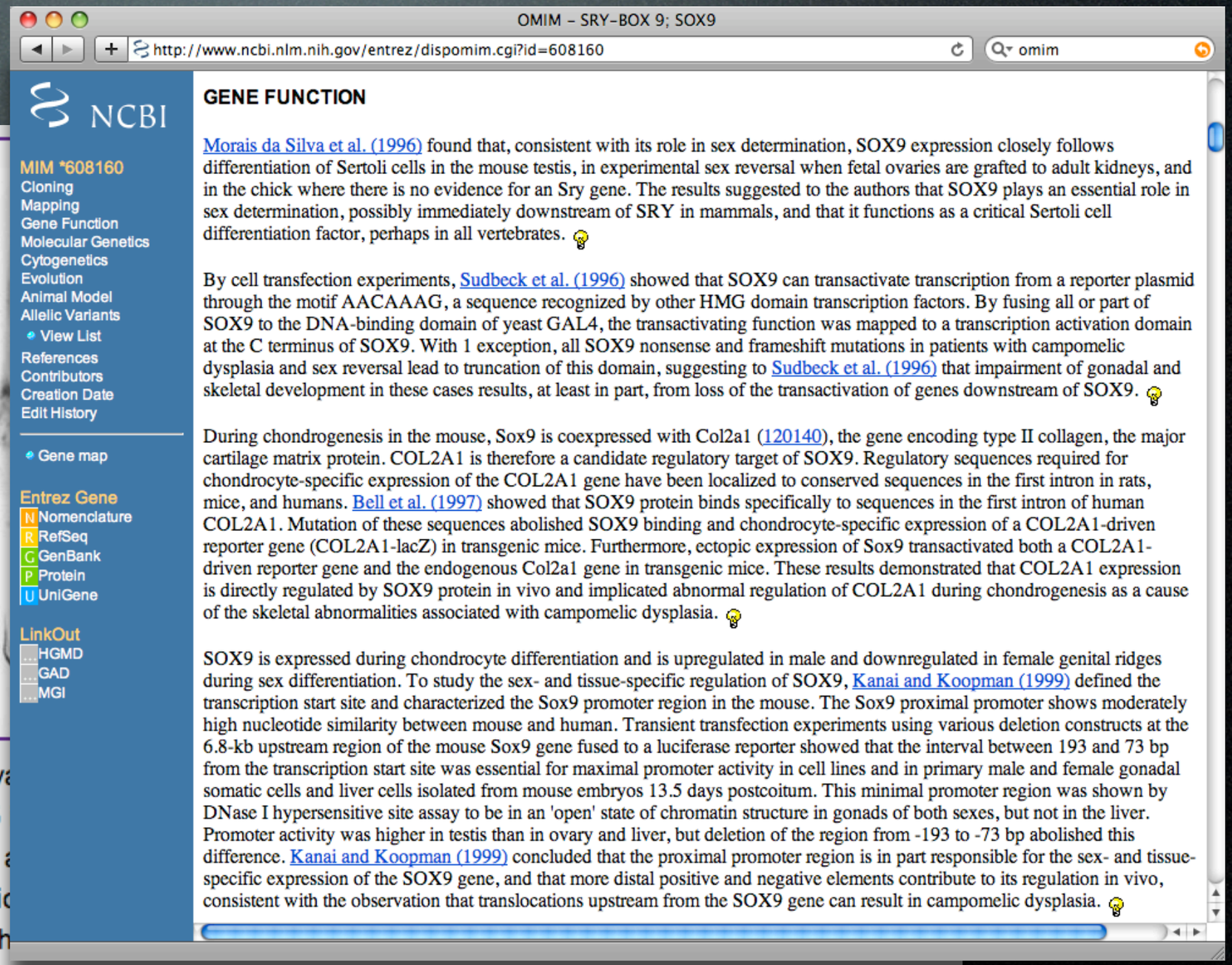
Kimmel et al, 2003

Phenotype observations important to many fields



this stage), the hyomandibula (hm) and ceratohya
ceratobranchial 5 (cb5). Overlaying ceratobranchial 5
pectoral girdle. Two other craniofacial bones present a
the endopterygoid. (B) Many of the anterior ossificatic
and the cleithrum are present, shortened and somewh
severely malformed, an example of the `wicket' phenotype discussed in the text (see also Fig. 3). In the hyoid arch the opercle is present and its joint region (upper part of the bone) is markedly expanded, a mild example of the `opercle-gain' phenotype described in the text and other Figures. Scale bar: 100 μm.

Kimmel et al, 2003



As complex, free text phenotypes are resistant to computing

5. Aortic canal on compound Weberian centrum: ventrally open groove [0]; bony tube [1] (Lundberg et al., 1991b). Within the family a uniquely derived and unreversed synapomorphy of Pimelodidae exclusive of *Steindachneridion* and *Phractocephalus-Leiarius* group, CCI = 1.
6. Position of right cardinal vein foramen (or open trough): vertebra 5 [0]; vertebra 6 [1] (Nass, 1991). Within the family a uniquely derived and unreversed synapomorphy of Pimelodidae exclusive of *Steindachneridion* and *Phractocephalus-Leiarius* group, CCI = 1.
7. Ethmoid plate form: rounded [0]; quadrangular [1] (Lundberg et al., 1991b). Within Pimelodidae a uniquely derived and unreversed synapomorphy of *Pimelodus* group plus *Calophysus* group, CCI = 1.
8. Mesethmoid cornua deflection: none [0]; abruptly deflected ventrally [1] (Lundberg et al., 1991b). Within Pimelodidae a uniquely derived and unreversed synapomorphy of *Pimelodus* group plus *Calophysus* group, CCI = 1.
9. Coronoid process: shallow to moderately inclined [0]; steeply inclined, tall, and a little recurved [1] (Lundberg et al., 1991b). Within Pimelodidae a uniquely derived and unreversed synapomorphy of *Pimelodus* group plus *Calophysus* group, CCI = 1.

47. *Shape of posterior dorsomedian fontanelle.* An elongate-rectangular posterior fontanelle of most catfishes appears to be the plesiomorphic condition (see also Tilak 1963, 1964, 1965a; Lundberg 1982; Arratia 1987; Grande 1987). An irregularly-shaped or rounded

rowing posteriorly [2] (Buitrago, pers. comm.). Within Pimelodidae uniquely derived basally in *Brachyplatystoma* but reversed in *B. tigrinum*, CCI = 0.5; state 2 is independently derived in *Steindachneridion* and *Sorubim* group.

17. Mandibular nerve canal in articular: absent [0]; present [1] (Nass, 1991). Highly homoplastic within Pimelodidae; uniquely derived basally in *Brachyplatystoma* but reversed in *B. tigrinum*, CCI = 0.2.
18. Urohyal vertical keel and posterior process: smooth sided [0]; with lateral ridges [1]. Within Pimelodidae a uniquely derived and unreversed synapomorphy of *Brachyplatystoma* exclusive of *B. vaillantii*, CCI = 1.
19. Orbital notch: present [0]; absent, eye not bounded by lateral ethmoid and sphenotic projections [1]. Within Pimelodidae a uniquely derived and unreversed synapomorphy of *Brachyplatystoma* exclusive of *B. vaillantii*, CCI = 1.
20. Ectopterygoid: present [0]; absent [1]. Within Brachyplatystomatini an unambiguously derived and unreversed synapomorphy of *Brachyplatystoma* exclusive of *B. vaillantii*, but independently derived in some Pimelodidae and other catfishes, CCI = 0.33.

State 0 = frontal broad anteriorly and moderately narrow posteriorly, anterior space reduced (adults) and arms moderately wide; 1 = frontal moderately broad posteriorly, anterior space moderately enlarged; 2 = frontal broad posteriorly, anterior arms narrow, space enlarged.

49. *Laminar bone over the anterior vertebrae.* The laminar bone is usually continuous medianly in ariids (except in *Galeichthys* and *Ancharius*) and is more extensive in larger individuals, an ontogenetic change evidenced in most taxa. However, the excavation of the laminar bone posteromedially and the overlapping of the transverse process bases laterally is variable. I consider that a minimal cover over the aortic groove is plesiomorphic in ariids and interpret a 'minimal cover' as exposed transverse process bases and a deep median excavation on the ventral surface. The laminar bone in ariids extends over four to eight vertebra centra. Some ariids possess apomorphic modifications in the laminar shelf, such as depressions (e.g., *Guiritinga barbus, Cinetodus froggatti*) or median single keel (e.g., high and acute in *Batrachocephalus, Nemapteryx armiger*) or double keel (e.g., *Bagre marinus*).

(Lundberg and Akama 2005)

Finding similar information in free-text is difficult

“lacrymal bone…flat”	Mayden 1989
“lacrimal…small, flat”	Grande and Poyato-Ariza 1999
“lacrimal…triangular”	Royero 1999
“first infraorbital (lachrimal) shape…flattened”	Kailola 2004
“fourth infraorbital…anterior and posterior margins…in parallel”	Zanata and Vari 2005

Meaning of words depends on context

- Burrowing insectivorous mammals in the family Talpidae
- A spy buried secretly within an organization or country
- The SI unit used in chemistry for the amount of a substance
- A small, sometimes raised area of skin, usually with darker pigment
- A Mexican sauce made from chili peppers and other spices, including chocolate
- A massive structure, usually of stone, used as a pier, jetty, or breakwater between places separated by water

Meaning of words depends on context

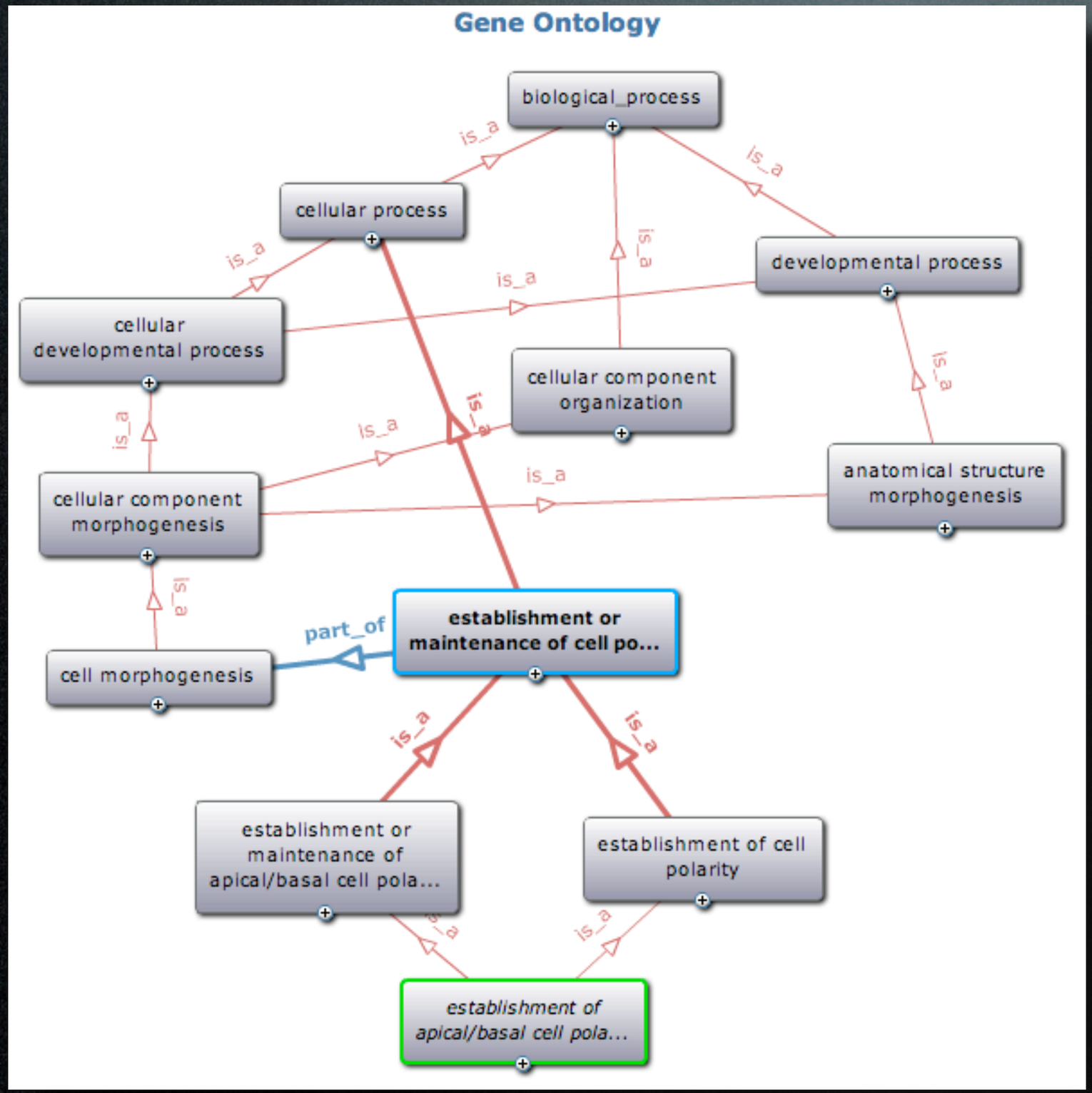
Mole:

- Burrowing insectivorous mammals in the family Talpidae
- A spy buried secretly within an organization or country
- The SI unit used in chemistry for the amount of a substance
- A small, sometimes raised area of skin, usually with darker pigment
- A Mexican sauce made from chili peppers and other spices, including chocolate
- A massive structure, usually of stone, used as a pier, jetty, or breakwater between places separated by water

What is an ontology?

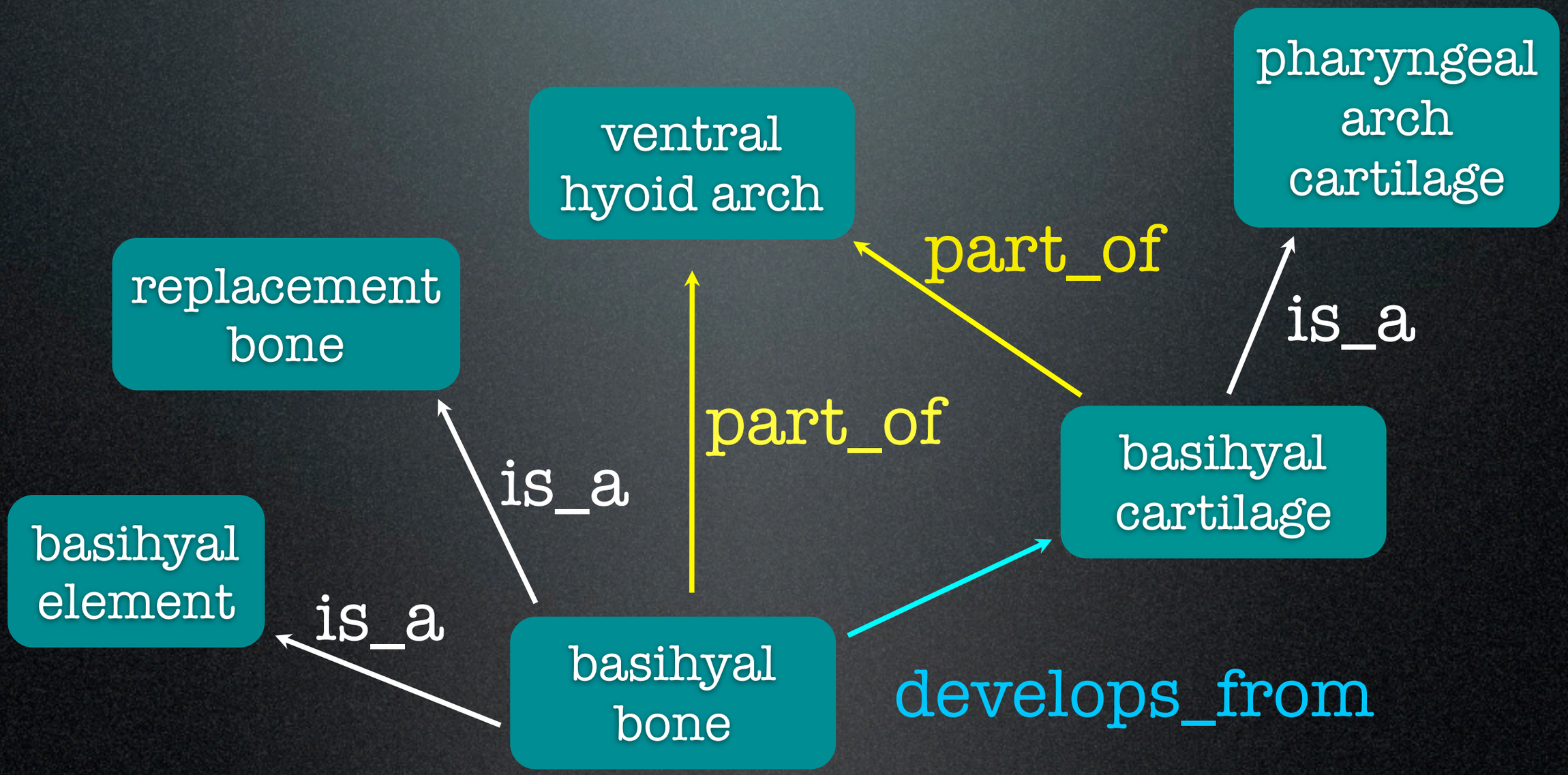
- An ontology is a type of vocabulary with well-defined terms and the logical relationships that hold between them.
- An ontology represents the knowledge about its subject domain.

Ontologies support reasoning

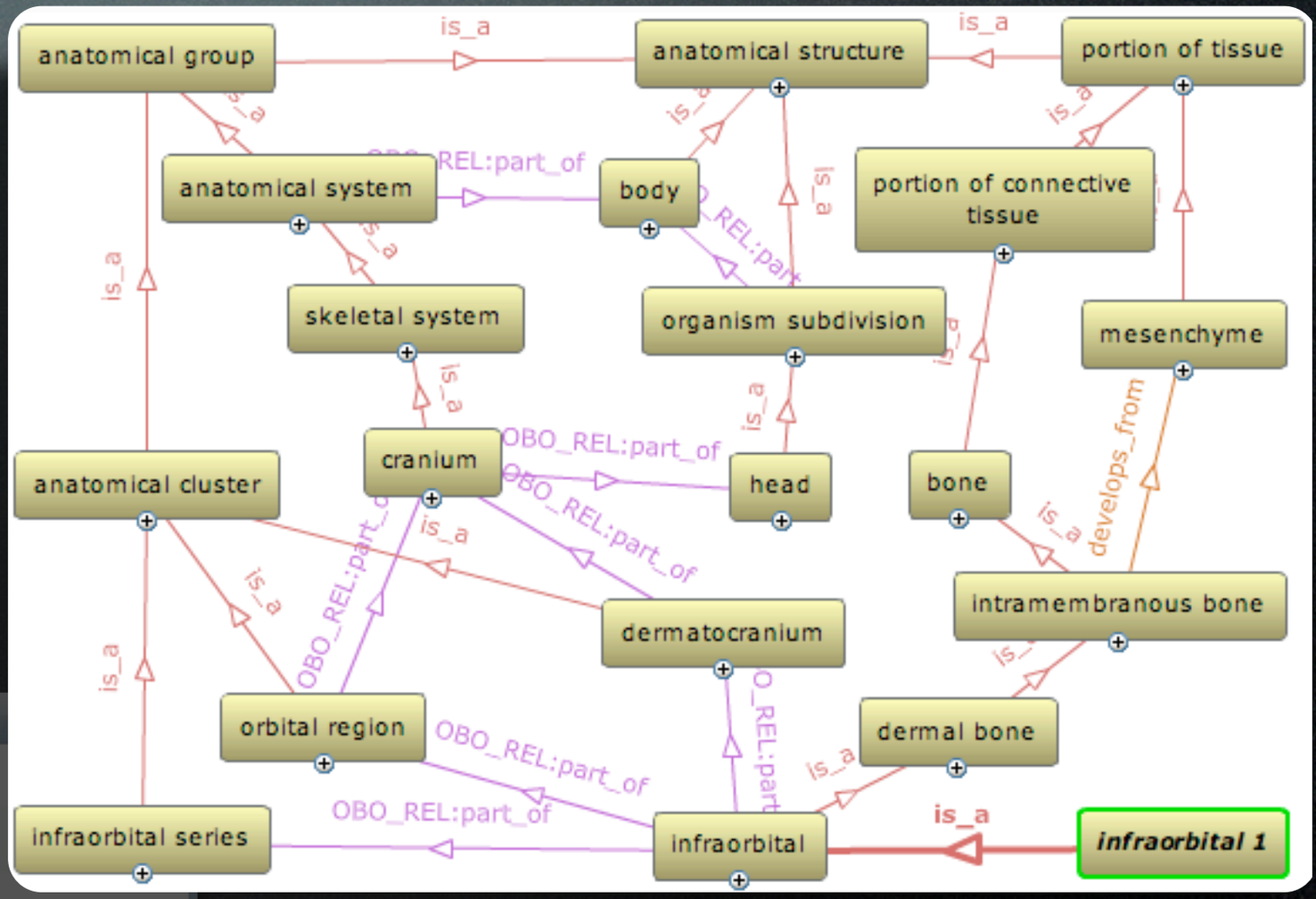


- Relationships ("assertions") induce a hierarchical structure.
- Ontologies can be processed by machines to make inferences.

The same principles apply to anatomy



The same principles apply to anatomy



Details for infraorbital 1 [Teleost Anatomy Ontology]

ID	TAO:0000223
Name	infraorbital 1
Children	0
Definition	Infraorbital that is the first (anteriormost) bone of the infraorbital series.
xref_definition	ZFIN:curator
RELATED SYNONYM	lachrymal bone
RELATED SYNONYM	lacrimal bone
RELATED SYNONYM	lacrymal bone

Integrating across studies?

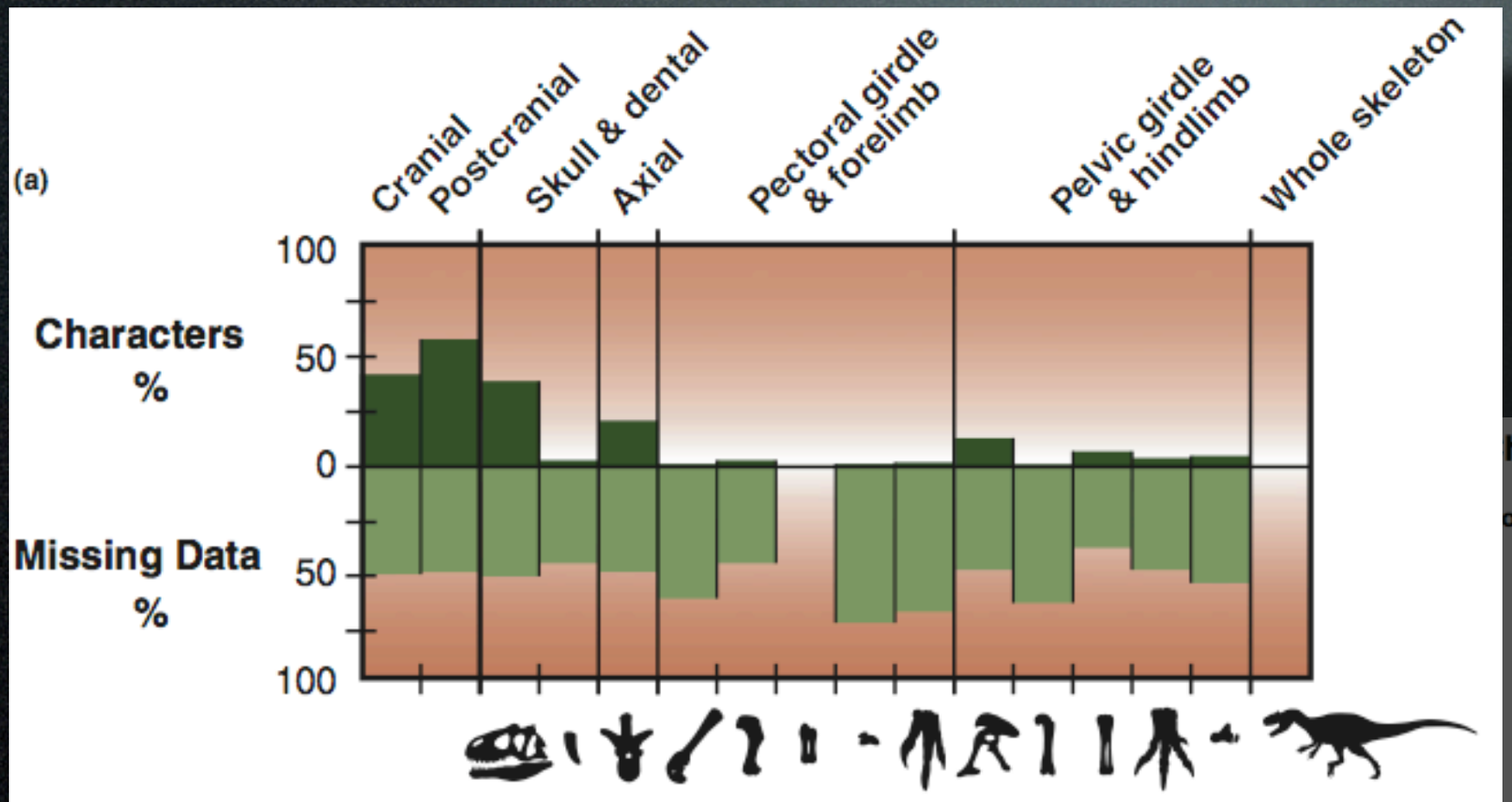
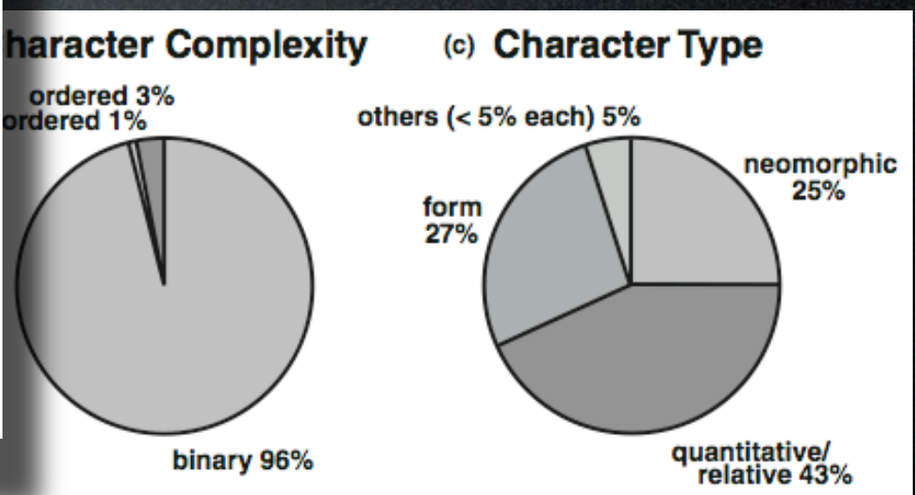
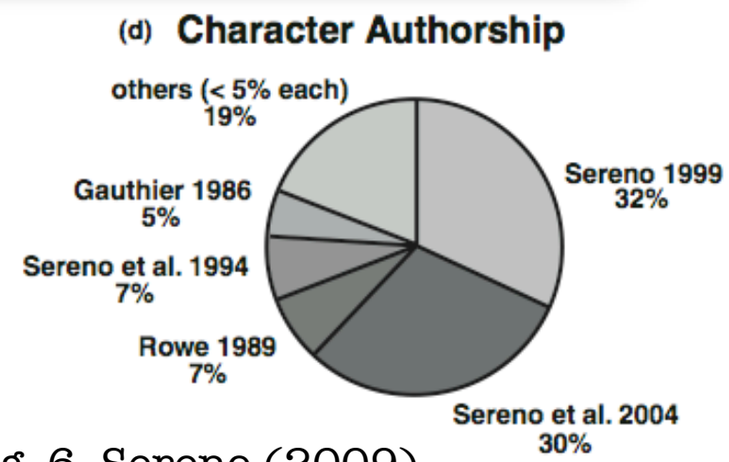


Fig. 7, Sereno (2009)





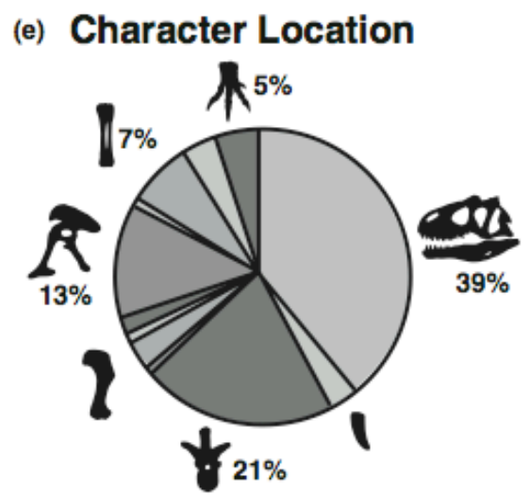


Fig. 6, Sereno (2009)

Computing example: Search by Similarity

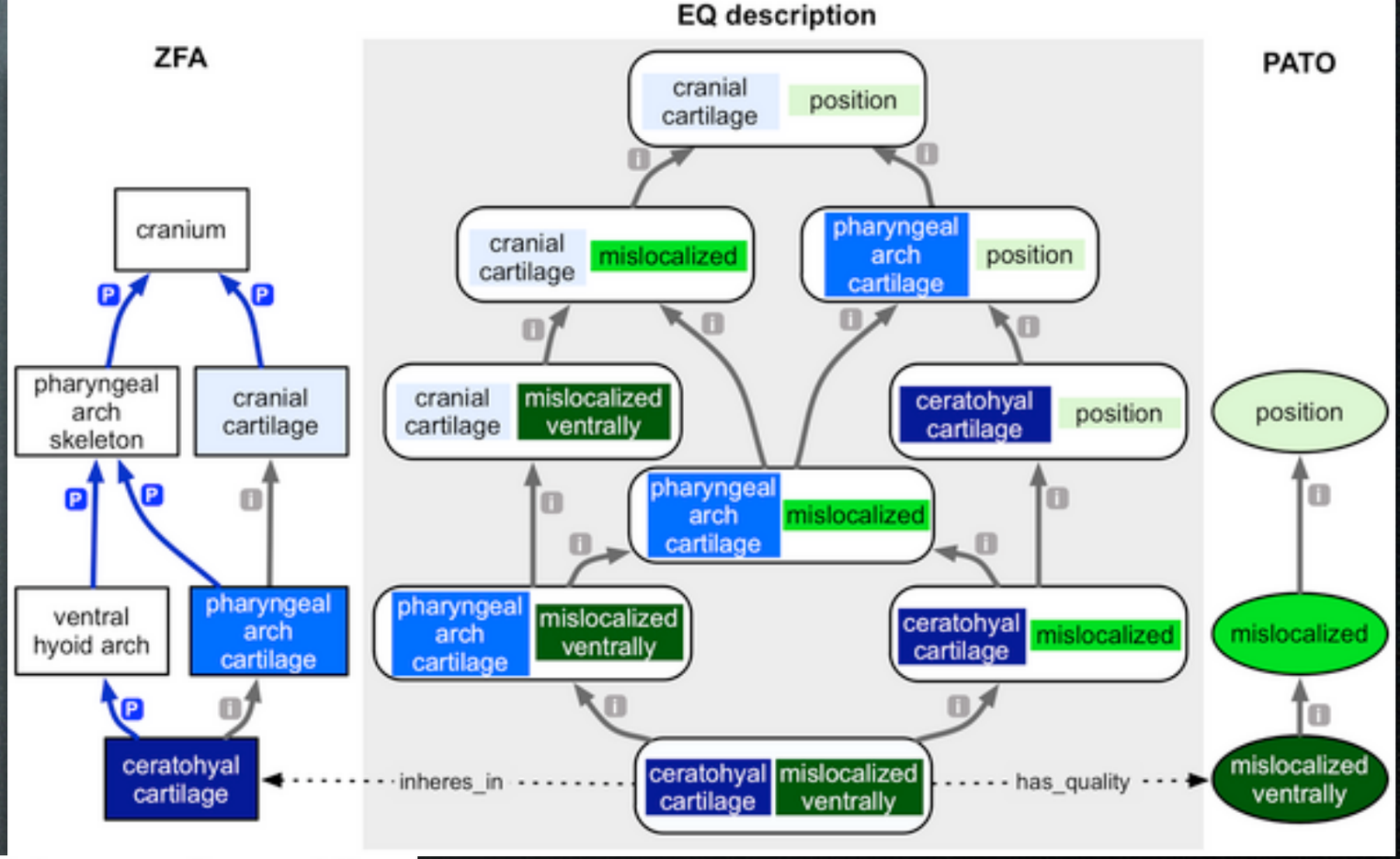


Fig. 3, Washington et al (2009)

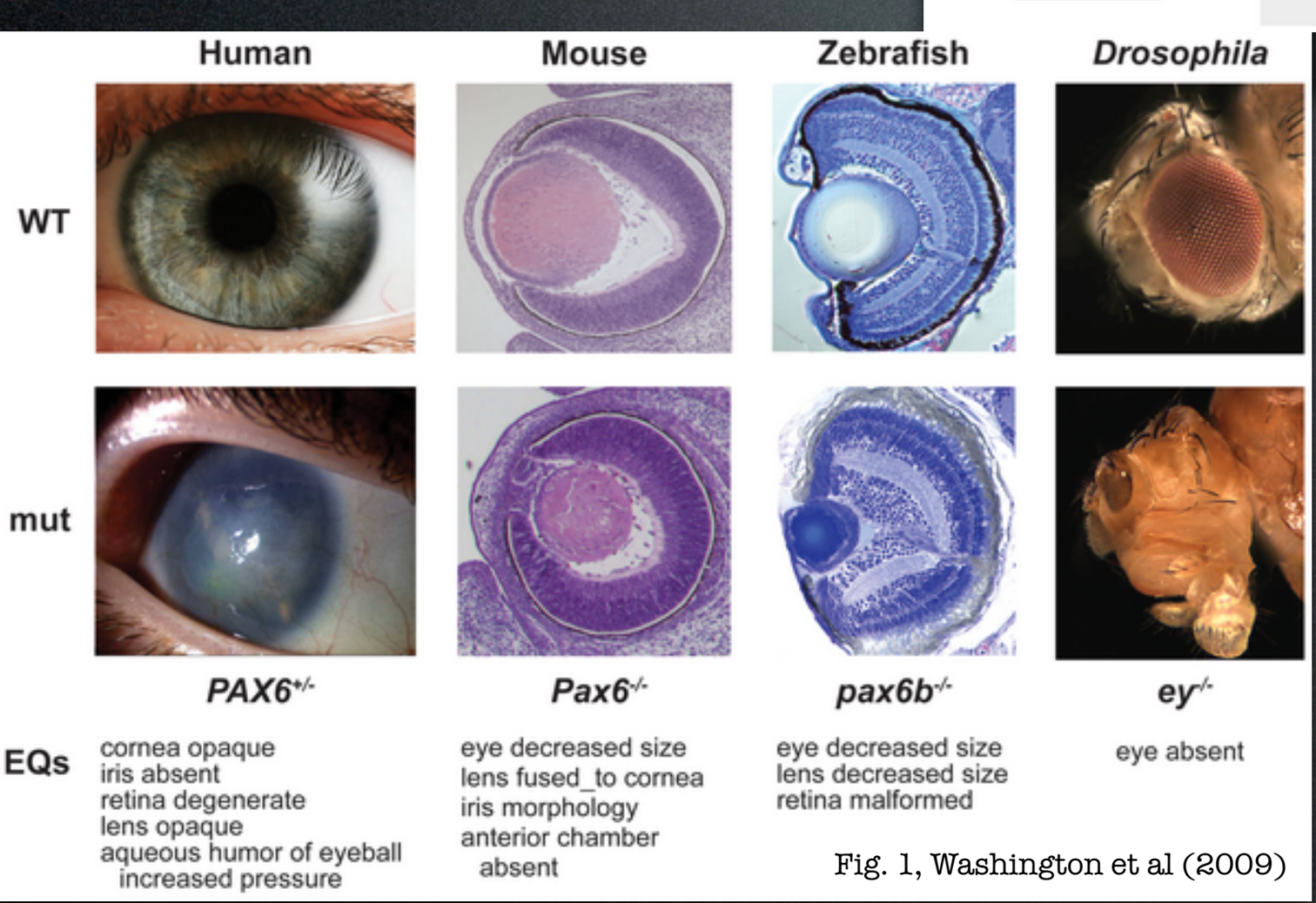
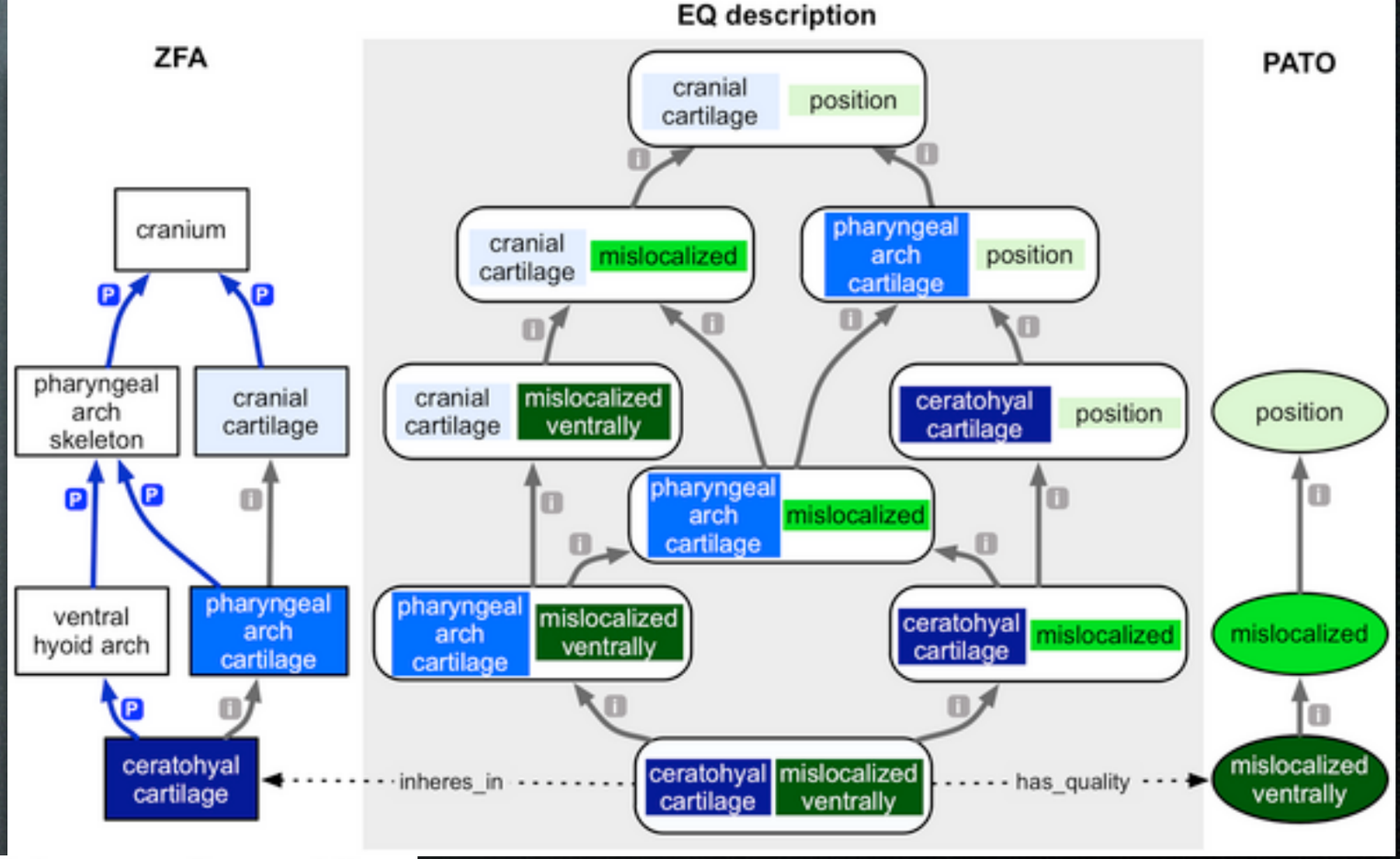


Fig. 1, Washington et al (2009)

Computing example: Search by Similarity



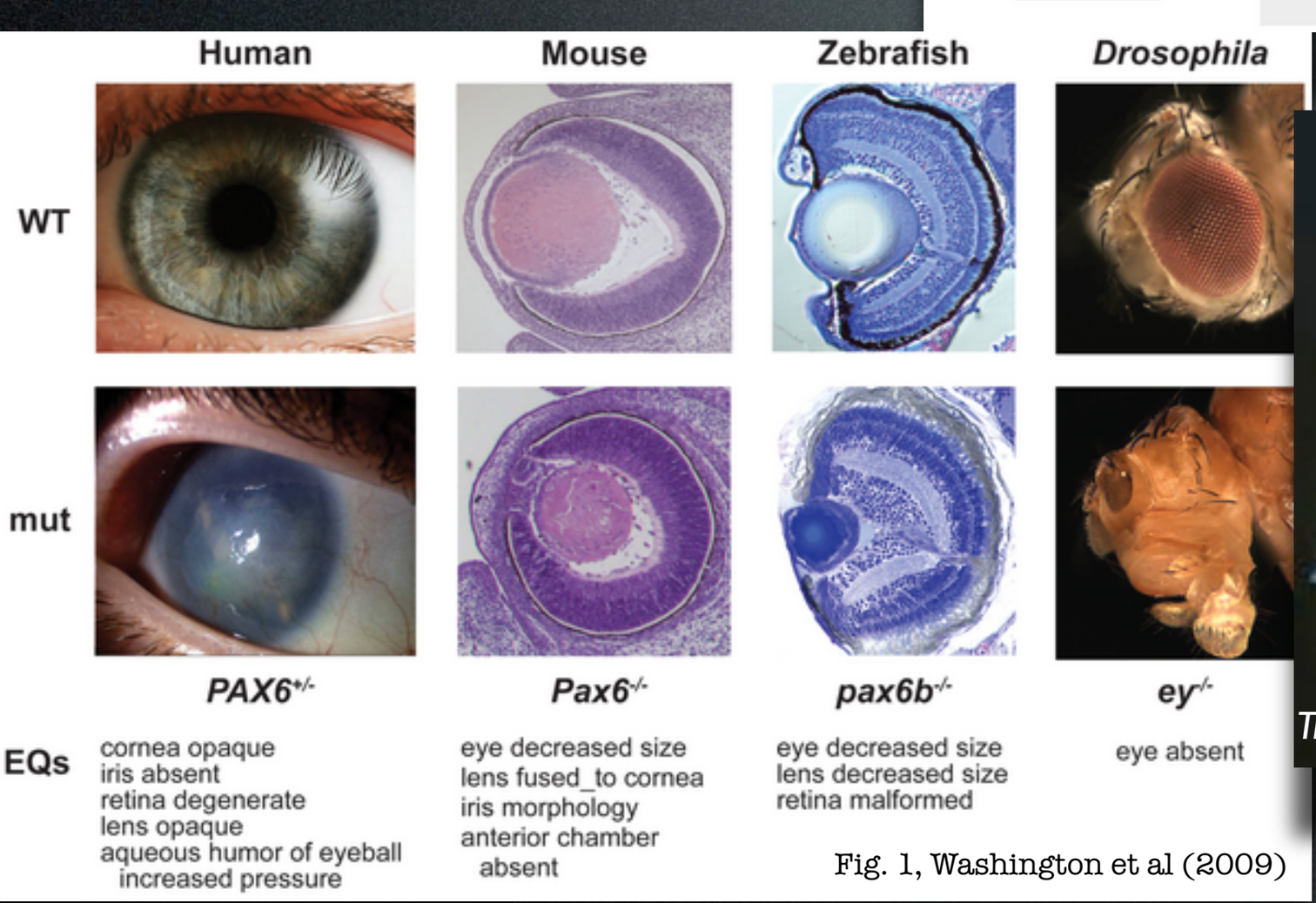


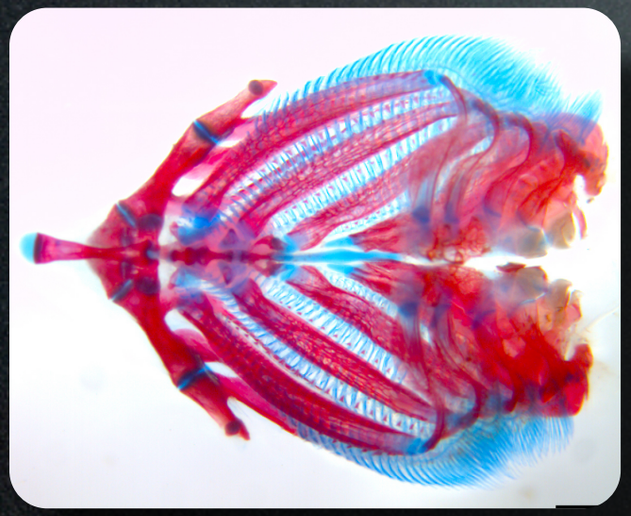
Fig. 1, Washington et al (2009)

Fig. 3, Washington et al (2009)

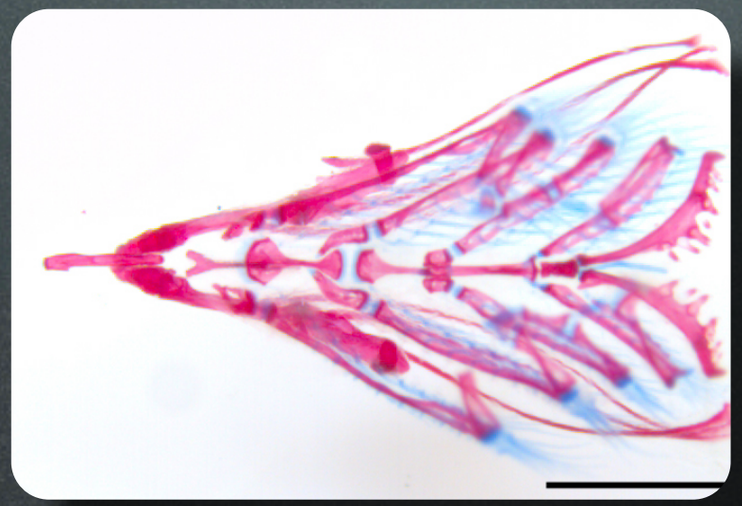
Trogloglanis pattersoni - a blind catfish

http://tolweb.org/Trogloglanis/69910

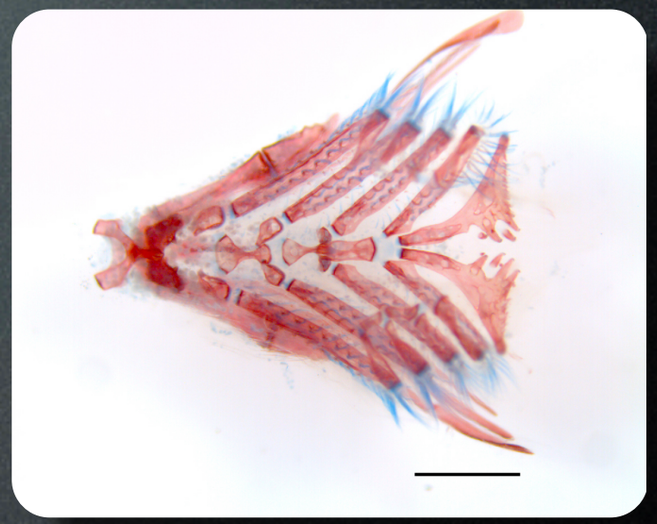
Computing over comparative morphology?



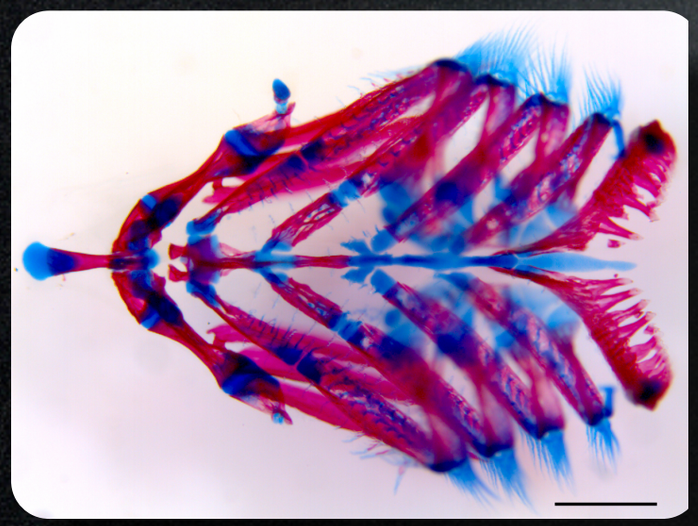
Cyprinus carpio



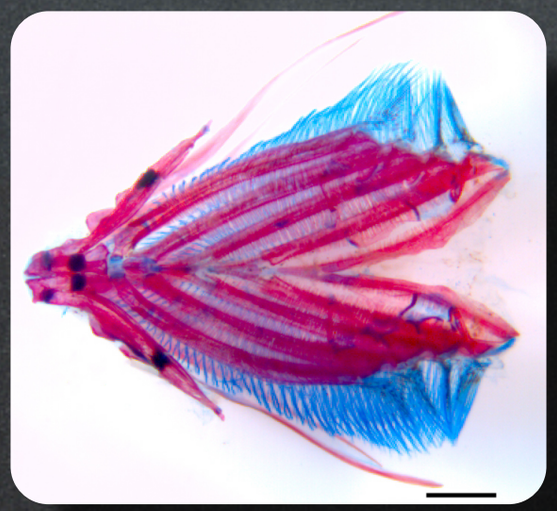
Pangio anguillaris



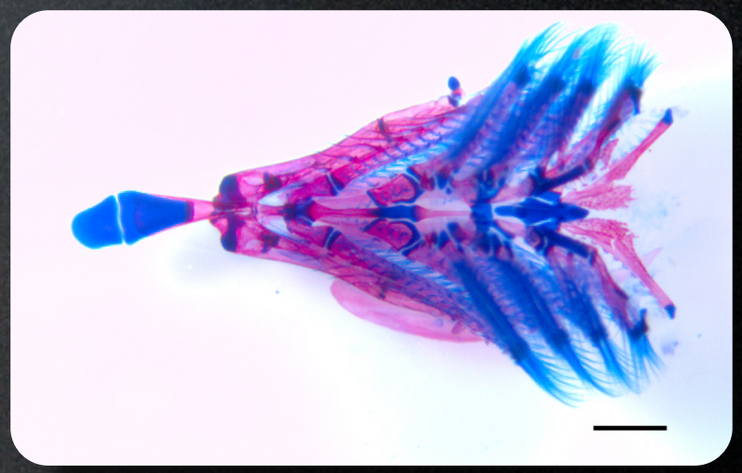
Nemacheilus fasciatus



Catostomus commersoni



Gyrinocheilus aymonieri



Phenacogrammus interruptus

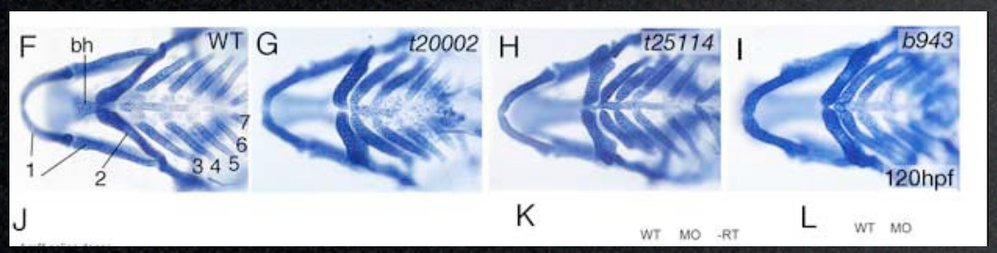
Knowledge mining & hypothesis generation

Model Organism

Mutagenesis

Mutant or missing protein at specific developmental stage

Phenotype change(s) to wildtype



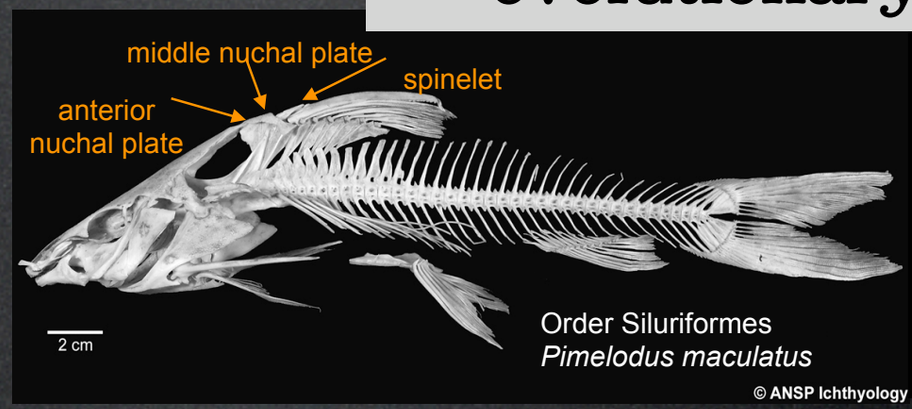
Laue et al (2008)

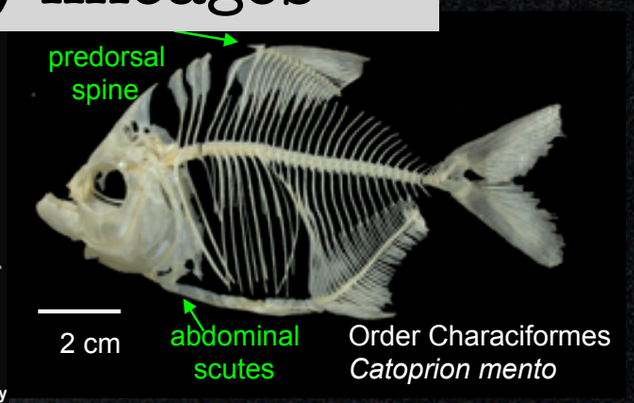
Non-model organisms

Mutation, selection, drift, gene flow

Altered expression or function of protein

Phenotype changes between evolutionary lineages

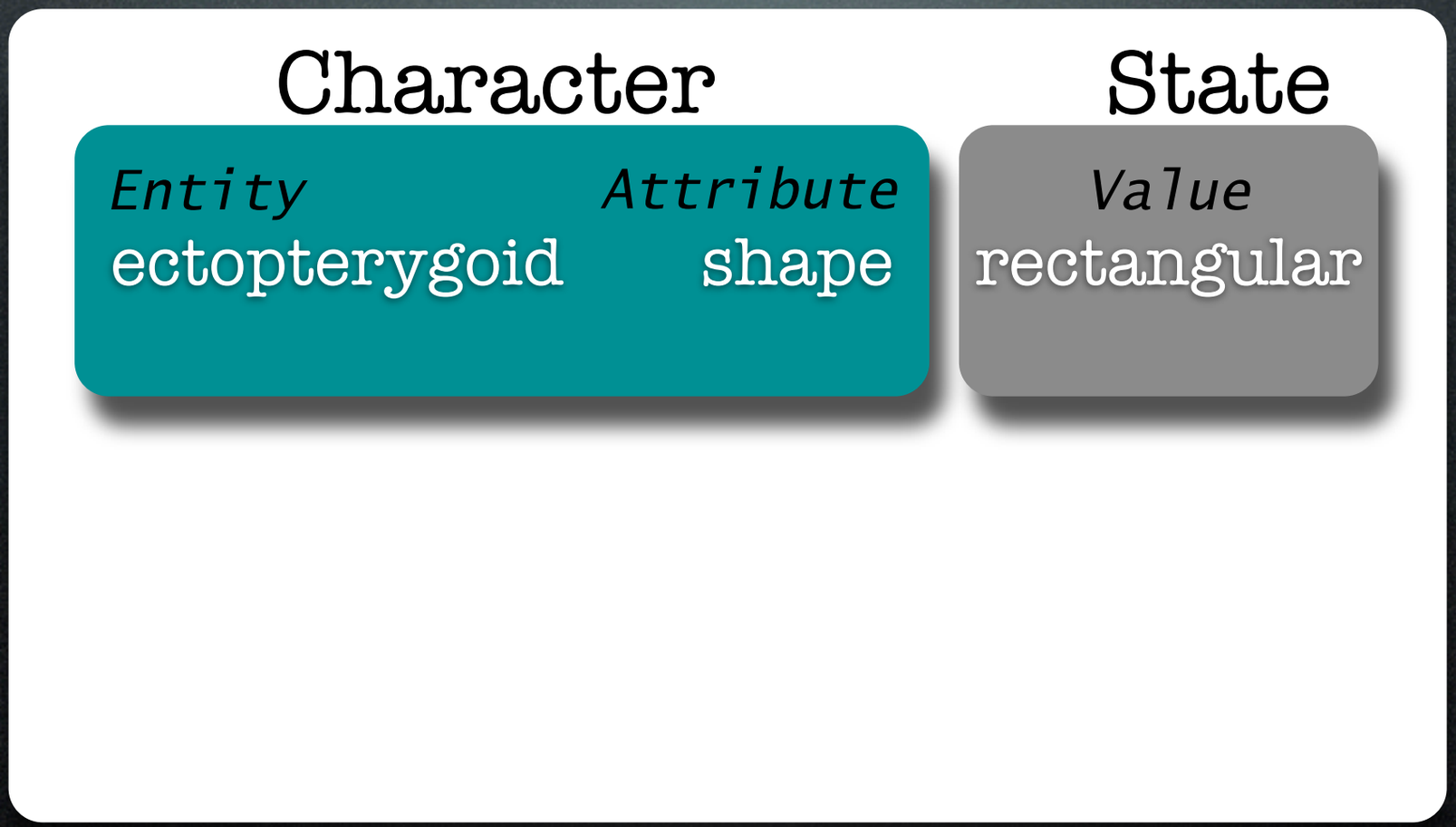




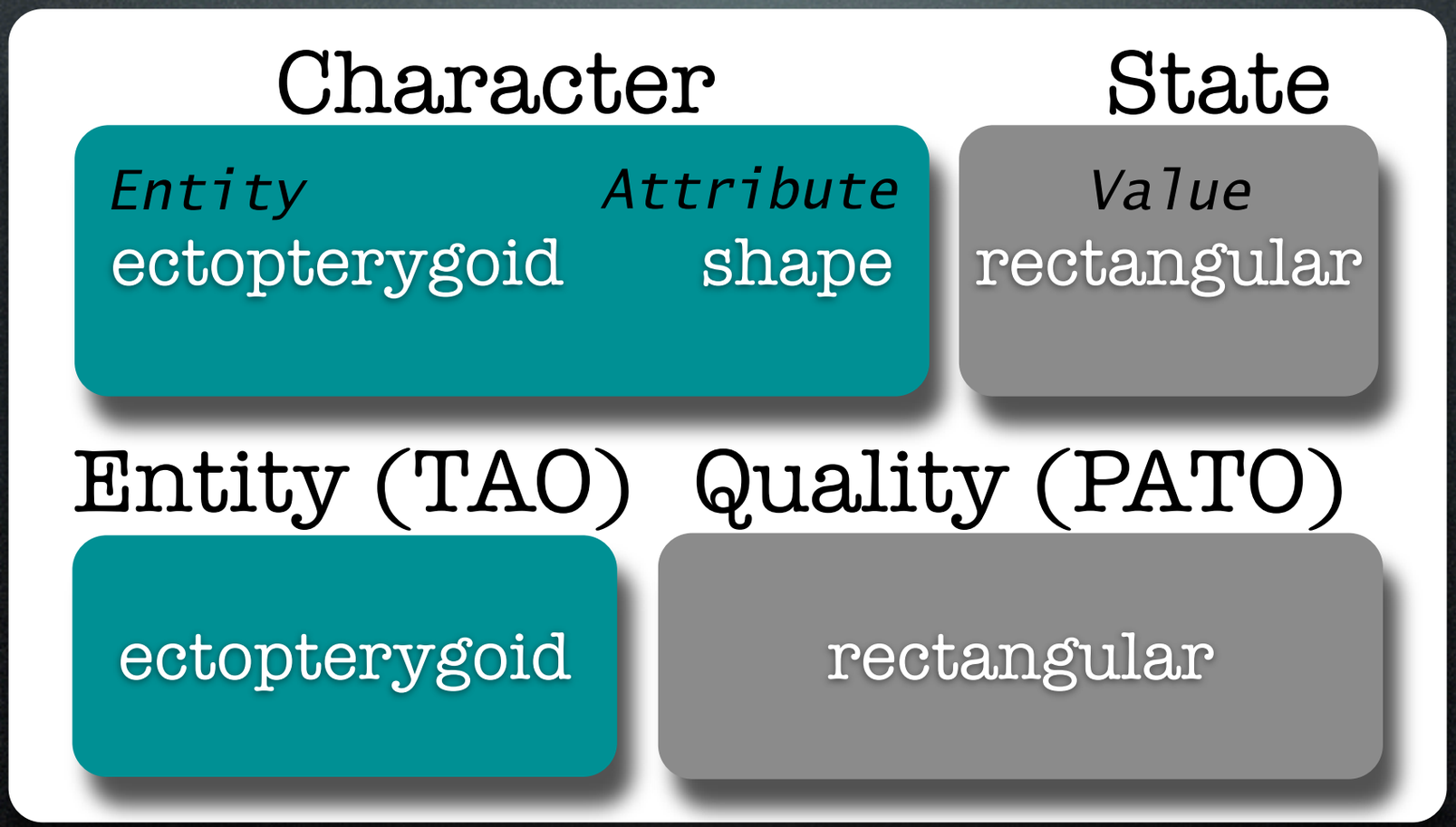
Phenoscape

- Collaboration between P. Mabee (PI, U. South Dakota), M. Westerfield (ZFIN), and Todd Vision (UNC, NESCent)
- Aim: Foster devo-evo synthesis by
  - Prototyping a database of curated, machine-interpretable evolutionary phenotypes.
  - Integrating these with mutant phenotypes from model organisms.
  - Enabling data-mining and discovery for candidate genes of evolutionary phenotype transitions.
- Informatics for the project is developed and hosted at NESCent

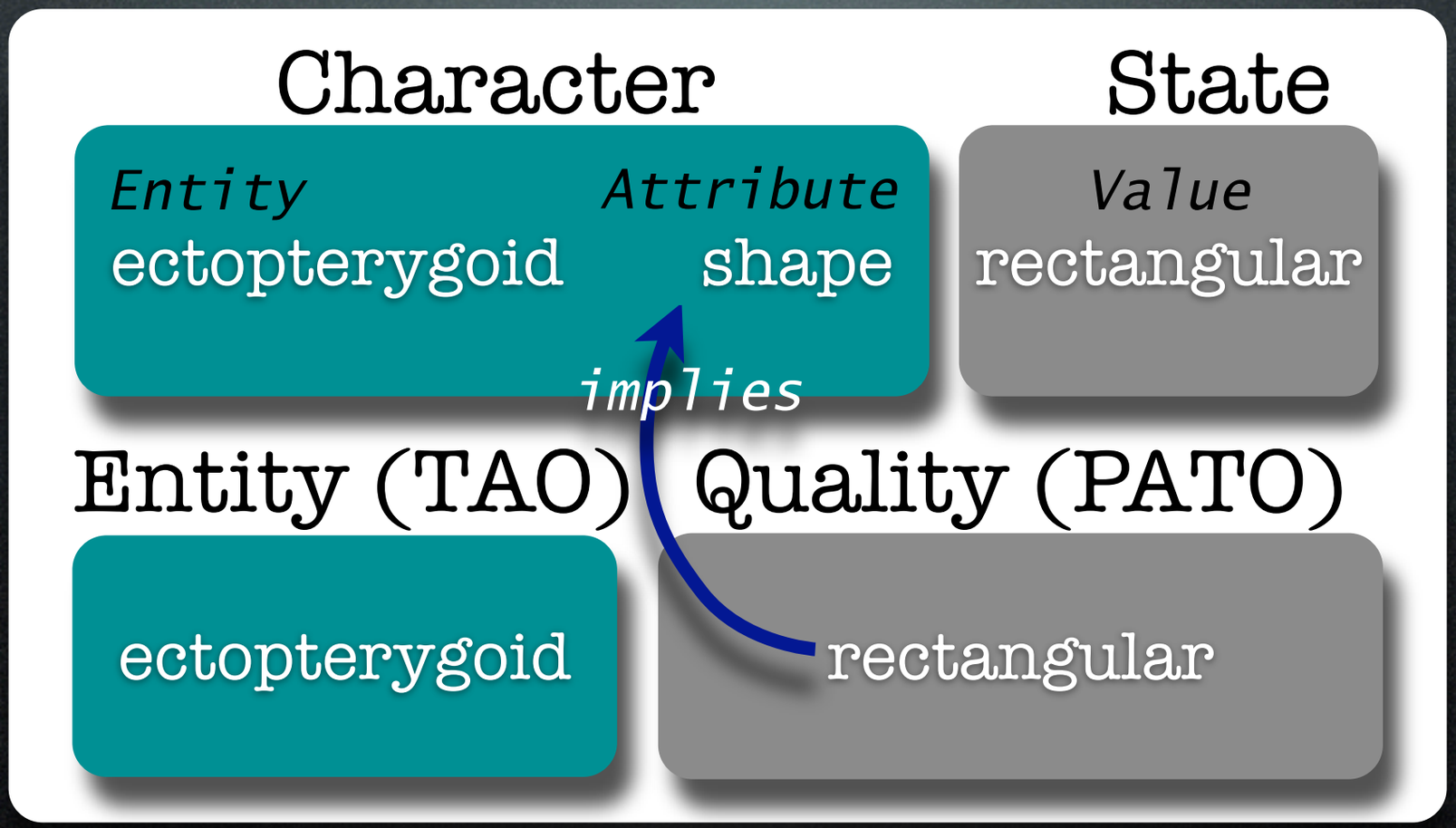
Entity-Quality Model for Evolutionary Phenotypes



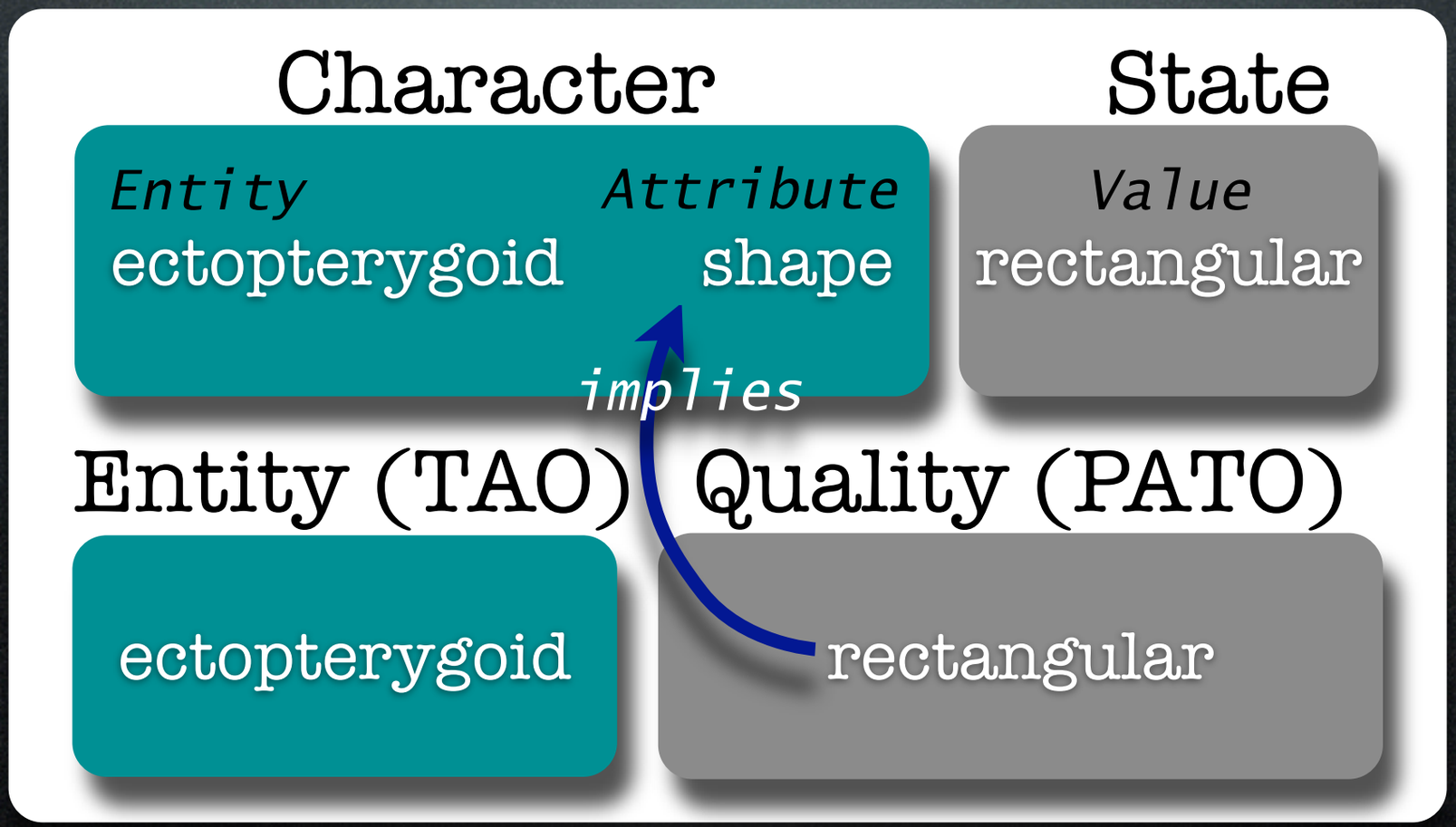
Entity-Quality Model for Evolutionary Phenotypes



Entity-Quality Model for Evolutionary Phenotypes



Entity-Quality Model for Evolutionary Phenotypes



Phenotype

Taxon phenotype assertion

Links a taxon to a phenotype

Links a quality to the entity that is its bearer

Phenotypic Quality ontology term

some Batrachoglanis raninus

exhibits some

rectangular ***inheres_in*** some ectopterygoid

Taxon ontology term

Anatomy ontology term

Taxon phenotype assertion

Links a taxon to a phenotype

Links a quality to the entity that is its bearer

Phenotypic Quality ontology term

some Batrachoglanis raninus

exhibits some

rectangular ***inheres_in*** some ectopterygoid

Taxon ontology term

Anatomy ontology term

Curator

Evidence Code

Specimen

Publication

Gene phenotype assertion

Links a genotype to a phenotype

Links a quality to the entity that is its bearer

Phenotypic Quality ontology term

edn1 tf216b/tf216b

influences some

curvature ***inheres_in*** some maxilla

Genotype

Anatomy ontology term

Publication

Full workflow: free-text → EQ → integrated KB

legacy free-text character data

56. *Naked body.* A scaleless body is a derived condition in the ostariophysans. Most cypriniforms, characiforms and most primitive teleosts and gonorhynchiforms possess scales on all or part of the body, while the majority of catfishes (including ariids) and gymnotoids lack scales – although scales are often represented by ossified lateral line tubes (Roberts 1973; Fink and Fink 1981). Some catfishes (doradids,

Kailola (2004)

© Jean Ricardo Simões Vitule

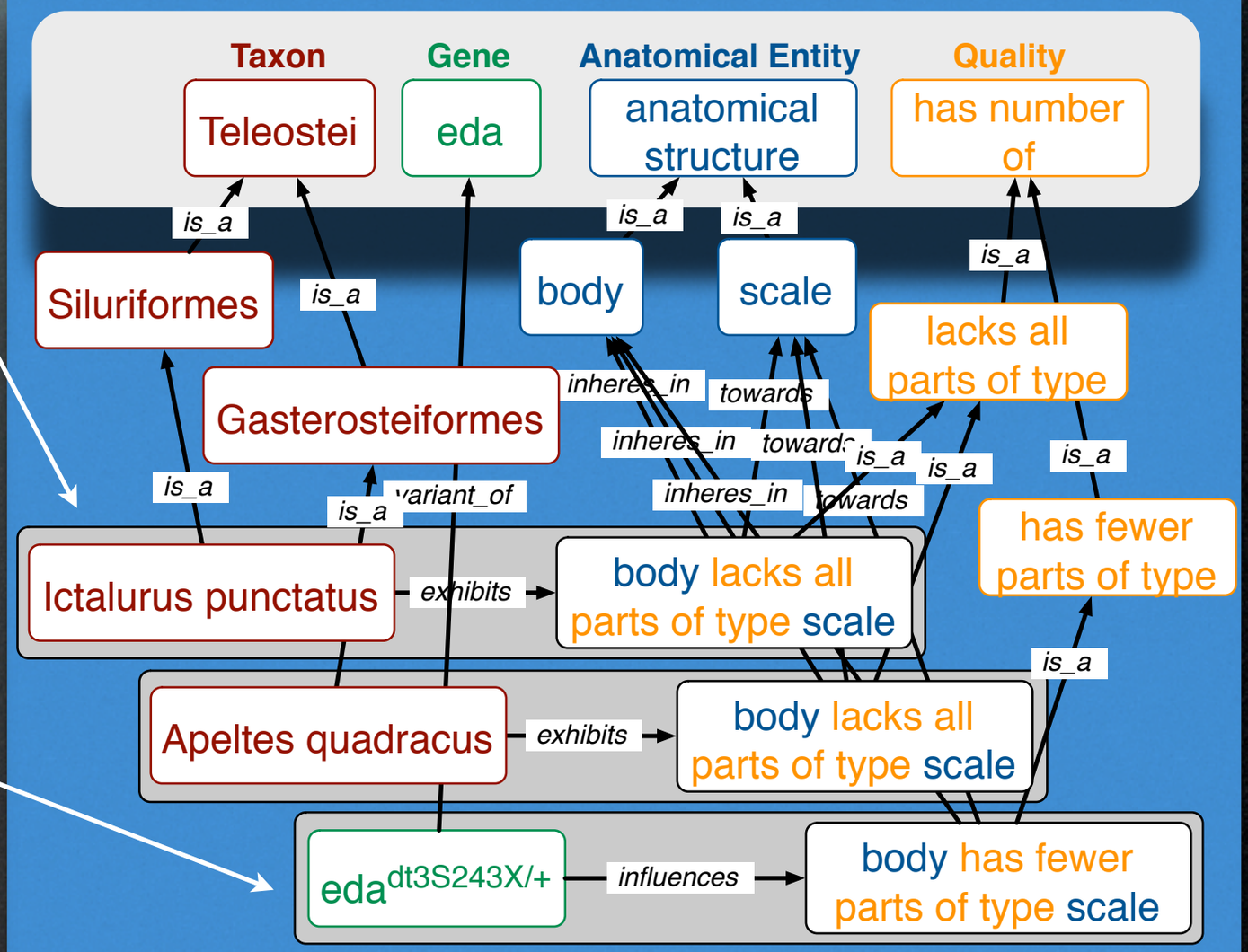
mutant phenotype

Here, we describe the phenotypic and molecular characterization of a set of mutants showing loss of adult structures of the dermal skeleton, such as the rays of the fins and the scales, as well as the pharyngeal teeth. The mutations represent adult-viable, loss of function alleles in the *ectodysplasin* (*eda*) and *ectodysplasin receptor* (*edar*) genes.

Harris et al. (2008)

EQ = body lacks all parts of type scale

EQ = body has fewer parts of type scale



Full workflow: free-text → EQ → integrated KB

legacy free-text character data

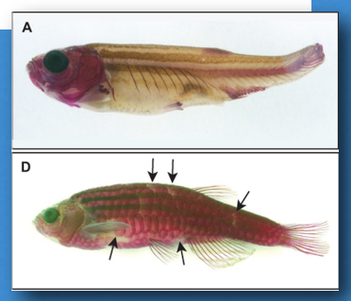
56. *Naked body.* A scaleless body is a derived condition in the ostariophysans. Most cypriniforms, characiforms and most primitive teleosts and gonorhynchiforms possess scales on all or part of the body, while the majority of catfishes (including ariids) and gymnotoids lack scales – although scales are often represented by ossified lateral li
Fink and Fink 1981). Some

Kailola (2004)

mutant phen

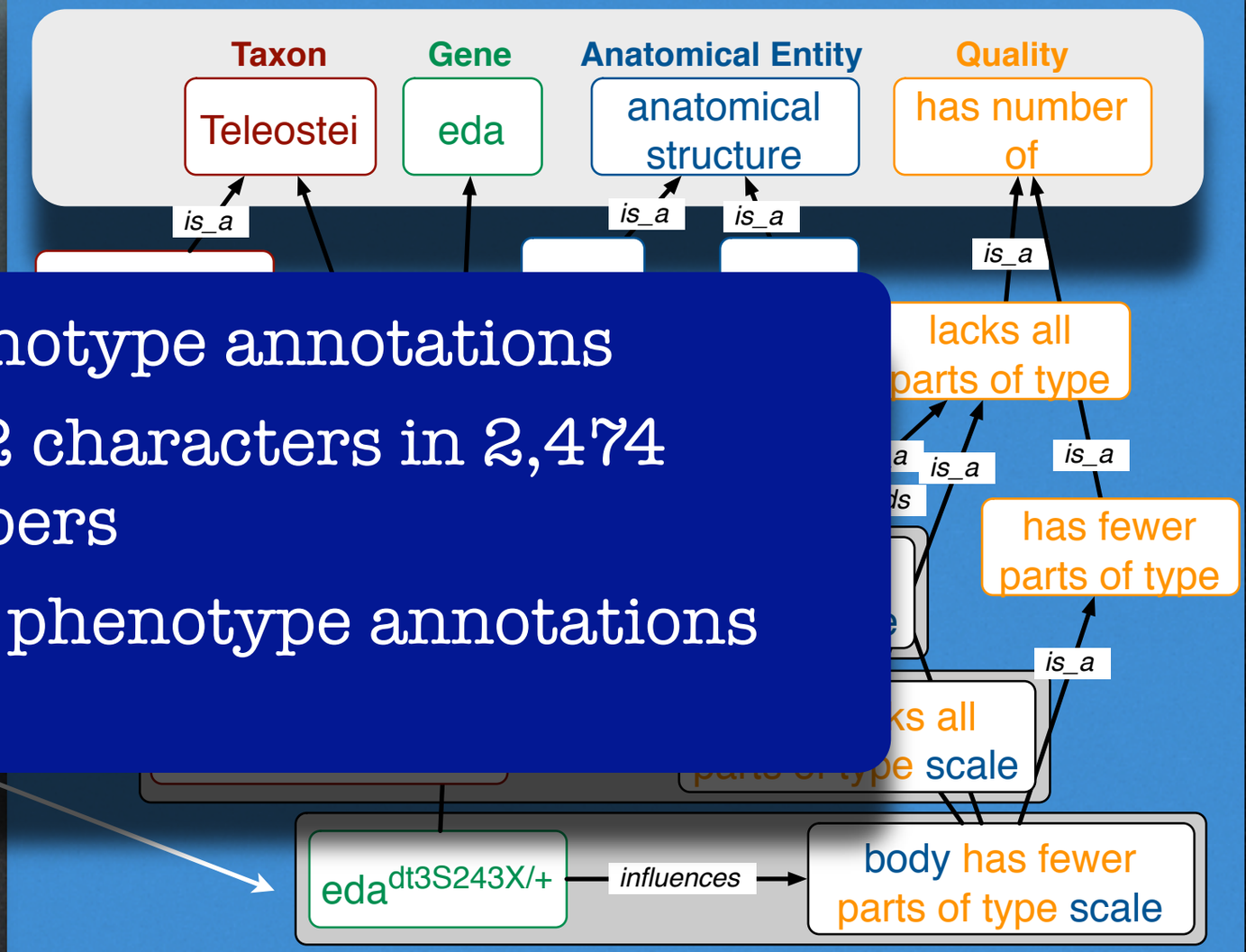
Here, we describe the p
molecular characterizati
mutants showing loss of
the dermal skeleton, such as the rays of the fins and the scales, as well as the pharyngeal teeth. The mutations represent adult-viable, loss of function alleles in the *ectodysplasin* (*eda*) and *ectodysplasin receptor* (*edar*) genes.

Harris et al. (2008)

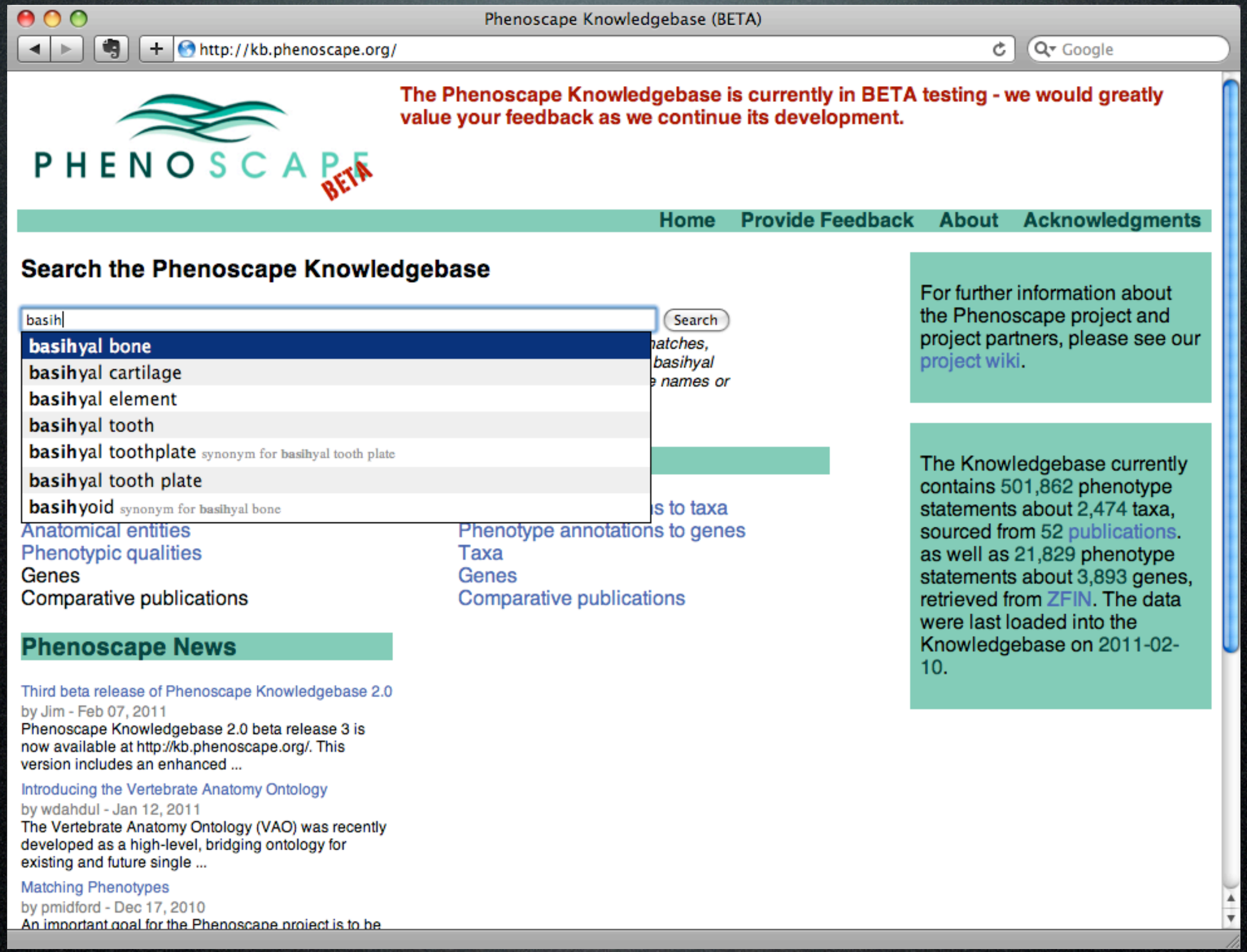


EQ = body lacks all parts of type scale

- 501,862 taxon phenotype annotations
- Curated from 4,732 characters in 2,474 species from 52 papers
- From ZFIN: 21,829 phenotype annotations about 3,893 genes



Phenoscape Knowledgebase



PHENOSCAPE BETA

The Phenoscape Knowledgebase is currently in BETA testing - we would greatly value your feedback as we continue its development.

Site search: Go

Enter entity terms (e.g. basihyal bone), phenotypic qualities (e.g. shape, size), taxonomic names (e.g. Ictaluridae), gene names or symbols (e.g. cadherin 6, cdh6), or publications.

Query for:

Phenotype annotations to taxa

Taxon is: Add

Ictalurus

and

Phenotype is: Add

Any

and

Publication is: Add

Any

Include inferred annotations [help]

Apply Filter

« Previous Results 1 - 20 of 2450 Next »

	Phenotype [help]			
Taxon	Entity	Quality	Related Entity	Source
Ictalurus australis	posterior region of supraoccipital crest	bifid		
Ictalurus australis	process of occipital region	shape		
Ictalurus australis	facial foramen	position		
Ictalurus australis	pectoral fin spine	shape		
Ictalurus australis	anatomical margin of process of dorsal side of cranium	round		
Ictalurus australis	Weberian apparatus	depth		
Ictalurus australis	lateral ethmoid wing	increased length		
Ictalurus australis	frontal bone	structure	adductor mandibulae complex	
Ictalurus australis	epihyal-ceratohyal joint	cartilaginous		

PHENOSCAPE BETA

The Phenoscape Knowledgebase is currently in BETA testing - we would greatly value your feedback as we continue its development.

Site search: Go

Enter entity terms (e.g. basihyal bone), phenotypic qualities (e.g. shape, size), taxonomic names (e.g. Ictaluridae), gene names or symbols (e.g. cadherin 6, cdh6), or publications.

Query for:

Phenotype annotations to taxa

Taxon is: Add

Ictalurus

and

Phenotype is: Add

mesethmoid bone • shape

[broader/refine]

including parts

and

Publication is: Add

Any

Include inferred annotations

[help]

Apply Filter

« Previous Results 1 - 11 of 11 Next »

	Phenotype [help]			
Taxon △	Entity	Quality	Related Entity	Source
Ictalurus australis	mesethmoid cornu	bifurcated		
Ictalurus balsanus	mesethmoid cornu	bifurcated		
Ictalurus dugesii	mesethmoid cornu	bifurcated		
Ictalurus furcatus	mesethmoid cornu	bifurcated		
Ictalurus lupus	mesethmoid cornu	bifurcated		
Ictalurus meridionalis	mesethmoid bone	shape		
Ictalurus mexicanus	mesethmoid cornu	bifurcated		
Ictalurus pricei	mesethmoid cornu	bifurcated		
Ictalurus punctatus	mesethmoid cornu	bifurcated		
Ictalurus punctatus	mesethmoid bone	shape		
Ictalurus punctatus	medial region of anterior margin of mesethmoid bone	notched		

Phenoscape Knowledgebase (BETA)

http://kb.phenoscape.org/taxon_annotations?filter%5Btaxa%5D%5B0%5D=TTO%3A101020&filter%5Bphenc

Google

PHENOSCAPE BETA

The Phenoscape Knowledgebase is currently in BETA testing - we would greatly value your feedback as we continue its development.

Site search: Go

Enter entity terms (e.g. basihyal bone), phenotypic qualities (e.g. shape, size), taxonomic names (e.g. Ictaluridae), gene names or symbols (e.g. cadherin 6, cdh6), or publications.

Home | Provide Feedback | About | Acknowledgments

Query for:

Phenotype annotati

Taxon is:

Ictalurus

and

Phenotype is:

mesethmoid bo

[broaden/refine]

including parts

and

Publication is:

Any

Include inferre

[help]

Apply

mesethmoid bone

Source: Teleost Anatomy Ontology
Synonyms: ethmoid
Definition: Endochondral bone that extends forward from the frontal bones and articulates posterolaterally with the lateral ethmoids and the vomer and parasphenoid ventrally. The mesethmoid is an unpaired median bone.
ID: TAO:0000323
overlaps: mesethmoid-frontal joint, mesethmoid-lateral ethmoid joint, mesethmoid-nasal joint, mesethmoid-premaxillary joint, mesethmoid-vomer joint, neurocranium
develops from: ethmoid cartilage, neurocranial trabecula
is part of: chondrocranium, olfactory region
is a type of: endochondral bone
may have part: lateral mesethmoid wing, mesethmoid cornu, mesethmoid ventral diverging lamella

View details for **mesethmoid bone**

« Previous

Results 1 - 11 of 11

Next »

Phenotype [help]			
Entity	Quality	Related Entity	Source
mesethmoid cornu	bifurcated		
mesethmoid cornu	bifurcated		
mesethmoid cornu	bifurcated		
mesethmoid cornu	bifurcated		
mesethmoid cornu	bifurcated		
mesethmoid bone	shape		
mesethmoid cornu	bifurcated		
mesethmoid cornu	bifurcated		
mesethmoid cornu	bifurcated		
mesethmoid bone	shape		
egion of anterior margin of mesethmoid bone	notched		

Phenoscape Knowledgebase (BETA)

http://kb.phenoscape.org/taxon_annotations?filter%5Btaxa%5D%5B0%5D=TTO%3A101020&commit=Apply

Google

PHENOSCAPE BETA

The Phenoscape Knowledgebase is currently in BETA testing - we would greatly value your feedback as we continue its development.

Site search: Go

Enter entity terms (e.g. basihyal bone), phenotypic qualities (e.g. shape, names or symbols (e.g. cdh6), or publications.

Source Data

Ictalurus australis	anatomical margin of process of dorsal side of cranium	round	

Lundberg 1992

character 13: Sagittal crests on skull roof

state: broad with sculptured surface restricted to supraoccipital, and rounded margins near fronto-supraoccipital suture

Query for:

Phenotype annot

Taxon is:

Ictalurus

and

Phenotype is:

Any

and

Publication is:

Any

Include infer

[help]

App

knowledgments

Next »

Entity	Source
Ictalurus australis	frontal bone	structure	adductor mandibulae complex	
Ictalurus australis	epihyal-ceratohyal joint	cartilaginous		

publication: Lundberg 1992

source: Phenoscape-annotated publications

Overview | Data Matrix | Specimens

Original character-by-taxon data matrix from publication **Lundberg 1992**. You can also query for all phenotype annotations curated from this matrix.

	1. Posterior flap of adipose fin	2. Gill membranes	3. Orbital rim	4. Caudal fin
Amelurus brunneus	free from back and caudal fin	divergent	free	scarcely emarginate to rou
Amelurus catus	free from back and caudal fin	divergent	free	forked with rounded lobes
Amelurus melas	free from back and caudal fin	divergent	free	scarcely emarginate to rou
Amelurus natalis	free from back and caudal fin	divergent	free	scarcely emarginate to rou
Amelurus nebulosus	free from back and caudal fin	divergent	free	scarcely emarginate to rou
Amelurus platycephalus	free from back and caudal fin	divergent	free	scarcely emarginate to rou
Amelurus serracanthus	free from back and caudal fin	divergent	free	scarcely emarginate to rou
Ictalurus australis	free from back and caudal fin	divergent	free	forked with pointed lobes
Ictalurus balsanus	free from back and caudal fin	divergent	free	forked with pointed lobes
Ictalurus dugesii as Ictalurus dugesi	free from back and caudal fin	divergent	free	forked with pointed lobes
Ictalurus furcatus	free from back and caudal fin	divergent	free	forked with pointed lobes
Ictalurus lupus	free from back and caudal fin	divergent	free	forked with pointed lobes
Ictalurus mexicanus	free from back and caudal fin	divergent	free	forked with pointed lobes
Ictalurus pricei	free from back and caudal fin	divergent	free	forked with pointed lobes
Ictalurus punctatus	free from back and caudal fin	divergent	free	forked with pointed lobes
Noturus flavus	adnate to back and broadly joined to caudal fin	divergent	united to cornea, eye subcutaneous	scarcely emarginate to rou
Noturus insignis	adnate to back and broadly joined to caudal fin	divergent	united to cornea, eye subcutaneous	scarcely emarginate to rou
Noturus stigmosus	adnate to back and partly joined to caudal fin	divergent	united to cornea, eye subcutaneous	scarcely emarginate to rou
Prietella phreatophila	adnate to back and broadly joined to caudal fin	divergent	?	scarcely emarginate to rou

publication: Lundberg 1992

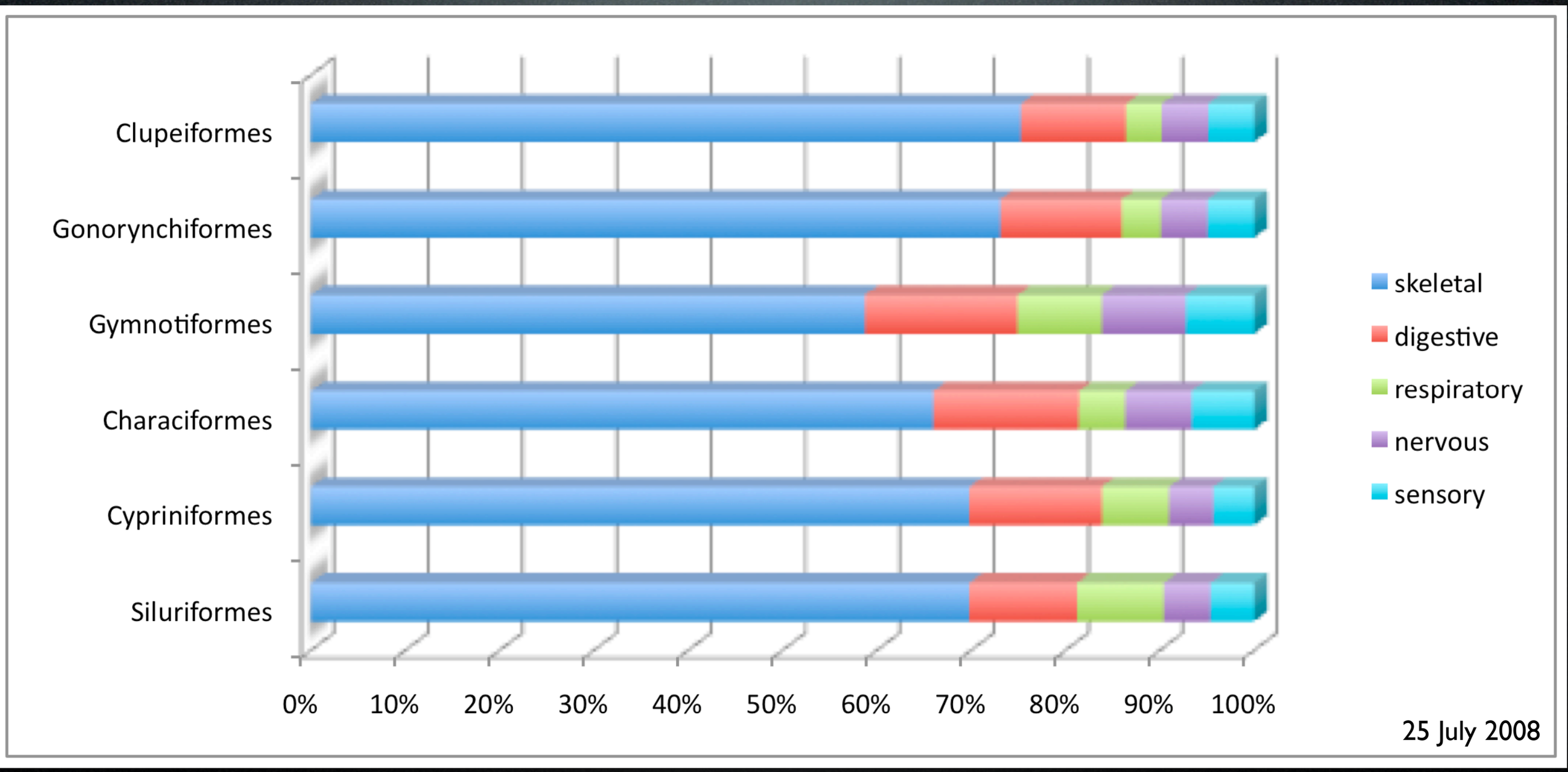
source: Phenoscape-annotated publications

Overview | Data Matrix | Specimens

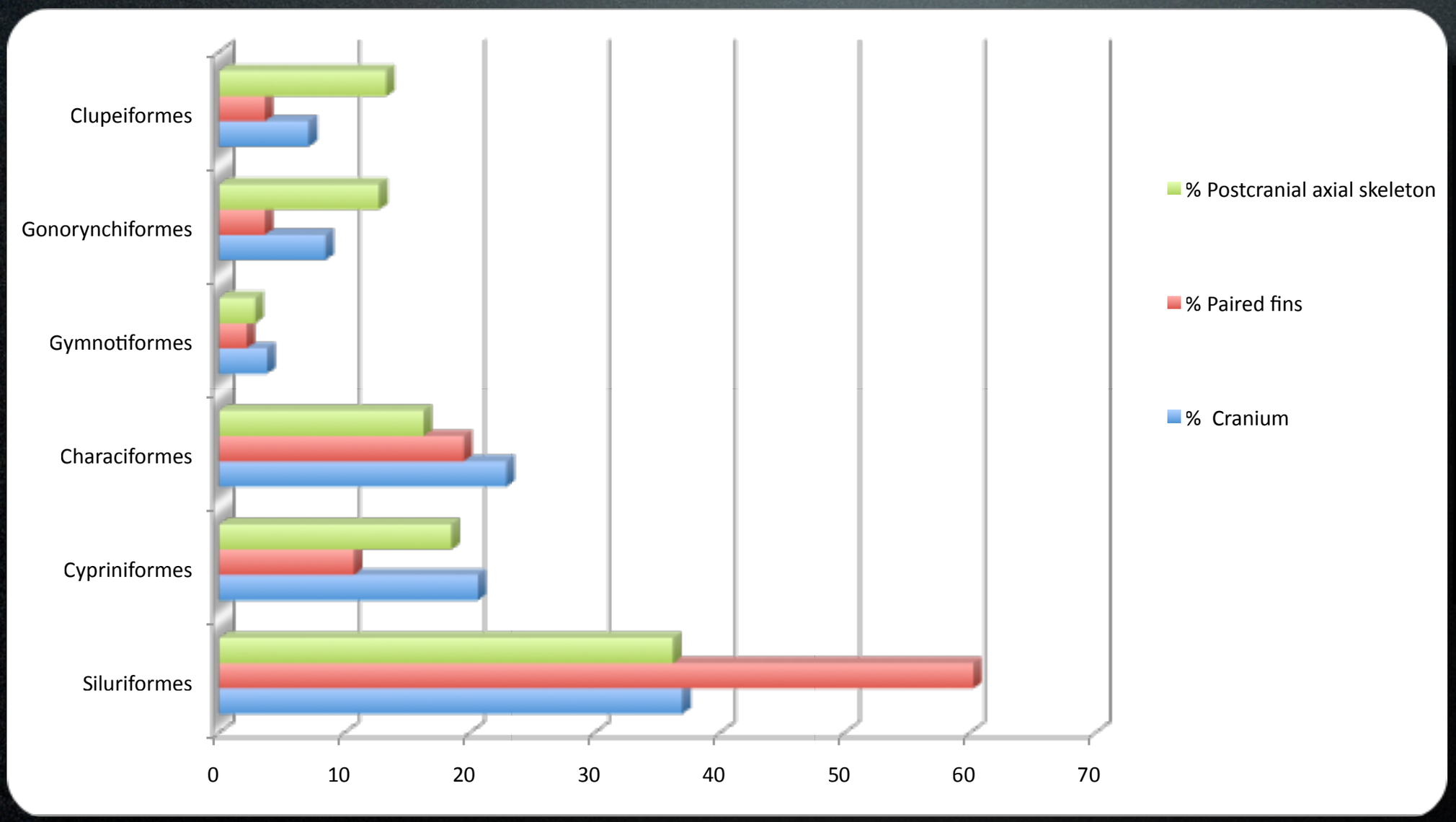
Taxa and specimens used in this study

Taxon name used in publication	Valid taxon	Specimens	Comment
Noturus nocturnus	*Noturus nocturnus*	UMMZ 165883, UMMZ 167214	
Satan eurystomus	*Satan eurystomus*		WMM not in collection code list
Noturus flavus	*Noturus flavus*	UMMZ 126366, UMMZ 126365, UMMZ 182039, UMMZ 111724, UMMZ 165852, UMMZ 165842, UMMZ 165833, UMMZ 189178	
Ictalurus lupus	*Ictalurus lupus*	UMMZ 169619, UMMZ 17???4, UMMZ 186503, UMMZ 179822	One of the specimen ID numbers for UMMZ was partially unreadable. I replaced the unreadable numbers with question marks.
Pylodictis olivaris	*Pylodictis olivaris*	UMMZ 186652, UMMZ 173452, UMMZ 169029, UMMZ 103107, UMMZ 152549, UMMZ 160732, UMMZ 170129, UMMZ 97069, UMMZ 186266	
		UW 3276, UW 3275, AMNH 9499, USNM 3983, USNM 3982, USNM 3981, USNM 3985, USNM 8122, AMNH 6387, AMNH 6388, USNM 167588, USNM 167589, USNM 167590, USNM 2179	PF not in list of

Major taxonomic groups have similar distribution of entities among phenotypes



Some notable differences for skeletal characters



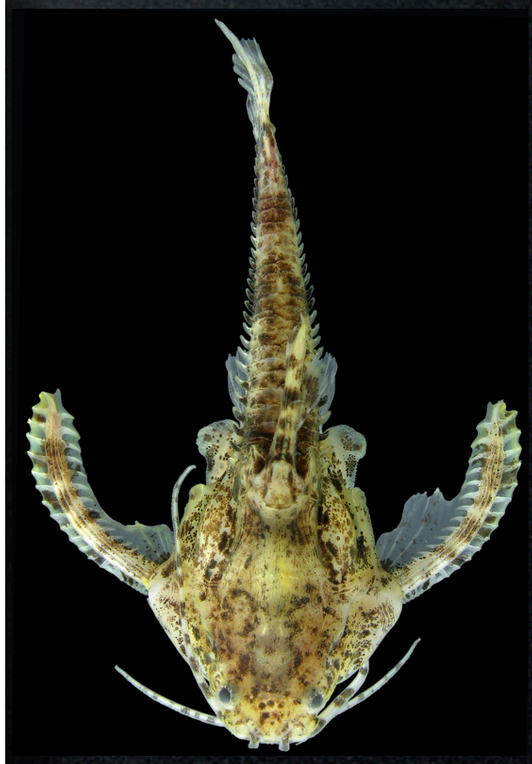
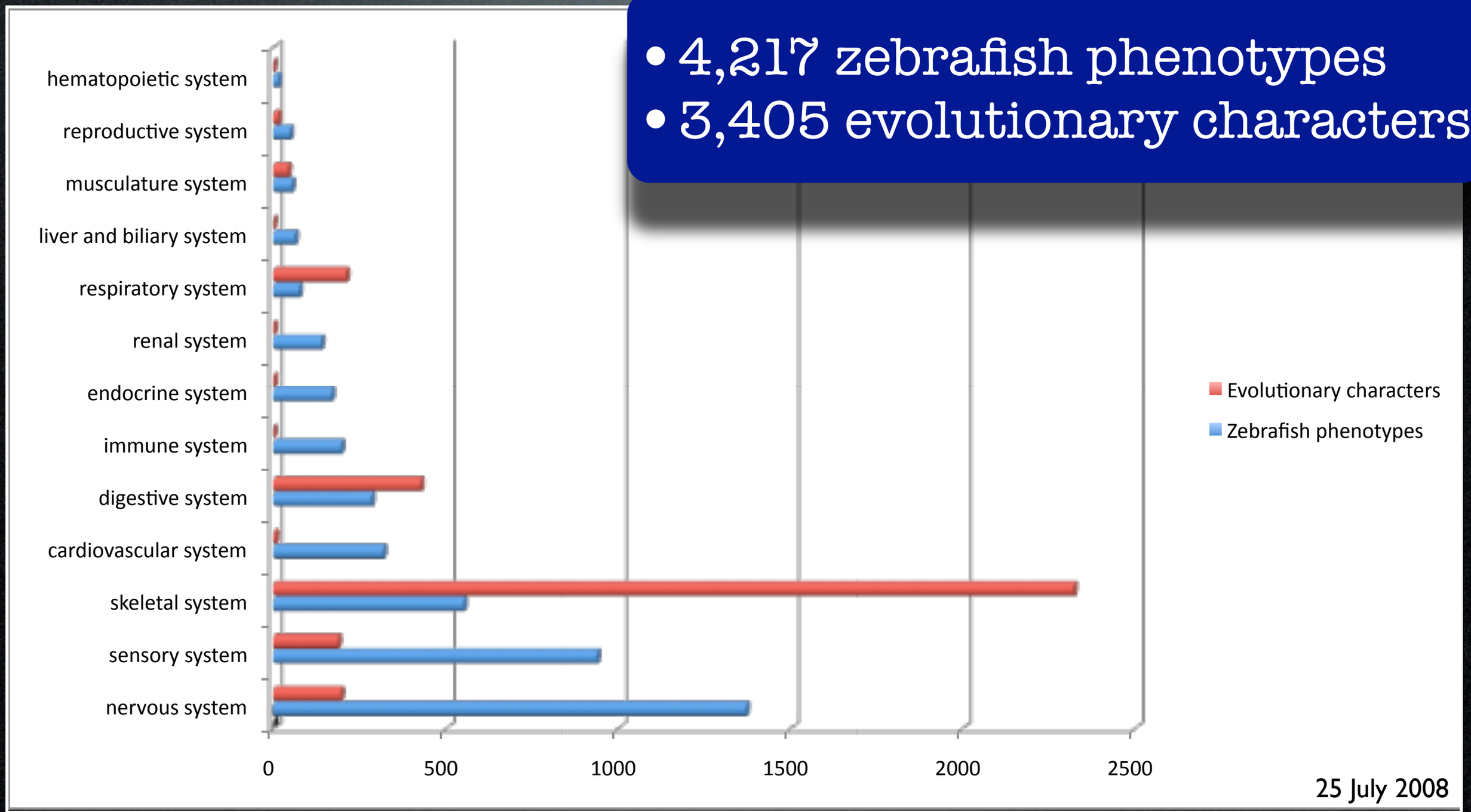


Image from Sabaj-Perez

25 July 2008

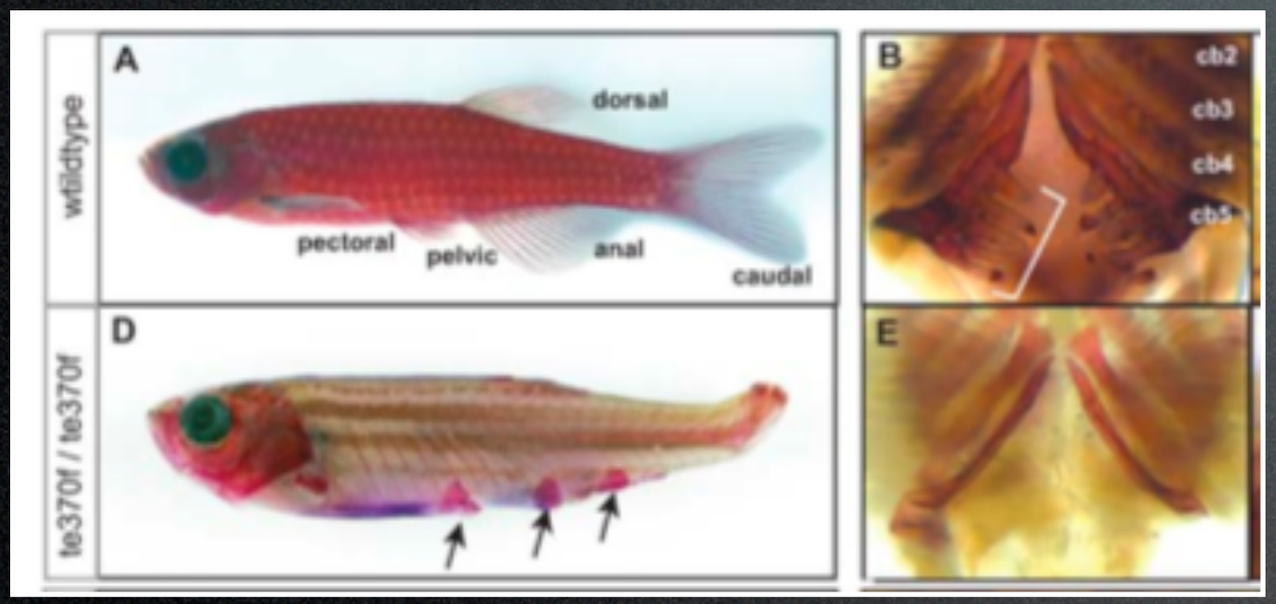
Substantial overlap between model organism and evolutionary phenotypes

- 4,217 zebrafish phenotypes
- 3,405 evolutionary characters



Hypothesis generation: Genetic basis for scale loss in Siluriformes

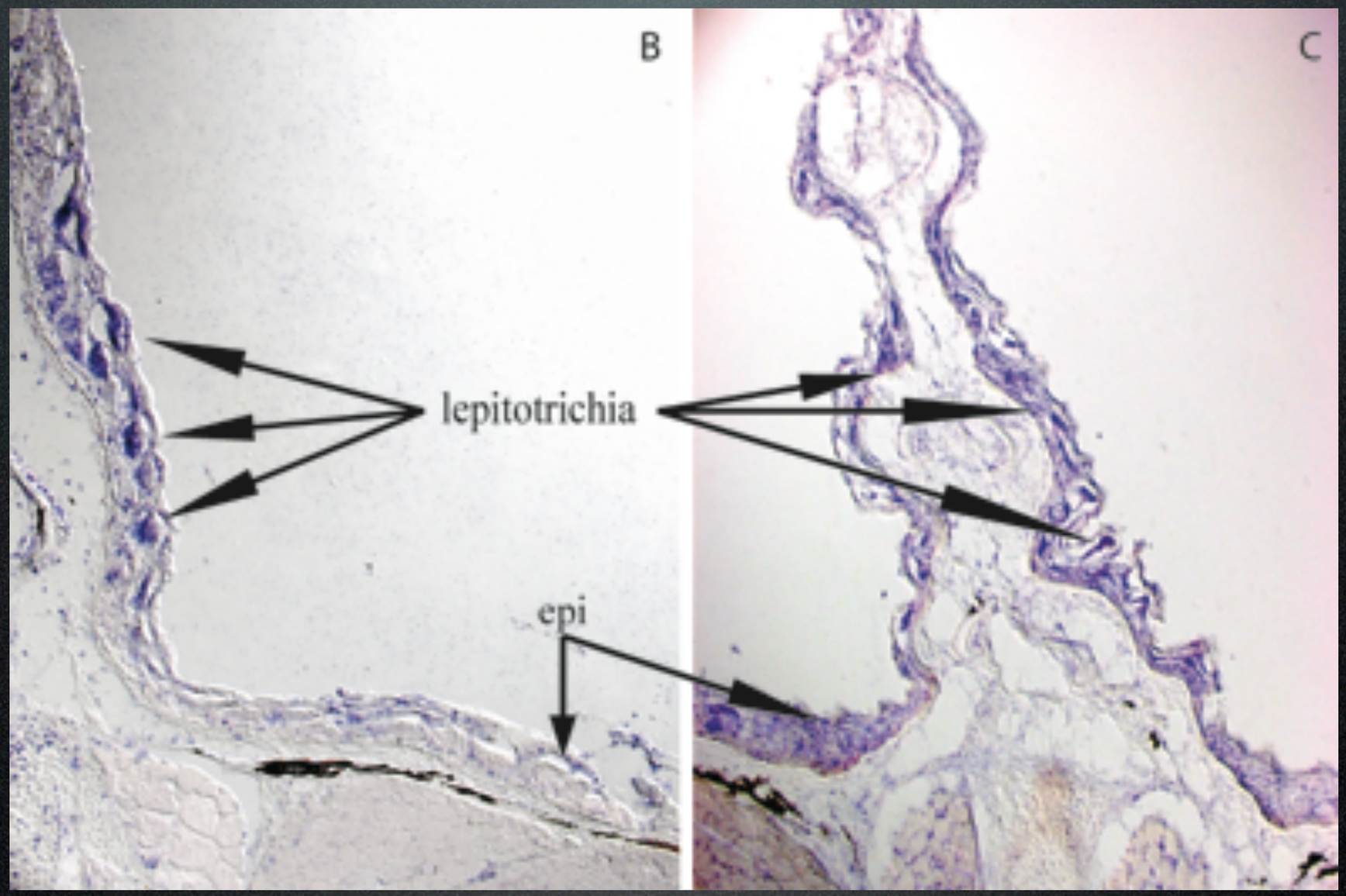
Mutation of *eda* gene in *Danio*:



Harris et al., 2007

Ictalurus punctatus:

Wet lab test
(Work by Richard Edmunds)
Ictalurus punctatus



eda expression is lacking in the epidermis

Hypothesis generation: Genetic basis for absence of the basihyal bone in Siluriformes

Mutation of *brpf1* gene in *Danio*:

PHENOTYPE:

Genotype(s): *brpf1*$^{b943/b943}$, *brpf1*$^{t20002/t20002}$, *brpf1*$^{t25114/t25114}$

Observed In: basihyal

Stage Range : Day 5

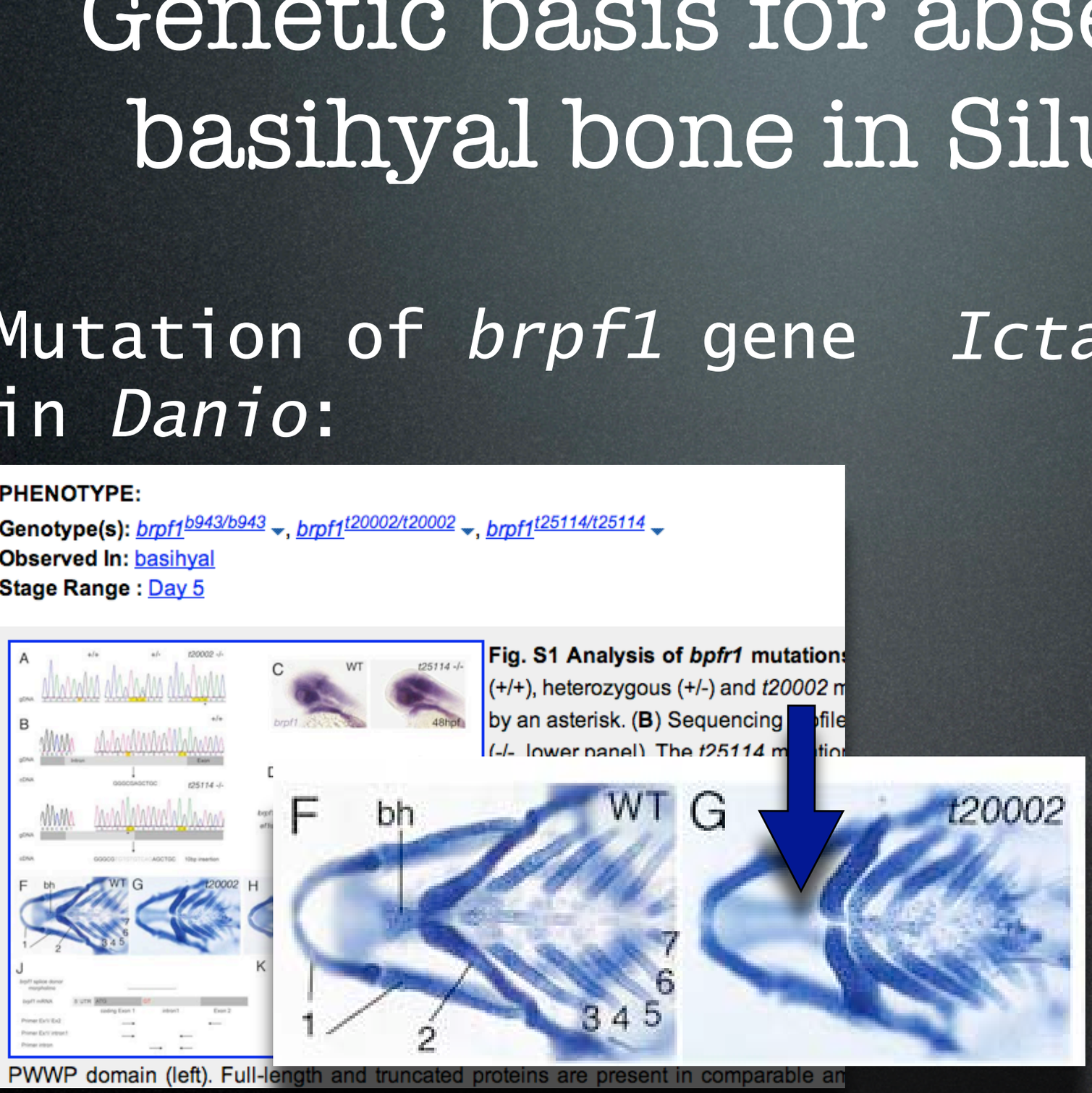


Fig. S1 Analysis of *bpfr1* mutations
(+/+), heterozygous (+/-) and *t20002* m
by an asterisk. (**B**) Sequencing
(-/- lower panel). The *t25114* m

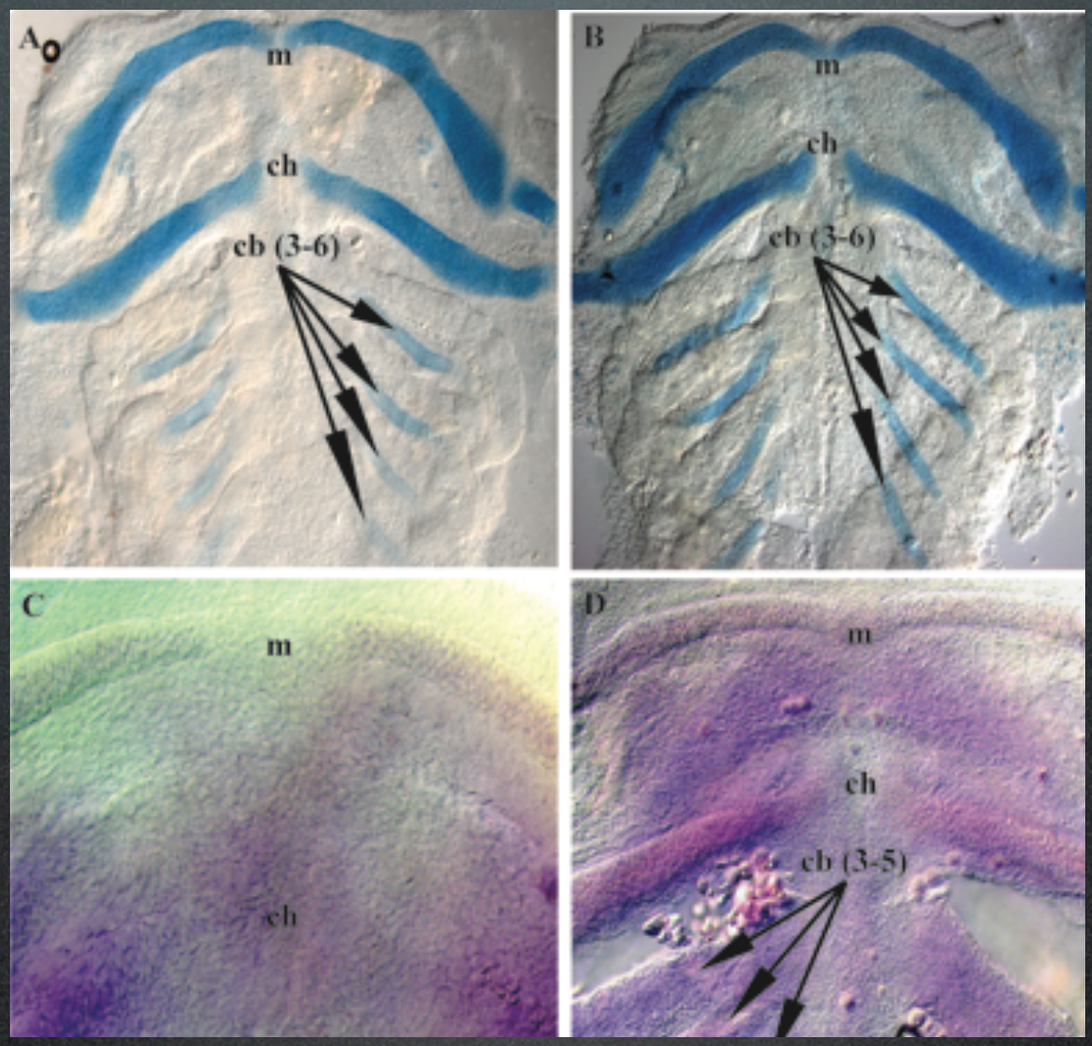
PWWP domain (left). Full-length and truncated proteins are present in comparable an

Laue et al (2008)

Ictalurus punctatus:

Wet lab test
(Work by Richard Edmunds)

Ictalurus punctatus

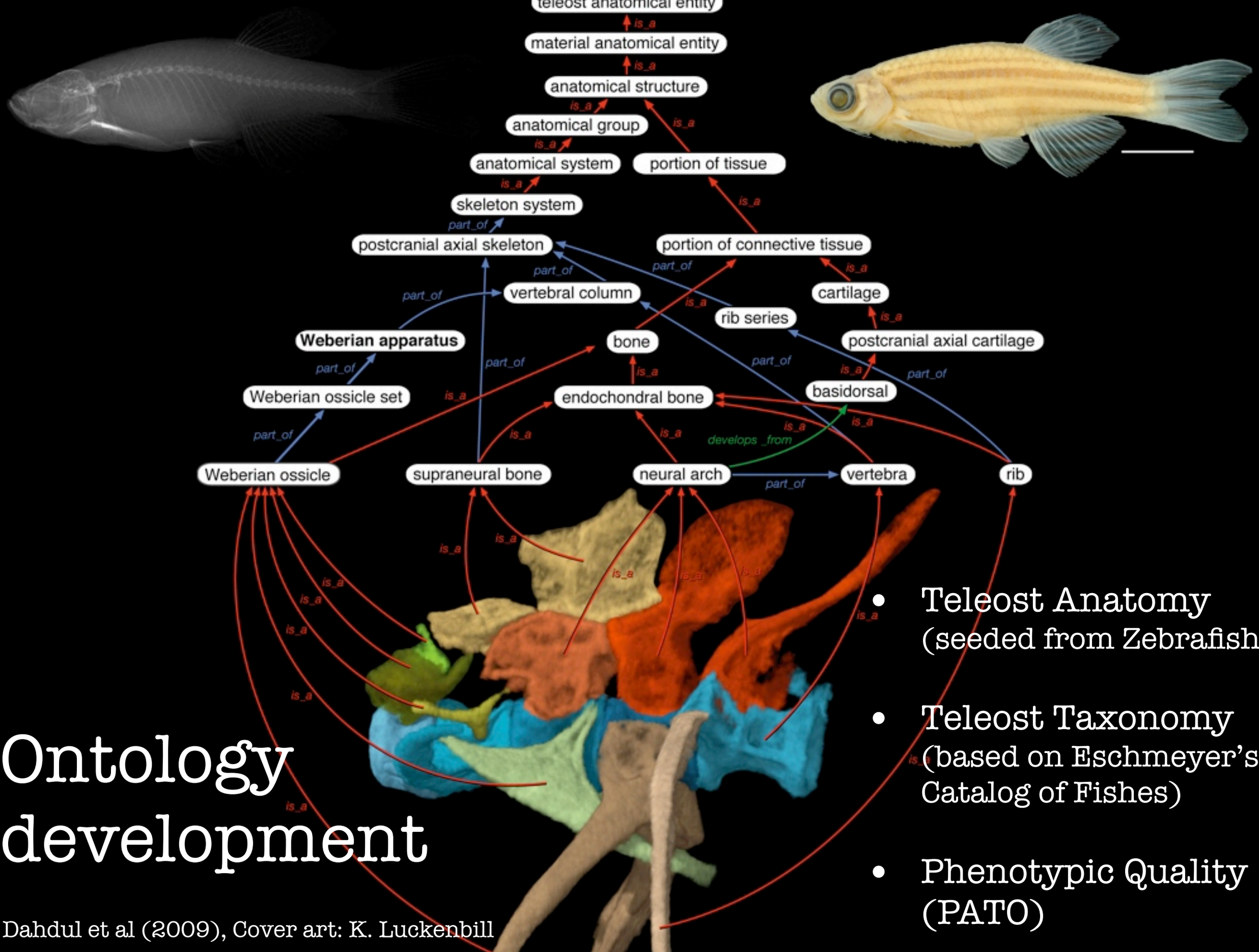


78 hpf 86 hpf

brpf1 lacks expression in the basihyal

The parts to make this work

- Ontologies that capture the knowledge domains
- Efficient data curation workflow
- Expressive and scalable inference engine

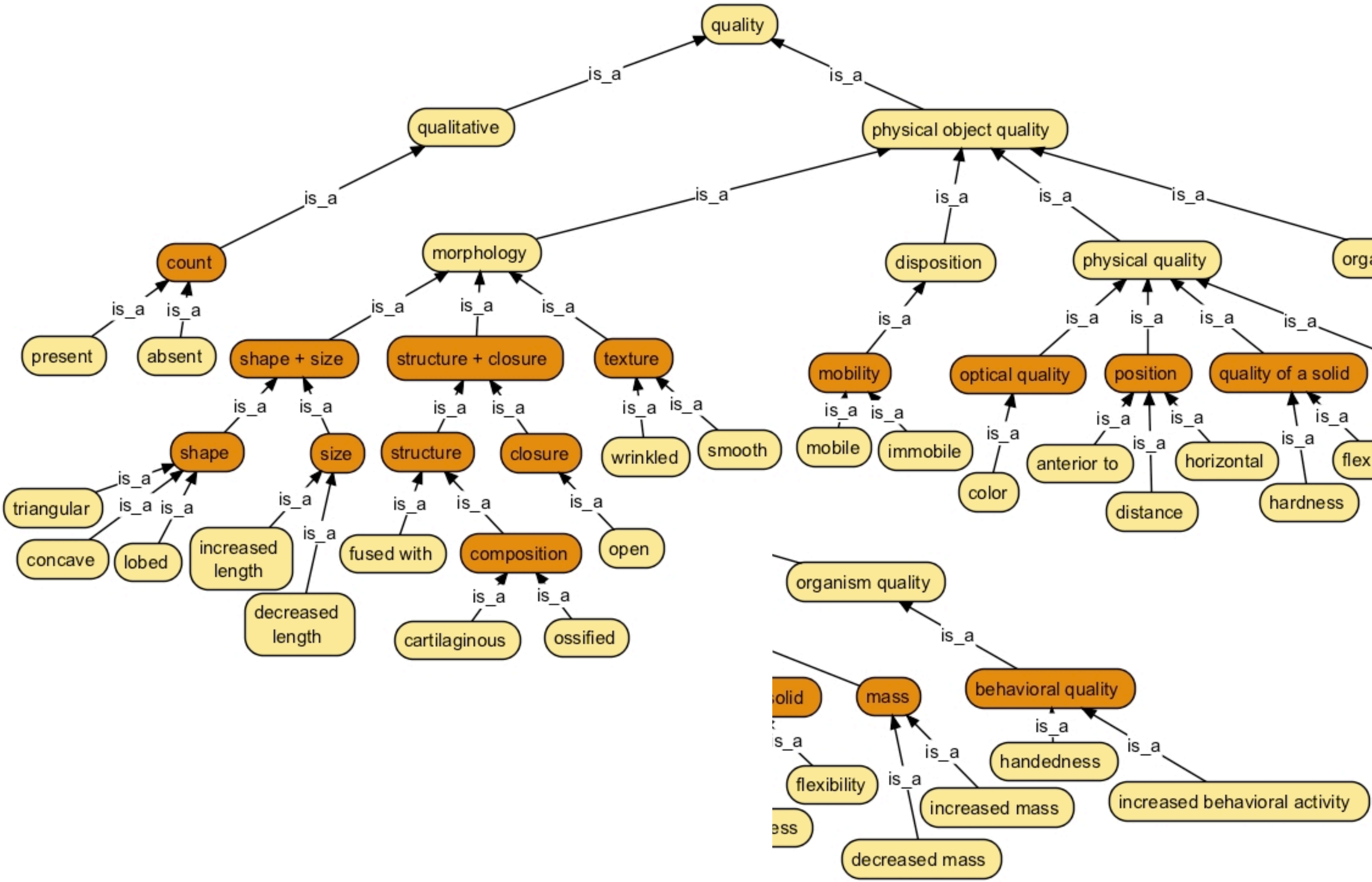


Ontology development

Dahdul et al (2009), Cover art: K. Luckenbill

- Teleost Anatomy (seeded from Zebrafish)
- Teleost Taxonomy (based on Eschmeyer's Catalog of Fishes)
- Phenotypic Quality (PATO)

Making PATO usable for evolutionary data



Getting ontologies right is a challenge

- What is the right axis of classification?
  - Structure versus function
  - Relational vs monadic qualities
  - PATO: shape and size vs natural language

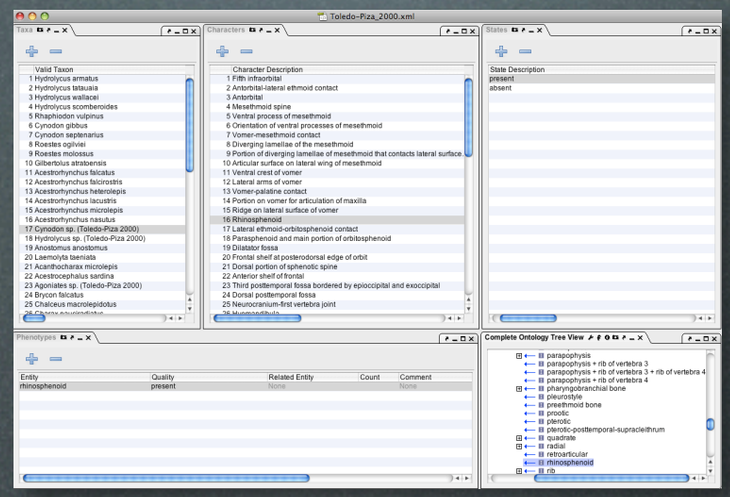
    ”Interopercle shape: expanded posteroventrally”

- Different ways to observe or generate a phenotypic quality
  - Color as color hue (radiation quality) or pigmentation (structural quality)
  - Relative sizes don’t have a universal reference

Curation

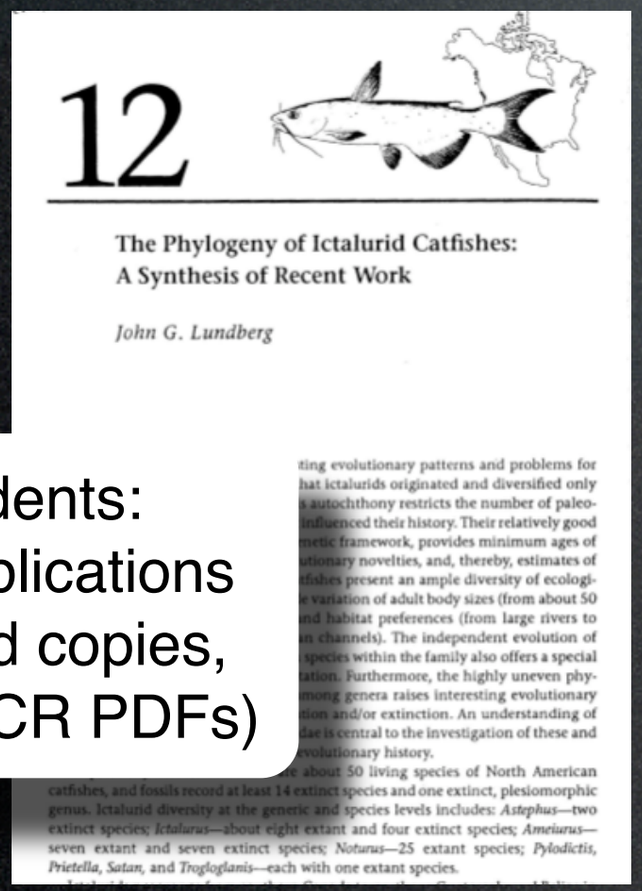
Dahdul et al., 2010 PLoS ONE

2. Students: Manual entry of free text character descriptions, matrix, taxon list, specimens and museum numbers using ***Phenex***



3. Character annotation by experts: Entry of phenotypes and homology assertions using ***Phenex***

1. Students: gather publications (scan hard copies, produce OCR PDFs)



12

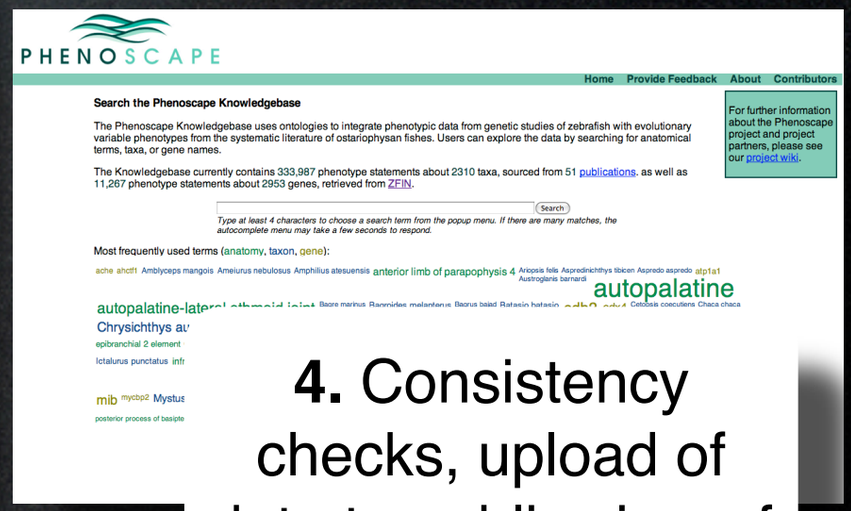
The Phylogeny of Ictalurid Catfishes: A Synthesis of Recent Work

John G. Lundberg

ting evolutionary patterns and problems for
hat ictalurids originated and diversified only
s autochthony restricts the number of paleo-
influenced their history. Their relatively good
metic framework, provides minimum ages of
utionary novelties, and, thereby, estimates of
fishes present an ample diversity of ecologi-
e variation of adult body sizes (from about 50
nd habitat preferences (from large rivers to
n channels). The independent evolution of
species within the family also offers a special
ation. Furthermore, the highly uneven phy-
mong genera raises interesting evolutionary
tion and/or extinction. An understanding of
dae is central to the investigation of these and
volutionary history.

e about 50 living species of North American catfishes, and fossils record at least 14 extinct species and one extinct, plesiomorphic genus. Ictalurid diversity at the generic and species levels includes: *Astephus*—two extinct species; *Ictalurus*—about eight extant and four extinct species; *Ameiurus*—seven extant and seven extinct species; *Noturus*—25 extant species; *Pylodictis*, *Prietella*, *Satan*, and *Trogloglanis*—each with one extant species.

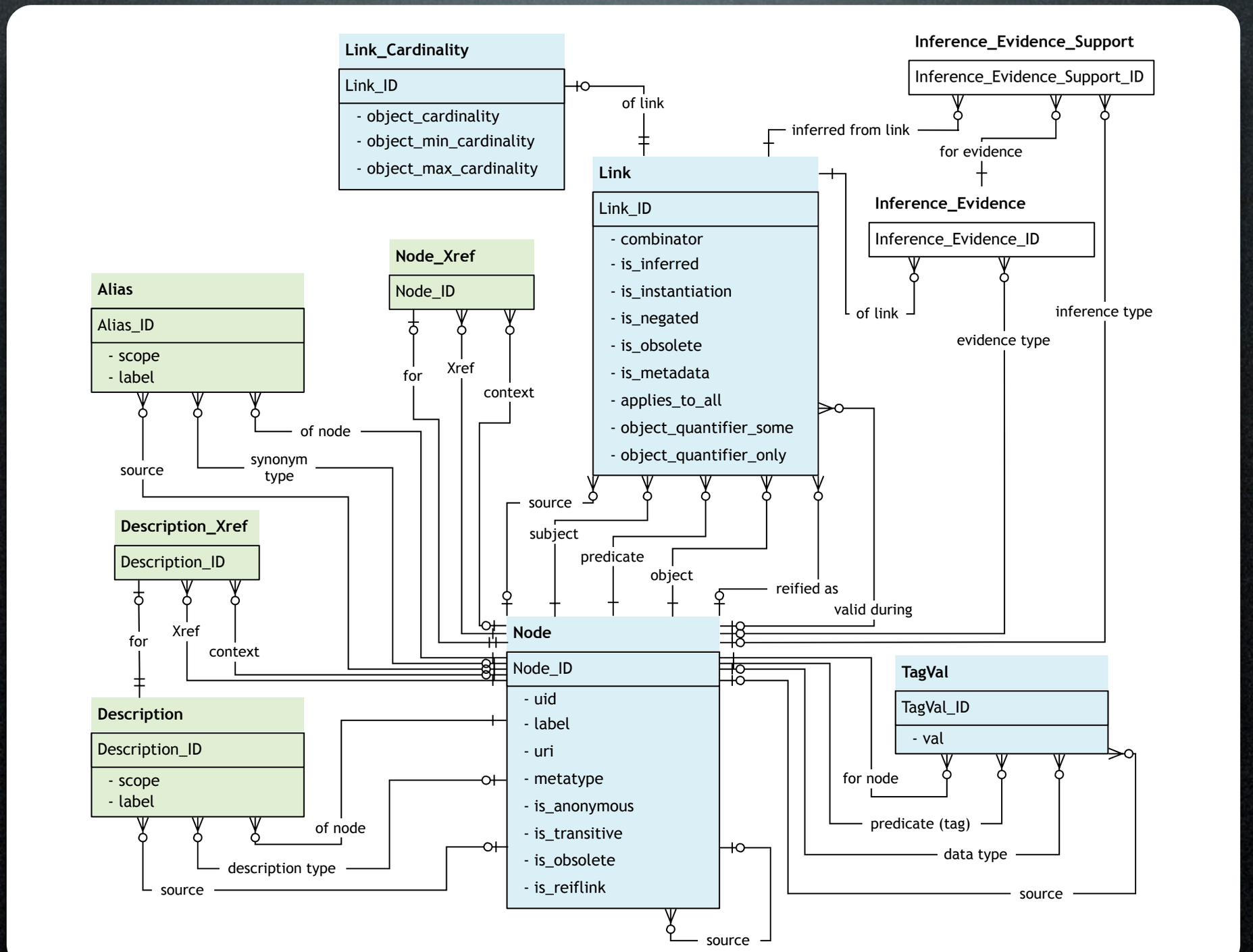
Curators:
Wasila Dahdul
Miles Coburn
Jeff Engemen
Terry Grande
Eric Hilton
John Lundberg
Paula Mabee
Richard Mayden
Mark Sabaj Pérez



4. Consistency checks, upload of data to public view of Phenoscape KB

KB is based on OBD (Ontology-Based Database)

(C. Mungall, LBL)



ECO:evidence

curator(s)

PATO:quality

Measurement
-value/max/min
-unit

ZFIN:Publication
uid = ZFIN ID

TTO:taxon
uid = TTO ID

has_evidence

dc:creator

OBO_REL:is_a

has_measurement

OBO_REL:posited_by

PHENO:exhibits

Phenotype
(class expression)

OBO_REL:influences

Genotype
uid = ZFIN ID

OBO_REL:towards

OBO_REL:inheres_in

{TAO,ZFA}:entity
uid = {TAO,ZFA} ID

OBO_REL:variant_of

PHENO:asserted_for_otu

PHENO:has_taxon

OBO_REL:posited_by

{TAO,ZFA}:entity
uid = {TAO,ZFA} ID

Gene
uid = ZFIN ID

OBO_REL:posited_by

CDAO:TU
name = Publication Taxon

CDAO:CharacterStateDatum

CDAO:has_State

CDAO:CharacterStateDomain
name = state text

CDAO:has_TU

PHENO:has_comment

dwc:individualID

CDAO:CharacterStateDataMatrix

CDAO:has_Character

CDAO:has_Datum

PHENO:has_comment

comments
(literal text)

Specimen

CDAO:Character
name = character text

comment
(literal text)

PHENO:has_publication

PHENO:has_comment

PHENO:has_comment

dwc:catalogID

dwc:collectionID

PHENO:Publication
-dc:abstract
-dc:bibliographicCitation
-dc:date

publication notes
(literal text)

comment
(literal text)

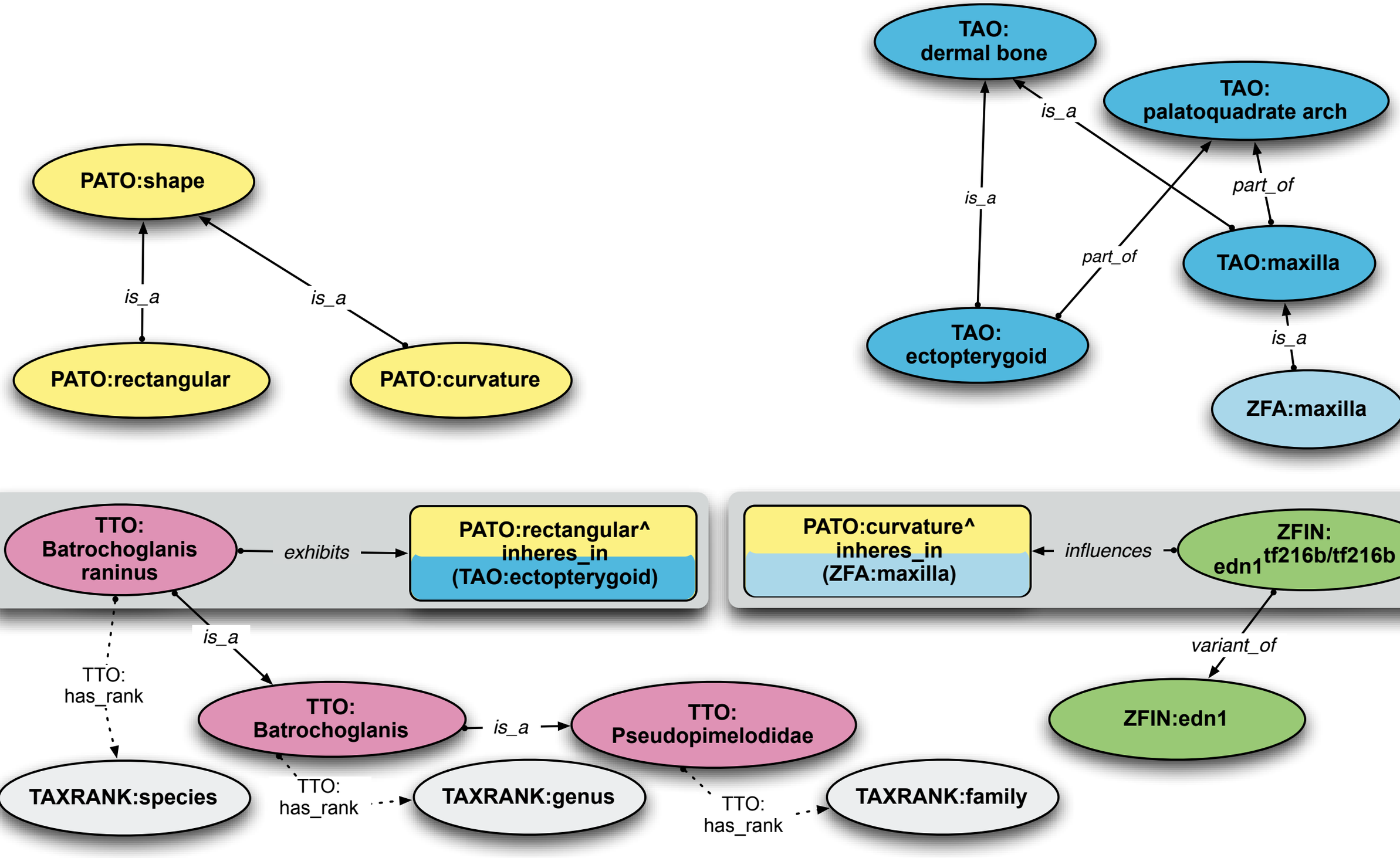
catalog number
(literal text)

COLLECTION

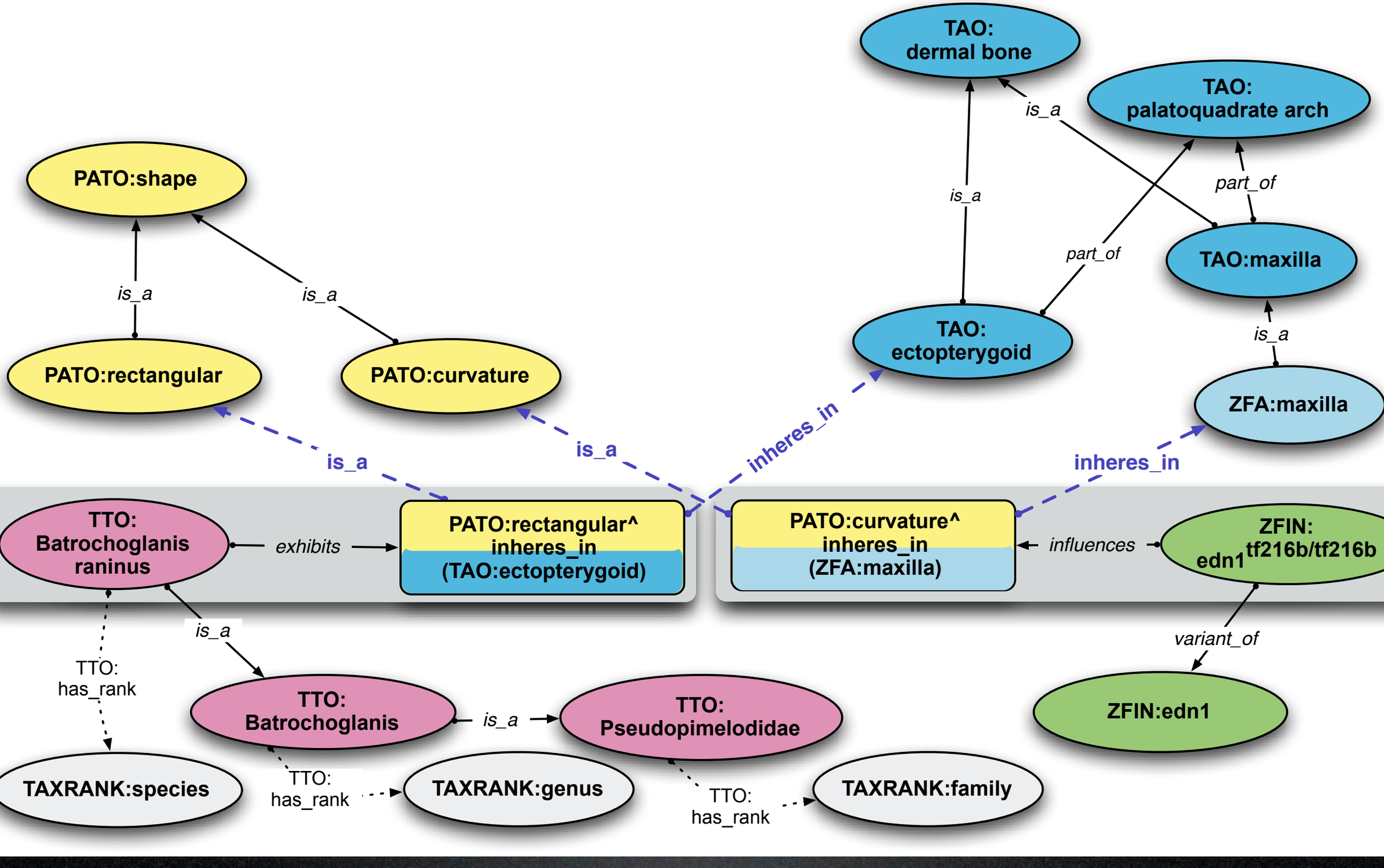
How does reasoning work?

How does reasoning work?

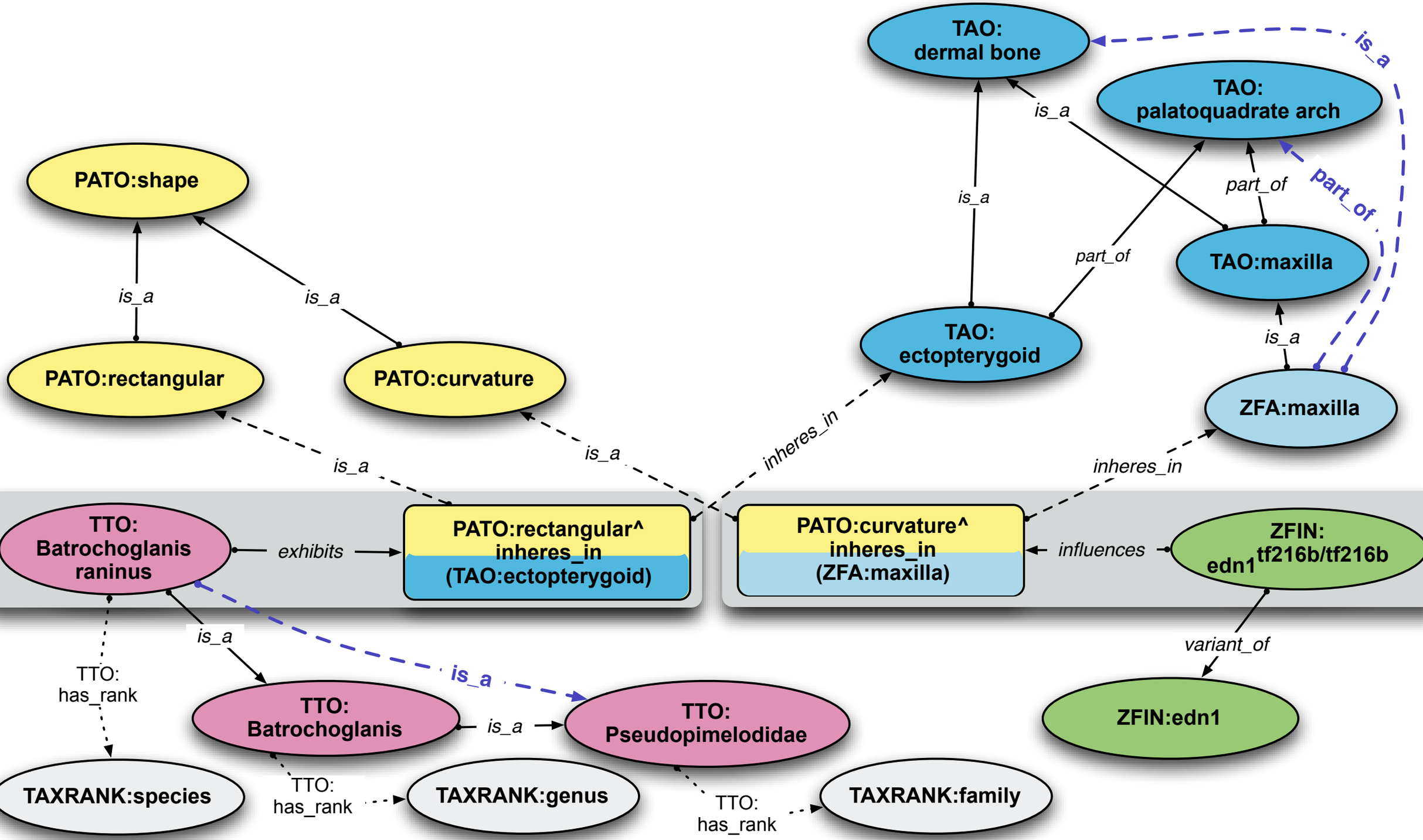
How does reasoning work?



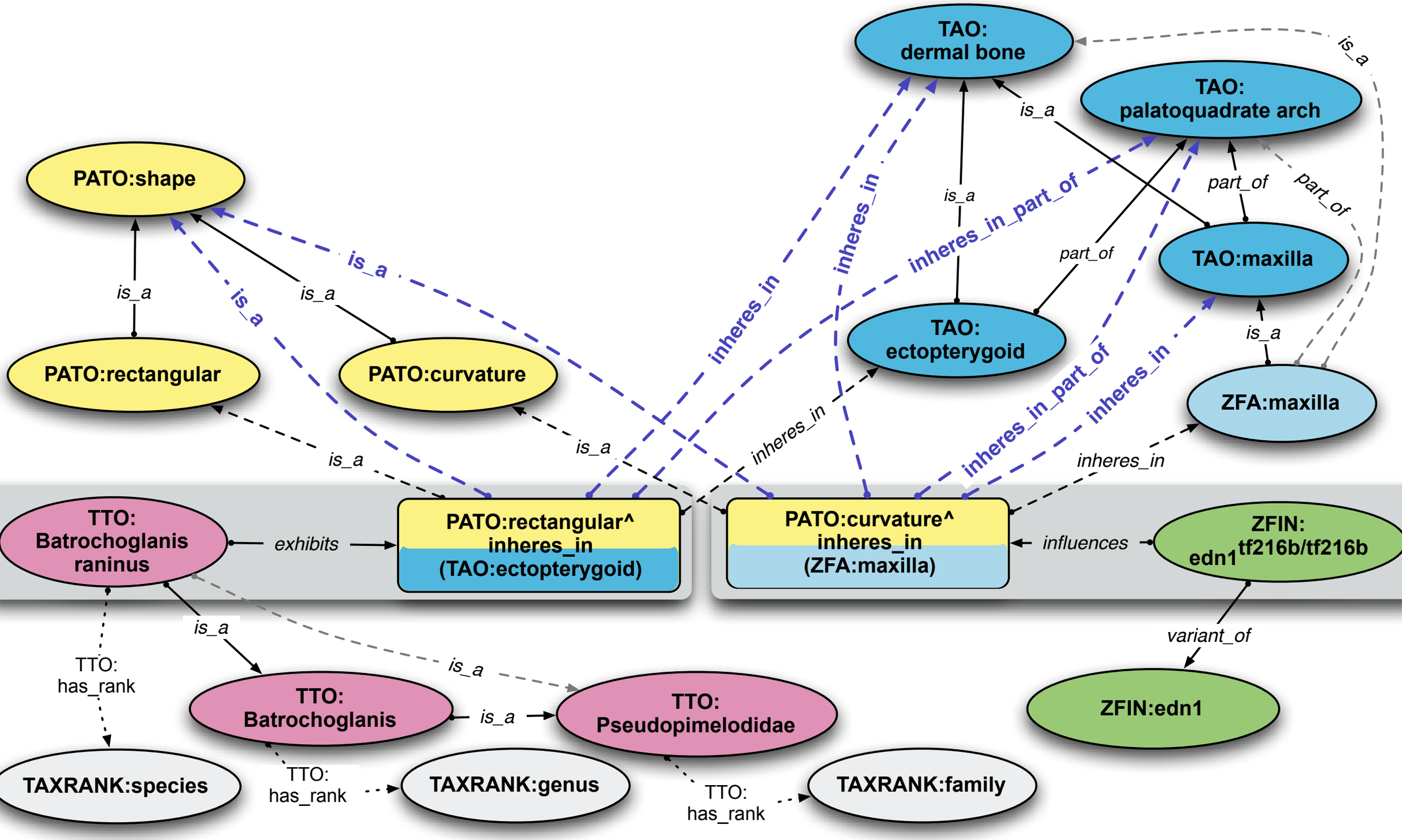
How does reasoning work?



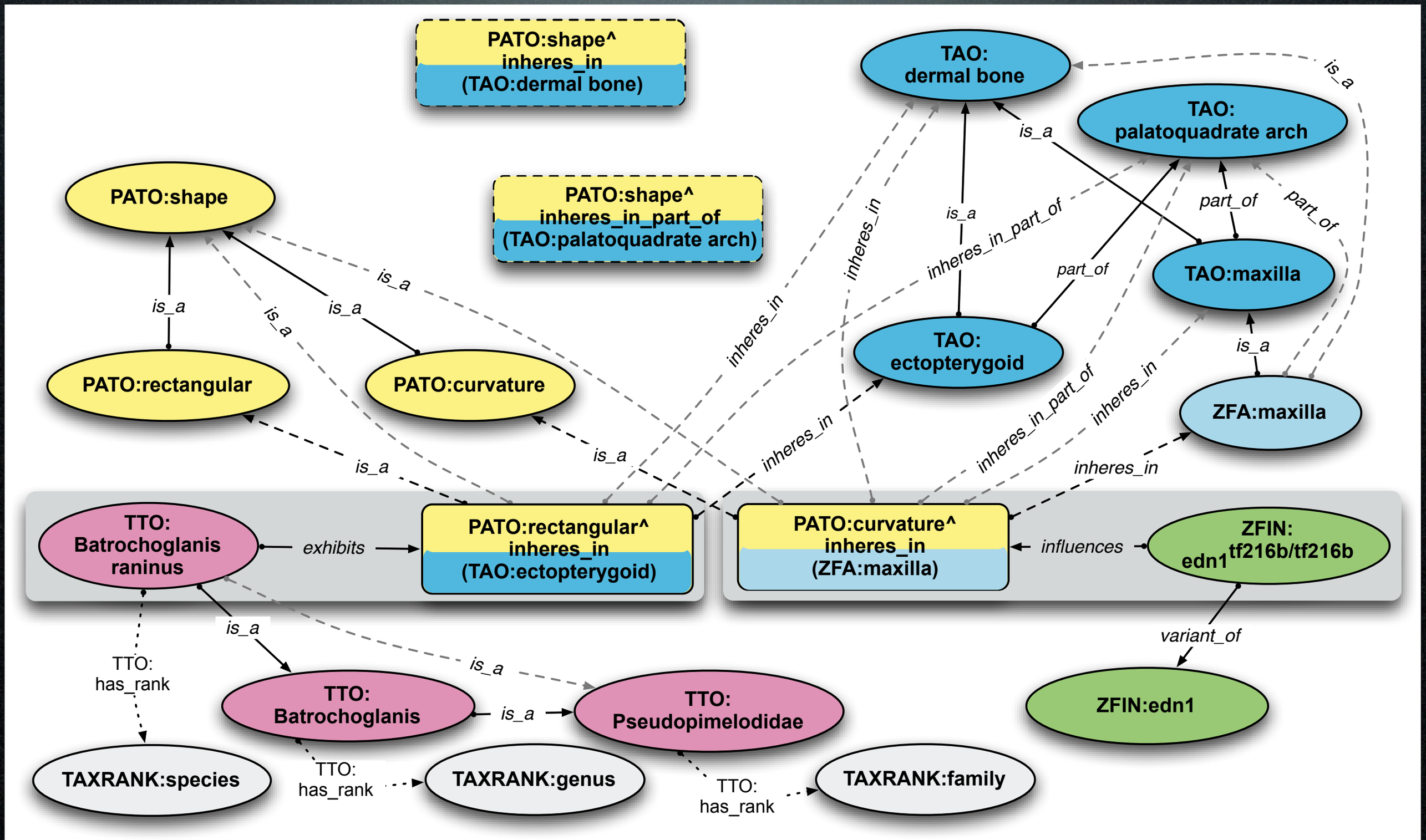
How does reasoning work?



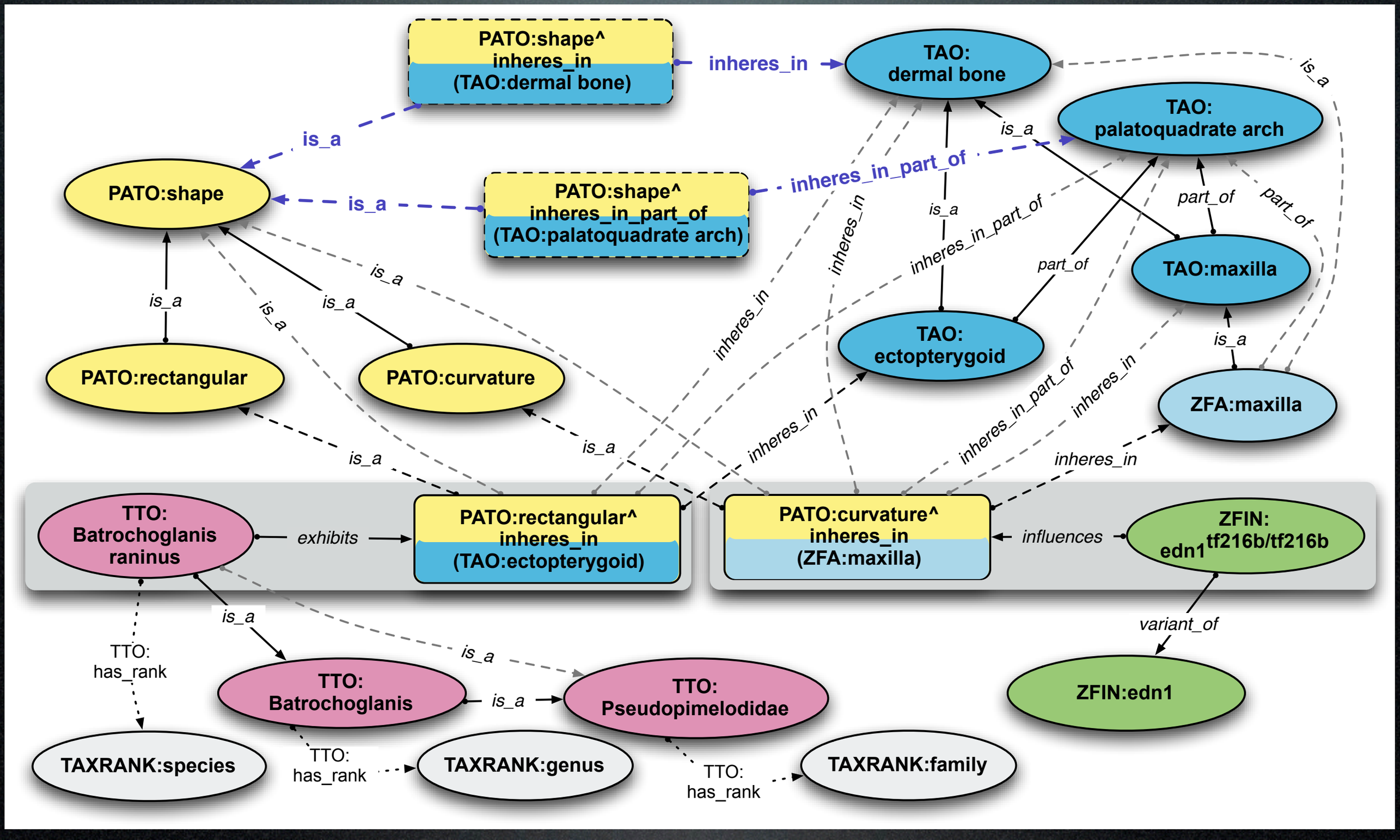
How does reasoning work?



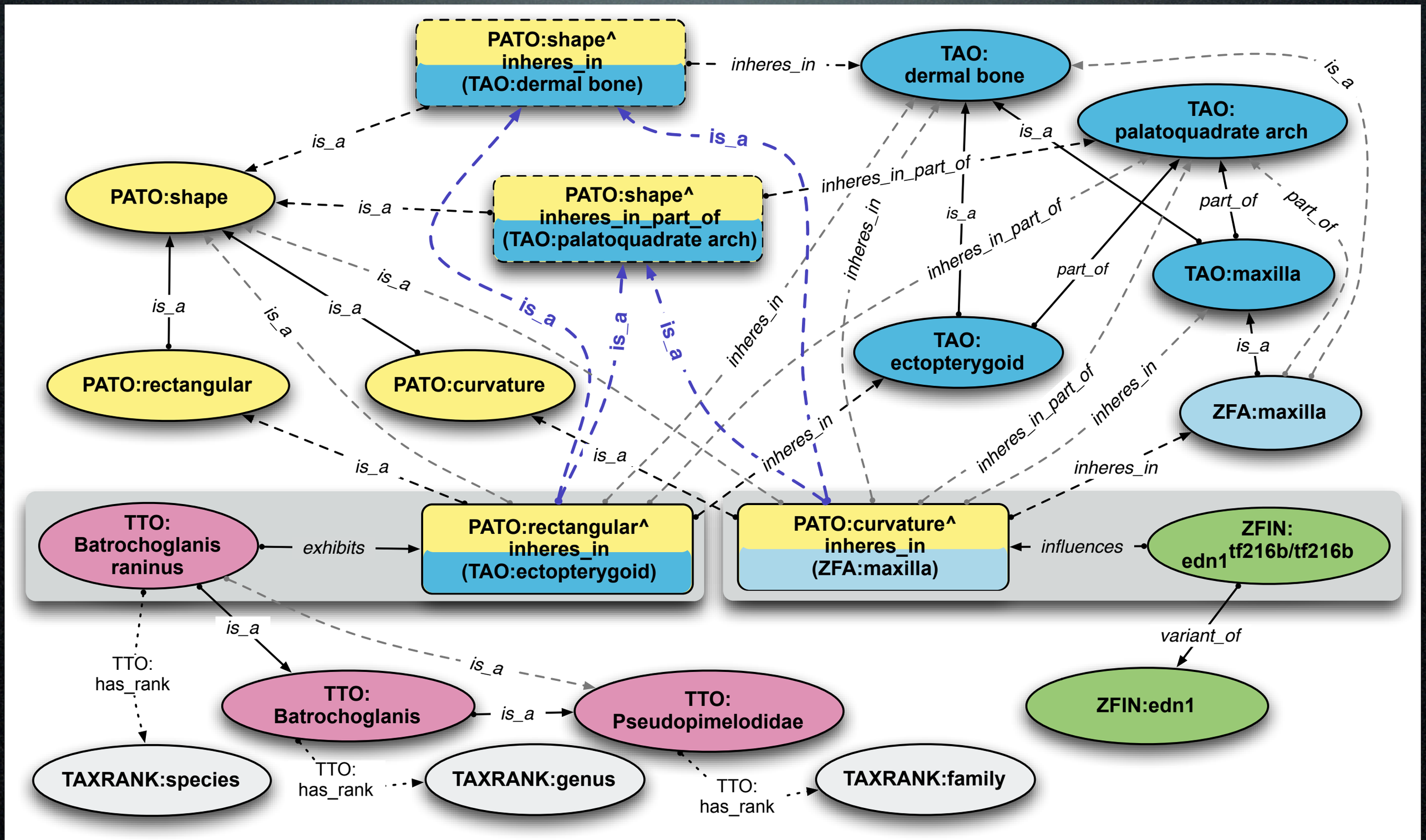
How does reasoning work?



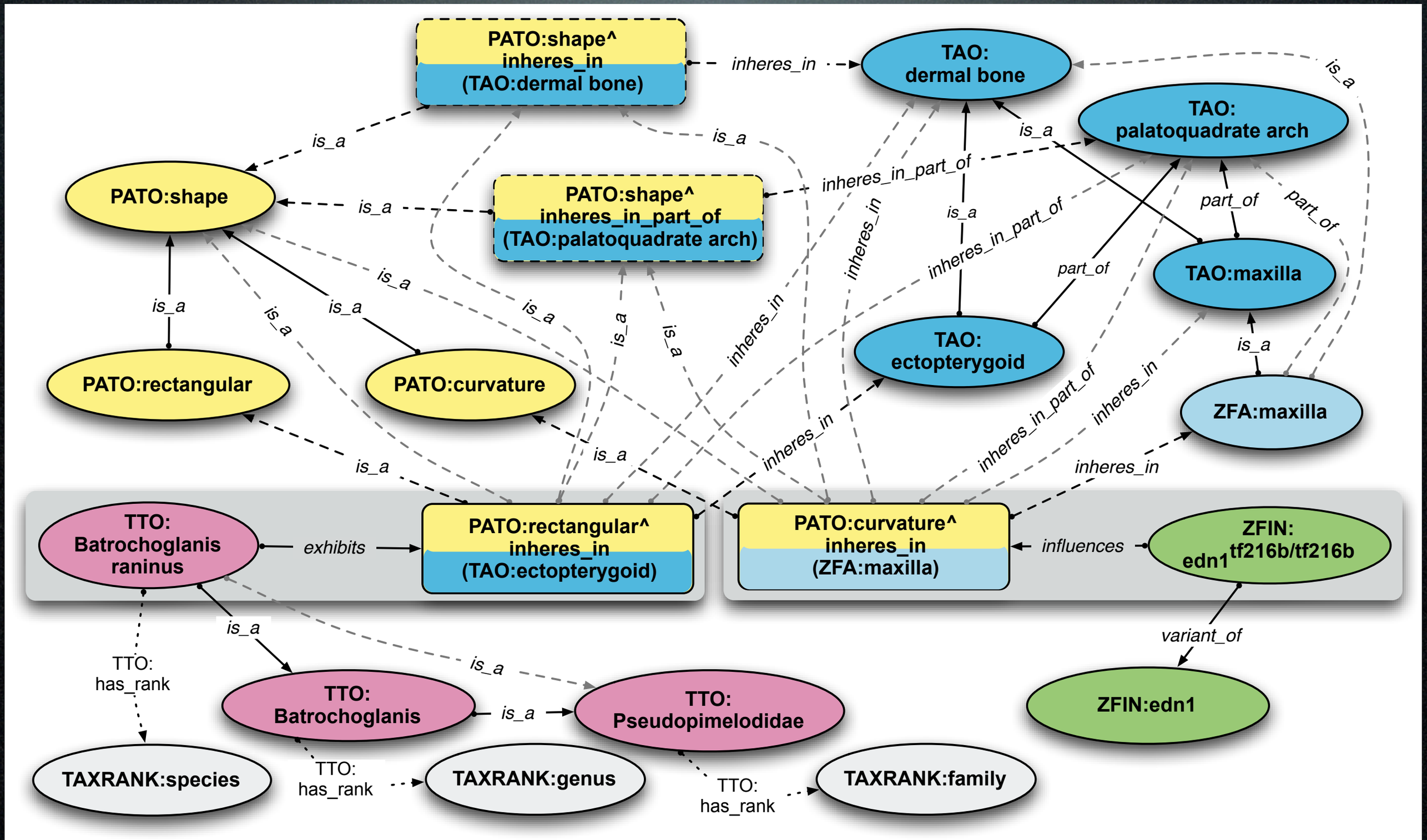
How does reasoning work?



How does reasoning work?

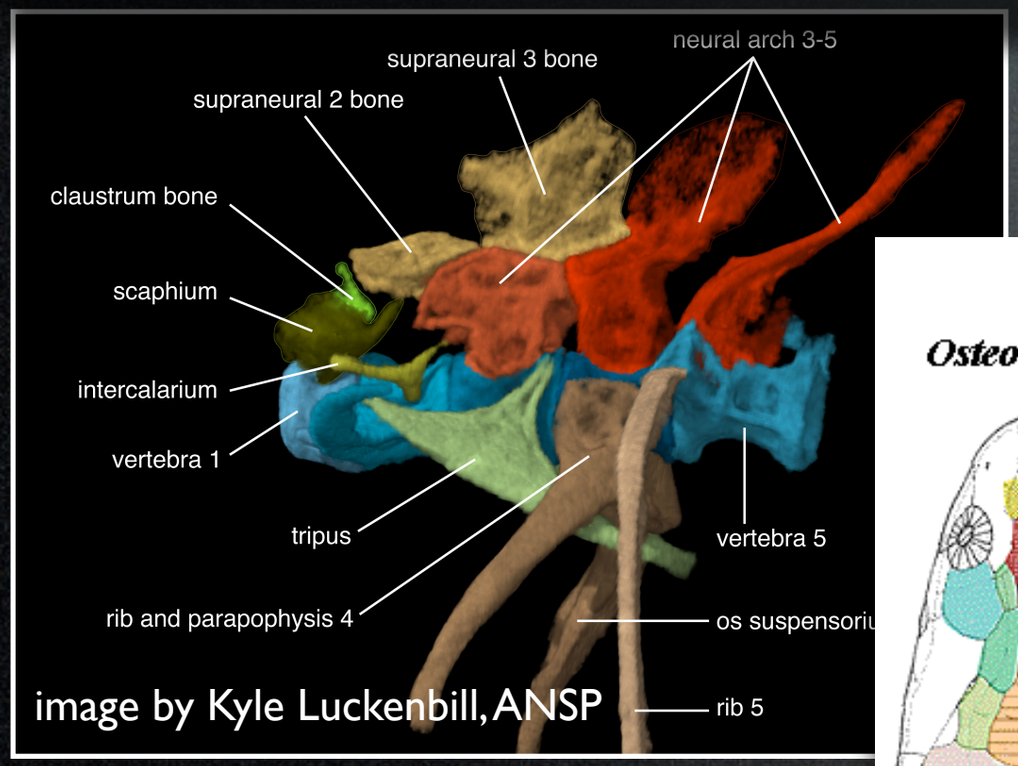


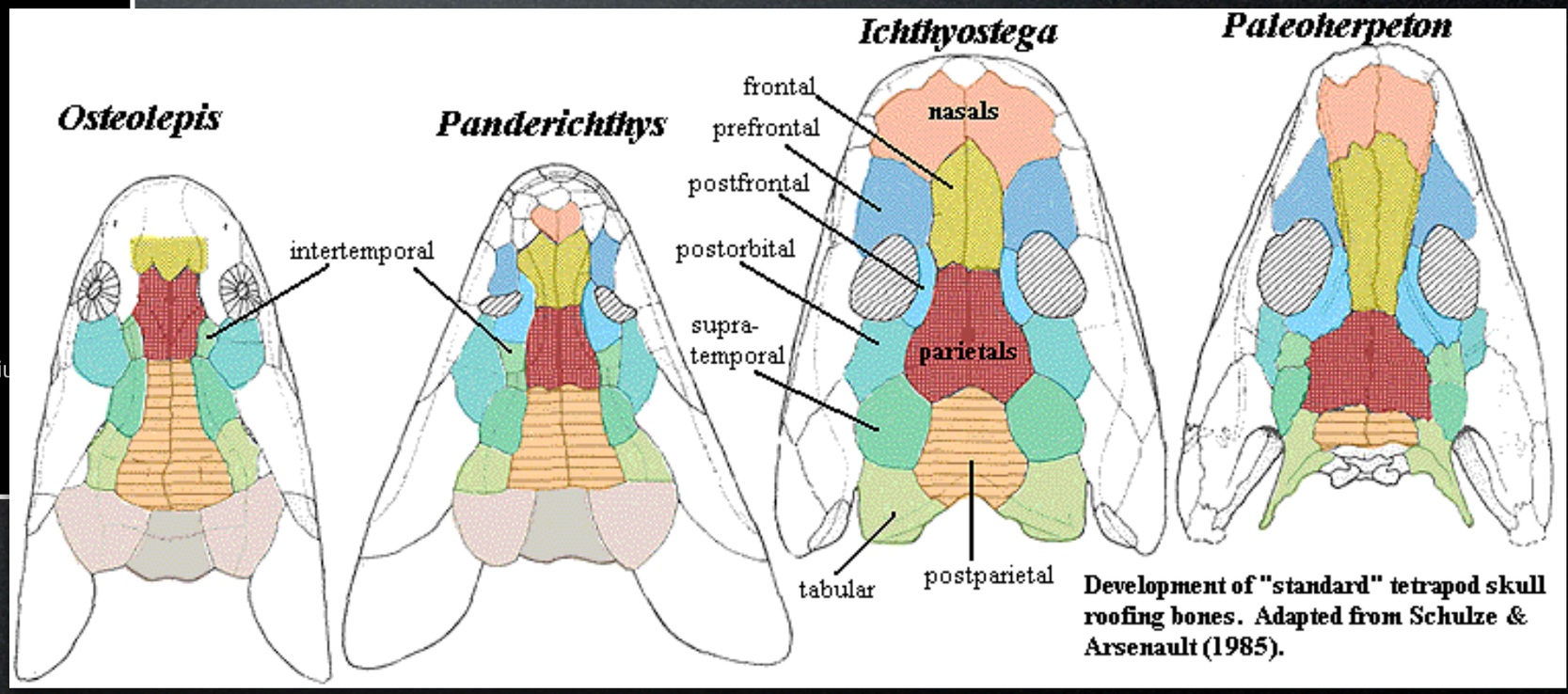
How does reasoning work?



Future Directions: 1. Reasoning over homology

Entity 1	Taxon 1	Relationship	Entity 2	Taxon 2	Evidence	Reference(s)
scaphium	Otophysi	homologous_to	neural arch 1	Teleostei	IDS, IMS, IPS	(Fink and Fink, 1981; Rosen and Greenwood, 1970)
intercalarium	Otophysi	homologous_to	neural arch 2 (ventral portion)	Teleostei	IDS, IMS, IPS	(Rosen and Greenwood, 1970)
intercalarium	Otophysi	homologous_to	neural arch 2	Teleostei	NAS	(Fink and Fink, 1981)
intercalarium	Otophysi	homologous_to	neural arch 2	Teleostei	IMS	(Hora, 1922)
intercalarium	Otophysi	homologous_to	rib of vertebra 2	Teleostei	TAS	(Hora 1922)
tripus	Otophysi	homologous_to	parapophysis + rib of vertebra 3	Teleostei	IDS, IMS, IPS	(Fink and Fink, 1981; Rosen and Greenwood, 1970)

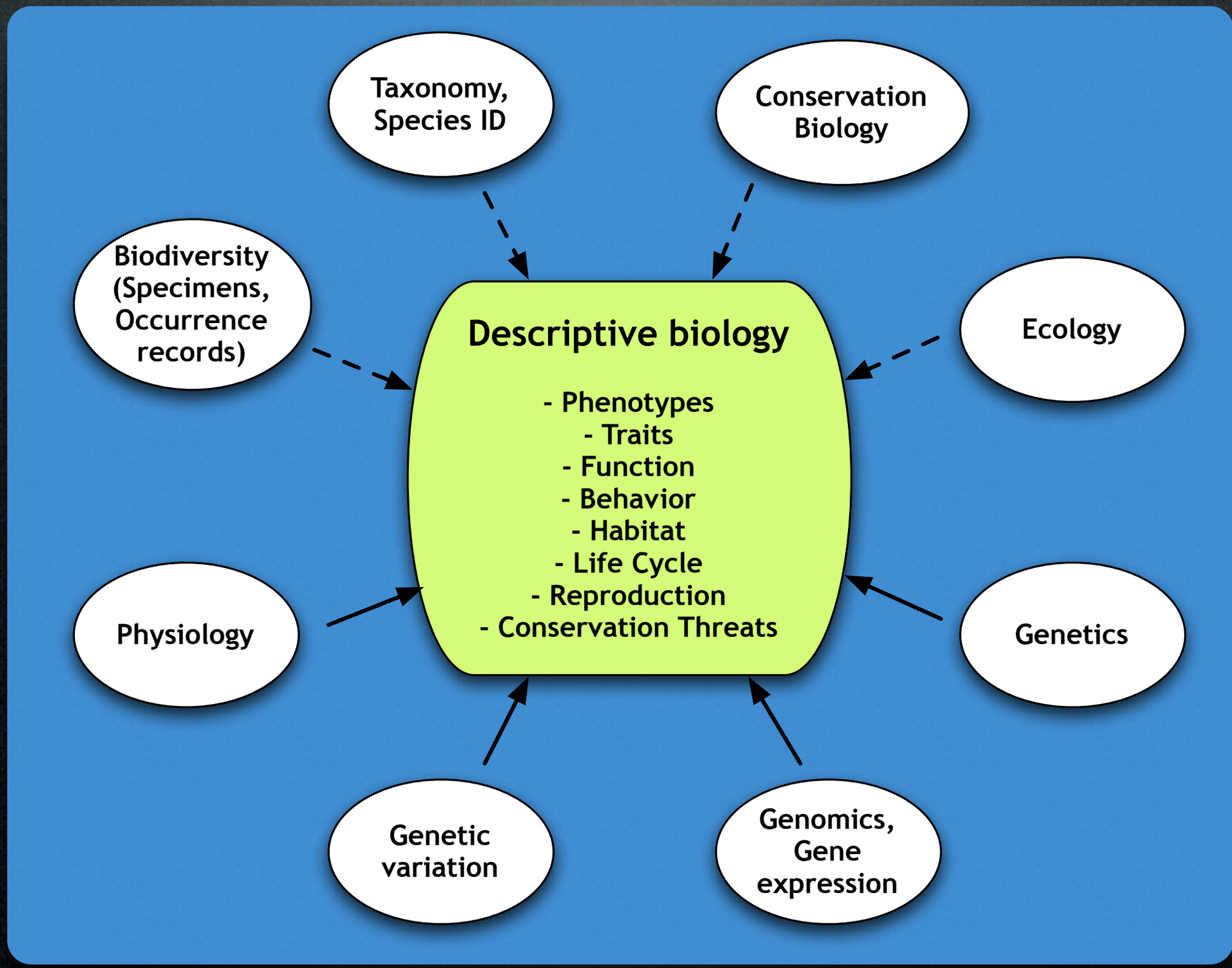




What next?

- Modeling and reasoning over homology
- Efficient searching and scoring of semantic similarity
- Reducing the bottlenecks in data curation

Opening descriptive biological data to computing can enable new science



Acknowledgements

- Phenoscape Personnel & PIs: P. Mabee, M. Westerfield, T. Vision, J. Balhoff, C. Kothari, W. Dahdul, P. Midford
- Phenoscape curators & workshop participants
- Berkeley Bioinformatics & Ontologies Project (BBOP): C.Mungall, S.Lewis
- National Evolutionary Synthesis Center (NESCent)
- NSF (DBI 0641025)

Phenotypic similarity matches taxa to candidate genes

Similarity (IC)	Taxon (subsuming taxon with variable phenotypes in subsumed taxa)	Taxon phenotype (one of two or more subsumed variable taxon phenotypes)	Candiate Gene(s) (zebrafish)	Gene phenotype	Subsuming phenotype
15.16	*Danio*	*Danio rerio*: epural separated from urostyle	trpm7	epural, composition	epural, structure
14.45	Otophysi	Siluriformes: scales, absent	eda	scales, absent	scales, count
13.25	Siluriformes	Siluriformes: basihyal cartilage, absent	brpf1, disc1 and 10 more	basihyal cartilage, absent	basihyal cartilage, count
10.0	*Tachysurus*	process of Meckel's cartilage, adjacent to coronoid process	edn1, foxd3 and 22 more	Meckel's cartilage, mislocalized posteriorly	Meckel's cartilage, position

Mapping EQs back to characters is a challenge

- Properties of “good” phylogenetic characters:
  - Exclusivity of states
  - Distinguishability of states
  - Independence of characters
- Finding exclusive states requires incompatible phenotypes. How to determine incompatibility?
  - Two phenotypes are incompatible iff they cannot both inhere in the same specimen.
  - Two qualities are incompatible iff an entity cannot bear both.

Which EQs and qualities are incompatible?

- Incompatible Qs
  - present vs. absent
  - triangular vs. round
  - absent vs. any other quality
- Incompatible EQs
  - (Q inheres_in bone E) vs (cartilage E absent)
- Compatible Qs
  - present vs. any other quality (except absent)
  - serrated vs. round
  - some colors

Detecting phenotype change and variation

Hemiodus argenteus

{**shape:bent** *inheres_in* supraorbital bone,
count:absent *inheres_in* upper pharyngeal 5 tooth}

Hemiodus

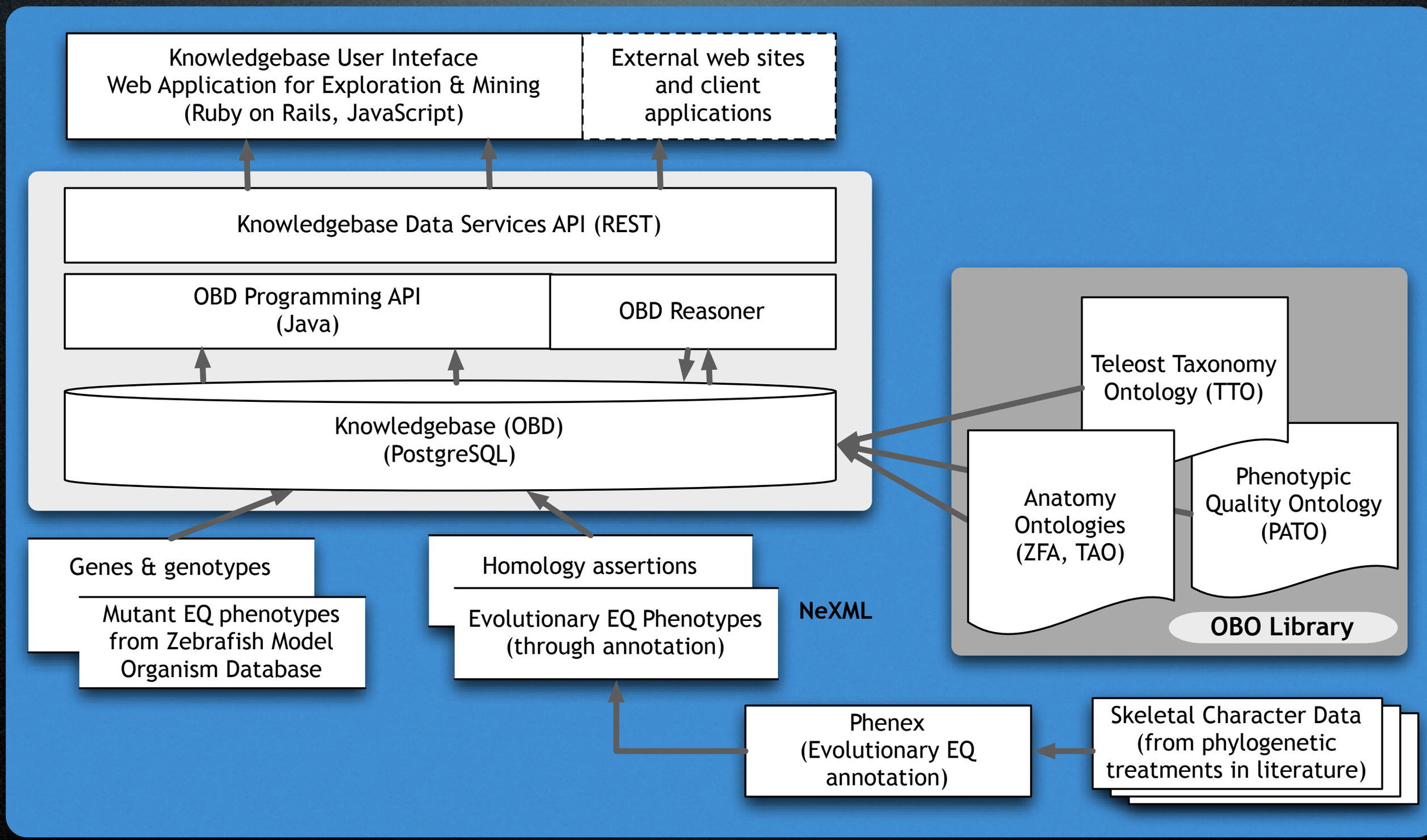
{**shape:bent** *inheres_in* supraorbital bone,
shape:straight *inheres_in* supraorbital bone,
count:absent *inheres_in* upper pharyngeal 5 tooth,
count:present *inheres_in* upper pharyngeal 5 tooth}

Hemiodus unimaculatus

{**shape:straight** *inheres_in* supraorbital bone,
count:present *inheres_in* upper pharyngeal 5 tooth}

{Change in: **shape** *inheres_in* supraorbital bone,
Change in: **count** *inheres_in* upper pharyngeal 5 tooth}

System architecture



Formalizing homology relationships

- Formal pattern is ternary:

  `E1 in_taxon T1 homologous_to E2 in_taxon T2 as E3 in_taxon T3`

- Classifying homology relationships
  - 1-1 homology (phylogenetic homology)
  - serial homology

- `A iso_homologous_to B as C`
  `⇒ all A derived_by_descent_from some`
  `(C and has_derived_by_descendent some B)`
  `and`
  `all B derived_by_descent_from some`
  `(C and has_derived_by_descendent some A)`

- `shares_ancestor_with` as a relation chain:
  `derived_by_descent_from o has_derived_by_descendent`

Option 1: Asserting homology at higher-level taxa

homology-relations (http://purl.obolibrary.org/obo/homology-relations) - [/Users/lapp/Devel/phenoscape/trunk/vocab/homology/utest_homol-exp-mod.owl]

homology-relations (http://purl.obolibrary.org/obo/homology-relations)

Active Ontology | Entities | Classes | Object Properties | Data Properties | Individuals | OWLViz | DL Query

Asserted class hierarchy | Inferred class hierarchy

Asserted class hierarchy: 'forelimb Mammalia'

- Thing
  - file:/Users/lapp/Devel/phenoscape/trunk/voca
  - root
  - taxonomic_rank
  - 'anatomical entity'
    - 'material anatomical entity'
      - 'anatomical structure'
        - 'multi-cellular organism'
        - 'organism subdivision'
          - wing
            - 'wing Aves'
            - 'wing Gallus gallus'
          - appendage
            - limb/fin
              - limb
                - forelimb
                  - 'forelimb Mammalia'
                  - 'forelimb Mus musculus'
                  - 'forelimb Homo sapiens'
                - hindlimb
                  - 'hindlimb Mus musculus'
                  - 'hindlimb Gallus gallus'
                  - 'hindlimb Homo sapiens'
            - 'pelvic spur'
              - 'pelvic spur Python'
          - 'anterior region of body'
          - 'posterior region of body'
        - 'vestigial structure'

Usage: 'forelimb Mammalia'

Show: this disjoints named sub/superclasses

Found 5 uses of 'forelimb Mammalia'

- 'forelimb Mammalia' equivalentTo forelimb and PHENOSCAPE_in_taxon some Mammalia
- 'forelimb Mammalia' *label* "forelimb Mammalia"@en
- Class('forelimb Mammalia')
- 'forelimb Mammalia' subClassOf derived_by_descent_from some (has_derived_by_descendant some 'forelimb Mammalia' and has_derived_by_descendant some 'wing Aves')
- 'wing Aves'
- 'wing Aves' subClassOf derived_by_descent_from some (has_derived_by_descendant some 'forelimb Mammalia' and has_derived_by_descendant some 'wing Aves')

Description: 'forelimb Mammalia'

Equivalent classes

- forelimb and PHENOSCAPE_in_taxon some 'Mammalia'

Superclasses

- derived_by_descent_from some (has_derived_by_descendant some 'wing Aves' and has_derived_by_descendant some 'forelimb Mammalia')
- forelimb

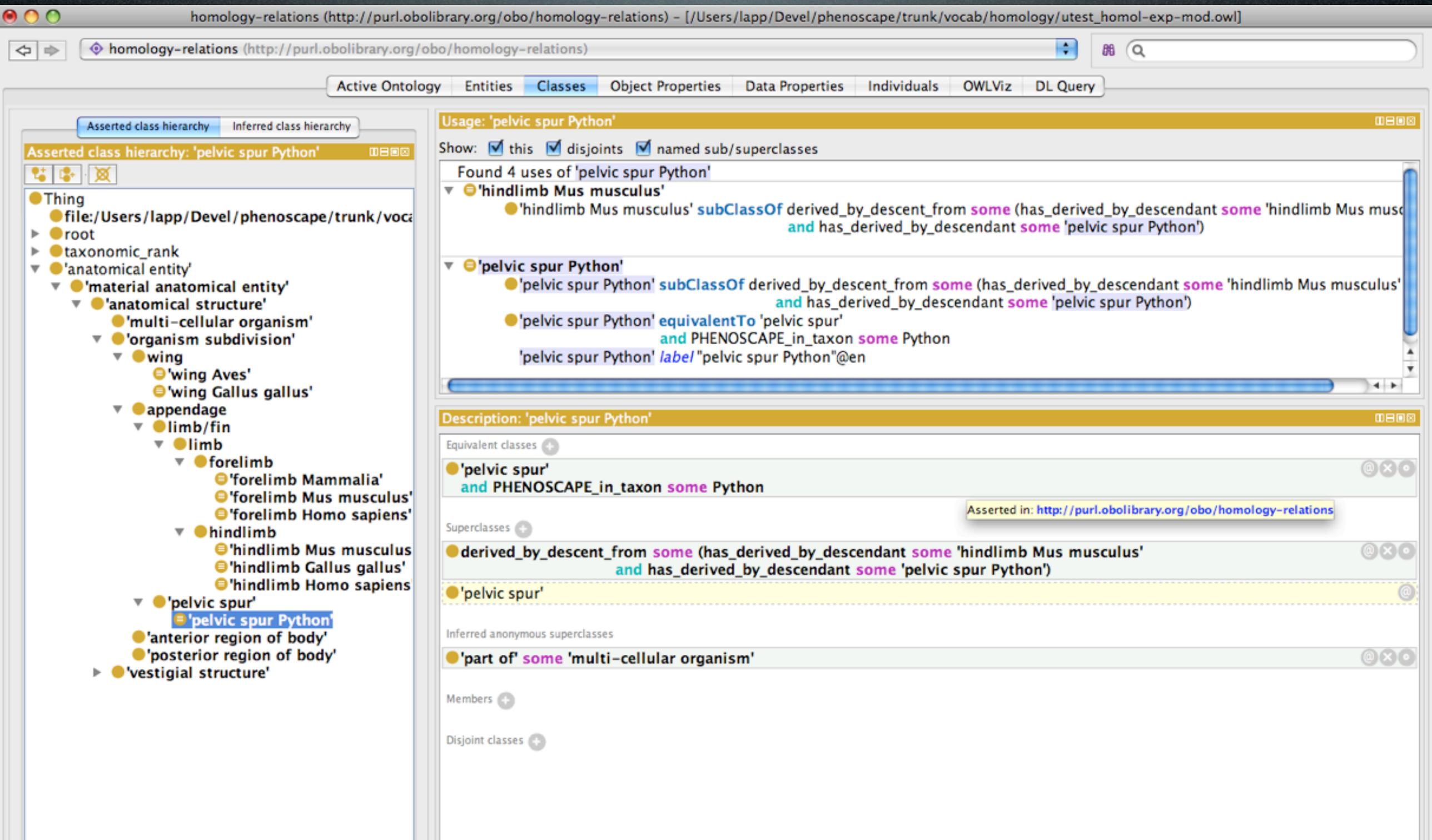
Inferred anonymous superclasses

- 'part of' some 'multi-cellular organism'
- 'part of' some 'anterior region of body'
- anterior_to some hindlimb

Members

Disjoint classes

Option 2: Asserting homology at species level



Validation through standard OWL-DL reasoning

Query (class expression)

shares_ancestor_with some ('forelimb' and PHENOSCAPE_in_taxon some 'Amniota')

Execute | Add to ontology

Query results

Sub classes (2)

- 'forelimb Mammalia'
- 'wing Aves'

Descendant classes (5)

- 'forelimb Homo sapiens'
- 'forelimb Mammalia'
- 'forelimb Mus musculus'
- 'wing Aves'
- 'wing Gallus gallus'

Query (class expression)

shares_ancestor_with some ('hindlimb' and PHENOSCAPE_in_taxon some 'Amniota')

Execute | Add to ontology

Query results

Sub classes (4)

- 'hindlimb Gallus gallus'
- 'hindlimb Homo sapiens'
- 'hindlimb Mus musculus'
- 'pelvic spur Python'

Descendant classes (4)

- 'hindlimb Gallus gallus'
- 'hindlimb Homo sapiens'
- 'hindlimb Mus musculus'
- 'pelvic spur Python'