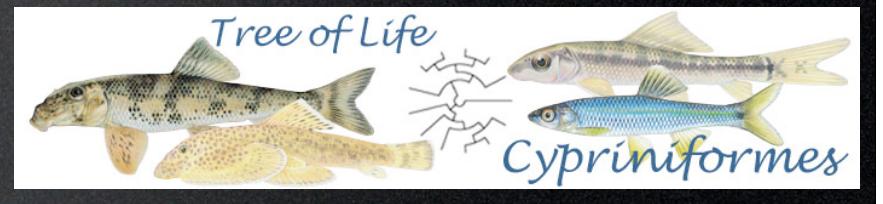
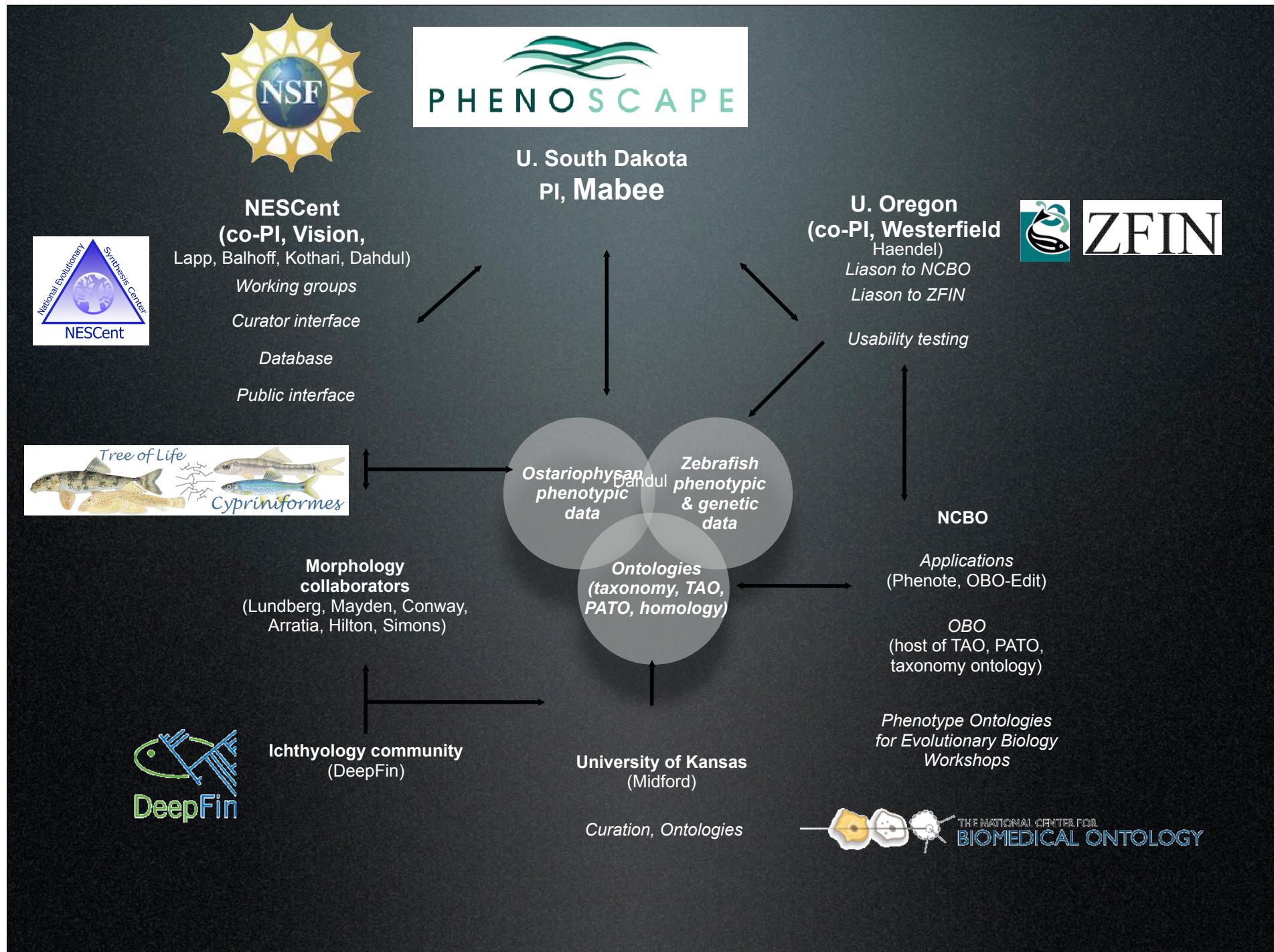


Phenoscape: Bridging systematics and genetics with ontologies

Paula Mabee
University of South Dakota

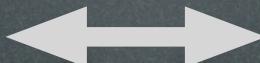






Goal: Connect these data using ontologies

Model
organisms
(biomed)



Comparative
evolutionary
biology

- Phenotypes
- Genetics

zebrafish

- Morphology
- Phylogeny

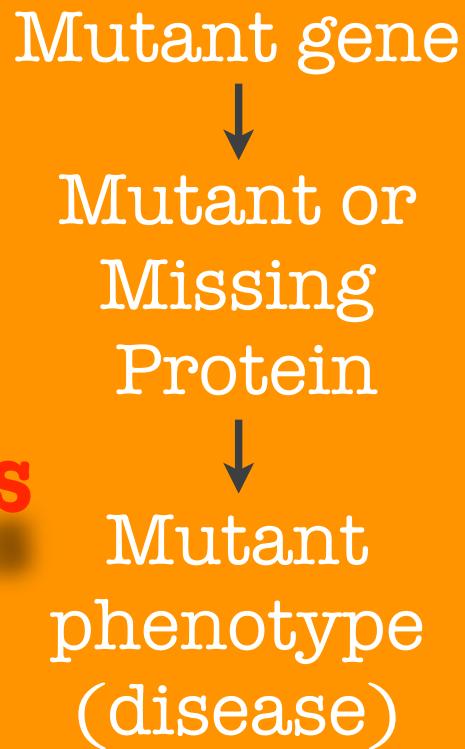
ostariophysan fishes

Ontologies can connect human disease to candidate genes

Animal models



Humans

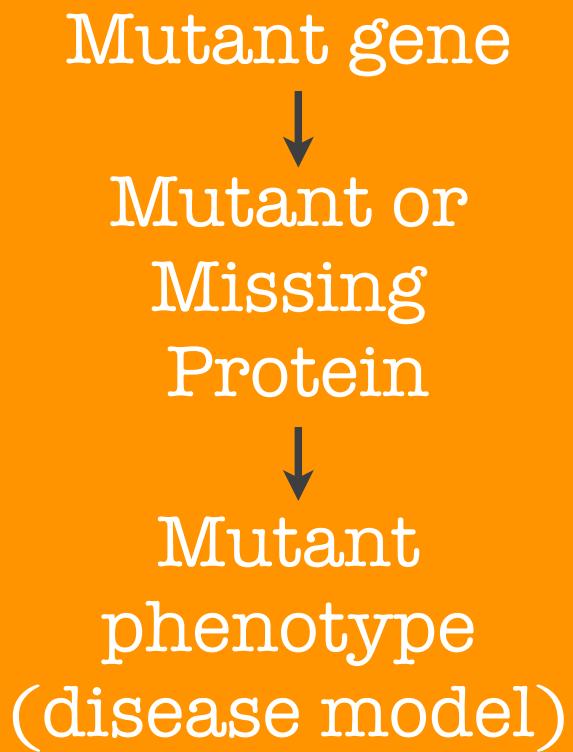


ONTOLOGIES

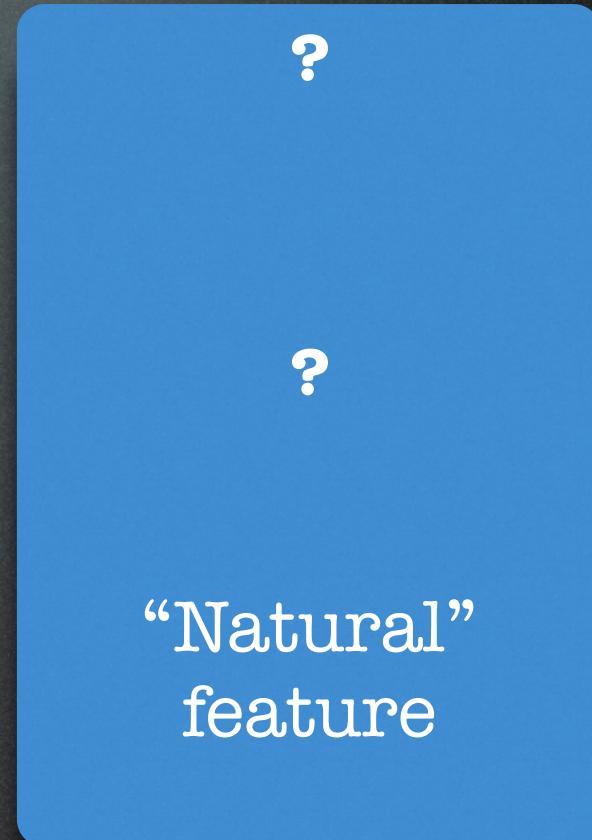


Ontologies can connect species variation to candidate genes

Animal models

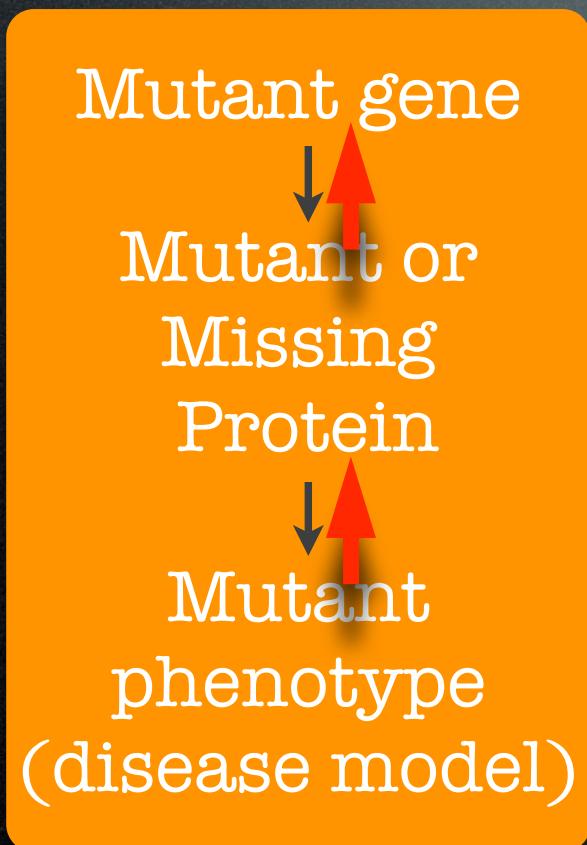


Evolutionary Species



Ontologies can connect species variation to candidate genes

Animal models



Evolutionary Species

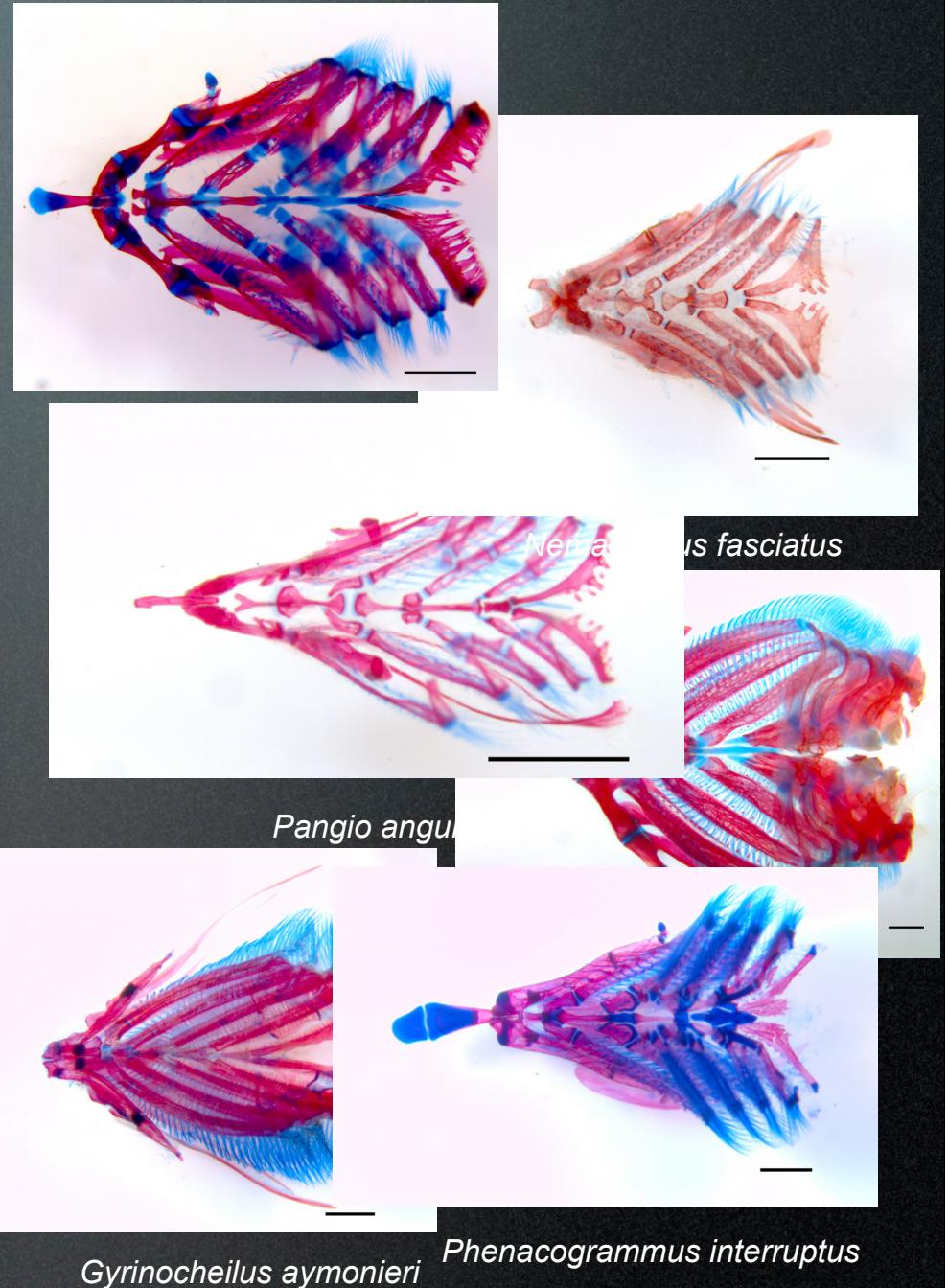
Candidate gene

Natural phenotype



Phenoscape goals:

- identify taxa in which specific features vary
- identify candidate genes for evolutionary changes
- discover similar phenotypes and similar OTUs (phenotypic BLAST)
- correlation matrices of traits & genes



E.g. Basihyal lost in catfishes
(Basihyal primitively present)



Sample query: What are all the possible genes underlying evolutionary loss in the basihyal?

Results

GENES

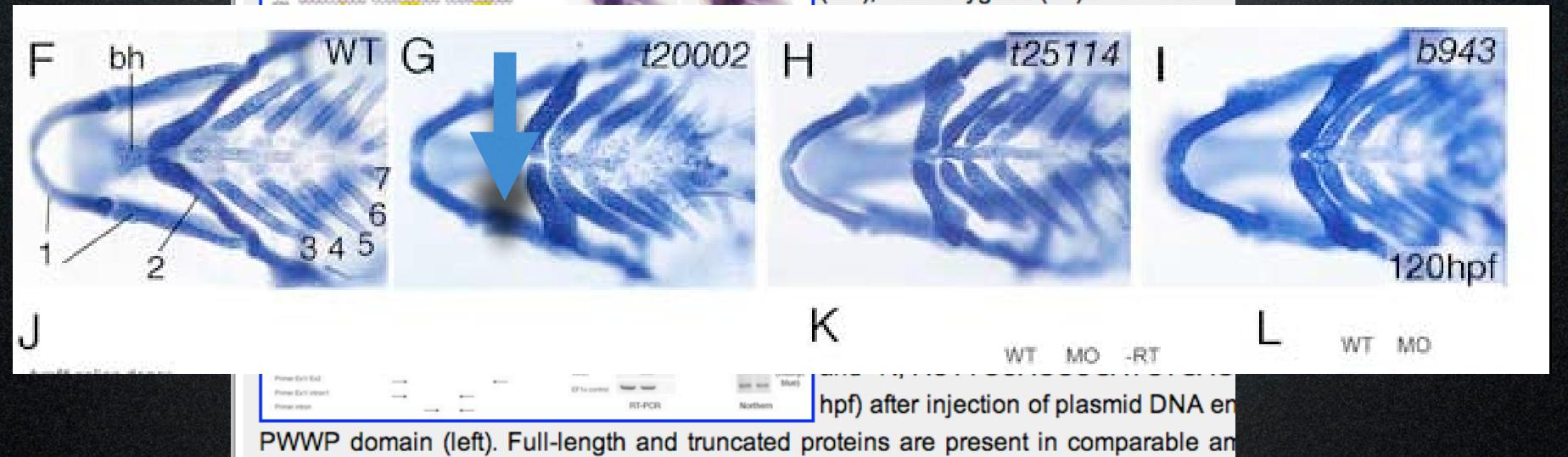
ANATOMY

IMAGES



PHENOTYPE affecting basihyal
Mutant and Transgenic Lines

Genotype (Background)	Affected Genes	Phenotype	Figures
<u>brpf1</u> ^{b943/b943}	<u>brpf1</u>	absent, hypoplastic	<u>2 figures</u> from 1 publication
<u>brpf1</u> ^{t20002/t20002}	<u>brpf1</u>	present	<u>4 figures</u> from 1 publication
<u>brpf1</u> ^{t25114/t25114}	<u>brpf1</u>	absent, hypoplastic	<u>2 figures</u> from 1 publication
<u>sox9a</u> ^{tw37/+}	<u>sox9a</u>	aplastic	<u>1 figure</u> from 1 publication
<u>sox9a</u> ^{tw37/tw37}	<u>sox9a</u>	aplastic	<u>1 figure</u> from 1 publication



Basihyal loss in zebrafish due to mutation in brpf1

ZFIN ID: ZDB-FIG-080604-30

[Laue et al., 2008](#) - The multidomain protein Brpf1 binds histones and is required for Hox gene expression and segmental identity.
Development 135(11):1935-1946 - [Full text @ Development](#)

Evolutionary loss of
basihyal in catfishes due
to changes in this gene:
brpf1?

Requirements:

- **Develop ontologies**
- Develop appropriate syntax
- Curate phenotypes (characters)

Traditional comparative study: survey morphological variation



Cyprinus carpio



Pangio anguillaris



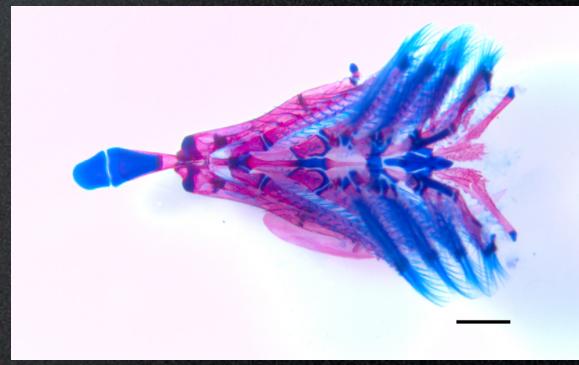
Nemacheilus fasciatus



Catostomus commersoni



Gyrinocheilus aymonieri



Phenacogrammus interruptus

Morphological data in free text format

- not a computable format
- cannot be compared among species
- cannot be linked to underlying genetics
- cannot be reasoned across

APPENDIX 1: CHARACTERS USED FOR PHYLOGENETIC ANALYSIS

Unless otherwise indicated, terminology follows Ronquist (1995a) and Ronquist and Nordlander (1989). Transformation series hypotheses are given for multi-state characters. Following each character is the character's consistency index and retention index on the preferred tree (Fig. 4). Observed character states are given in Table 3.

General Body Sculpture

1. Microsculpture on vertex, lateral surface of pronotum and mesoscutum: (0) absent, surface not dull (Figs. 9A–9D and 10A–10C); (1) present, linear, making the surface dull (not illustrated): (CI = 1.00, RI = 1.00, goodness of fit (G-fit) = 10).

Head

2. Shape of head in anterior view: (0) rounded, approximately as high as broad (Figs. 8A, 8B, and 9C); (1) elongate, higher than broad (Figs. 8C, 8D, 9A, and 9B): (CI = 0.25, RI = 0.82, G-fit = 5).

3. Relative position of eye: (0) close to ocelli, ratio of distance between compound eye and posterior mandibular articulation to distance between posterior ocellus and compound eye ≥ 1.18 (Figs. 8B and 8C); (1) removed from ocelli, ratio ≤ 1.13 (not illustrated); (CI = 0.20, RI = 0.50, G-fit = 4.3).

4. Size of ocelli: (0) small, ratio of maximum diameter of a lateral ocellus to shortest distance between lateral ocelli 0.22–0.40 (not illustrated); (1) large, ratio 0.44–0.65 (Figs. 8B and 8D); (CI = 0.11, RI = 0.62, G-fit = 2.7).

(Figs. 8B and 8D); (1) long (not illustrated); (CI = 0.20, RI = 0.33, G-fit = 4.3).

7. Shape of compound eyes in dorsal view: (0) rounded, distinctly protruding from the surface of the head, particularly anteriorly (Figs. 8B and 8D); (1) less rounded, not distinctly protruding from the surface of the head (not illustrated); (CI = 0.25, RI = 0.73, G-fit = 5).

8. Lateral frontal carina: (0) absent (Fig. 8D); (1) present (Fig. 8B, more easily seen in dorsal view); (CI = 0.50, RI = 0.50, G-fit = 7.5).

9. Hair punctures on lateral part of vertex: (0) indistinct or absent (Figs. 8B and 8D); (1) present, distinctly enlarged (not illustrated); (CI = 0.33, RI = 0.60, G-fit = 6).

10. Sculpture on posterior part of vertex (seen in dorsal view, not illustrated): (0) smooth or punctate, without linear component; (1) with parallel or slightly radiating, transverse strigae; (CI = 0.50, RI = 0.75, G-fit = 7.5).

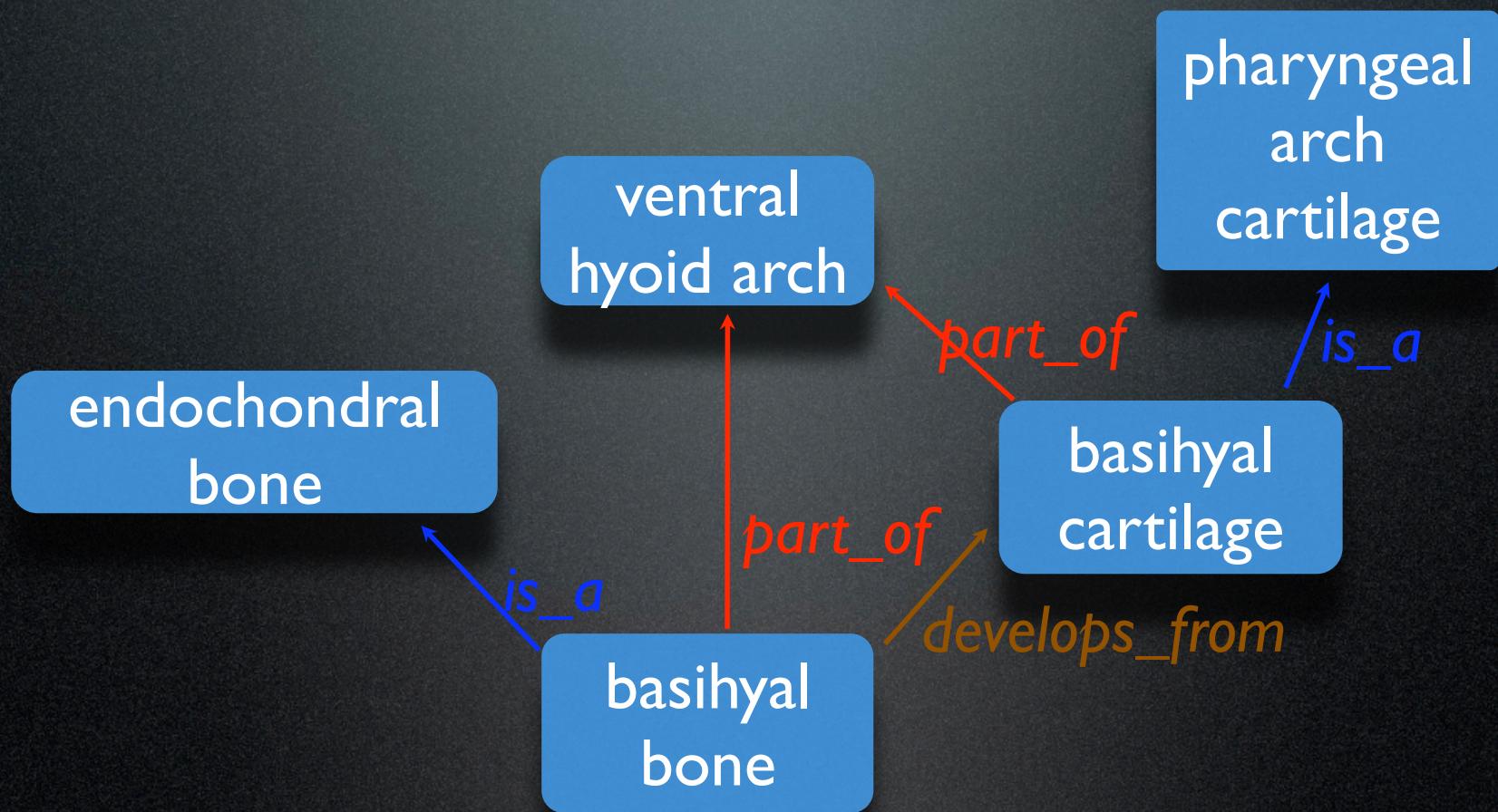
11. Relative position of antennal sockets: (0) close to ocelli; ratio of vertical distance between inner margin of antennal foramen and ventral margin of clypeus to vertical distance between anterior ocellus and antennal rim < 2.0 (not illustrated); (1) intermediate, ratio 2.25–4.1 (Figs. 8B and 8D); (2) far from ocelli, ratio > 4.4 (not illustrated). Ordered 012; (CI = 0.08, RI = 0.33, G-fit = 12).

12. Vertical carina adjacent to ventral margin of antennal socket: (0) absent (Fig. 8B); (1) present (Fig. 8D); (CI = 0.50, RI = 0.86, G-fit = 7.5).

13. Vertical delineations on lower face: (0) absent (Figs. 8B and 8D); (1) single carina or ledge (not illustrated); (2) several parallel or subparallel carinae (not illustrated). Unordered; (CI = 0.29, RI = 0.54, G-fit = 3.8).

14. (Subdivision of 13:1) Shape of single vertical delineation of lower face (not illustrated): (0) rounded divergent ledges running from antennal sockets to dor-

Teleost Anatomy Ontology



Phenoscape ontologies

New:

Teleost Anatomy
Ontology
(2233 terms; 387
skeletal)

Teleost
Taxonomy
Ontology
(36,060 terms;
38,000 synonyms)

Taxonomic
Rank
Ontology
(8->31 terms)

Zebrafish
Anatomy
Ontology
(2196 terms; 310
skeletal)

Existing:

Phenotype and
Trait Ontology
(1,075 terms)

Spatial
Ontology
(106 terms)

Evidence
Code
Ontology

Requirements:

- Develop ontologies
- **Develop appropriate syntax**
- Curate phenotypes (characters)

Requirements:

- Develop ontologies
- Develop appropriate syntax
- **Curate phenotypes (characters) from [prototype Otophysi] fishes**

Creating prototype by curating evolutionary data

Taxon	# Species	# Papers	# Characters
Cypriniformes (Mayden; Coburn)	3,268	70	1125
Siluriformes (Lundberg)	2,867	87	1200
Characiformes (Dahdul)	1,674	124	800
Gymnotiformes (Arratia)	134	2	200
Gonorynchiformes (Arratia)	37	80	75
Clupeiformes (Hilton)	364	60	380
TOTAL	8,344	423	3,780

63,080, 640 phenotypic data points

Phenex curation tool

The screenshot displays the Phenex curation tool interface with three main windows:

- Taxa Window:** A list of taxon names under two columns: "Valid Taxon" and "Publication Taxon". The row for *Astyanax scabripinnis* is selected and highlighted in blue.
- Term Info Window:** A detailed view for the selected taxon, *Astyanax scabripinnis*.
 - Basic Info:** Term: *Astyanax scabripinnis*, ID: TTO:1051209, Ontology: teleost-taxonomy, Definition: (no definition provided).
 - Synonyms (3):** RELATED *Astyanax scabripinnis laticeps*, *Astyanax scabripinnis paranae*, *Tetragonopterus scabripinnis*.
 - Links (1):**
 - Parents:** *Astyanax*
 - Children:** None listed.
 - Other Properties (1):** has_rank TTO:species
- Specimens Window:** A table showing specimen details for the selected taxon.

Collection	Catalog ID
MZUSP	44539

Phenex curation tool

The Phenex curation tool interface consists of five windows:

- Characters**: A list of 19 characters with descriptions:
 - 1 Degree of development of ventral diverging lamellae of
 - 2 Foramen on posterior portion of lateral wing of mesethmoid
 - 3 Presence and degree of development of posterior portion
 - 4 Orientation of distal portion of lateral wing of mesethmoid
 - 5 Form of contact between vomer and mesethmoid
 - 6 Form of anterior projection of vomer overlapping posterior
 - 7 Presence or absence of a dorsolateral expansion of vomer
 - 8 Form of contact between vomer and palatine
 - 9 Form of anterior process of lateral ethmoid
 - 10 Form of nasal
 - 11 Presence or absence of antorbital
 - 12 Snout length
 - 13 Presence or absence of rhinosphenoid
 - 14 Well-developed bony expansion on ventral margin of rhinosphenoid
 - 15 Presence or absence of a frontal fontanel
 - 16 Presence or absence of a parietal fontanel
 - 17 Epiphyseal bar
 - 18 Dorsal portion of sphenotic spine
 - 19 Orientation of sphenotic spine
- States for Character**: A table showing symbols (0 for present, 1 for absent) and state descriptions:

Symbol	State Description
0	present
1	absent
- Term Info**: Details for the term "epiphyseal bar".
 - Basic Info**: Term: epiphyseal bar, ID: TAO:0001502, Ontology: teleost_anatomy, Definition: Bilaterally paired medial bar of cartilage formed at midorbit through medial chondrification from triangles of cartilage that later form the junction of the taeniae marginales anterior, taeniae marginales posterior and the epiphyseal bar. Definition ref 1: ZFIN:curator.
 - Links (1)**: Parents: is_a [chondrocranium cartilage](#). Children: None.
 - DBxrefs (2)**: None.
- Phenotypes for State: 0 - present**: A table showing phenotypes for the present state of the epiphyseal bar.

Entity	Quality	Related Entity	Comment
epiphyseal bar	present	None	None
- Complete Ontology Tree View**: A hierarchical tree view of biological entities:
 - intestinal villus
 - mesenchyme
 - muscle
 - nerve
 - notochord
 - portion of connective tissue
 - bone
 - cartilage
 - cranial cartilage
 - chondrocranium cartilage
 - auditory capsule
 - basal plate cartilage
 - epiphyseal bar
 - ethmoid cartilage
 - kinethmoid cartilage
 - lamina orbitonasalis
 - neurocranial trabecula
 - occipital arch cartilage
 - parachordal cartilage

Morphological image databases



CToL images deposited in “Morphbank” data repository

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

About Browse Search Tools Help m

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)

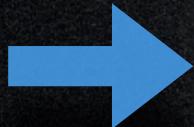


Image Record: [460945] Chromobotia macracanthus

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

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

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

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

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

Specimen Specimen id: 460920 Basis of record: [S] - Specimen Sex: unknown Form: unknown Stage: Juvenile/Adult Catalog number: 199848 Collector: Date collected:	Locality Locality Id: Continent ocean: Country: Locality: Latitude: Longitude: Elevation (m):
Determination Class: Actinopterygii Order: Cypriniformes Family: Cobitidae Genus: Chromobotia Species: Chromobotia macracanthus	Determination annotations (Add Annotation...)
External links/identifiers External identification: CToL-S:0001165 External identification: CToL-I:0000501 Ontology: Teleost Anatomy Ontology TAO:0000316	Other Annotations (Add Annotation...)

More information: Image + ontology

THE NATIONAL CENTER FOR BIOMEDICAL ONTOLOGY

 BioPortal 2.0

Home Browse Search Projects All Mappings Teleost anatomy and development [Sign In](#) [Register](#)

basihyal ([Link To Concept](#))

[Details](#) [Visualization](#) [Marginal Notes](#) [Mappings](#) [Resources](#)

Class Name: basihyal
ID: TAO:0000316

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

Database References: ZFIN

Is A: endochondral bone

Develops From: basihyal cartilage

Part Of: ventral hyoid arch

Related Synonym: "basihyoid", "glossohyal"

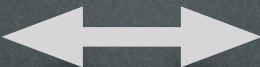
Phylogenetic tree on the left showing relationships between various pharyngeal structures, with 'basihyal' highlighted at the bottom.

Summary

- Evolutionary morphology and genetic data can be linked using ontologies
- **Phenoscape**: prototype of a generalizable system for making this connection
- Crosses disciplines, facilitates new discoveries through data mining

Future goal: Connect many new data types using ontologies

Model
organisms
(biomed)



Comparative
evolutionary
biology

- Phenotypes
- Genetics

- Morphology
- Phylogeny
- Ecology
- Locality

Biodiversity informatics

Vouchers

Species
morphology



Model organism
phenotypes

Genetics

Biomedical informatics

Species
Genetics
Ecology
Morphology
Locality
Medicine

Phenoscape training opportunity

- 10 week internships
 - travel + stipend
 - typically summer (May-August)
- Mabee lab or National Evolutionary Synthesis Center
- informatics; morphological data; ontologies