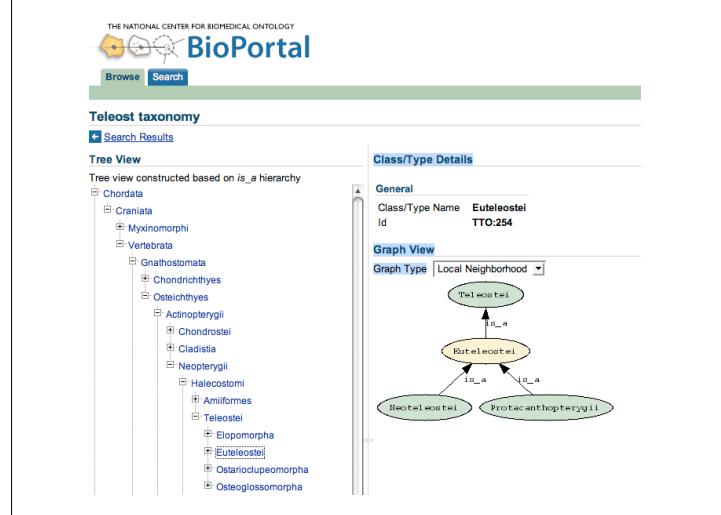


A hypothetical query

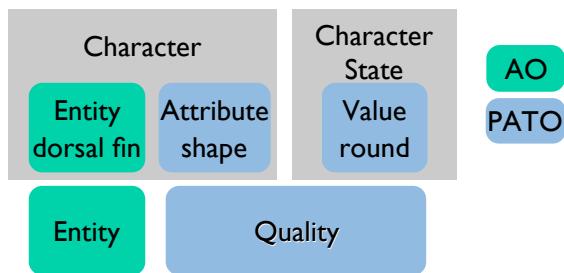
- You note a reduction in the number of branchiostegal rays in a zebrafish mutant and ask “is this bone variable in number among species in nature?”
- You go to a Phenoscape to visualize character states mapped onto a phylogenetic tree
 - Most Ostariophysi have 3 rays, but you see that a reduction has occurred multiple times: in solenostomids and syngnathids (ghost pipefishes and pipefishes), giganturids (telescope fishes) and saccopharyngoid (gulper and swallower) eels
- You then ask “what parallel changes have occurred?”
 - Loss of the swimbladder, pelvic fins, and scales
 - Elongation of the mandibular or hyoid arches
 - Reduction or loss of the opercle in syngnathids and saccopharyngoids.
 - A variety of other bones and soft tissues are lost or greatly modified

How to enable computable phenotypes?

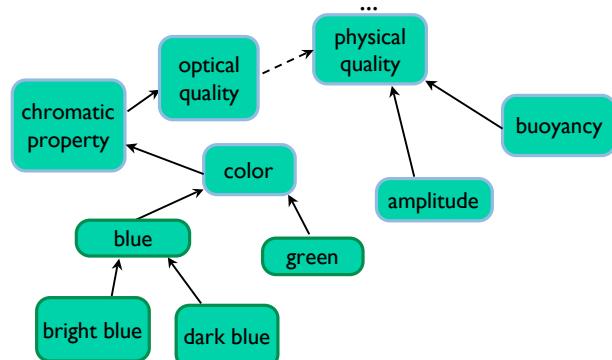
- Semantics via ontologies
- Syntactic convention is EQ
 - Entity terms: **Teleost Anatomy Ontology (TAO)**
 - Quality terms: **Phenotype and Trait Ontology (PATO)**
 - e.g. Entity=dorsal fin, Quality=round (shape)
- Phylogenetic relationships
 - Teleost taxonomy ontology**
- Curated phenotype data
 - Mutant phenotypes from zebrafish (14K in ZFIN)
 - Cypriniform “evolutionary phenotypes” from the literature
 - Ontology editor: **Phenote**
- Down the road: database, interface & reasoning engine



Character Matrix vs. EQ syntax



PATO slim for attributes

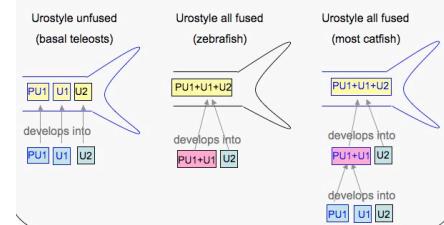


Anatomy ontology challenges

- Synchronization of anatomy ontologies
 - TAO and ZAO
- Different types of synonyms
 - True synonyms (fin rays = lepidotrichia)
 - Spelling variants (hyomandibular = hyomandibula)
 - Nonhomologous structures with the same name (lacrimal)
- Homology

Homology

- Ontologies are inherently typological
- “homologous_to” relation



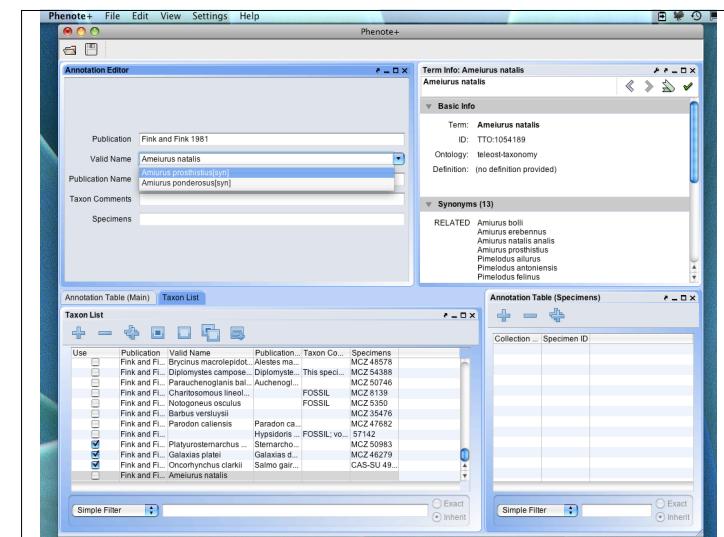
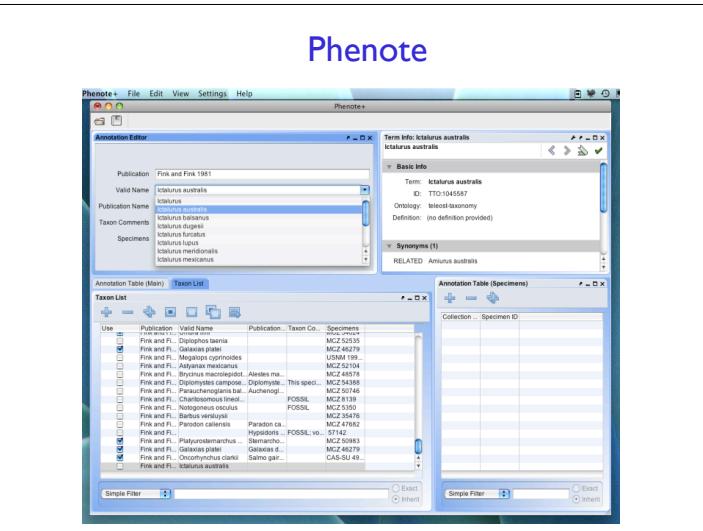
Taxonomy ontology

- The only previous NCBO taxonomy ontology was based on the NCBI taxonomy
 - TTO uses names from CoF, relations from CTOL
 - Have separated the taxonomic rank ontology
- Assignments are to specimens
 - Species may be not yet determined
 - Species may be not yet named
 - Specimen be from a mixed collection

Evidence Code Ontology

- Inferred from Phenotypic Similarity
 - Inferred from Compositional Similarity (ICS)
 - Inferred from Developmental Similarity (IDS)
 - Inferred from Morphological Similarity (IMS)
 - Inferred from Positional Similarity (IPS) -
 - Inferred from Gene Expression Similarity (IGES)
- Inferred from Computational Analysis
 - Inferred from Phylogeny (IP)

Phenote



Phenote+ File Edit View Settings Help

Annotation Editor

Publication: Fink and Fink 1981
Valid Name: *Opsarichthys uncirostris*
Publication Name:
Taxon Comments:
Entity: opercle
Quality: rectangular
Spatial Aspect:
Additional Entity:
Measurement:
Unit:

Term Info: opercle
opercle

Synonyms (1)
RELATED: opercles

Links (4)

Parents: *is_a* [dermal bone](#)
part of [dermal bone](#), [cranium](#), [opercular flap](#), [dorsal hyoid arch](#)

Children:

DBxrefs (2)
ZFA:0000250
ZFIN:70RA-NAT-011113-1K1

Annotation Table (Main) **Taxon List**

Annotation Table (Main)

Collection	Specimen ID	Entity	Quality	Additional Entity	Measure
MCZ	54388	kinosternum	present		
Xenocharax	spilurus	kinosternum	absent		
<i>Opsarichthys</i>	<i>uncirostris</i>	preethmoid bone	present		
<i>Diplomyces</i>	<i>chilensis</i>	ceratobranchial 5 bone	increased size		
<i>Diplomyces</i>	<i>chilensis</i>	opercle	triangular		
<i>Stompgyrus</i>	<i>macrurus</i>	opercle	rectangular		
<i>Xenocharax</i>	<i>spilurus</i>	opercle	triangular		
<i>Diplomyces</i>	<i>chilensis</i>	bassipterygium	united		
<i>Opsarichthys</i>	<i>uncirostris</i>	scaphium	anteriorodorsal to	vertebra 1	
<i>Rhoadia</i>	<i>alpina</i>	scaphium	dorsal to	vertebra 1	
		scaphium	anteriorodorsal to	vertebra 1	

Simple Filter:

Annotation Table (Specimens)

Collection	Specimen ID
MCZ	54388

Simple Filter:

Phenote+ File Edit View Settings Help

Annotation Editor

Publication: Fink and Fink 1981
Valid Name: *Opsarichthys uncirostris*
Publication Name:
Taxon Comments:
Entity: opercle
Quality: rectangular
Spatial Aspect:
Additional Entity:
Measurement:
Unit:

Term Info: dermal bone
dermal bone

Parents: *is_a* [bone](#)

Children: [caudal procurent ray](#), [anguloarticular](#), [parasphenoid](#), [suprareopercle](#), [epipharyngeal](#), [preopercle](#), [postopercle](#), [anterior](#), [horizontal limb](#), [preopercle lateral shelf](#), [epipharynx](#), [interopercle](#), [frontal bone](#), [anterior](#), [posterior](#), [supracleithrum](#), [subopercle](#), [epipharyngeal](#), [kinethmoid bone](#), [suprareopercle](#), [anterior](#), [posterior](#), [anterior](#), [principal ray](#), [suprareopercle](#), [maxilla](#), [anterior](#), [flange](#), [posttemporal](#), [epipharyngeal](#), [vertical strut](#), [anterior](#), [posterior](#), [anterior](#), [posterior](#), [anterior](#), [flange](#), [inferior](#), [postopercle](#), [branchiostegal ray](#), [extrascapular](#), [infrascapular](#), [flange](#), [maxilla](#), [epipharyngeal](#), [anterior](#), [posterior](#)

Annotation Table (Main) **Taxon List**

Annotation Table (Main)

Collection	Specimen ID	Entity	Quality	Additional Entity	Measure
MCZ	54388	kinosternum	present		
Xenocharax	spilurus	kinosternum	absent		
<i>Opsarichthys</i>	<i>uncirostris</i>	preethmoid bone	present		
<i>Diplomyces</i>	<i>chilensis</i>	ceratobranchial 5 bone	increased size		
<i>Diplomyces</i>	<i>chilensis</i>	opercle	triangular		
<i>Stompgyrus</i>	<i>macrurus</i>	opercle	triangular		
<i>Xenocharax</i>	<i>spilurus</i>	opercle	triangular		
<i>Diplomyces</i>	<i>chilensis</i>	bassipterygium	united		
<i>Opsarichthys</i>	<i>uncirostris</i>	scaphium	anteriorodorsal to	vertebra 1	
<i>Rhoadia</i>	<i>alpina</i>	scaphium	dorsal to	vertebra 1	
		scaphium	anteriorodorsal to	vertebra 1	

Simple Filter:

Annotation Table (Specimens)

Collection	Specimen ID
MCZ	54388

Simple Filter:

Phenote+ File Edit View Settings Help

Annotation Editor

Publication: Fink and Fink 1981
Valid Name: *Diplomyces chilensis*
Publication Name: *Diplomyces papilliferus*
Taxon Comments:
Specimens: "MCZ 36195", "MCZ 8290"

Term Info: dermal bone
dermal bone

Parents: *is_a* [bone](#)

Children:

Annotation Table (Main) **Taxon List**

Annotation Table (Main)

Collection	Specimen ID	Entity	Quality	Additional Entity	Measure
MCZ	36195	kinosternum	present		
MCZ	8290	kinosternum	absent		

Simple Filter:

Annotation Table (Specimens)

Collection	Specimen ID
MCZ	36195
MCZ	8290

Simple Filter:

Phenote+ File Edit View Settings Help

Annotation Editor

Publication: Fink and Fink 1981
Valid Name: *Multiple Values*
Publication Name: *Multiple Values*
Taxon Comments: *Multiple Values*
Entity: dorsal fin
Quality:
Spatial Aspect:
Additional Entity:

Term Info: dorsal fin
dorsal fin

Term: **dorsal fin**
ID: TAO:0001173
Ontology: teleost_anatomy
Definition: (no definition provided)

Links (6)

Parents:

Annotation Table (Main) **Taxon List**

Annotation Table (Main)

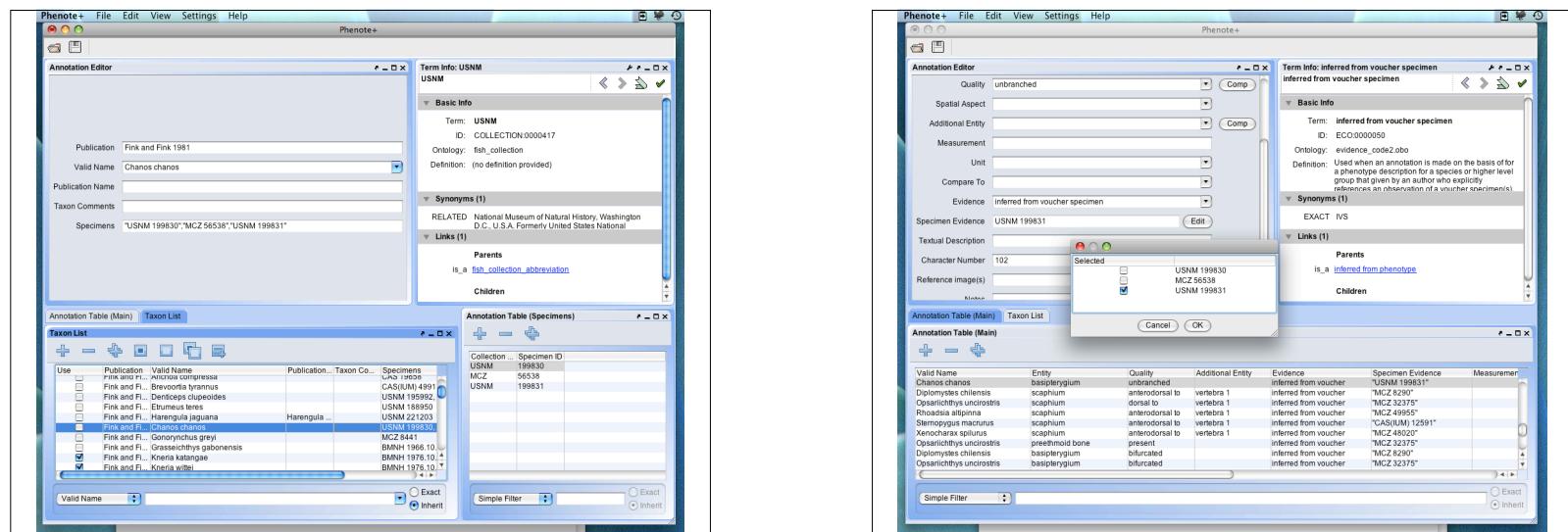
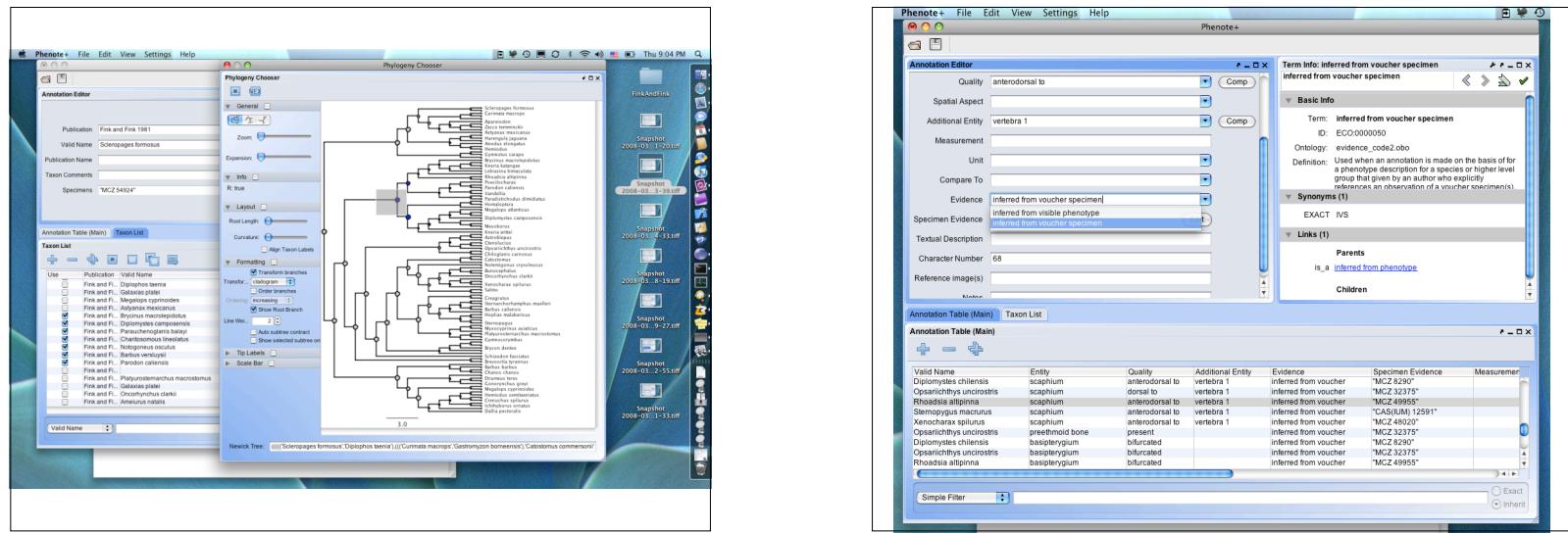
Collection	Specimen ID	Entity	Quality	Additional Entity	Measure
MCZ	36195	bassipterygium	bifurcated		
MCZ	8290	bassipterygium	bifurcated		

Simple Filter:

Annotation Table (Specimens)

Collection	Specimen ID
MCZ	36195
MCZ	8290

Simple Filter:



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- www.phenoscape.org
- www.phenote.org
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 - Data & ontology curation or software development
- 1-day workshop at the Evolution meetings in Minneapolis

