



Advisory Board Meeting

(electronic)

December 4, 2012

- I. Introductions
- II. Project updates
- III. Upcoming plans
- IV. Q&A and discussion
- V. Spring 2013 AB meeting



Phenoscape Advisory Board

- Members
 - John Day-Richter (Google)
 - Brian K. Hall (Dalhousie University)
 - Cyndy Parr (Encyclopedia of Life)
 - Alan Ruttnerberg (stepping down)
 - Paul Schofield (University of Cambridge)
 - Peter Vize (University of Calgary)
- Roles
 - Evaluate goals, plans, progress
 - Share knowledge, suggest opportunities
 - Help prioritize activities, ameliorate risks
- Meetings
 - Annual virtual AB meeting
 - Annual F2F project meeting



Phenoscape project team

- National Evolutionary Synthesis Center (NESCent)
 - Todd Vision
 - Hilmar Lapp
 - Jim Balhoff
 - Peter Midford
 - Prashanti Manda (starting Feb 2013)
- University of South Dakota
 - Paula Mabee
 - Wasila Dahdul
 - Alex Dececchi
- University of Oregon (Zebrafish Information Network)
 - Monte Westerfield
 - Yvonne Bradford
 - Ceri Van Slyke
- Oregon Health & Science University
 - Melissa Haendel
- University of Chicago
 - Paul Sereno
 - Nizar Ibrahim
- Mouse Genome Informatics
 - Judith Blake
 - Terry Hayamizu
- Cincinnati Children's Hospital Medical Center
 - Aaron Zorn (Xenbase)
 - Christina James-Zorn (Xenbase)
- California Academy of Sciences
 - David Blackburn
- University of Arizona
 - Hong Cui
 - Zilong Chang
- Lawrence Berkeley National Labs
 - Suzi Lewis
 - Chris Mungall

NSF: July 1, 2011-June 30, 2015

The image shows a screenshot of the PhenoscAPE website. At the top right, the word "PHENOSCAPE" is written in a green, sans-serif font. Below the header, there is a horizontal navigation bar with several items:

- ABOUT**
- PHENOSCAPE KNOWLEDGEBASE**
- PHENOTYPES**
- SOFTWARE**
- RESOURCES**
-PARTNERSHIPS
- ONTOLOGIES**

On the left side of the page, there is a vertical list of links under the "ABOUT" category:

- About
- Acknowledgements ▾
- Background
- Learn more ▾
- News
- Opportunities ▾
- Outreach
- People ▾
- Project intranet

The background of the page features stylized illustrations of various plant life stages, including seed pods, flowers, and fruits, arranged along a blue and yellow gradient.

Phenoscape activities overview

The screenshot shows a Trello board titled "Phenoscape Roadmap" with the following columns:

- In Progress:**
 - CharaParser installation simplification.
 - Improve EQ generation performance of CharaParser
 - Planning for February Phenoscape collaboration workshop
 - Recruitment of postdoc at UNC
 - Edit phenoscape-ext to a more final state
 - VTO Release 1.1
 - Migrate VTO to OWL
 - Improved Phenotype Annotation syntax
 - Homology reasoning use-cases and requirements
- Next up:**
 - Document OWL representation model for KB content.
 - Annotation data QA
 - Recruitment of postdoc at USD
 - Improved CharaParser performance
 - CharaParser ontology access improvements
 - Reasoning rules for default homology
 - Sandbox KB for evolutionary data
- Planned for Year2:**
 - Document KB content model changes for model organism data
 - Implement new KB backend technology
 - Homology reasoning test data and environment
 - Initial collection of homology assertions in KB
 - Integration of homology reasoning and provenance into KB backend
 - Integration of homology reasoning and provenance into KB UI
 - Integration of homology reasoning into semantic similarity match
 - Statistics for semantic similarity match with desirable properties
 - Add ontology versioning to phenex data files
 - KB import of mouse phenotypes
- Planned for Year3:**
 - Comprehensive collection of homology assertions
 - KB UI support for new MOD content
 - Usability testing of KB UI for new MOD content
 - Usability testing of KB UI for homology reasoning and provenance
 - Scalable computation of similarity match statistics
 - Integration of semantic similarity search into KB UI
 - Usability testing of semantic similarity search UI
 - Correlated phenotype exploration in KB UI
 - Regularly updated OWL dumps of the KB content
 - Interface for bulk requesting of unmatched terms from CharaParser run
- Planned for Year4:**
 - Evaluation of knowledge discovery and hypothesis generation
- Completed:**
 - Consolidated ontology development plan
 - Vertebrate Taxonomy Ontology 1.0
 - Integrate CharaParser output into Phenex
 - Develop collaborative ontology editor technology plan
 - Develop workflow and training plan for switch to protege
 - New front page(s) for Phenoscape website
 - Phenex/KB support for annotation of fin/limb evolutionary data
 - Determining suitable term

Phenoscape roadmap on Trello: <http://bit.ly/HDniSS>



Report of Phenoscape Scientific Advisory Board (27th April 2012)

1. Consider other large scale sources of structured data (Todd)
2. Further analyze candidate genes by incorporating gene set and pathway analysis for additional value to users (Todd)
3. Outreach, identification of user groups and real-life use-cases (Todd)
4. VAO and Uberon merge into one cross-species anatomy (Melissa)
5. Quality of the phenotype data capture, annotations (Alex)
6. Capturing inter-individual variation within taxa (Alex)
7. Practicality and utility of capturing data from birds and humans (Melissa & Monte)
8. Distribution of effort into developing tools vs. data capture & curation (Todd)
9. Concern over ontology term brokering with the NCBO tools (Jim)
10. Improvement in the granularity of phenotype descriptions, PATO (Alex)



II. Project updates

1. Curation workflow (Hong Cui, Jim Balhoff)
2. Ontology development (Melissa Haendel, Peter Midford)
3. Phenotype curation (Alex Dececchi, Monte Westerfield)
4. Homology reasoning (Hilmar Lapp)
5. Semantic similarity (Todd Vision)
6. Outreach (Nizar Ibrahim)
7. Knowledgebase development (Jim Balhoff)

1. Curation workflow

1a. Scalable Workflow: NLP

- Main goal
 - To speed the curation of phenotype descriptions
 - To improve the specificity of the annotation (“shape” -> “U-shaped”)
- Progress made since the last meeting
 - Removing CharaParser’s dependency on relational database, Perl etc.
 - Addressing issues identified in the BioCreative workshop , April 2012
 - Issues
 - Group 1: From plain text to XML (machine understanding text description)
 - Group 2: From XML to EQ (ontologizing the understanding)
 - Addressed most Group 1 issues that have been identified.

Issues Addressed

- Self-referred terms
 - Eg. “counterpart” -> the entity itself
- Adjective -> Noun +absent/present
 - Eg. Unossified -> E: ossification of ...; Q: absent
- Connection/separation
 - Eg. **Connection** between A and B presents-> A **in contact with** B

Issues Addressed

- BSPO (Spatial Ontology)
 - Mapping to non-synonyms in ontology
 - Eg. proximal portion -> proximal region
 - Implicit part_of relation
 - Eg. **anterior** coracoid process ->
anterior region(part_of (coracoid))
 - Spatial terms used as adverbs
 - Eg. ... red **dorsally** -> **dorsal region** of ... is red

Currently Working on

- Relational Slim
 - Providing methods to retrieve all terms in relational slim -- optimization of ontology lookup
 - Enforce: If entity and related entity are both present, the quality must be from relational slim
- Binary statements (absent/present, yes/no)
 - “quadrate , dorsoventrally oriented crest located on the posterior side ”
 - absent
 - present

Future Plans

- Working with others (Melissa, Jim, Wasila, Alex, etc.) to identify curation patterns
- Implement these patterns in CharaParser.EQ Generator
- Address group 2 issues (i.e., XML to EQ) by better utilizing the knowledge coded in ontologies

1. Curation workflow

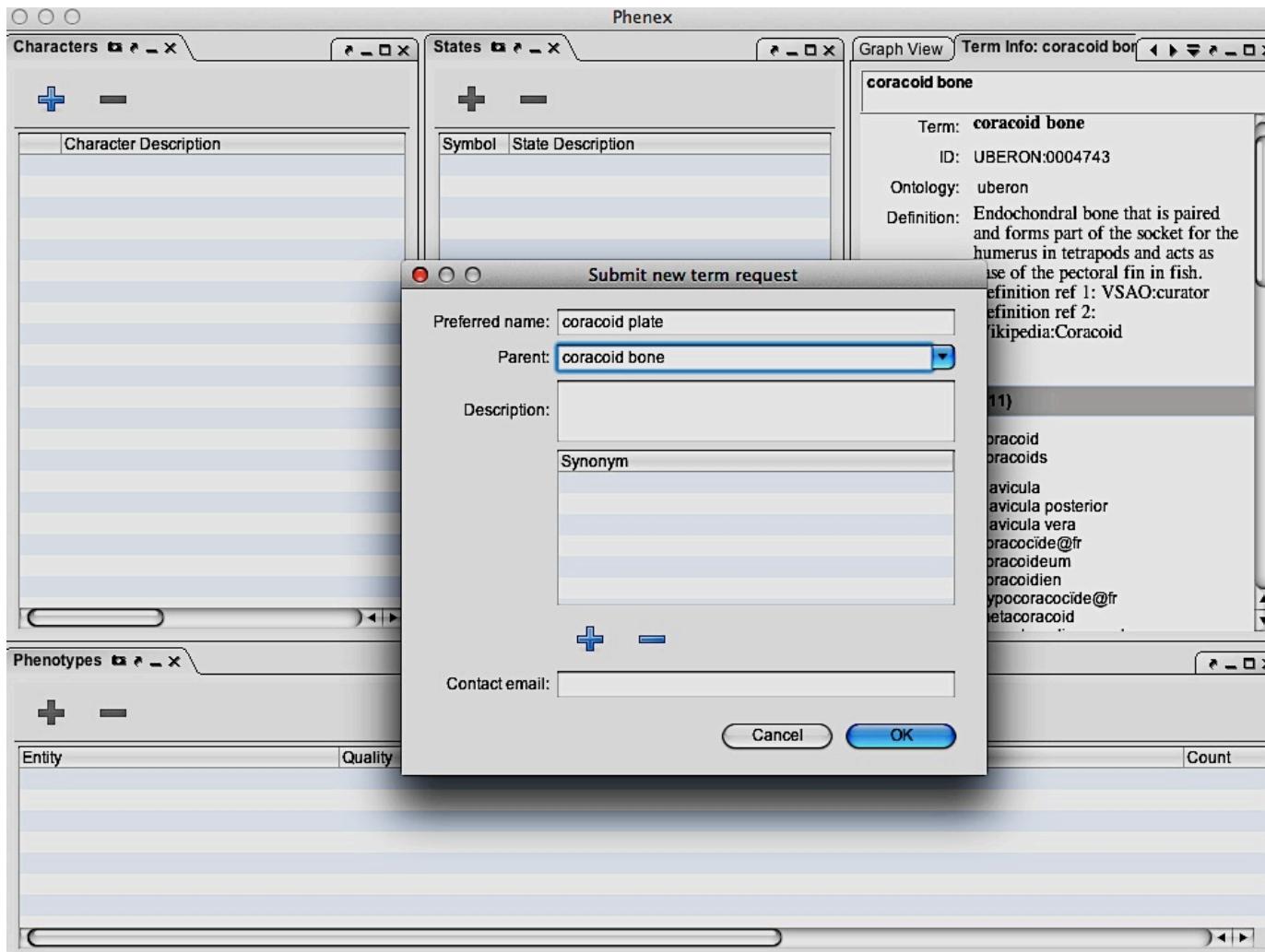
1b. Ontology Term Broker

- Goal: remove bottleneck created by encountering missing ontology terms during annotation work
- Progress since April 2012: working system is in place⁹
 - Phenex request interface
 - Provisional term management web interface
 - Phenex auto-update of implemented provisional terms
 - Lightweight solution: meets our needs, avoids development of elaborate back-end by using Bioportal provisional term services

Term broker workflow

- Request missing terms from within annotation interface
- Receive provisional term ID to use in annotation
- Asynchronously, edit ontology to add new terms and update provisional term record with permanent ID
- References to provisional terms are automatically migrated to permanent IDs

Request a term, immediately available for use in annotation



View requests in web interface

Term requests

SUBMITTED	PROVISIONAL IRI	LABEL	DESCRIPTION	PARENT	PERMANENT IRI
2012-10-26 10:55:52.342 PDT	http://purl.bioontology.org/ontology/provisional/064dfc58-8143-491f-ab74-374507502f35	inter-clavicle joint	Joint that articulates the left and right clavicles. Requested by: wasila.dahdul@usd.edu	joint of girdle	Update
2012-09-26 12:37:50.410 PDT	http://purl.bioontology.org/ontology/provisional/5810ff9f-b481-40a8-97da-362bb9c943cc	coracoid plate	Broad, convex expansion of the coracoid bone below the level of the glenoid. Based on Romer 1956 Requested by: alex.deecchi@gmail.com	coracoid bone	Update
2012-08-31 13:17:06.385 PDT	http://purl.bioontology.org/ontology/provisional/1585a400-3465-4b98-8539-1d5c21ed6cfb	equal	Is a quality that denotes the state where two or more entities possess the same number, value or measurement for a given trait of interest. ?Equal? is a child of count whose direct parent is present. ?Equal? is primarily associated with measurement or count data as opposed to ?same? which is more closely associated with a qualitative description. Requested by: alex.deecchi@gmail.com	quality	Update
2012-11-14 07:48:12.457 PST	http://purl.bioontology.org/ontology/provisional/2cad732a-d955-44c7-9ded-e086c7314c7b	hypochordal lepidotrichium	Lepidotrichium located ventral to the notochord Requested by: wasila.dahdul@usd.edu	http://purl.obolibrary.org/obo/UBERON_4000172	Update
2012-08-28 11:44:47.102 PDT	http://purl.bioontology.org/ontology/provisional/43c83028-4821-437f-9e4c-4163b0d379ea	single	A derivative of count that signifies the presence of one and only one example of the given character of interest. The sole representative of a state or trait. Requested by: alex.deecchi@gmail.com	count	Update
2012-09-04 08:38:23.451 PDT	http://purl.bioontology.org/ontology/provisional/00de8892-a538-450f-bb2c-131a0eb235be	parallel-sided	Having two edges parallel to each other. Requested by: alex.deecchi@gmail.com	shape	Update
2012-09-13 11:49:22.750 PDT	http://purl.bioontology.org/ontology/provisional/16e8dadf-1512-4a6f-90f9-6b234314200a	subdermal	Located or placed beneath the skin; subcutaneous. Reference: http://medical-dictionary.thefreedictionary.com/subdermal Requested by: alex.deecchi@gmail.com	shape	Update
2012-09-30 15:13:07.65 PDT	http://purl.bioontology.org/ontology/provisional/d46a4834-296a-43f2-ba70-325ffa57e07	articular surface	Surface of articulation between skeletal elements - not necessarily a joint. Requested by: nibrahim@uchicago.edu		Update

Edit ontology

The screenshot displays four panels of an ontology editor interface:

- Class hierarchy:** Shows the inheritance path of the class 'coracoid plate'. It includes nodes like 'webbing of bone', 'zone of bone organ', and various anatomical features.
- Annotations:** Lists annotations for 'coracoid plate':
 - label** [language: en]: coracoid plate
 - created_by** [type: string]: A.Decechci
 - definition** [type: string]: Broad, convex expansion of the coracoid bone below the level of the glenoid. Based on Romer 1956
- Description:** Lists sub-classes and super-classes:
 - Equivalent To**: None listed.
 - SubClass Of**:
 - 'zone of bone organ'
 - part_of some 'coracoid bone'
 - SubClass Of (Anonymous Ancestor)**:
 - part_of some organ
 - part_of some bone
 - part_of some 'skeletal system'
 - part_of some 'anatomical system'
- Members**: A list of entities associated with the class, including 'coracoid process of scapula', 'cribriform plate', and various bone structures.

Update permanent IRI in web interface; Phenex will auto-update references to the provisional ID

Term requests

SUBMITTED	PROVISIONAL IRI	LABEL	DESCRIPTION	PARENT	PERMANENT IRI
2012-10-26 10:55:52.342 PDT	http://purl.bioontology.org/ontology/provisional/064dfc58-8143-491f-ab74-374507502f35	inter-clavicle joint	Joint that articulates the left and right clavicles. Requested by: wasila.dahdul@usd.edu	joint of girdle	[Update]
2012-09-26 12:37:50.410 PDT	http://purl.bioontology.org/ontology/provisional/5810ff9f-b481-40a8-97da-362bb9c943ec	coracoid plate	Broad, convex expansion of the coracoid bone below the level of the glenoid. Based on Romer 1956 Requested by: alex.dececcchi@gmail.com	coracoid bone	http://purl.obolibrary.org/obo/UBERON_4200002 [Update]
2012-08-31 13:17:06.385 PDT	http://purl.bioontology.org/ontology/provisional/1585a400-3465-4b98-8539-1d5c21ed6cfb	equal	Is a quality that denotes the state where two or more entities possess the same number, value or measurement for a given trait of interest. ?Equal? is_a child of count whose direct parent is present. ?Equal? is primarily associated with measurement or count data as opposed to ?same? which is more closely associated with a qualitative description. Requested by: alex.dececcchi@gmail.com	quality	[Update]
2012-11-14 07:48:12.457 PST	http://purl.bioontology.org/ontology/provisional/2cad732a-d955-44c7-9ded-e086c7314c7b	hypochordal lepidotrichium	Lepidotrichium located ventral to the notochord Requested by: wasila.dahdul@usd.edu	http://purl.obolibrary.org/obo/UBERON_4000172	[Update]
2012-08-28 11:44:47.102 PDT	http://purl.bioontology.org/ontology/provisional/43c83028-4821-437f-9e4c-4163b0d379ea	single	A derivative of count that signifies the presence of one and only one example of the given character of interest. The sole representative of a state or trait. Requested by: alex.dececcchi@gmail.com	count	[Update]
2012-09-04 08:38:23.451 PDT	http://purl.bioontology.org/ontology/provisional/00de8892-a538-450f-bb2c-131a0eb235be	parallel-sided	Having two edges parallel to each other. Requested by: alex.dececcchi@gmail.com	shape	[Update]
2012-09-13 11:49:22.750 PDT	http://purl.bioontology.org/ontology/provisional/16c8dadf-1512-4a6f-90f9-6b234314200a	subdermal	Located or placed beneath the skin; subcutaneous. Reference: http://medical-dictionary.thefreedictionary.com/subdermal Requested by: alex.dececcchi@gmail.com	shape	[Update]
2012-09-30 15:13:07.65 PDT	http://purl.bioontology.org/ontology/provisional/d46a4834-296a-43f2-ba70-325ffffa57e07	articular surface	Surface of articulation between skeletal elements - not necessarily a joint. Requested by: nibrahim@uchicago.edu		[Update]

Next six months

- Improve provisional term web interface
 - Add ability to correct typos in labels, definitions
- Interaction of web interface with ontology trackers
 - Each ontology has a different request system
 - Add field to note which terms have been submitted to an ontology tracker

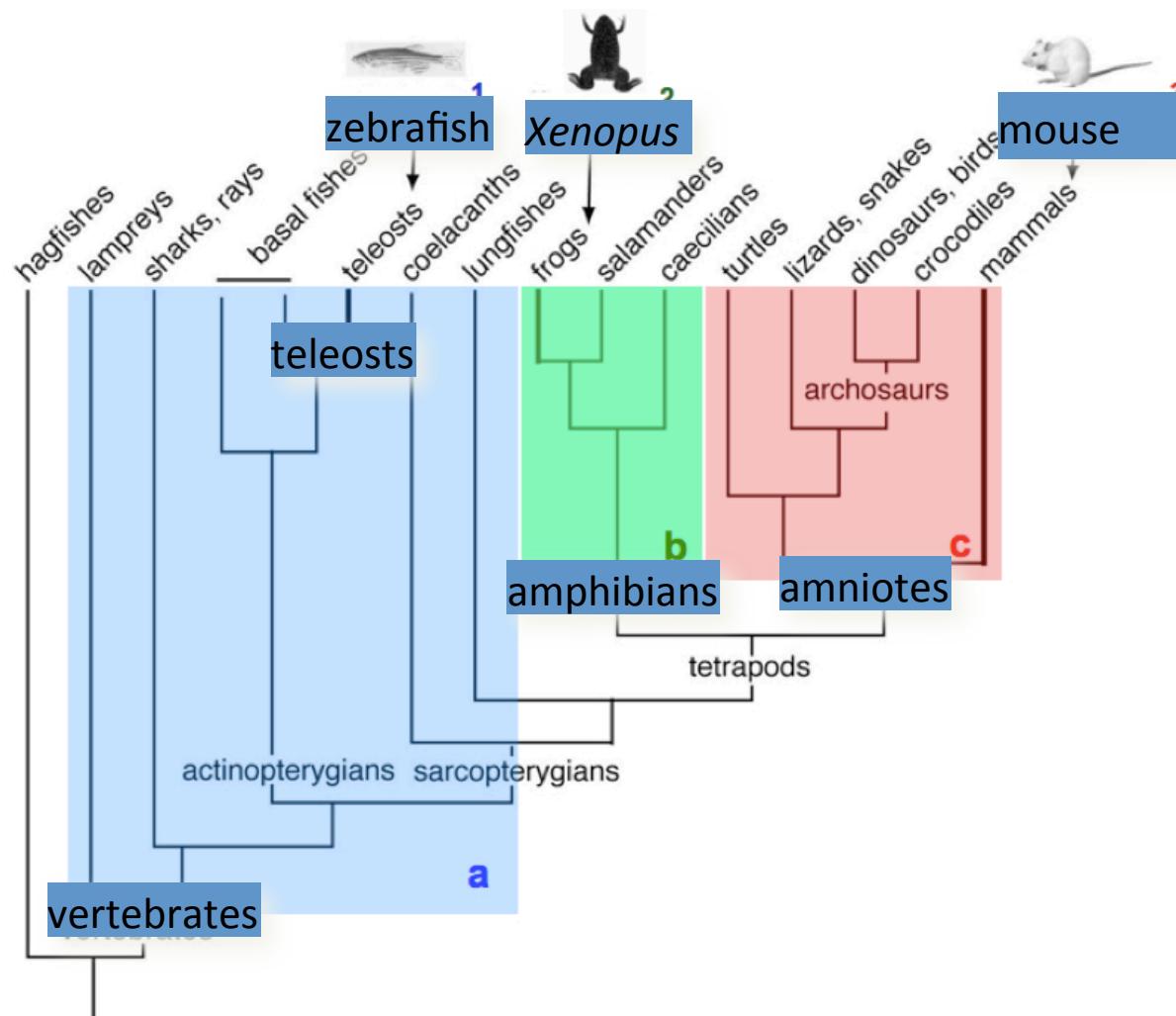
2. Ontology Development

2a. Anatomy

Objectives: Coordinate the development and alignment of vertebrate anatomy ontologies, with focus on the limb/fin skeleton

- Anatomy ontologies provide the entity term in phenotype annotations
- Limb/fin transition is key Phenoscape use case
- Integration across the various ontologies is required to support cross-species queries
- Logical definitions required for inference and analysis

Taxa/model organism anatomy relevant to Phenoscape



Recommendations from April Advisory Board Meeting

4. VAO and Uberon merge into one cross-species anatomy ontology

Coordination and allocation of responsibilities and edit rights should be sorted out

7. Examine the practicality and utility of capturing data from birds and humans using new VAO/Uberon ontology

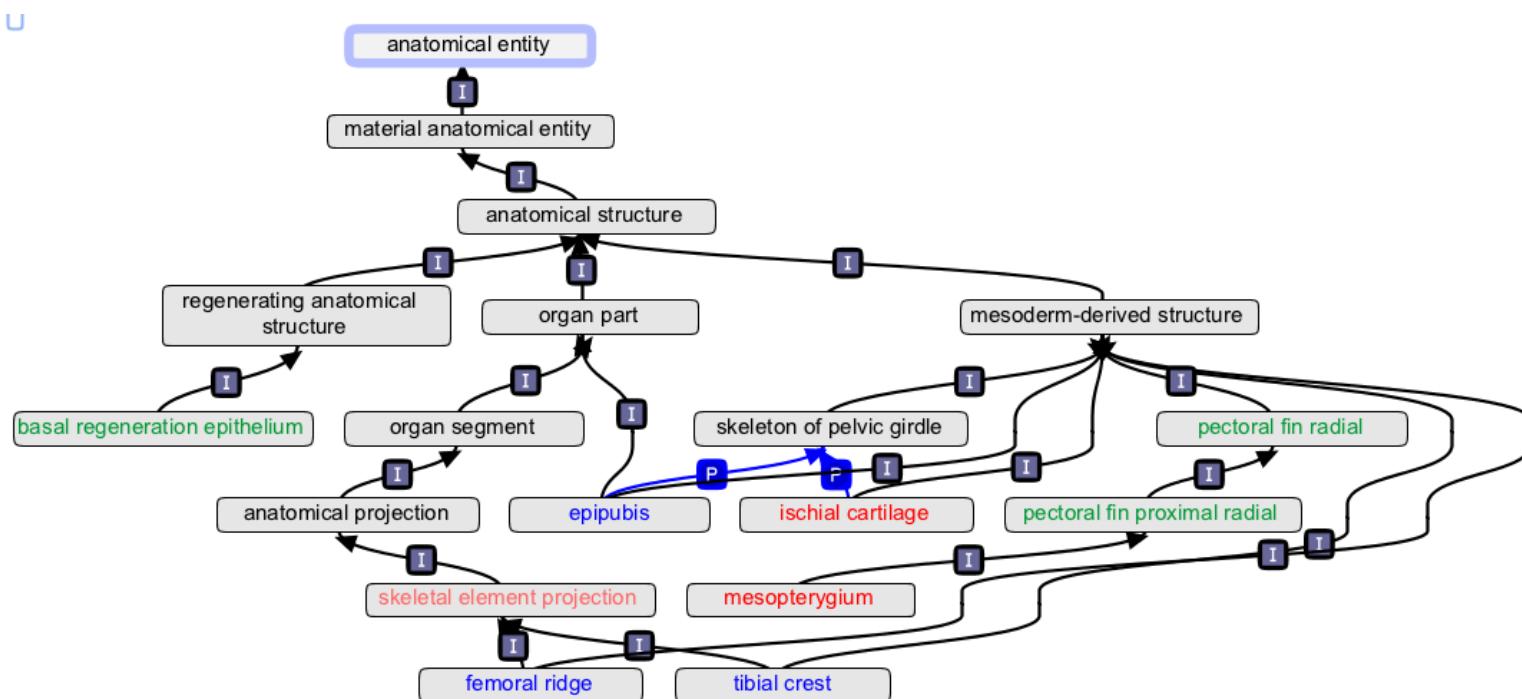
What have we achieved?

- Released VAO as VSAO (Vertebrate Skeletal Anatomy Ontology) to be consistent with our PloS One paper in press (see PONE-D-15442R2 “A unified anatomy ontology of the vertebrate skeletal system”)
- Created one ontology file “phenoscape-ext” with distributed editing

[http://phenoscape.svn.sourceforge.net/viewvc/phenoscape/
trunk/vocab/edit/phenoscape-ext.owl?view=log](http://phenoscape.svn.sourceforge.net/viewvc/phenoscape/trunk/vocab/edit/phenoscape-ext.owl?view=log)

Phenoscape-ext

- Where there existed an equivalence axiom in uberon to a TAO, AAO, or VSAO class, the classes were rolled up into an uberon-core class.
- Subclasses from these ontologies are retained as uberon subclasses within specific ID ranges for each
- Existing defs from TAO, AAO, VSAO are preserved using ‘external definition’ property and annotated with the source



Ontology workflow

- Adopted Protégé 4 ontology editor and OWL2 format
- Trained ontology curators in use of SmartSVN, Protégé, and phenoscape-specific ontology editing workflow (also they attended NESCent Anatomy Ontology course)
- Ongoing weekly hands-on ontology edit training
- Documented workflows for ontology editing
http://phenoscape.org/wiki/Ontology_workflow
- Fast access during annotation to changes in phenoscape-ext by Phenex, immediate access to temporary terms via the Term Broker

Ontology Attribution

One concern was how to property attribute ontology contributions. We now have the following:

- Definitions are annotated with source
- ‘created_by’ property to indicate term editor
- Specific property ‘external_definition’ that is annotated with the source

Next six months

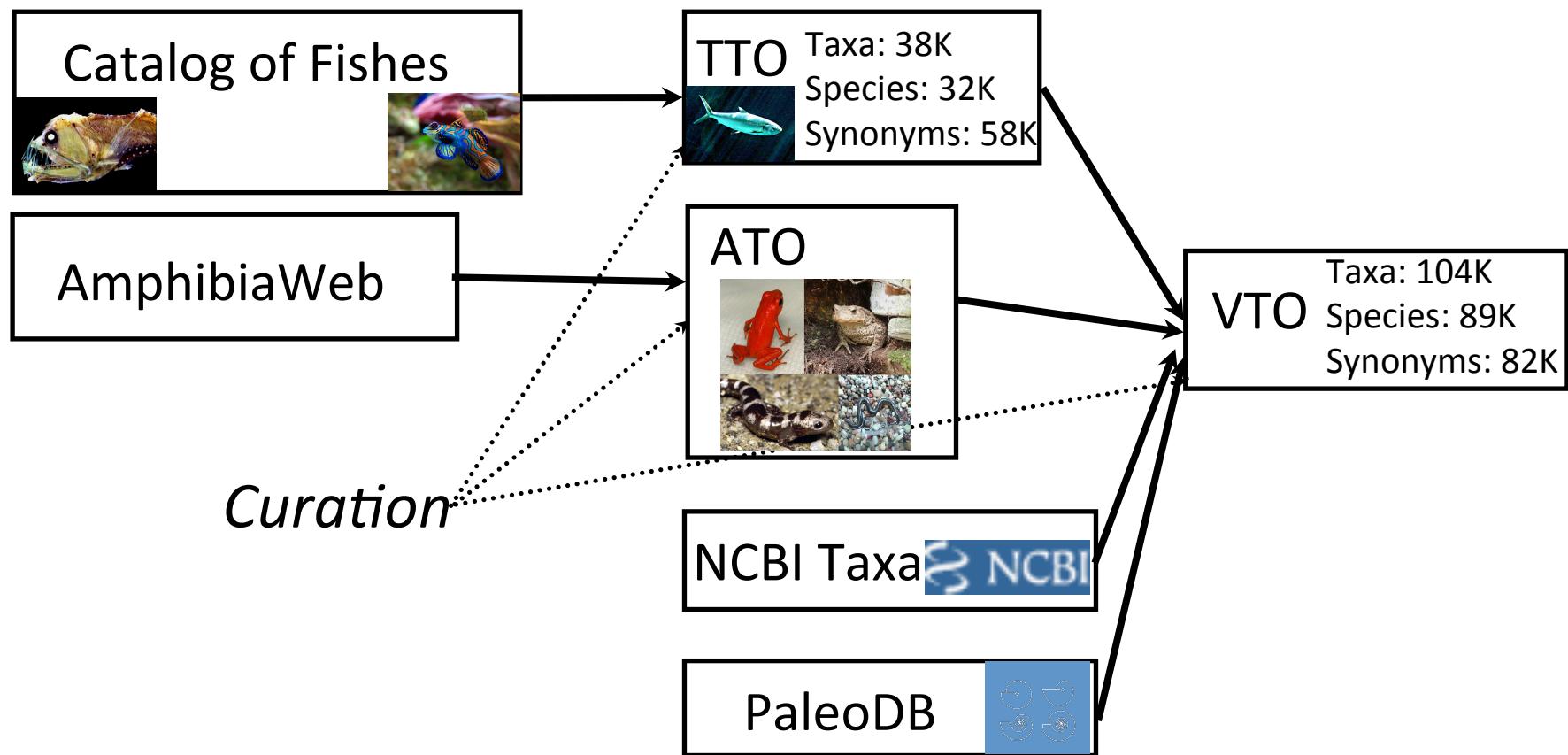
- Address outstanding TAO tracker items
- Continue further defining ingested classes
- Obsoleting redundant or incorrectly generalized classes
- Updates based on serial homology discussions at RCN (e.g. digit count, etc.)
- Enhance further avian-specific anatomical structures (many in uberon already)
- Use ORCID IDs to for author annotation (as per developing OBO Foundry best practices)

2. Ontology development

2b. Taxonomy

- Vertebrate Taxonomy Ontology (VTO)
 - Not itself an authoritative taxonomy
 - But derived from authoritative and widely used sources
- Participants: **Peter**, Jim, David, Alex, Nizar
- Since April
 - Interoperability with NCBI identifiers
 - Integration with PaleoDB
 - Add xrefs to Fishbase species pages

VTO building blocks



<http://purl.obolibrary.org/obo/vto.owl>

Releases

- July 2012
 - VTO - no VTO identifiers; incorporated PBDB
- November 2012
 - VTO - NCBI, PBDB update; Fishbase links; resolve some release process issues
- December 2012
 - ATO - AmphibiaWeb update
 - TTO - update from CoF, resolve a few problems found in VTO release
 - VTO - updates from NCBI, TTO, ATO, PBDB, resolve basal tetrapod issues, go though release process with Jim

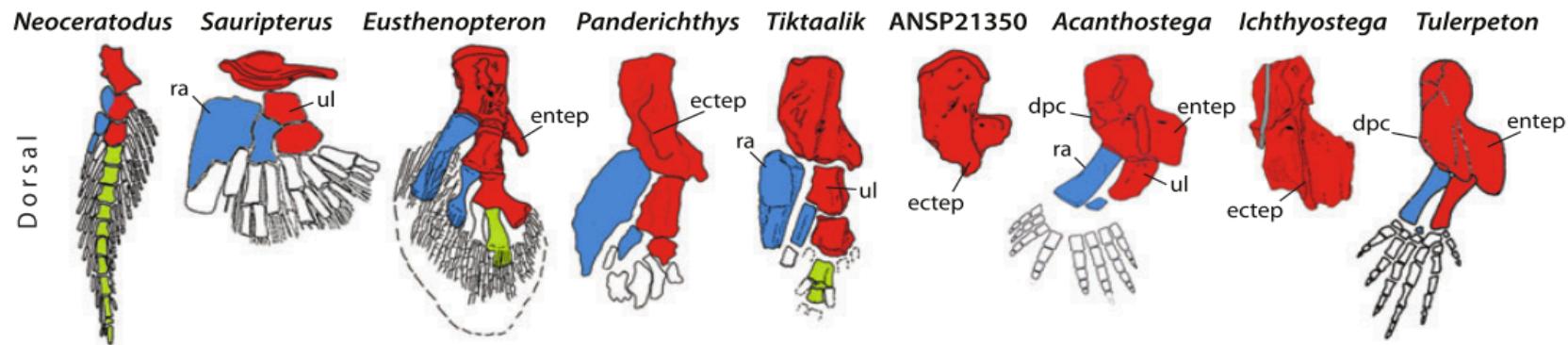
Next steps

- Stable tool ready to be used
 - VTOTool can generate VTO and ATO updates using appropriate scripting
 - Now generates OBO that is compatible with the OBO Ontology Release Tool
- Updating wiki documentation in December
 - How to maintain and execute scripts

3. Phenotype curation

3a. Evolutionary data

Motivation: fin/limb species phenotypes from published literature required for capstone test case



Clack, J. A. (2009). The Fin to Limb Transition: New Data, Interpretations, and Hypotheses from Paleontology and Developmental Biology. *Annual Review of Earth and Planetary Sciences*, 37(1), 163-179

Participants: Wasila, **Alex**, Nizar, Melissa, Paula, David, Paul

Phenotype annotation of taxon

Vertebrate Taxonomy
Ontology
(104,746 terms)



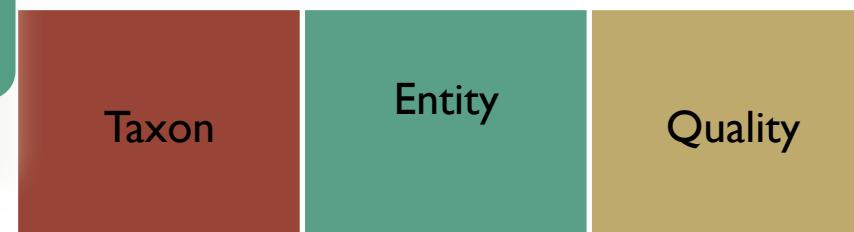
species 1

Uberon Anatomy
Ontology
(10520 terms; 27743
synonyms)



species 2

Quality Ontology
(PATO)
(1,363 terms, 753
synonyms)



species 1

species 2

Head

Head

cylindrical

flat

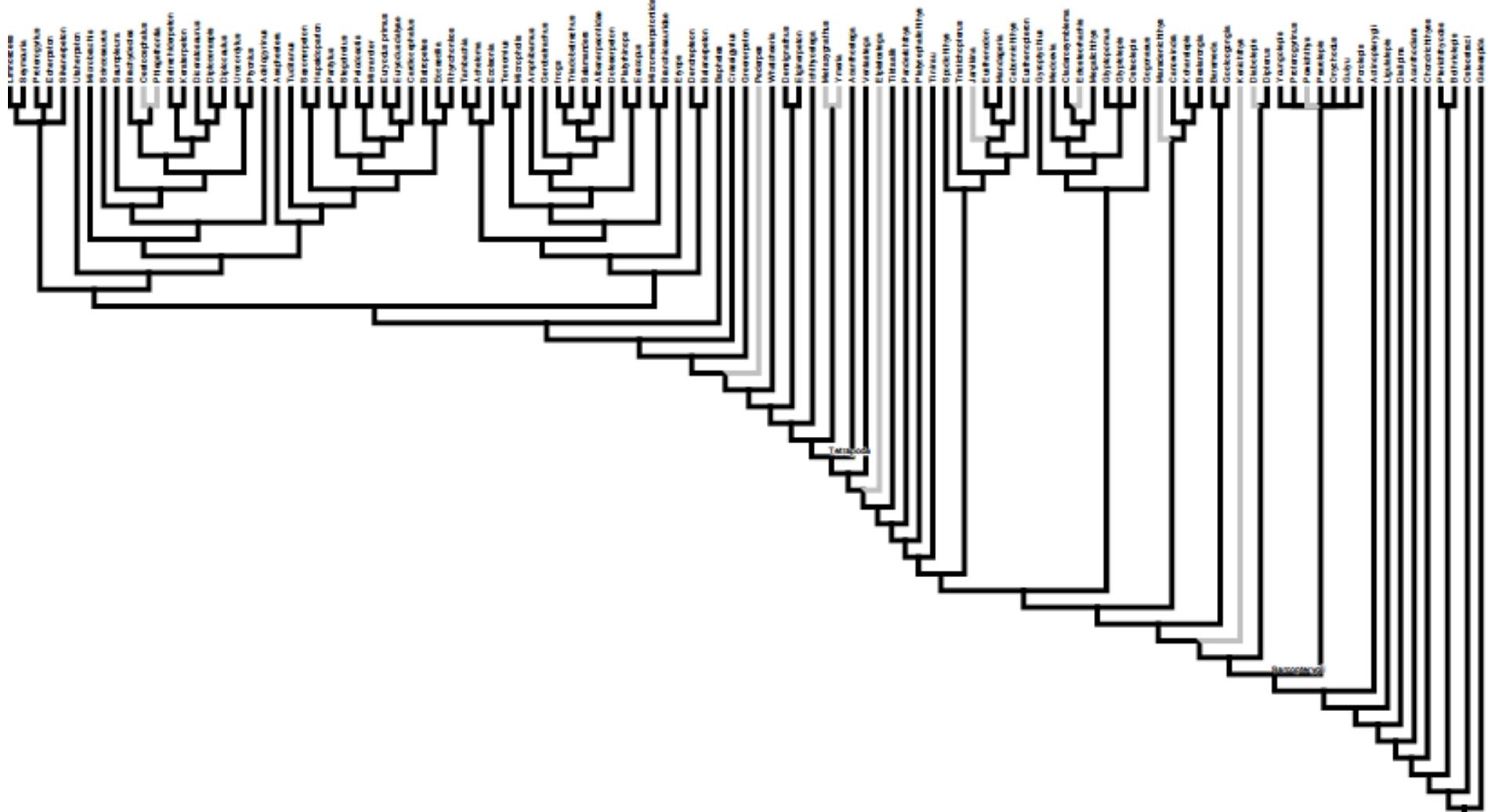
Taxonomic scope of annotation

Tetrapoda

Fishes

Amniotes

Amphibians



Curation objectives

Taxon	curator	# of papers	# of fin/ limb characters	# characters curated to date
Fishes	Wasila	19	218	108 (49%)
Amphibians	Alex	41	888	584 (66%)
Amniotes	Nizar	6	295	164 (56%)

- Amphibian dataset: 653 OTUs
- Minimum of 41318 annotations to OTU
- First draft of curation is targeted for completion by January 1st 2013

Curation software improvements

- ORB (ontology request broker) function permitting the creation of temporary term id's has increased curation rate.
- Polymorphism support⁶.
- Requested term addition to Phenoscape ext file ongoing

Annotation quality control⁵

- Goal: improve inter-curator consistency in annotation
- Items completed:
 - Relational qualities: Analyzed annotations to determine which relational qualities (e.g., ‘fused with’) should be used in phenotype syntax for EQE
 - Reviewed all annotations for consistency in use of relational qualities
 - Proposed “annotation checker” panel for Phenex; provides user with feedback on use of relational qualities

Annotation quality control⁵

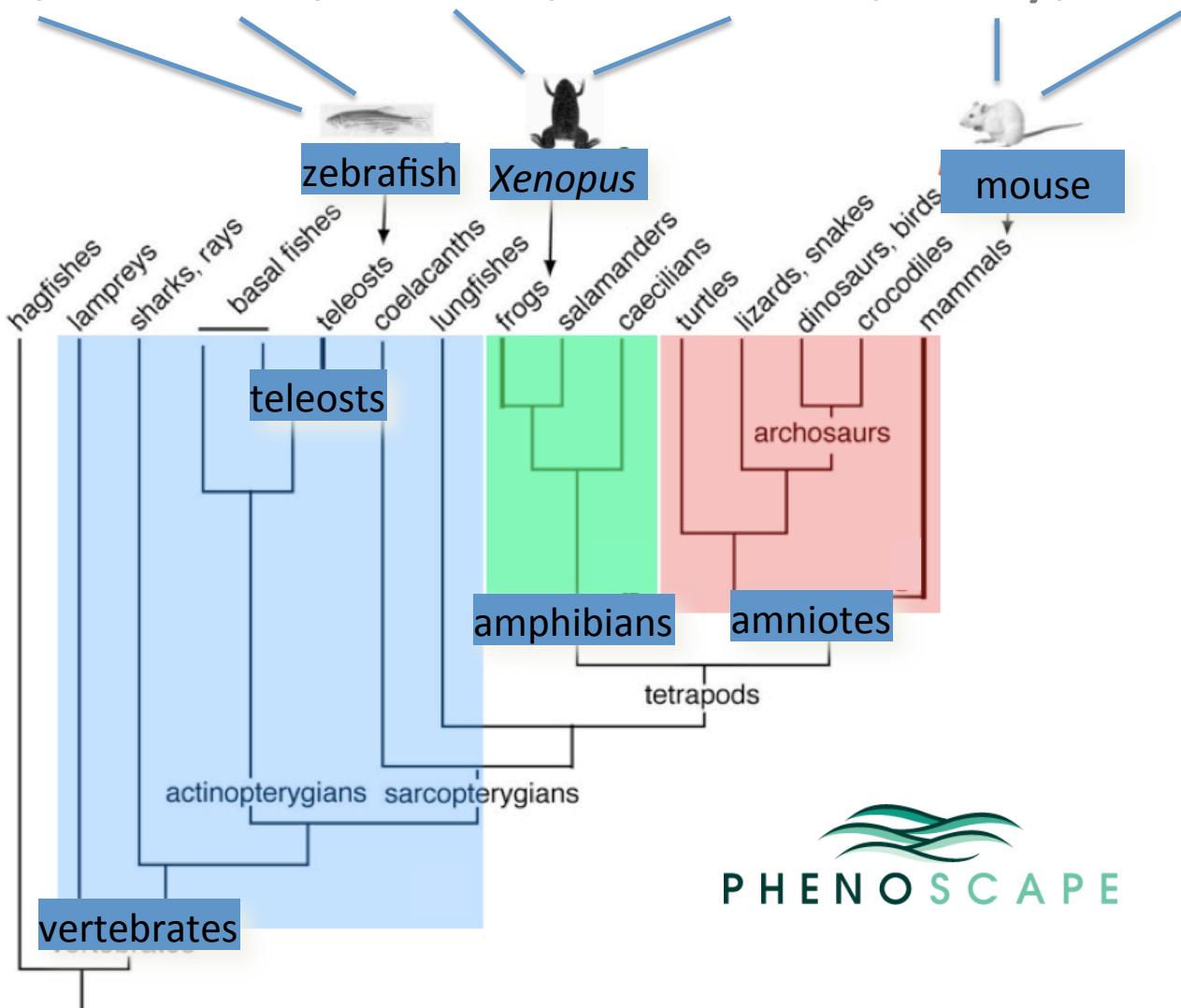
- Relations used in post-composition (in progress)
 - Determine which relations (e.g., *anterior_to*) should be used for post-composed entities
 - Analyze list of relations used in annotations and relations used in Uberon
 - Analyze annotations for consistency in use of relations
 - Constrain the post-composition editor in Phenex to the list of “approved” relations

PATO term additions¹⁰

- PATO
 - 14 PATO terms, 2 synonyms added since June 2011
 - 2 other changes (updated definition)
 - 38 provisional terms created (ORB)
- Met with George Gkoutos in RCN meeting, October 2012

3. Phenotype curation

3b. Model organisms: (Monte, Ceri, Yvonne, Aaron, Christina, Judy, Terry)



PHENOSCAPE

Model organism phenotypes

- **Aim**
 - Annotate skeletal phenotypes for fin and limb for genetic mutants from zebrafish, Xenopus, and mouse
- **Milestones**
 - Curation of phenotypes & gene expression
 - Finalize what phenotype data will be imported into Phenoscape KB
 - Develop pipelines for uploading Xenbase and MGI data to Phenoscape
 - Incorporate phenotype & gene expression into KB

Model organism phenotypes

Progress – ZFIN (April - November 2012)

- 65,708 phenotypes for ~4543 genes (+14%)
 - 852 fin phenotypes
 - 2167 publications with phenotypes
 - Includes 7 direct submissions with 11,624 phenotypes

Model organism phenotypes

Progress – MGI

- 2000 limb phenotypes for ~450 genes
- Phenotypes curated as ~150 MP (mammalian phenotype) terms
- MP terms mapped to EQ (complete for limb)
- Mappings are maintained up to date

Model organism phenotypes

Progress – Xenbase

- Released new XAO v2.0.8 (in OBO foundry 10/23/2012)
- Ongoing annotation of limb/fin gene expression
 - 202 papers, 81 genes with expression images
- Established Phenote configuration (with Jim Balhoff) to annotate limb/fin phenotypes

Model organism phenotypes

Immediate future plans – ZFIN

- Continue curation
- Import gene expression data into Phenoscape

Model organism phenotypes

Immediate future plans – MGI

- Continue curation
- Maintain limb MP->EQ mappings
- Develop pipeline to import mappings into Phenoscape (nearly ready)
- Import anatomy terms & developmental stages (EMAPA) into Phenoscape (soon)
- Import gene expression data into Phenoscape

Model organism phenotypes

Immediate future plans – Xenbase

- Continue curation
- Finalize import of gene expression data into Phenoscape KB
- Begin curating limb phenotypes

Model organism phenotypes

Deliverables

- Complete & up-to-date phenotype & gene expression data for 3 model organisms
- Add human (HPO) and bird data (RCN)⁷
- Paper – methods & challenges of integrating such data

4. Homology Reasoning

- Enable integration of phenotypes across the fin-to-limb transition in vertebrate evolution.
 - Fin and tetrapod limb skeletons represented by different classes
 - Yet parts are biologically the “same” by common origin, or by developmental program.
- Enable computing over homology assertions as hypotheses.
 - Subclassing is often too strong: assertions conflict with each other and vary in acceptance.
 - Require attribution, evidence, and provenance.
- Participants: Hilmar (coordination), Chris (logic formalization); Paula, David, Paul, Alex, Nizar (Comparative vertebrate morphology)

Deliverables

1. Comprehensive collection of vertebrate fin/limb homology assertions.
2. Capture rigorous homology assertions in the KB.
3. OWL/RDF data model for representing homology in the KB.
4. Integrate homology-based results in KB user-interface.
5. Integrate homology into semantic similarity scoring.

Progress Since April 2012

- Met with experts in vertebrate limb development and evolution to document homology use-cases and reasoning requirements.
 - Requirements consistent with subsumption reasoning.
 - Obviates special treatment of “default homology”.
- Developed and contrasted alternatives to represent homology assertions in OWL, including the vHOG and VBO approaches.
 - Revealed deficiencies of previously developed OWL model.
- Curated a test-set of cross-vertebrate fin/limb iterative and phylogenetic homologies.

Goals for next six months

1. Capture mechanism for homology assertions:
 - Explore tools for data entry (Open Refine, Rightfield)
 - Automatic conversion into OWL and loaded into KB.
2. Preliminary collection of fin/limb homologies for testing
3. OWL/RDF data model for homology in the KB

5. Semantic similarity

- Goals
 - A statistical method for identifying similar phenotype profiles among or between genotypes and lineages,
 - Incorporate ontology structure, annotation density, partial knowledge of evolutionary variability, and homology
 - Address challenges of speed and scalability
- Participants
 - Jim, Judy, Chris, Hilmar, Paula, (Prashanti), Peter, Todd

Semantic similarity

Pristigaster (123)

Dorsal fin position
Orbitosphenoid present
Lateral line length
decreased
...

sub (4)

Anatomical system abnormal
Neuromuscular process
controlling balance
abnormal
Otolith abnormal
Sensory system abnormal

Account for ontology structure, information content, messy biology

- Terms need not necessarily match lexically
- There need not be a one-to-one match between phenotypes, for biological and methodological reasons
- Similarity between common phenotypes is less informative
- A match in quality alone is not meaningful

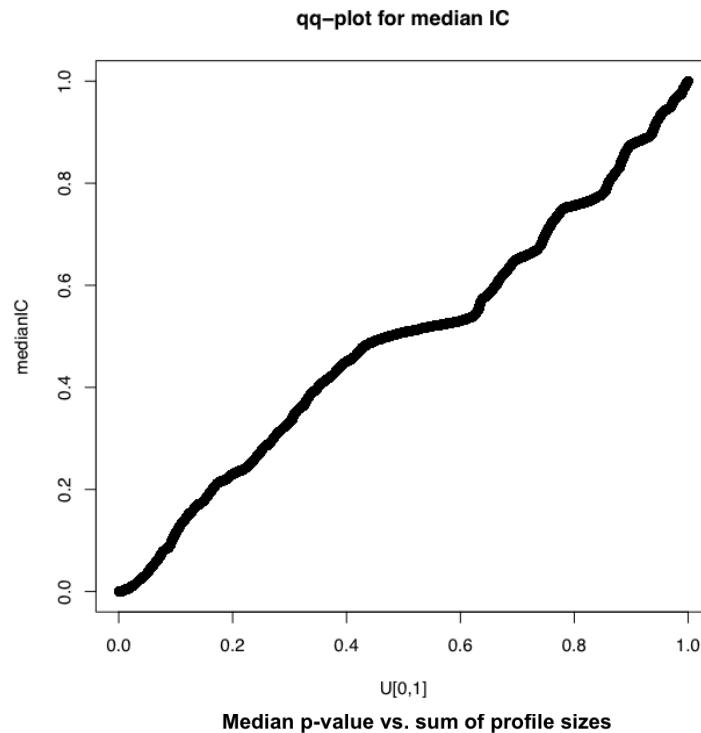
Semantic similarity

Progress to date

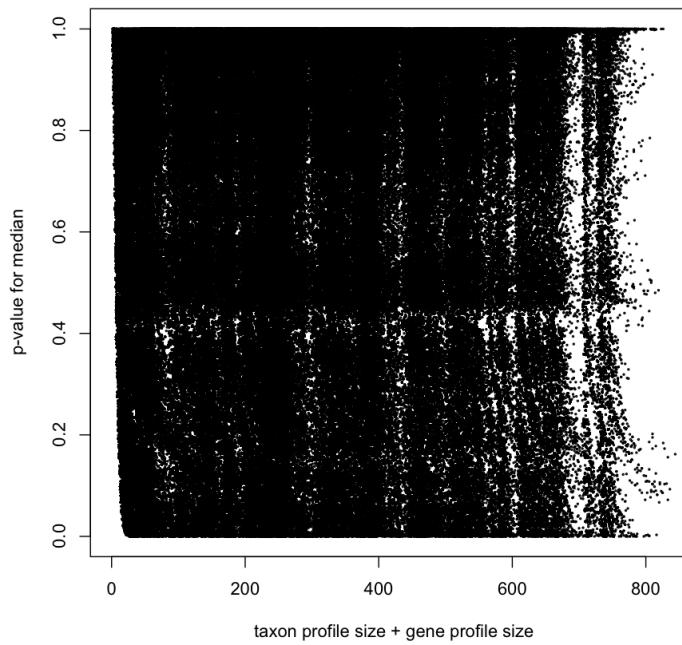
- Initially planned yrs, 1-2, mostly shifted to yrs. 2-3
- Results from Phenoscape I motivated us to develop stats robust to profile size differences
- Method:
 - medianIC: median pairwise information content
 - Used resampling to derive p -values for each possible profile size comparison

Key statistical results

1. p -values approximately uniformly distributed



2. p -value distribution is independent of profile sizes



Application to Phenoscape I data

- 4,159 gene profiles from ZFIN
- 378 evolutionary profiles from Ostariophysi
- medianIC calculated for all 1,572,102 profile pairs
- qualities compared within attributes
- *p*-values derived from 100K permutations
- 16 significant pairs expected under H_0
- 1,362 significant pairs at $p < 1 \times 10^{-5}$
- False Discovery Rate = 1.15%

An example

Hexanematichthys

1. gill raker of posterior margin of gill arch 2 skeleton - count
2. lateral line of caudal peduncle - shape
3. posttemporal fossa - size

cacna1da

1. behavior - quality
2. musculoskeletal movement - quality
3. neuromast hair cell - quality
4. neuromuscular process controlling balance - quality
5. neuron-neuron synaptic transmission - quality
6. posterior lateral line neuromast hair cell development - quality
7. plasma membrane of neuromast hair cell - shape

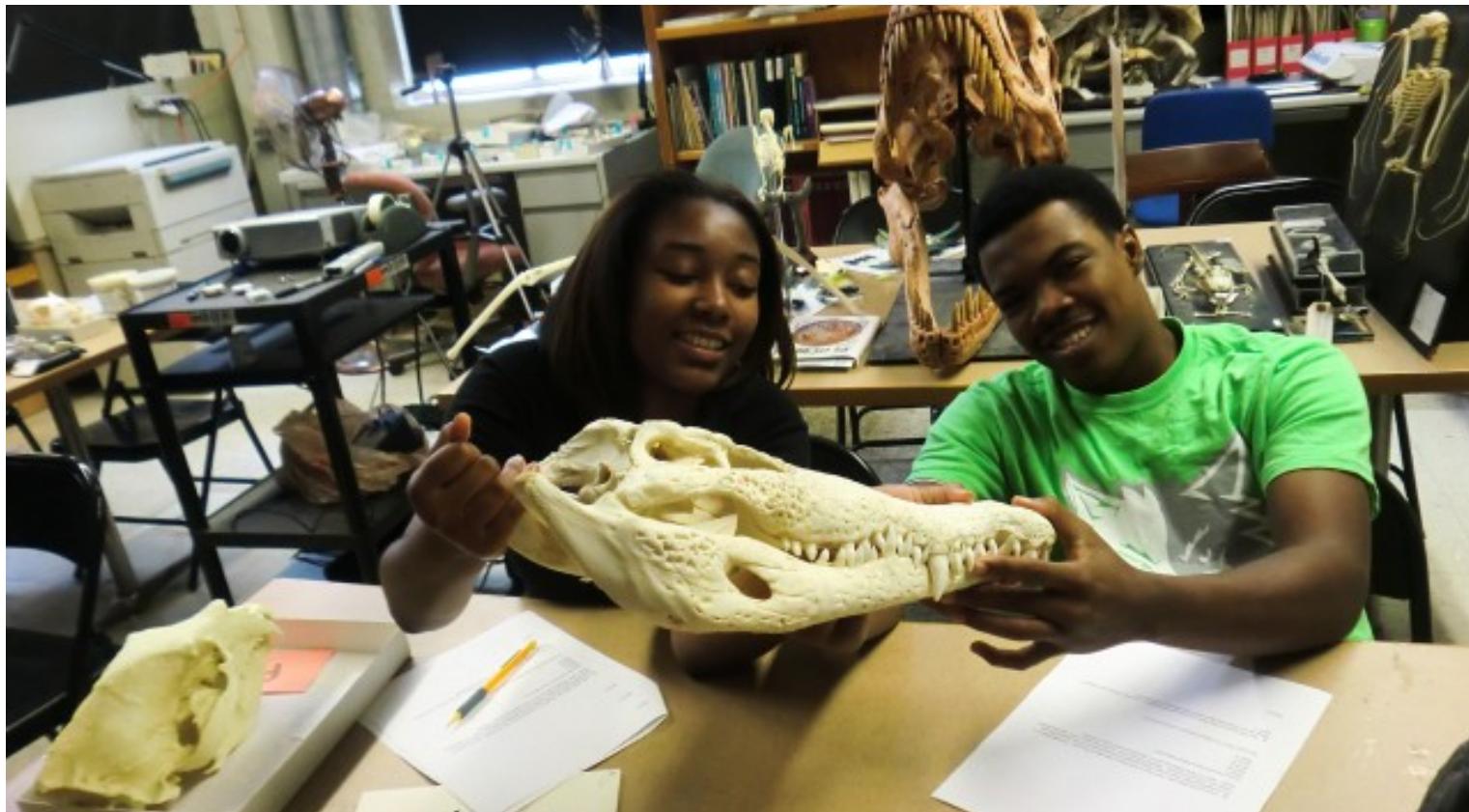
medianIC = 11.8, $p < 1 \times 10^{-5}$

Future plans (six months and beyond)

- Develop
 - Validation tests
 - Methods for meta-interpretation (e.g. enrichment, filtering, visualization, integration with external genetic data)²
 - Methodology for including homology statements
- Explore areas for improvement
 - Better statistics for highly improbable matches
 - Precomputed database representation
- Consider alternative approaches
 - Bauer *et al.* (2012) Bayesian network approach
- KB implementation

6. Outreach

- Participants: **Nizar, Paul, Paula**



Junior Biocurator program, Summer 2012

Junior Biocurator Program

- 6 week program at the University of Chicago, in collaboration with Project Exploration (Sereno, Ibrahim)
 - 5 students
 - Phase 1: Introduction to Morphological Biocuration
 - 2 weeks
 - Phase 2: Biocuration Imaging (Photography)
 - 3 weeks
 - Phase 3: Biocuration Imaging (Scanning)
 - 1 week
- Images to be used in Phenoscape anatomy ontology

SURE Program

- 10 week student internship program for American Indian undergraduate students at the University of South Dakota (Mabee)
 - 2 students
 - Anatomy, whole mount preparation, dissection (fishes)
 - Data entry (Phenex), databases (Phenoscape KB; OMIM), literature reviews
 - Weekly lectures on fin/limb development, genes
 - Human limb phenotype related to fin phenotypes
 - Poster presentations
- Paired fin loss data available in Phenoscape KB



Development of the hindlimb and pelvic fin

Char Green, Alyssa Block, Dr. Paula Mabee
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Introduction

When we as humans see something out of the ordinary it triggers many questions as to why, or how that could have happened. That is why fin and limb development and the associated genes are well studied. The gene *tbx4* is involved in hindlimb development in humans and mouse and pelvic fin development in zebrafish¹. Using biocurator software and tools, I annotated data to a new evolutionary phenotype database that is linked to genes of model organisms (Phenoscope Knowledgebase; kb.phenoscope.org), and soon to be connected to human phenotypes. I researched and related human forelimb genes and genetic phenotypes as found in the human genetic OMIM database to the pelvic fin anatomy that I studied in zebrafish.

Anatomy and development of limb

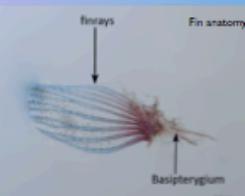


Figure 1: Pelvic girdle and fin of zebrafish consisting of the basipterygium and fin rays.

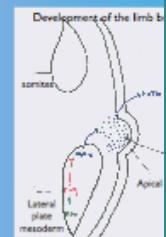


Figure 2: Genes needed for limb development in lateral plate mesoderm is where *tbx5* is expressed (AER = Apical Ectodermal Ridge).

Genes for hindlimb/ pelvic fin

The gene *tbx4* participates in hindlimb development and is expressed in the lateral plate mesoderm. When *tbx4* is not expressed in the hindlimb/ pelvic fin they cause malformations in humans².

Conclusions

When *tbx4* and *ptx1* are not expressed in the hindlimb/ pelvic fin they cause malformations in humans².

Your Inner Fin: Studying Zebrafish Pectoral Fin Development to Understand Human Forelimb Mutations



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Introduction

Fin and limb development is well-studied in humans and other animals, and many of the genes involved can be related to anatomy. Some genes, such as *tbx5*, which are involved in pectoral fin development in fishes, have a conserved function in human limbs; e.g., humans with defects in the *tbx5* gene have malformations in their upper limbs (Holt-Oram Syndrome). Using biocurator software and tools, I annotated data to a new evolutionary phenotype database that is linked to genes of model organisms (Phenoscope Knowledgebase; kb.phenoscope.org), and soon to be connected to human phenotypes. I researched and related human forelimb genes and genetic phenotypes as found in the human genetic OMIM database to the pectoral fin anatomy that I studied in zebrafish.

Materials and Methods

To learn the anatomy of the pectoral girdle, cleared and stained specimens of the zebrafish, *Danio rerio*, were examined using a Wild M5 stereomicroscope. The pectoral girdle was dissected from the specimens, and images were captured using a Leica DFC280 camera and processed using Adobe Photoshop CS3 Extended. The databases ZFIN, MGI, OMIM, Phenoscope, PhenExplorer, and NCBI were used to gather gene data as well as data on human syndromes. Curation and annotation of the scientific literature concerning fin/limb loss was done using Mendeley, Mesquite, and Phenex.

You Are A Mutant!

Everyone is a mutant! Some of your 43,441 genes³ are likely duplicated, deleted, or otherwise mutated, making you a mutant with varying phenotypic attributes. Giving each of us a variety of abnormalities with varying severity.

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7. Knowledgebase reasoning & development

- Participants: **Jim**, Chris, Hilmar
- Goal:
 - Integrate the ontologies, homology assertions, EQ phenotype annotations, model organism genes and phenotypes, wild-type gene expression, and semantic search tool into a knowledgebase (KB).
 - The KB will enable discovery through multiple machine and human interfaces.

kb.phenoscapes.org

- Current “production” system
 - OBD relational database
 - Java-based web services application: JSON data
 - Web interface: Ruby on Rails
- Interface subjected to several rounds of user testing
- Reasoning process: Perl/SQL materialization of inferred statements
 - Limitations on semantics, scalability

Phenoscape Knowledgebase

Go Duke SSL VPN Service Google Wikipedia FogBugz Phenoscape NESCent Intranet Import to Mendeley mx SourceForge Hudson Java 1.6 > +

 The Phenoscape logo features the word "PHENOSCAPE" in a bold, sans-serif font. Above the letter "P", there is a stylized graphic of three wavy lines in shades of teal and green.

The Knowledgebase currently contains 565,158 phenotype statements about 2,527 taxa, sourced from 57 publications, as well as 37,004 phenotype statements about 4,640 genes, retrieved from ZFIN. The data were last loaded into the Knowledgebase on 2012-11-15.

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

Feedback

Workspace About Acknowledgments

You can use the tools below to explore the Knowledgebase in different ways. Click the help  icons below to learn more about each tool.

Or, try the [Phenoscape Knowledgebase tutorial](#) to get started.

Find a specific term

Type at least 3 characters to select from matching ontology terms. If there are many matches, the autocomplete menu may take a few seconds to respond. Search 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.

Browse data (faceted view)

[Phenotypes](#)

Browse ontologies (tree view)

[Taxa](#)
[Anatomical entities](#)
[Phenotypic qualities](#)

Query data

[Phenotypes](#)
[Phenotype annotations to taxa](#)
[Phenotype annotations to genes](#)
[Taxa](#)
[Genes](#)
[Comparative publications](#)

Visualize data

[Phenotypic profile tree](#)
[Phenotype variation tree](#)

Phenoscape News

Society of Vertebrate Paleontology Annual Meeting 2012 (Raleigh, NC)
by carcharodontosaurus - Nov 27, 2012
The Phenoscape project had a strong presence at the largest Vertebrate Paleontology/Comparative Anatomy conference in the world ...

Phenex 1.6 released
by Jim - Oct 10, 2012
Phenex 1.6 has been released. Updates: Support for entry of polymorphic values in matrix cells (documentation). Improvements to ...

Knowledgebase tutorial now available
by Jim - Oct 04, 2012
The Phenoscape team has created a tutorial introduction to the Knowledgebase. The tutorial is designed to introduce users to ...

Junior Biocurator
by carcharodontosaurus - Sep 11, 2012
This summer, with the help of non-profit organization Project Exploration (<http://www.projectexploration.org/>), we ran the ...

DILS 2012
by Jim - Aug 28, 2012
In June I had the opportunity to attend DILS 2012 (Data Integration in the

Browse phenotypes « Phenoscope

kb.phenoscope.org/phenotypes/facets?facet_paths%5Bentity%5D=TAO%3A0001114%2CTAO%3A00005478 C Reader

Duke SSL VPN Service Google Wikipedia FogBugz Phenoscope NESCent Intranet Import to Mendeley mx SourceForge Hudson Java 1.6 >> +

PHENOSCAPE

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.

Feedback

Home Browse phenotypes Workspace About Acknowledgments

Phenotypes involving any quality for mouth with any related entity in any taxon or any gene

View these results as a [phenotypes query](#)

Refine Search:

Phenotype	« Previous	Results 1 - 20 of 1139	Next »
Any entity (19097) <input type="button" value="Choose"/> head (6117) mouth (1139 phenotypes) buccal papilla (6) dental plate (8) lip (19) oral cavity (9) palatoquadrate arch (735) ventral mandibular arch (341)			
Any quality (1139 phenotypes) <input type="button" value="Choose"/> decreased quality (104) increased quality (99) occurrence quality (2) physical object quality (899) qualitative (433)			
Any related entity (1139 phenotypes) <input type="button" value="Choose"/> anatomical cluster (15) anatomical space (4) compound organ (152) multi-tissue structure (16) organism subdivision (10)			

	Entity ↓	Quality	Related Entity
	Meckel's cartilage	fused with	Meckel's cartilage
	Meckel's cartilage	overlap with	Meckel's cartilage
	Meckel's cartilage	attached to	coronomeckelian
	Meckel's cartilage	alignment	palatoquadrate cartilage
	Meckel's cartilage	fused with	palatoquadrate cartilage
	Meckel's cartilage	fused with	ceratohyal cartilage
	Meckel's cartilage	position	coronoid process
	Meckel's cartilage	angle increased in magnitude relative to normal	anterior/posterior axis of body
	Meckel's cartilage	protruding	
	Meckel's cartilage	kinked	

Query phenotype annotations to taxa < Phenoscaper
kb.phenoscaper.org/taxon_annotations?filter%5Btaxa_match_type%5D=any&filter%5Btaxa%5D%5B0%5D=TT

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.

Feedback

Home **Query phenotype annotations to taxa** **Workspace** **About** **Acknowledgments**

Query for:

Phenotype annotations to genes

Taxon is:
Siluriformes [Add](#)

and

Phenotype is:
mouth [broaden/refine] [X](#)
 including parts

and

Publication is:
 Any

Apply higher taxon annot all included species [X](#)

« Previous **Results 1 - 20 of 36844** **Next »**

Phenotype				
Taxon ↓	Entity	Quality	Related Entity	Source
<i>Acanthicus histrix</i>	entopterygoid	present		View
<i>Acanthicus histrix</i>	quadrate	structure	preopercular sensory canal	View
<i>Acanthicus histrix</i>	process of quadrate	present		View

Source Data

<i>Acanthicus histrix</i>	entopterygoid	present	
---------------------------	---------------	---------	--

Friel 1994
character 25: Endopterygoid
state: endopterygoid present
as taxon: *Acanthicus histrix*
curated by: Jeffrey Engeman

Query phenotype annotations to genes « PhenoscAPE

kb.phenoscAPE.org/gene_annotations?filter%5Btaxa%5D%5B0%5D=TTO%3A1380&filter%5Bphenotypes%5D%5B0%5D=acvr2aa

Duke SSL VPN Service Google Wikipedia FogBugz PhenoscAPE NESCent Intranet Import to Mendeley mx SourceForge Hudson Java 1.6

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.

PHENOSCAPE

Feedback

Home Query phenotype annotations to genes Workspace About Acknowledgments

Query for:

Phenotype annotations to genes

Gene is: Any Add

and

Phenotype is: mouth [broaden/refine] including parts

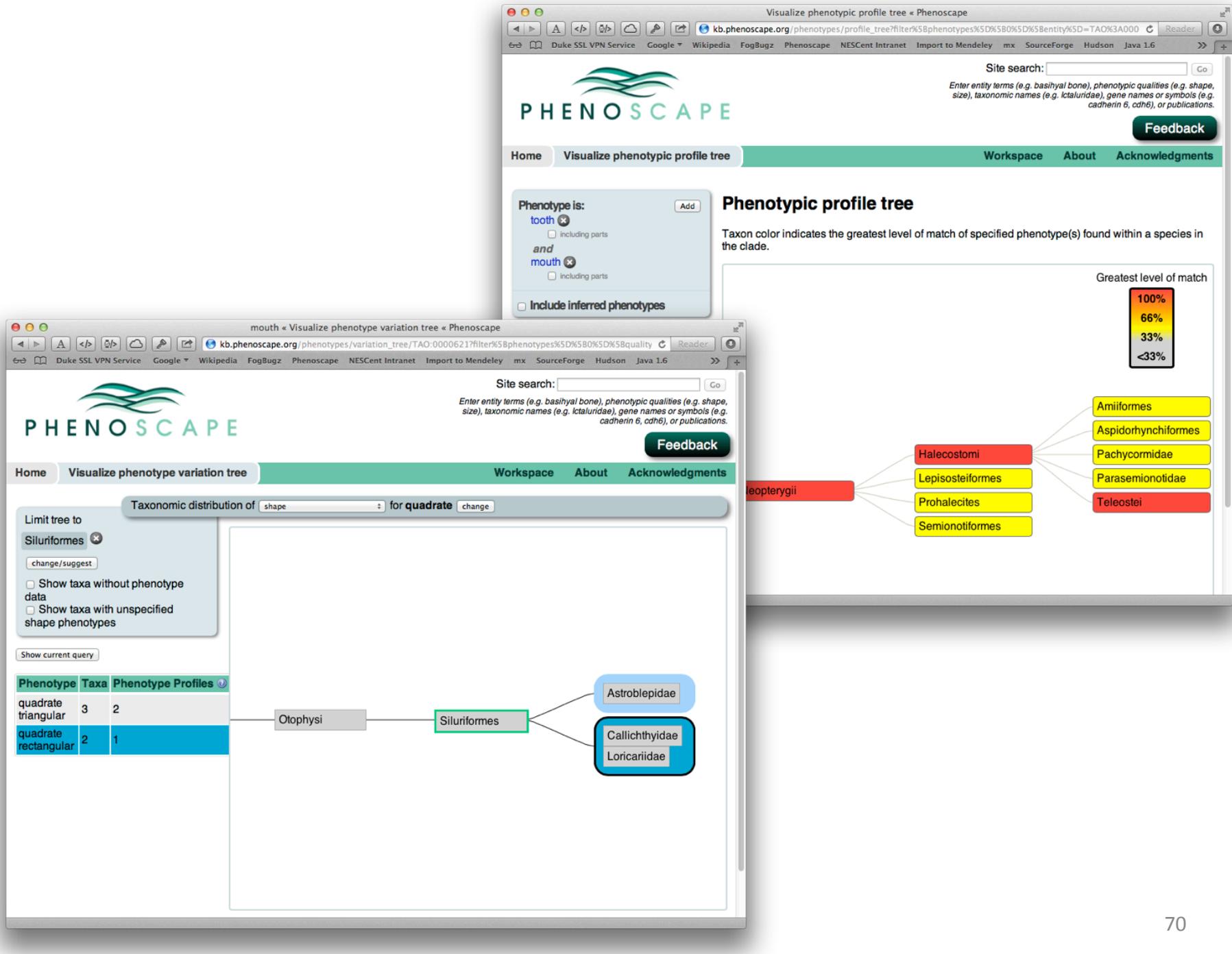
Results 1 - 20 of 509					
Phenotype					
	Gene ↓	Entity	Quality	Related Entity	Source
	acvr2aa	maxilla	fused with	maxilla	View
	acvr2aa	Meckel's cartilage	fused with	palatoquadrate cartilage	View
	acvr2aa	palatoquadrate cartilage	fused with	hyosympathetic cartilage	View

Source Data

acvr2aa	maxilla	fused with	maxilla	
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Genotype or morpholino: MO1-acvr2aa — Albertson, R.C., Payne-Ferreira, T.L., Postlethwait, J., and Yelick, P.C.. 2005. Zebrafish acvr2a and acvr2b exhibit distinct roles in craniofacial development. *Developmental Dynamics* 233(4):1405-1418.

[View all phenotypes for this gene at ZFIN](#)



Current focus

- Move data and reasoning to RDF/OWL
 - Address issues with scalability, expressivity, data standardization/reusability
 - All data downloadable in bulk:
<https://github.com/phenoscape/phenoscape-kb>
- Defer development of new complex user interfaces until driving projects emerge from collaboration workshop

Progress since April 2012

- RDF/OWL KB build pipeline maturing
 - Configurable system for downloading ontologies and data, converting to OWL, performing pre-reasoning, loading into triple-store
 - Now incorporates Uberon ontology, evolutionary data, ZFIN gene expression data
- Adopted BigData RDF triple-store
- Adaptation of web interfaces to SPARQL query service, in progress
- Active discussion of optimal data import strategies for combining data from model organism databases
- Added curator attribution to existing KB website

for: « Previous Results 1 - 20 of 514384 Next »

Phenotype annotations to taxa

is: Taxon ↓ Entity Quality Related Entity Source

type is: *Aramis brama* mesocoracoid bone separated from cleithrum

ation is:

Siebert 1987

character: Shoulder girdle [From Sawada 1982 character 37: relationship between 39: mesocoracoid and cleithrum]

state: absent [mesocoracoid and cleithrum separate]

curated by: Richard L. Mayden; Jeffrey Engeman

Aramis brama tripus shape

Aramis brama mesocoracoid bone separated from cleithrum

Next six months

- Complete addition of MOD data to OWL knowledgebase
 - ZFIN, MGI, Xenbase phenotypes and gene expression
 - HPO (human) annotations
- Advance from currently experimental SPARQL-based web interfaces to beta-quality website which provides much of the functionality of existing OBD-based system



III. Upcoming plans



Report of Phenoscape Scientific Advisory Board (27th April 2012)

1. Consider other large scale sources of structured data (Todd)
2. Further analyze candidate genes by incorporating gene set and pathway analysis for additional value to users (Todd)
3. Outreach, identification of user groups and real-life use-cases (Todd)
4. VAO and Uberon merge into one cross-species anatomy (Melissa)
5. Quality of the phenotype data capture, annotations (Alex)
6. Capturing inter-individual variation within taxa (Alex)
7. Practicality and utility of capturing data from birds and humans (Melissa & Monte)
8. Distribution of effort into developing tools vs. data capture & curation (Todd)
9. Concern over ontology term brokering with the NCBO tools (Jim)
10. Improvement in the granularity of phenotype descriptions, PATO (Alex)

Collaboration Workshop

- February 8-10, California Academy of Sciences
- NSF supplement obtained to involve expert facilitators (\$30K, KnowInnovation)
- Identified participants who bring data, ideas, time and interest in collaboration
- Confirmed outside participants (10)
 - Gill Berjerano, Stanford University
 - Stan Blum, California Academy of Sciences
 - Greg Carter, The Jackson Laboratory
 - Wyatt Clark, Indiana University
 - Matthew Harris, Harvard Medical School, Dept of Genetics
 - Hans Larsson, McGill University
 - Gavin Naylor, College of Charleston
 - David Parichy, Univ. Washington
 - Karen Sears, University of Illinois
 - Brian Sidlauskas, Oregon State
- Plus roughly equal number of Phenoscape participants



Publications & presentations (since April 2012)

Papers:

- Arighi CN *et al.* (*in press*) An Overview of the BioCreative 2012 Workshop Track III: Interactive Text Mining Task. Database
- Dahdul WM *et al.* (*in press*) A unified anatomy ontology of the vertebrate skeletal system. PLoS ONE
- Bertone MA *et al.* (*in press*) Matching arthropod anatomy ontologies to the Hymenoptera Anatomy Ontology: Results from a manual alignment. Database.

Posters, Conference talks, and Seminars:

- Sereno, P, Ibrahim N, Mabee P, Vision T, Lapp H. "Phenoscape: A New Anatomical Ontology of Vertebrates", Society of Vertebrate Paleontology, Raleigh NC, October 2012.
- Dececchi TA, Balhoff J, Dahdul W, Ibrahim N, Lapp H, Midford P, Sereno P, Vision T, Westerfield M, Mabee P Blackburn D. "Phenoscape: bridging the gap between fossils and genes", Society of Vertebrate Paleontology, Raleigh NC, October 2012.
- Mabee PM "Synthesis of evolutionary and developmental data for fishes using new informatics tools" (oral presentation) World Congress of Herpetology, Vancouver BC, August 2012.
- Balhoff JP, Dahdul WD, Lapp H, Mabee P, Midford P, Vision TJ, Westerfield M, and the Phenoscape Project Team. "The Phenoscape Knowledgebase: Integrating phenotypic data across taxonomy, from biodiversity to developmental genetics" (poster) DILS 2012, College Park, MD, June 2012.

See http://phenoscape.org/wiki/Publications_and_Posters



Larger questions. How do we...

... create a community of KB users?

... make sure there is a case to stakeholders for sustainability?

... support curation of additional taxa and genetic models?

... incentivize contributions of data from other researchers?

... promote community participation in maintenance of ontologies and software?