Some features of ontologies

- Controlled vocabulary (but oh so much more)
 - Enable machine communication
 - Can be used to annotate data
- · Logically defined relationships between terms
 - Enable logical reasoning
 - Expose data to generic query and analysis tools
- Serve as a community representation of knowledge



Phenotypes as structured text

AMERICAN MUSEUM NOVITATES

NO. 3286

APPENDIX 1. CHARACTER SUMMARY

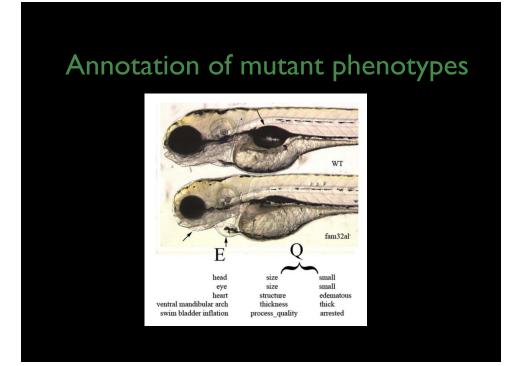
86

- Fifth infraorbital. 0, well developed, without contact between fourth and sixth infraorbitals;
 greatly reduced, with posteroventral margin of sixth infraorbital in contact with posterodorsal margin of fourth infraorbital.
- Antorbital-lateral ethmoid contact. 0, no contact; 1, antorbital contacting ventral wing of lateral ethmoid along its entire lateral edge.
- 3. Antorbital. 0, flat, platelike, without medial process; 1, with a short medial, vertically aligned process at its posterior edge that extends along posterior surface of ventral wing of lateral ethmoid; 2, with enlarged medial, vertically aligned process at its posterior edge that extends along posterior surface of ventral wing of lateral ethmoid.
- 4. Mesethmoid spine. 0, conical, or with a dif-

tilaginous surface at posterior portion of main body of vomer.

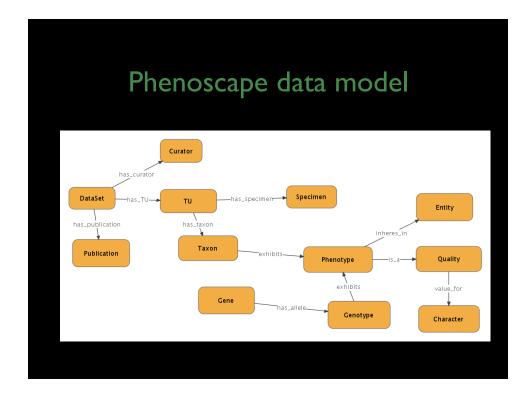
- 14. Portion on vomer for articulation of maxilla. 0, not modified in 1; 1, Presence of a shallow depression on its anterolateral surface where anterior tip of maxilla abuts.
- 15. Ridge on lateral surface of vomer. 0, absent;
 1, present.
 16 Phinesephere d, 0, present: 1, absent
- Rhinosphenoid. 0, present; 1, absent.
 Lateral ethmoid-orbitosphenoid contact. 0, absent; 1, present.
- Parasphenoid and main portion of orbitosphenoid. 0, well separated; 1, close to each other.
 Dilatator fossa. 0, not extending anteriorly on
- dorsal surface of frontal or if so, only to dorsoposterior edge of orbit; 1, highly developed, extending anteriorly on dorsal surface of frontal beyond dorsoposterior edge of orbit.

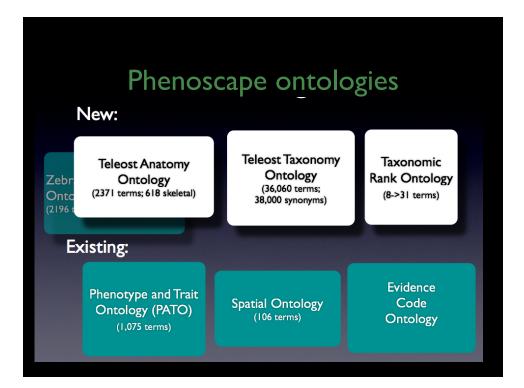
(Toledo-Piza 2000)



Reducing ontology complexity: use qualities that imply attributes

Α	Character		State
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	Entity	Q	uality
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В	triangular (PATO:0001875) type	e of (F	shape ATO:0000052)

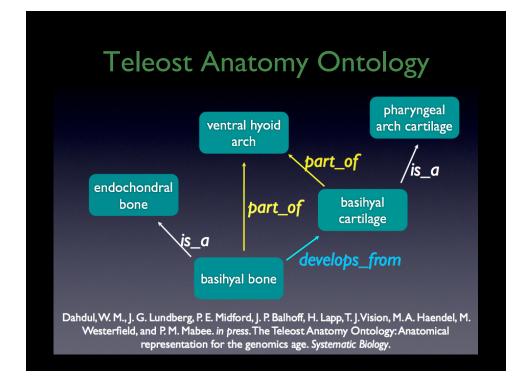






Teleost Anatomy Ontology

- Seeded from Zebrafish Anatomy Ontology
- Homology assertions are kept separately and attributed to an *authority* with an *evidence code*
- Participation is open
 - Mailing list with occasional jamborees
 - Ontology gatekeeper
- Ontology is built as needed for data annotation

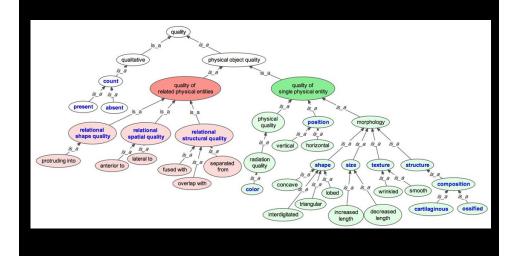


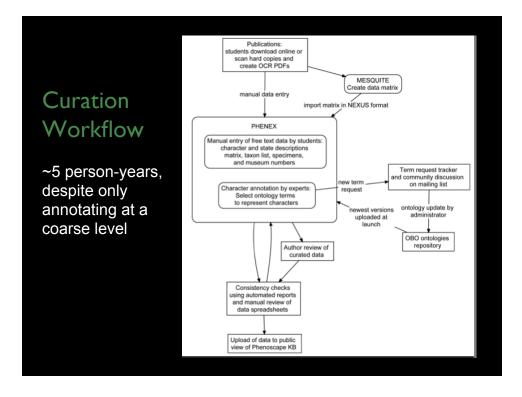
OBO Relations Ontology

- Foundational
 - is_a, part_of
- Spatial
 - Iocated_in, contained_in, adjacent_to
- Temporal
 - transformation_of, derives_from, preceded_by
- Participation
 - has_participant, has_agent

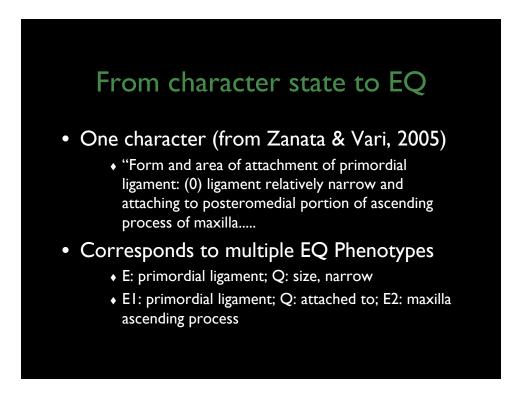
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Submitter: Paula Mabee	
Date Submitted: 2008-09-08	
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Magnification: NULL	
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Resolution (PPI): 100	
Submitted as: jpg	
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Angle: Dorsal	
Technique: Digital Camera	
Preparation: Cleared and counterstained for bone	
(Alizarin red) and cardlage (Alcian blue)	
Download: tiff (1.21 MB)	
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Copyright: Ericka Grey and Paula Mabee	
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Phenotype and Trait Ontology (PATO)



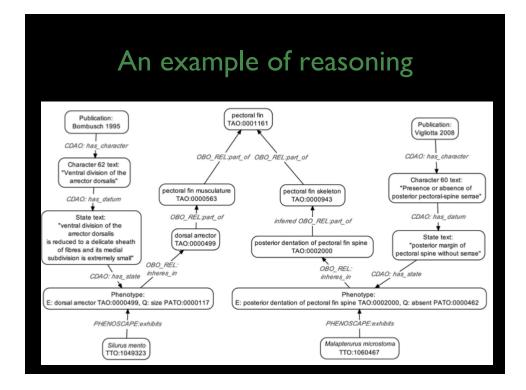


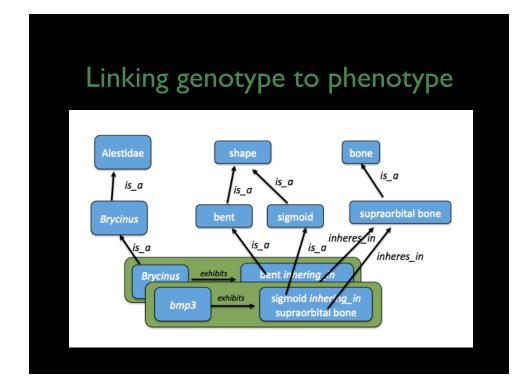
Complete Ontology Tree Vi Complete Ontology		C data c		ion	SO	ftware	
	Valid Taxon	Publication Taxon	Matrix Taxon	Comment	Figure		
	1 Acestrorhynchus lacustris 2 Brycinus lateralis	Acestrorhynchus lacustris	Acestrorhynchus	Buckup_19	198.xml		
	2 Brycinus lateralis 3 Boulengerella cuvieri	Characters D C = X		(*= 0 ×		r: Mesethmoid shape B (_ X)	(*= 0 ×
	4 Brycon guatemalensis			1.804			Treux
	5 Bryconops affinis				+ -		
	6 Characidium cf. zebra (Bucku)		Comment	Figure	Symbol State Desc	cription C	omment Figure
	7 Charax sp. (Buckup 1998) 8 Chilodus punctatus	1 Mesethmoid shape 2 Lateral ethmoidal wing	None	None	1 articular pr	poess of mesethmoid bone greatly reduced or missing N id trifurcate anteriorly, i.e. a pair of lateral processes proj N	one None
	9 Citharinus gibbosus	3 Ventral diverging lamellae of the mesethmoid bone		None	0 meseemo	o enurcate amenony, ne, a pair oriateral processes proj IV	one ivone
	10 Crenuchus spilurus	4 Region of mesethmoid-vomer joint	None	None			
	11 Ctenolucius hujeta	5 Mesethmoid-vomer joint 6 Attachment of the ectopeterygoid bone to vomer an	None d mesethmoid None	None			
	12 Cynopotamus argenteus	7 Rhinosphenoid bone	None	None			
	13 Distichodus maculatus	8 Frontal bone shape 9 Frontal fontanel	None	None			
	14 Hemiodus unimaculatus 15 Hepsetus odoe	10 Paired frontal foramina	None	None			
	15 Hepsetus odoe 16 Hoplias malabaricus	11 Anastomosis between supraorbital and pterotic ser		None			
	17 Piabucina panamensis	12 Frontal-pterotic joint 13 Dorsolateral margin of skull	None	None			
	18 Nannostomus unifasciatus	14 Canal of pterotic bone	None	None			
		15 Parietal fontanel 16 Parietal branch of supraorbital canal	None	None			
	Specimens for Taxon: Acestrorhy	17 Supratemporal laterosensory canal	None	None			
	n _R	18 Supraoccipital spine	None	None			
	* * -	19 Ventromedial opening of posttemporal fossa 20 Antorbital bone	None	None A			
(Collection	-31 Europachital hans	Mana	Mana T	-	,) 4 1
	UMMZ 2						24121
Graph View 🕨 🕈 🔞 🖬 🐔 🗕	· UMMZ 2	Phenotypes for State: 1 - articular process of meseth	mold bone greatly reduced or i	missing a ? _ x	(*=0×	Matrix B & _ X	(*=0×
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	UMM2 2	Entity Quality Related Entity	Count Comment	Unit	Measurement	3 Boulengerella cuvieri 1 7 1 0	1 1 0 0
		mesethmoid bone shape None	None	None		4 Brycon guatemalensis 1 0 0 0	0 0 1 0
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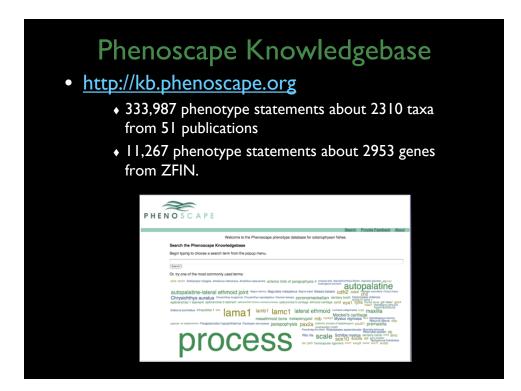


NeXML output from Phenex

- Original character and state definitions
- Taxa
 - Including specimen and collection IDs
- Character matrix
- Entity-quality phenotype assignments to taxa







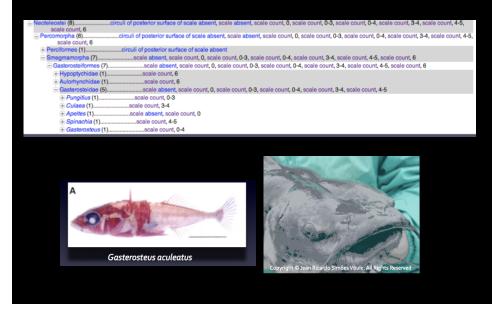
From character states to EQ statements

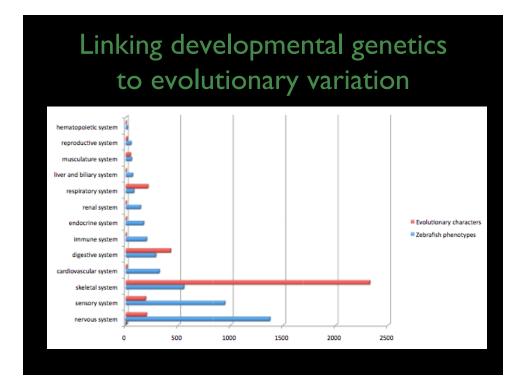
- Complexity of character definitions
 - I EQ
 - 2 EQ: 14%
 - 3 or more EQ: 2%
- Relational: 5%
- Binary: 69%
- Presence-absence: 28%
- Post-compositional: 35%

Search for ZFIN mutants affecting scale development

Phenotypes						10
Anatomy	Quality_				<u>Taxa</u>	46 genes
scale	relational spatial quality : angular placement, relational spatial quality towards process of parietal bone, relational spatial quality towards supraoccipital crest				<u>19</u>	
scale	texture : texture of				<u>91</u>	
scale	structure				<u>95</u>	
scale	position : inverted, spatial pattern				<u>79</u>	
scale	shape : round			17	372	
scale	quality : discontinuous, malformed, oc		A	dorsal		Bcb2
scale	count : absent, count of 0, count of 0-3	wtildtype			0	eb3 eb4
scale	size : decreased size	w	pectoral pelvic	lana	caudal	
	finless (eda)	te370f / te370f		7	,	
						Harris et al., 2007

Loss of scales in fish evolution





Application to plants?

- A large (but maybe not so large) legacy literature
- Abundance of mutant data
 - Some of which is now being annotated in EQ?
- A good foundational Plant Ontology
- Potential for linking G2P and iPToL grand challenge projects
- But this is not something that iPlant can do without leadership from morphologists
 - The work that needs to be done is almost 100% data curation and ontology refinement