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Skeletal Anatomy Jamboree

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NESCent 9 April 2010

Purpose of jamboree:

- Re-evaluate skeletal ontology for fishes (TAO), focussing at level of cell, tissue, and skeletal element
- Coordinate high-level terms with other vertebrate anatomy ontologies
- Goal to improve usefulness and accuracy of reasoning that is based on this ontology

Ontology important for:

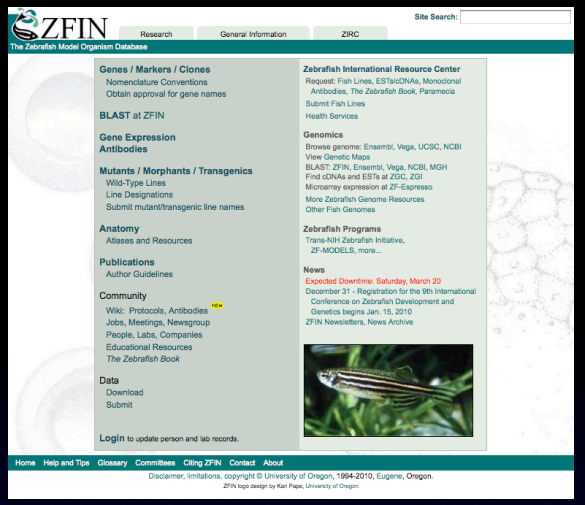
- answering specific questions
- discovery, data mining
- clear communication
- interoperability across databases

*need to keep in mind what it is we want to know and communicate, i.e., use

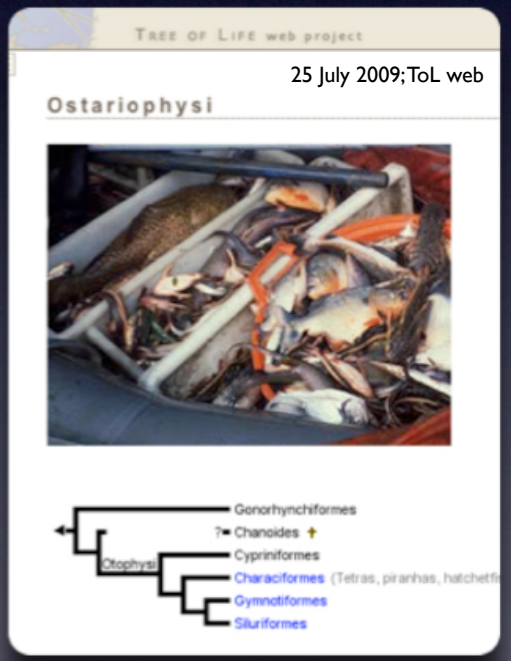
Phenoscape

2007

- Goal: To prototype a curated, ontology-based evolutionary 'phenotype' database that maps to genetic databases
- Foster devo-evo synthesis
- Enable data-mining and discovery for broad scale evolutionary patterns



Zebrafish (zfin.org)



Ostariophysan fishes

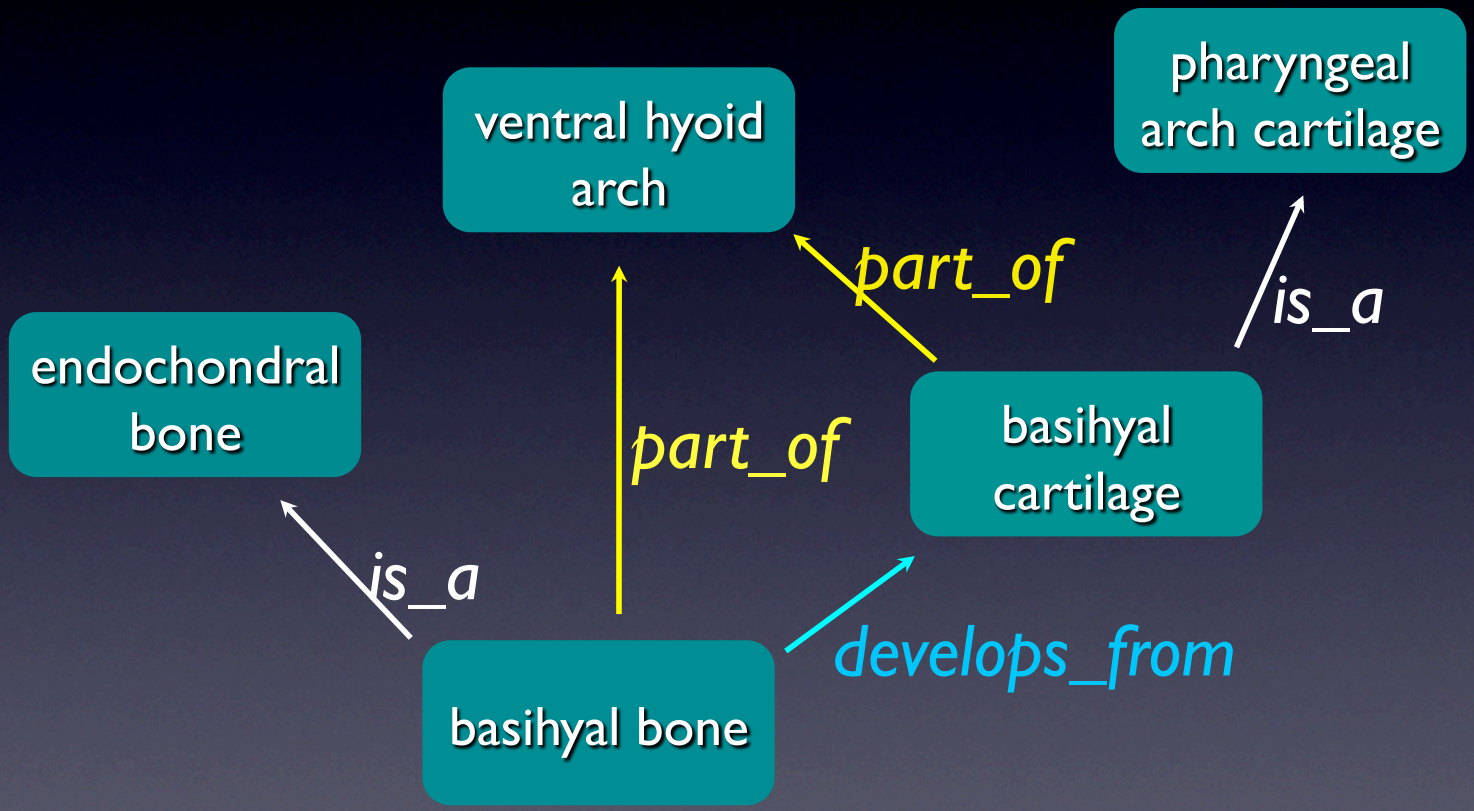
Search for:

- Search for candidate genes underlying evolutionary morphology
- Search for taxa with particular morphologies
- Aggregate morphological data across studies

Requirements:

1. Ontologies
2. Curation
3. Database

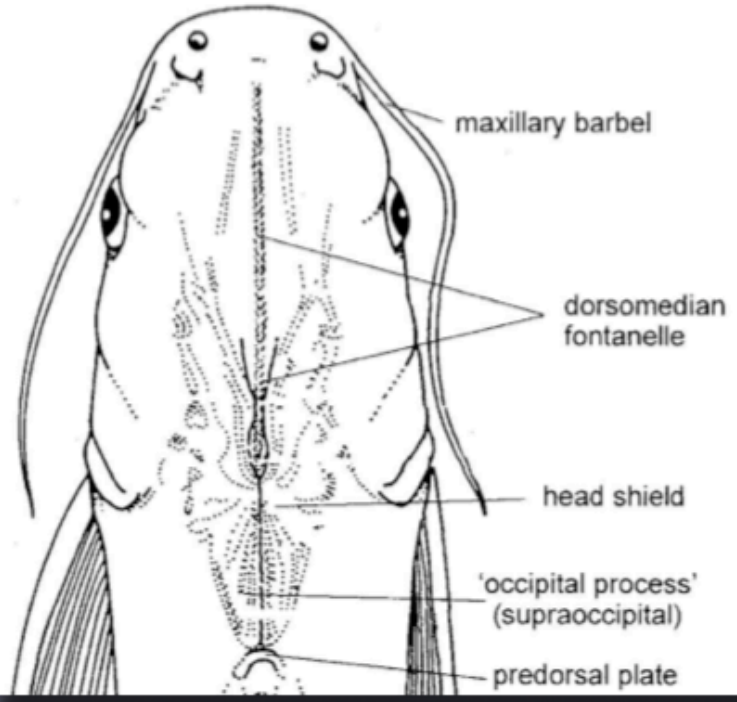
Ontology: Teleost Anatomy Ontology



Dahdul, W. M., J. G. Lundberg, P. E. Midford, J. P. Balhoff, H. Lapp, T. J. Vision, M. A. Haendel, M. Westerfield, and P. M. Mabee. 2010. The Teleost Anatomy Ontology: Anatomical representation

Curation: 'free-text' descriptions into machine-readable form using ontologies

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



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 arii cover' as exposed transv median excavation on th bone in ariids extends ov Some ariids possess apo laminar shelf, such as *barbus, Cinetodus frogga* high and acute in *Bat armiger*) or double keel

?	0	0	?	3	0	?	0	?	0	0	?	1	0	?
1	?	?	?	0	?	?	0	1	0	0	2	0	2	0
0	?	?	?	0	?	?	0	?	0	0	2	1	2	1
0	?	?	?	0	?	?	0	1	?	?	2	1	2	0
1	?	0	?	2	0	?	0	1	0	?	2	1	2	?
1	?	?	?	0	0	?	0	?	0	?	2	1	1	1
3	?	?	?	0	?	?	0	1	0	1	2	1	0	2
3	?	?	?	0	?	?	0	1	0	?	?	1	0	1

Curated 4,627 characters in 3,449 species from 51 papers

Taxon	# Papers	# Species	# Characters
Cypriniforms	8	676	794
Siluriforms	20	1724	2,110
Characiforms	10	754	1,156
Gymnotiforms	1	116	231
Gonorynchiforms	3	41	467
Clupeiforms	5	200	439
Euteleosts	3	145	582
Total	51	3,656	5,779
TOTAL ANNOTATED			

March 2010

Database: Phenoscape KB

kb.phenoscape.org

486,714 evolutionary phenotype assertions

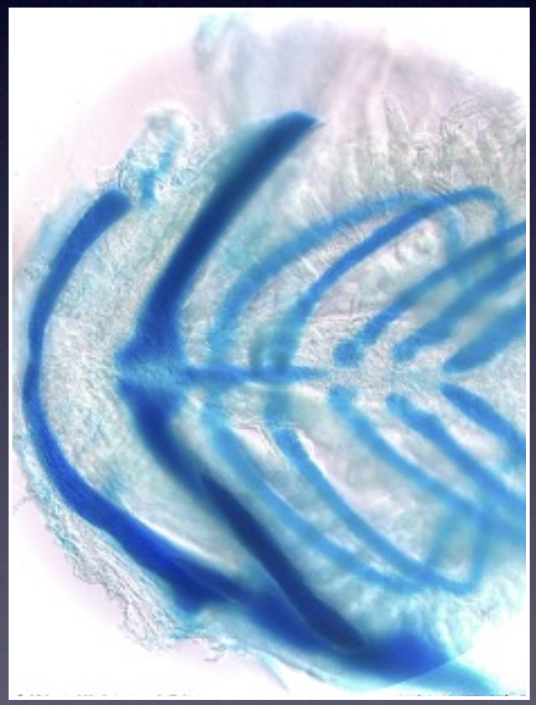
16,426 phenotype statements about 3,539 genes

25 July 2009

Kb enables:

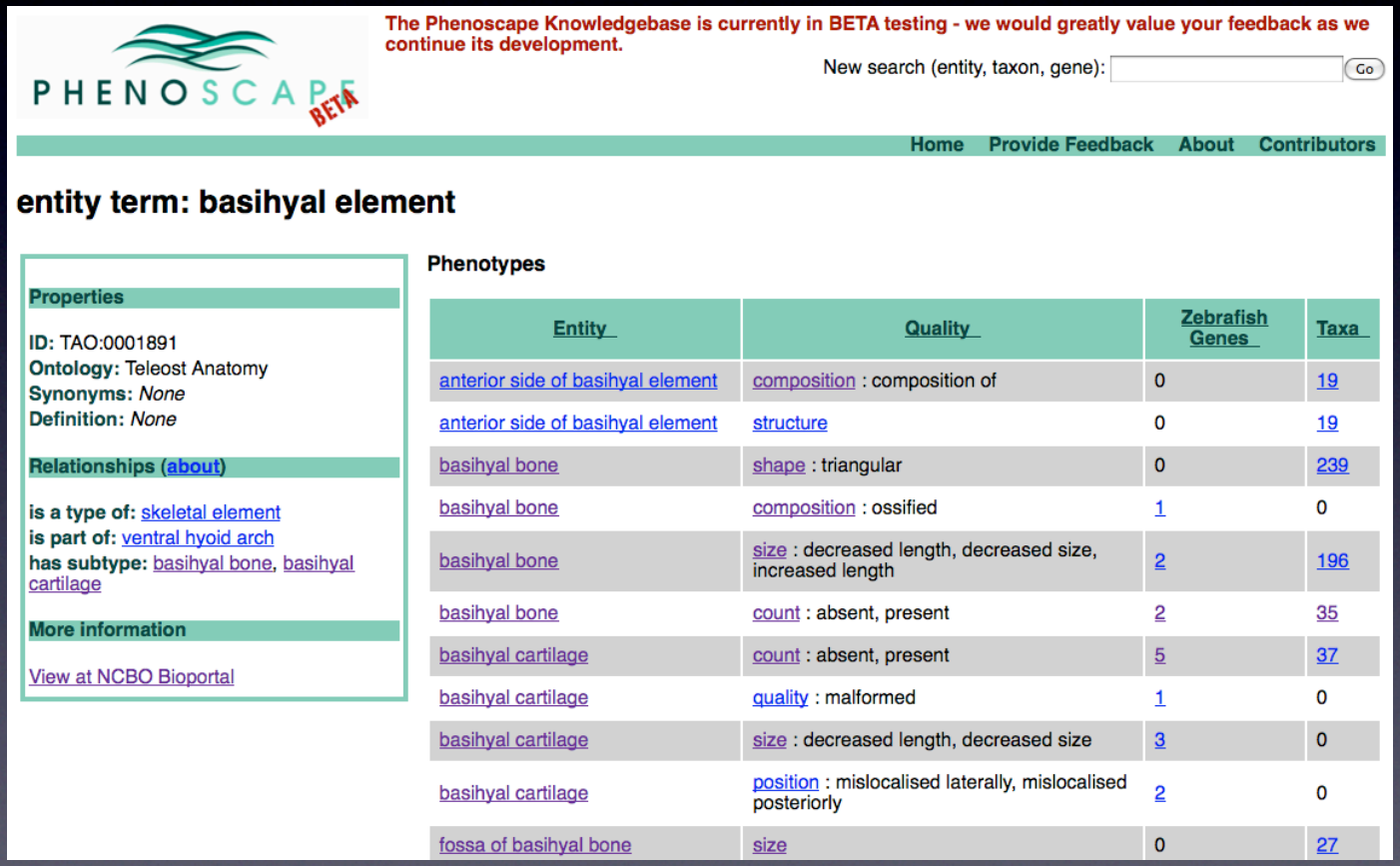
- Selection of candidate genes for evolutionary morphology
- Broad scale view, patterns of morphology across taxa
- E.g.,

1. Evolutionary phenotype: Loss of skeletal 'tongue' (basihyal) in all catfishes



Ictalurus punctatus

What genes are involved in the development of the basihyal?



PHENOSCAPE BETA

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

New search (entity, taxon, gene): Go

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entity term: basihyal element

Properties

ID: TAO:0001891
Ontology: Teleost Anatomy
Synonyms: *None*
Definition: *None*

Relationships (about)

is a type of: skeletal element
is part of: ventral hyoid arch
has subtype: basihyal bone, basihyal cartilage

More information

View at NCBO Bioportal

Phenotypes

Entity	Quality	Zebrafish Genes	Taxa
anterior side of basihyal element	composition : composition of	0	19
anterior side of basihyal element	structure	0	19
basihyal bone	shape : triangular	0	239
basihyal bone	composition : ossified	1	0
basihyal bone	size : decreased length, decreased size, increased length	2	196
basihyal bone	count : absent, present	2	35
basihyal cartilage	count : absent, present	5	37
basihyal cartilage	quality : malformed	1	0
basihyal cartilage	size : decreased length, decreased size	3	0
basihyal cartilage	position : mislocalised laterally, mislocalised posteriorly	2	0
fossa of basihyal bone	size	0	27

Results of query: list of basihyal element
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Mutations in *brpf1* result in basihyal cartilage loss (zebrafish phenotype)

PHENOTYPE:

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

Observed In: basihyal

Stage Range : Day 5

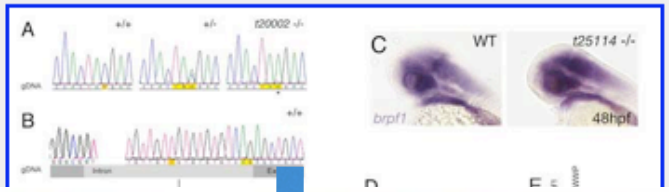
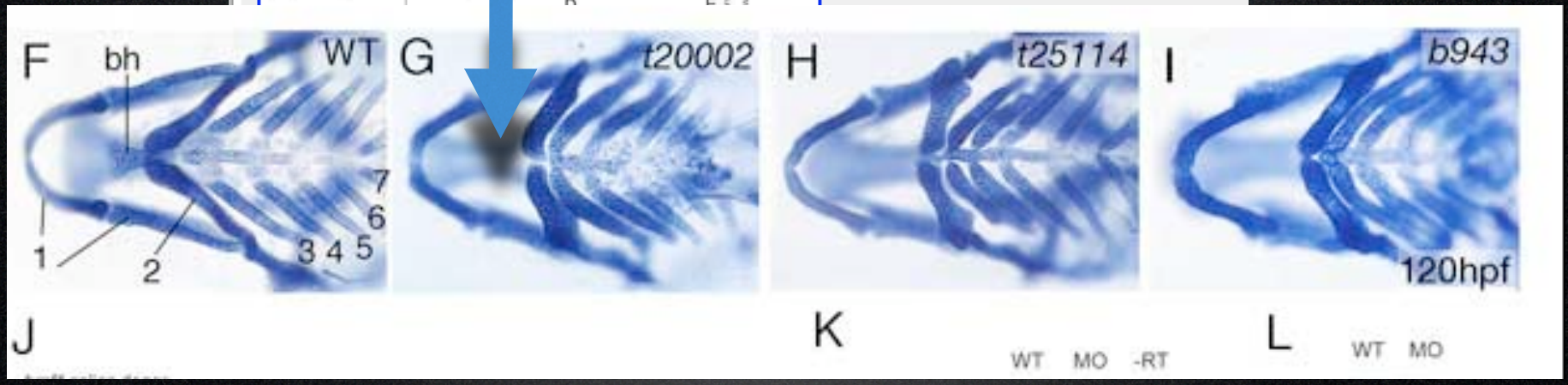


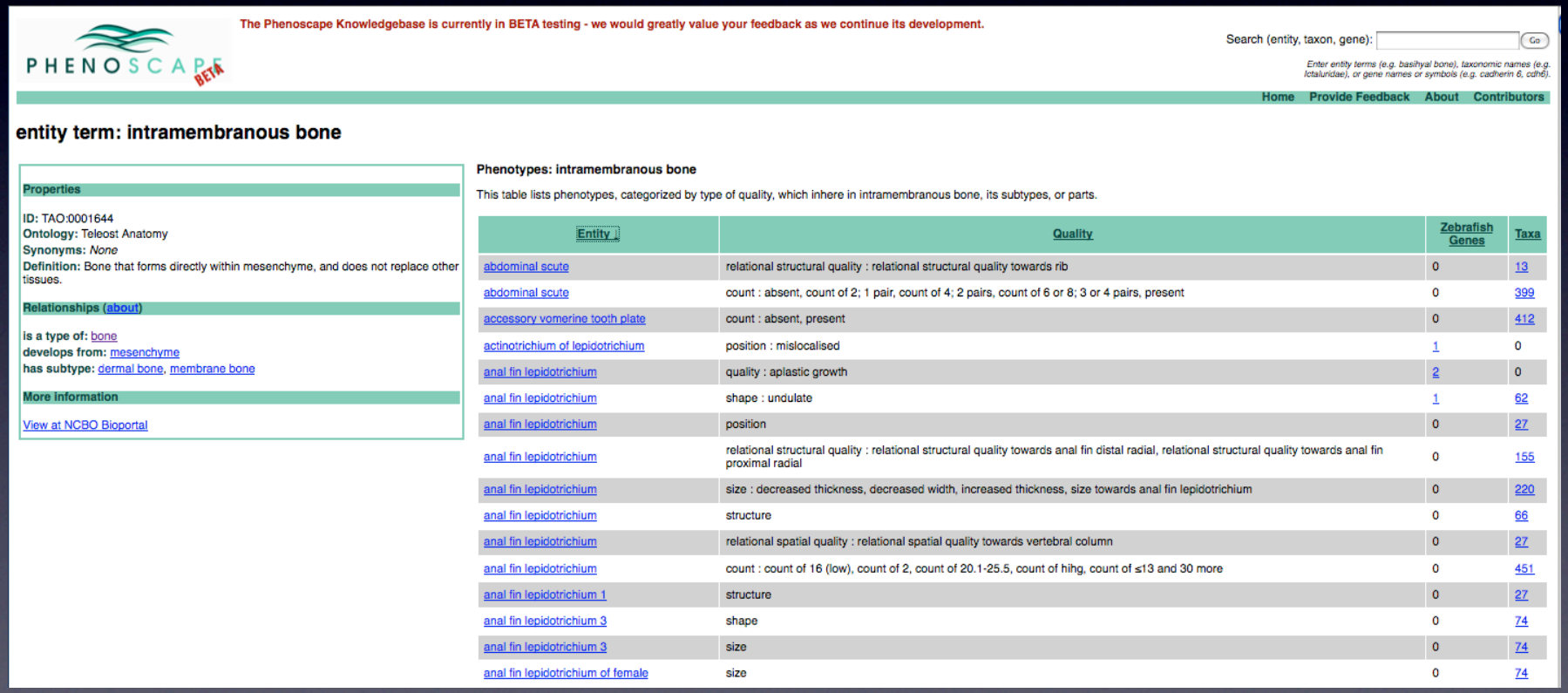
Fig. S1 Analysis of *bpfr1* mutation
(+/+), heterozygous (+/-) and *t20002* m
by an asterisk. (**B**) Sequencing profile
(-/-, lower panel). The *t25114* mutatio



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

2. What genes are involved in the development of intramembranous bones?



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Search (entity, taxon, gene): Go

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

Home | Provide Feedback | About | Contributors

entity term: intramembranous bone

Properties

ID: TAO:0001644
Ontology: Teleost Anatomy
Synonyms: *None*
Definition: Bone that forms directly within mesenchyme, and does not replace other tissues.

Relationships (about)

is a type of: bone
develops from: mesenchyme
has subtype: dermal bone, membrane bone

More information

View at NCBO Bioportal

Phenotypes: intramembranous bone

This table lists phenotypes, categorized by type of quality, which inhere in intramembranous bone, its subtypes, or parts.

Entity	Quality	Zebrafish Genes	Taxa
abdominal scute	relational structural quality : relational structural quality towards rib	0	13
abdominal scute	count : absent, count of 2; 1 pair, count of 4; 2 pairs, count of 6 or 8; 3 or 4 pairs, present	0	399
accessory vomerine tooth plate	count : absent, present	0	412
actinotrichium of lepidotrichium	position : mislocalised	1	0
anal fin lepidotrichium	quality : aplastic growth	2	0
anal fin lepidotrichium	shape : undulate	1	62
anal fin lepidotrichium	position	0	27
anal fin lepidotrichium	relational structural quality : relational structural quality towards anal fin distal radial, relational structural quality towards anal fin proximal radial	0	155
anal fin lepidotrichium	size : decreased thickness, decreased width, increased thickness, size towards anal fin lepidotrichium	0	220
anal fin lepidotrichium	structure	0	66
anal fin lepidotrichium	relational spatial quality : relational spatial quality towards vertebral column	0	27
anal fin lepidotrichium	count : count of 16 (low), count of 2, count of 20.1-25.5, count of hihg, count of ≤13 and 30 more	0	451
anal fin lepidotrichium 1	structure	0	27
anal fin lepidotrichium 3	shape	0	74
anal fin lepidotrichium 3	size	0	74
anal fin lepidotrichium of female	size	0	74

Results of query: list of intramembranous bones
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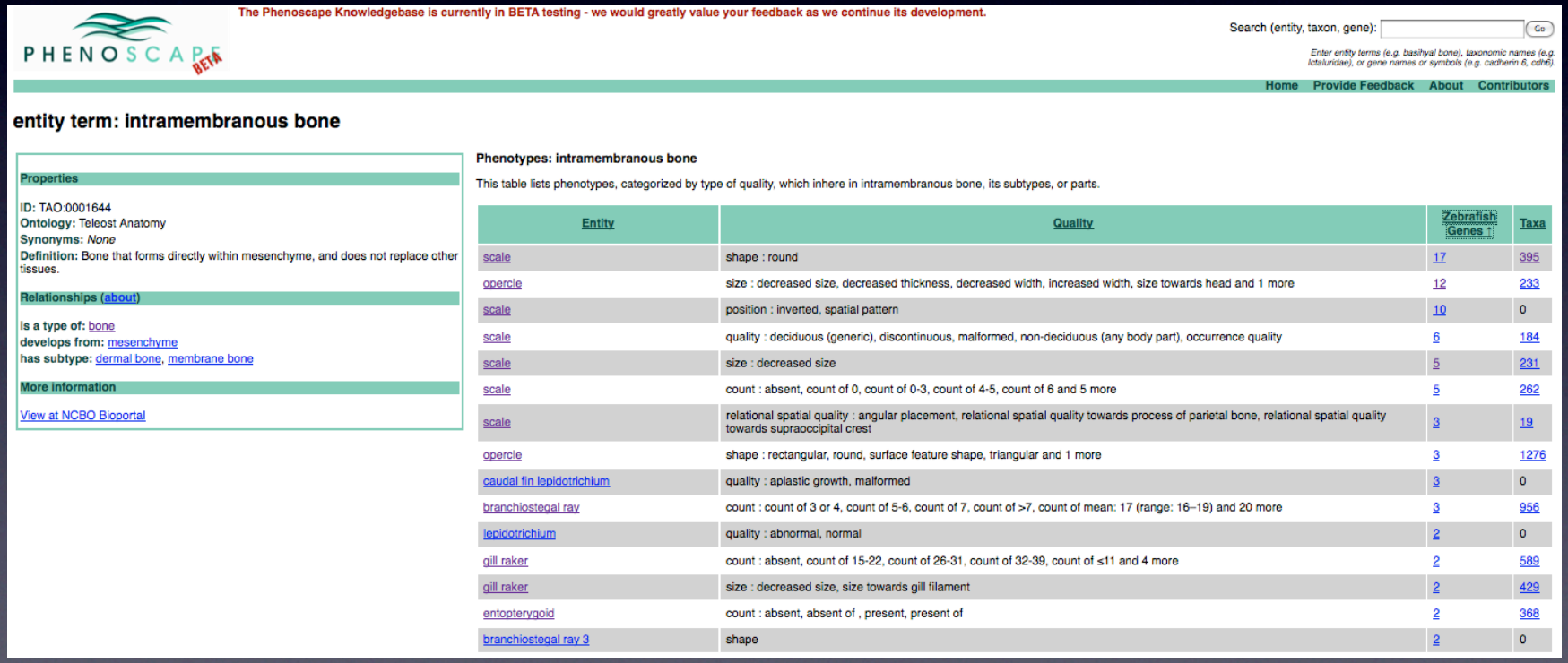
TAO: 'Intramembranous bone'

List sorted by genes



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entity term: intramembranous bone

Properties

ID: TAO:0001644
Ontology: Teleost Anatomy
Synonyms: *None*
Definition: Bone that forms directly within mesenchyme, and does not replace other tissues.

Relationships (about)

is a type of: bone
develops from: mesenchyme
has subtype: dermal bone, membrane bone

More information

View at NCBO Bioportal

Phenotypes: intramembranous bone

This table lists phenotypes, categorized by type of quality, which inhere in intramembranous bone, its subtypes, or parts.

Entity	Quality	Zebrafish Genes ↑	Taxa
scale	shape : round	17	395
opercle	size : decreased size, decreased thickness, decreased width, increased width, size towards head and 1 more	12	233
scale	position : inverted, spatial pattern	10	0
scale	quality : deciduous (generic), discontinuous, malformed, non-deciduous (any body part), occurrence quality	6	184
scale	size : decreased size	5	231
scale	count : absent, count of 0, count of 0-3, count of 4-5, count of 6 and 5 more	5	262
scale	relational spatial quality : angular placement, relational spatial quality towards process of parietal bone, relational spatial quality towards supraoccipital crest	3	19
opercle	shape : rectangular, round, surface feature shape, triangular and 1 more	3	1276
caudal fin lepidotrichium	quality : aplastic growth, malformed	3	0
branchiostegal ray	count : count of 3 or 4, count of 5-6, count of 7, count of >7, count of mean: 17 (range: 16–19) and 20 more	3	956
lepidotrichium	quality : abnormal, normal	2	0
gill raker	count : absent, count of 15-22, count of 26-31, count of 32-39, count of ≤11 and 4 more	2	589
gill raker	size : decreased size, size towards gill filament	2	429
entopterygoid	count : absent, absent of , present, present of	2	368
branchiostegal ray 3	shape	2	0

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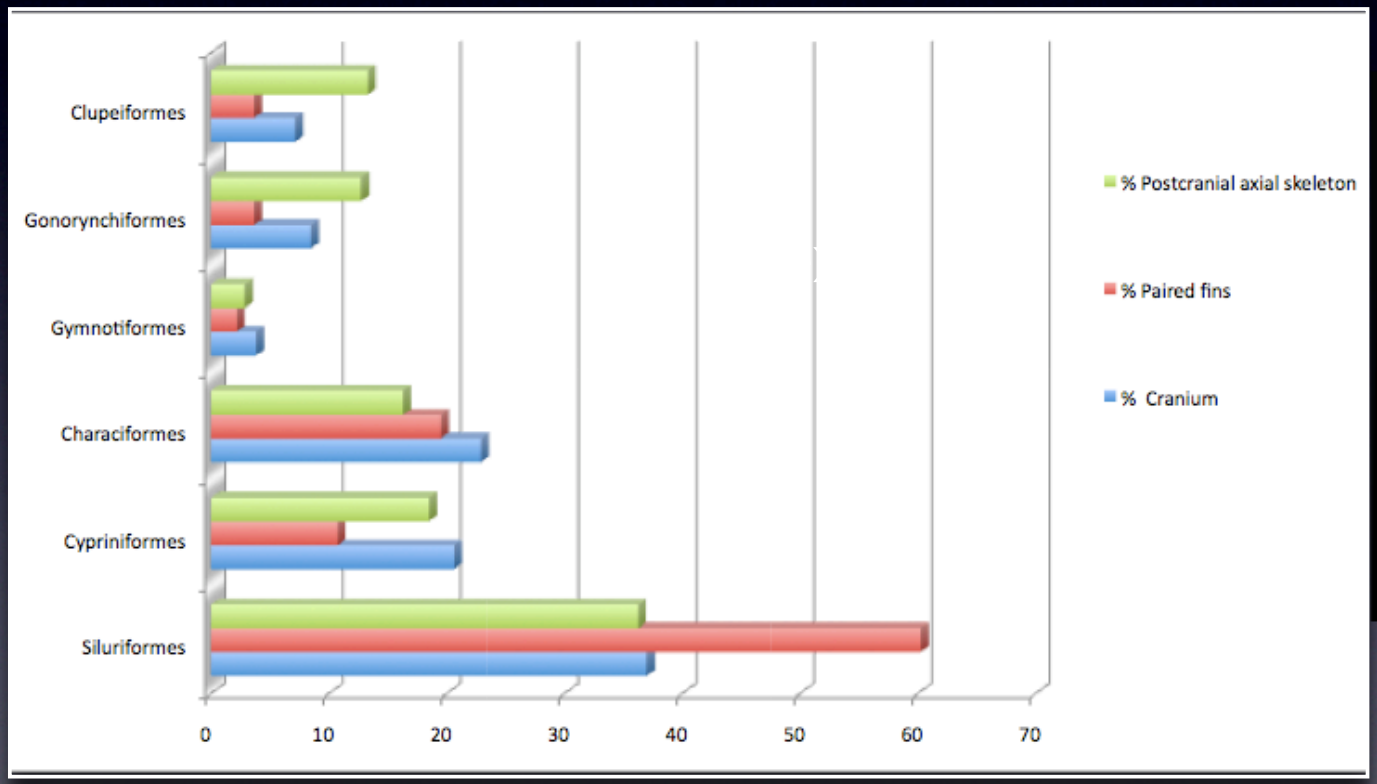
3. How do skeletal regions vary across taxa?



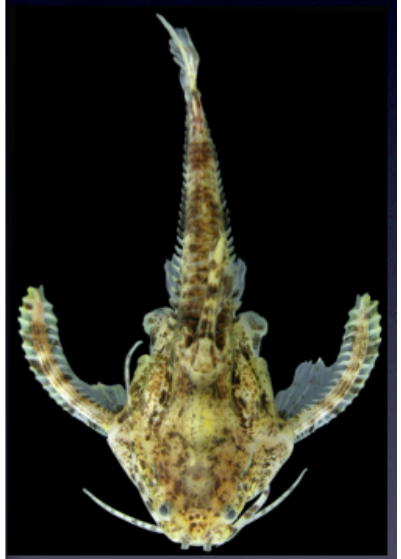


Image from Sabaj-Perez

TAO terms: 'Postcranial axial skeleton', 'Paired fins', 'Cranium'

Other use cases:

- Give me all the bones that come from neural crest vs. mesoderm
- Is there an enrichment in the neural crest vs. mesodermally derived bones in a particular clade?

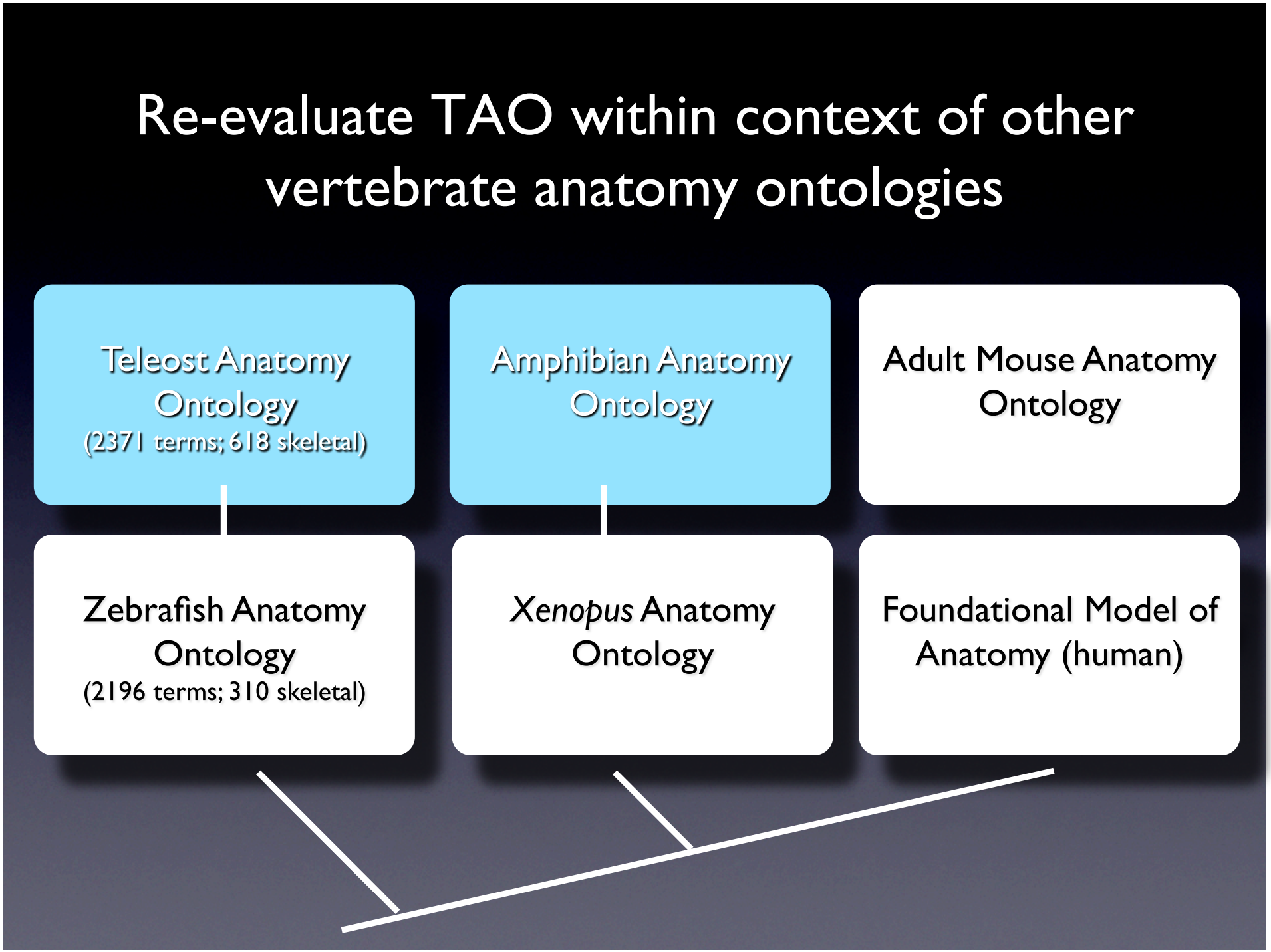
What we can NOT ask for currently:

- Give me a list:
  - of all the intramembranous bones in a mouse or *Xenopus* or any amphibian
  - of all the dermal bones, replacement bones in a mouse or *Xenopus* or any amphibian

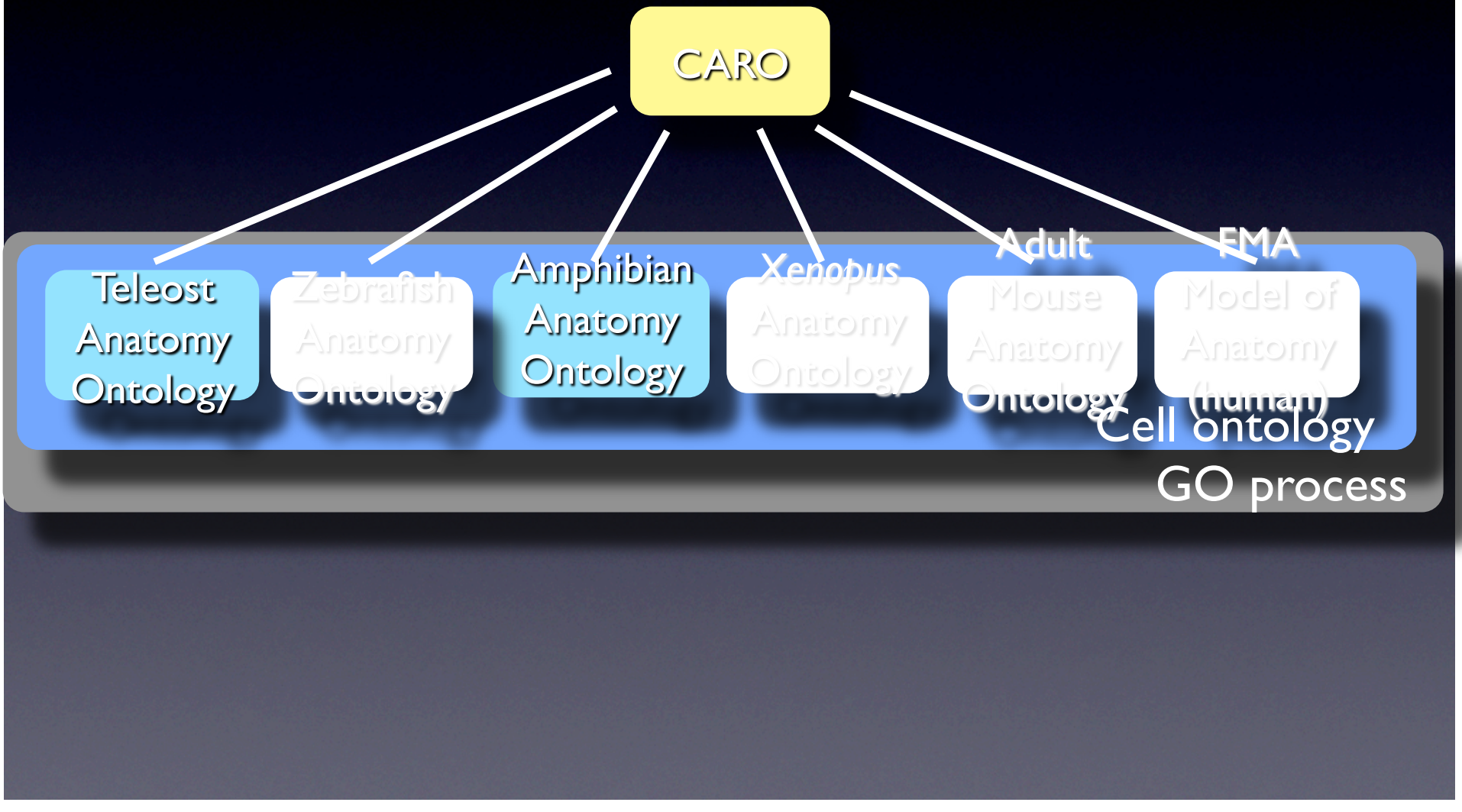
Purpose of jamboree:

- Re-evaluate skeletal ontology for fishes (TAO), focussing at level of cell, tissue, and skeletal element
- Goal to improve usefulness and accuracy of reasoning that is based on this ontology
  - to facilitate answering questions, 'use-driven' ontology

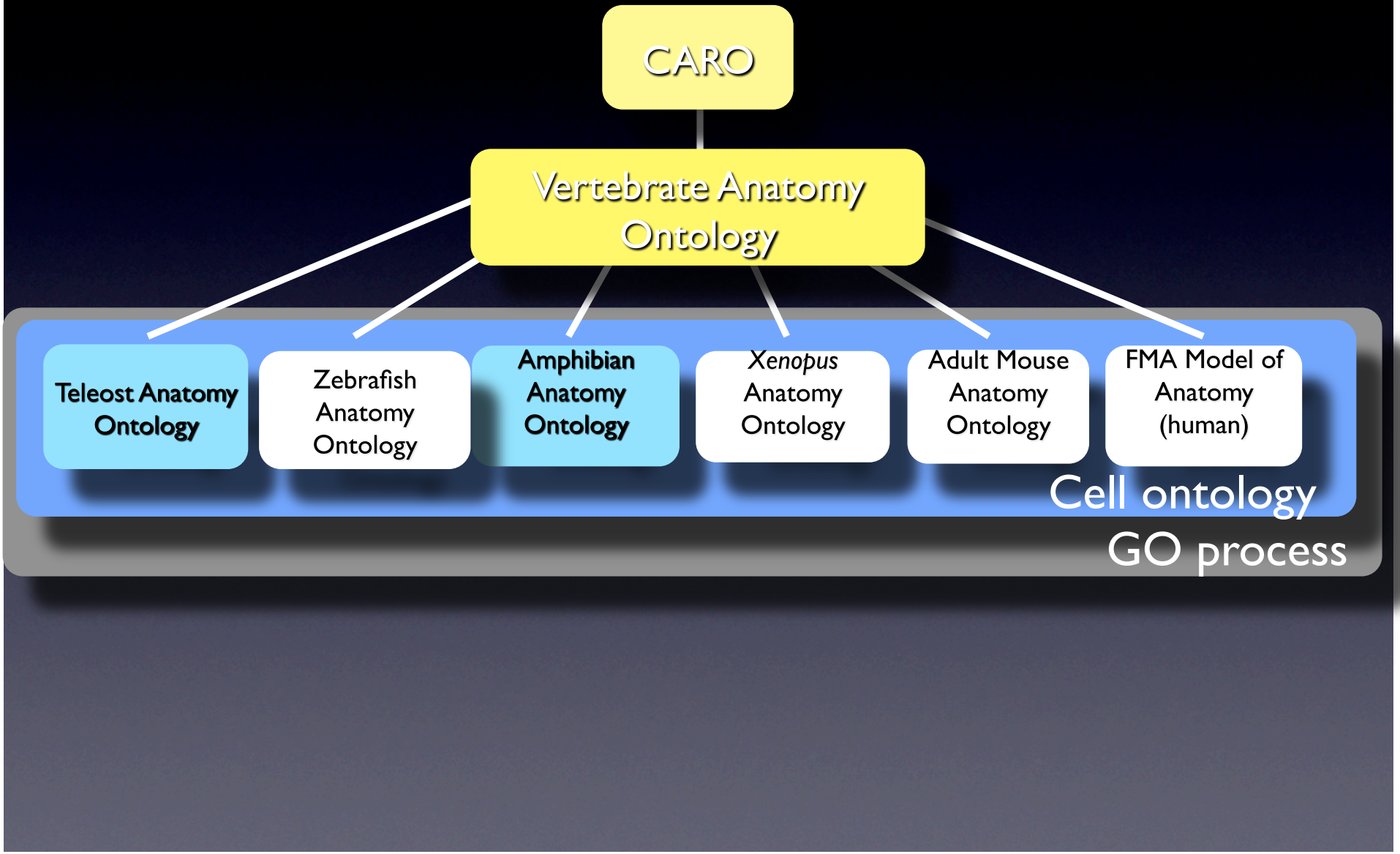
Re-evaluate TAO within context of other vertebrate anatomy ontologies



Re-evaluate TAO in relation to other relevant ontologies



Vertebrate Anatomy Ontology



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