

# A Gentle Introduction to Ontologies for Biology

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National Evolutionary Synthesis Center (NESCent)

Ontologies for Ichthyology and Herpetology Workshop  
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# Biology is data rich



## Specials

### Big Data

- EDITORIAL
- SPECIAL REPORT
- COLUMN: PARTY OF ONE
- FEATURES
- COMMENTARY
- BOOKS & ARTS
- ESSAY
- REVIEW
- PODCAST EXTRA



## 1000 Genomes

A Deep Catalog of Human Genetic Variation





# Data integration has to rely on computation

BOLD Systems

http://www.barcodinglife.org/views/login.php

## BARCODE OF LIFE DATA SYSTEMS

Advancing species identification and discovery through the analysis of short, standardized gene regions



Published Projects | Taxonomy Browser | Request an Account | Identify Specimen | Introductory Tutorial | Documentation | Citation

The Barcode of Life Data Systems (BOLD) is an online workbench that aids collection, management, analysis, and use of DNA barcodes. It consists of 3 components (MAS, IDS, and ECS) that each address the needs of various groups in the barcoding community.

### MANAGEMENT & ANALYSIS

**BOLD-MAS** provides a repository for barcode records coupled with analytical tools. It serves as an online workbench for the DNA barcode community.

Username  Password

[Request a new user account](#)  
[Forgot your username or password?](#)

### IDENTIFICATION ENGINE

**BOLD-IDS** provides a species identification tool that accepts DNA sequences from the barcode region and returns a taxonomic assignment to the species level when possible.



### EXTERNAL



NCBI

## Bramocharax caballeroi voucher ECO-CH-P5485A cytochrome oxidase subunit 1 (COI) gene, partial cds; mitochondrial

[Features](#) [Sequence](#)

LOCUS EU751721 652 bp DNA linear VRT 06-FEB-2009

DEFINITION Bramocharax caballeroi voucher ECO-CH-P5485A cytochrome oxidase subunit 1 (COI) gene, partial cds; mitochondrial.

ACCESSION EU751721

VERSION EU751721.1 GI:189164764

KEYWORDS BARCODE.

SOURCE mitochondrion Bramocharax caballeroi (Catemaco characin)


ORGANISM [Bramocharax caballeroi](#)  
Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Actinopterygii; Neopterygii; Teleostei; Ostariophysi; Characiformes; Characidae; Characidae incertae sedis; Bramocharax. de la Frontera Sur, Av. Centenario Km 5.5, Chetumal, Quintana Roo 77014, Mexico

FEATURES  
source Location/Qualifiers  
1..652  
/organism="Bramocharax caballeroi"  
/organelle="mitochondrion"  
/mol\_type="genomic DNA"  
/specimen\_voucher="ECO-CH-P5485A"  
/db\_xref="BOLD:MEFM051-05"  
/db\_xref="taxon:533334"  
/country="Mexico: Veracruz"  
/lat\_lon="18.418 N 95.111 W"  
/collection\_date="25-Jul-2005"  
/collected\_by="Martha Valdez"  
/identified\_by="Martha Valdez"

GLOBAL BIODIVERSITY INFORMATION FACILITY

SPECIES COUNTRIES DATABASE

... free and open access to biodiversity data





[illegible]



# Ontologies facilitate data integration

EMAGE

http://www.emouseatlas.org/emagewebapp/pages/emage\_general\_query\_result.jsf






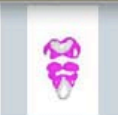


Quick Search Gene/Protein for \* Go

Home Search Analysis Data Submission EMAP Anatomy Atlas About Help

**Query: genes - detected - branchial arch (exact match to structures) - ts1 to ts26**

Select All On This Page | Deselect All On This Page Add/Remove Columns Display 10 entries per page

Page 3 of 70 Go to page: Go Flexible Scroll

Select	Gene/Protein (469)	Data Image (697)	Region (668)	Structures (568)	Theiler Stage (8)	Stage Given (41)	ID (697)	Mutant Allele (35)
<input type="checkbox"/>	Akr1a4			tail branchial arch otocyst fronto-nasal process	17	10.5 dpc	EMAGE:4189	wild-type
<input type="checkbox"/>	Alx1			medial-nasal process mesenchyme latero-nasal process mesenchyme 1st arch maxillary part 1st branchial arch	17	10.5 dpc	EMAGE:3606	wild-type
<input type="checkbox"/>	Alx1			1st arch mandibular part	17	10.5 dpc	EMAGE:3608	wild-type
<input type="checkbox"/>	Alx3			medial-nasal process embryo 1st arch mandibular part	17	10.5 dpc	EMAGE:122	wild-type

Link to original image EMAGE:3606

Open "http://www.emouseatlas.org/gxdb/dbImage/3606/3606.html" in a new window behind the current window



[illegible]

As of March 2009



# But phenotypes are complex, free text

OMIM - SRY-BOX 9; SOX9

http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=608160

NCBI

MIM \*608160

Cloning  
Mapping  
Gene Function  
Molecular Genetics  
Cytogenetics  
Evolution  
Animal Model  
Allelic Variants  
View List  
References  
Contributors  
Creation Date  
Edit History

## GENE FUNCTION

[Morais da Silva et al. \(1996\)](#) found that, consistent with its role in sex determination, SOX9 expression closely follows differentiation of Sertoli cells in the mouse testis, in experimental sex reversal when fetal ovaries are grafted to adult kidneys, and in the chick where there is no evidence for an Sry gene. The results suggested to the authors that SOX9 plays an essential role in sex determination, possibly immediately downstream of SRY in mammals, and that it functions as a critical Sertoli cell differentiation factor, perhaps in all vertebrates. 🧐

By cell transfection experiments, [Sudbeck et al. \(1996\)](#) showed that SOX through the motif AACAAAG, a sequence recognized by other HMG domain proteins, acts as a transcriptional activator. SOX9 to the DNA-binding domain of yeast GAL4, the transactivating factor, at the C terminus of SOX9. With 1 exception, all SOX9 nonsense and frameshift mutations lead to truncation of this domain, suggesting that skeletal development in these cases results, at least in part, from loss of the

86

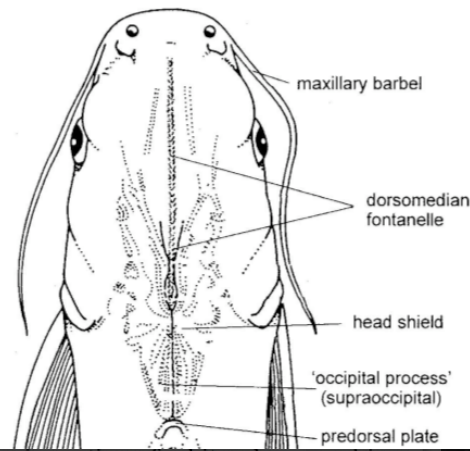
## AMERICAN MUSEUM NOVITATES

### APPENDIX 1. CHARACTER SUMMARY

1. Fifth infraorbital. 0, well developed, without contact between fourth and sixth infraorbitals; 1, greatly reduced, with posteroventral margin of sixth infraorbital in contact with posterodorsal margin of fourth infraorbital.
2. 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-

14. Portion on vomer. 0, not modified in depression on its anterior tip of maxilla.
15. Ridge on lateral. 1, present.
16. Rhinosphenoid. 0, present.
17. Lateral ethmoid. 0, present; 1, present.
18. Parasphenoid and main portion of orbitosphenoid. 0, well separated; 1, close to each other.
19. Dilator 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.

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 ariids and interpret a 'minimal cover' as exposed transverse process bases and a deep median excavation on the ventral surface. The laminar bone in ariids extends over four to eight vertebra centra. Some ariids possess apomorphic modifications in the laminar shelf, such as depressions (e.g., *Guiritinga barbus*, *Cinetodus froggatti*) or median single keel (e.g., high and acute in *Batrachoccephalus*, *Nemapteryx armiger*) or double keel (e.g., *Bagre marinus*).



# Integrating phenotypes can open up new science

Encyclopedia of Life

## *Ambystoma talpoideum* (Holbrook, 1838)

Mole salamander

Authoritative INFORMATION All

### TABLE OF CONTENTS

- Description
  - Morphology
  - Reproduction and Life History
  - Behavior
- Ecology and Distribution
  - Distribution
  - Habitat
  - Associations
  - Trophic Strategy
- Conservation
  - Conservation Status
- Relevance
  - Uses
  - Biodiversity Heritage Library
- References and More Information
  - Specialist Projects
  - Search the Web

### MORPHOLOGY

SWITCH TO COMMON NAMES

CLASSIFICATION : TEXT | GRAPHIC |

- Animalia +
- Chordata +
- Amphibia +
- Caudata +
- Ambystomatidae +
- Ambystoma +
- Ambystoma talpoideum* (Holbrook, 1838)

- Archaea +
- Bacteria +
- Chromista +
- Fungi +
- Plantae +
- Protozoa +
- Viruses +

#### Physical Description

SOURCE AND ADDITIONAL INFORMATION

AUTHOR Ashlee Behr

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SUPPLIER Animal Diversity Web

SOURCE URL [View original data object](#)

Mole salamanders are facultatively paedomorphic; they can either become terrestrial adults (metamorphic adults) or retain their aquatic larval form even as they become sexually mature (paedomorphic, branchiate, or neotenic adults). Paedomorphic adults can eventually undergo metamorphosis or they may remain in the aquatic form throughout life. Terrestrial adults live in areas surrounding breeding ponds while paedomorphic adults remain in permanent ponds. Environmental conditions present during larval development can determine which life form an individual salamander will become.

Both mole salamander morphs have short, stout bodies with broad, disproportionally large heads. Body size of terrestrial and aquatic males varies across populations and time. In some years and ponds, aquatic males will be larger, on average, than terrestrial males. In other years, the opposite can be



# Words often are polysemous

## Mole:

- Burrowing insectivorous mammals in the family Talpidae
- A spy buried secretly within an organization or country
- The SI unit used in chemistry for the amount of a substance
- A small, sometimes raised area of skin, usually with darker pigment
- A Mexican sauce made from chili peppers and other spices, including chocolate
- A massive structure, usually of stone, used as a pier, jetty, or breakwater between places separated by water



# Words often are polysemous

- “Replacement bone that is median and is the anterior-most bone of the ventral hyoid arch”

- basihyal bone
- basihyoid
- glossohyal

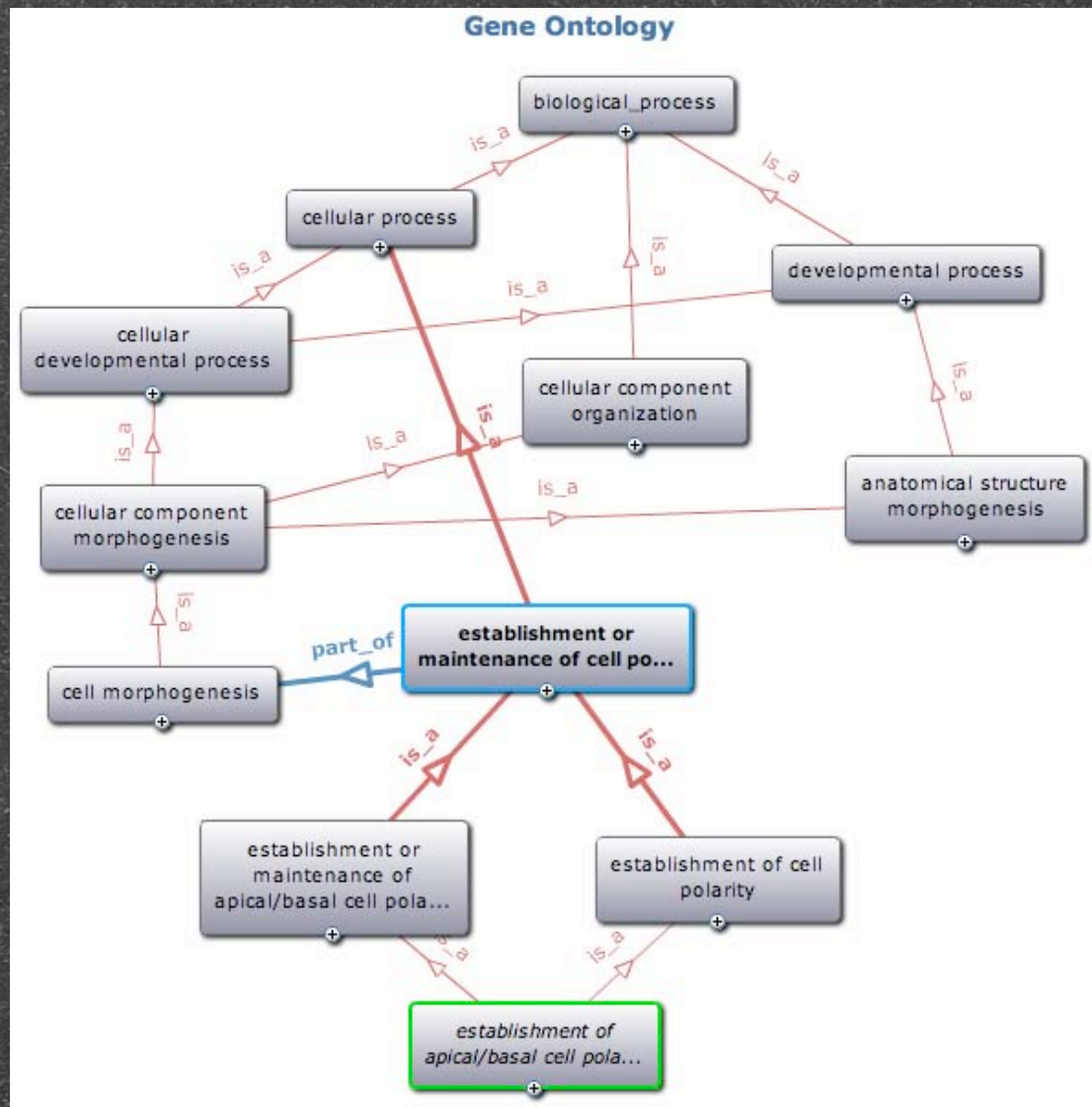


# What is an ontology?

- An ontology is a type of vocabulary with well-defined terms and the logical relationships that hold between them.
- An ontology represents the knowledge about its subject domain.



# Ontologies support reasoning



- The relationships (“assertions”) induce a hierarchical structure.
- Ontologies can be processed by machines to make inferences.



# What are ontologies not?

- An ontology is not a database.
  - Databases typically are silos, but ontologies can connect siloed data
- An ontology is not a terminology.
  - Terminologies are built ad-hoc to serve specific application needs.
- An ontology is not data.
  - Ontologies are used to annotate data and make data interoperable.



# Ontologies can address the problems with text

## List of characters

The list of characters, the analysis of certain morphological characters, and the phylogenetic relationships of certain teleosts are based on the features listed below. [0] represents the plesiomorphic character state and [1], [2], [3], and [4] the apomorphic character states. The outgroup used to polarize characters includes †*Watsonulus eugnathoides*, *Amia calva*, *Lepisosteus* spp., and others in different analyses.

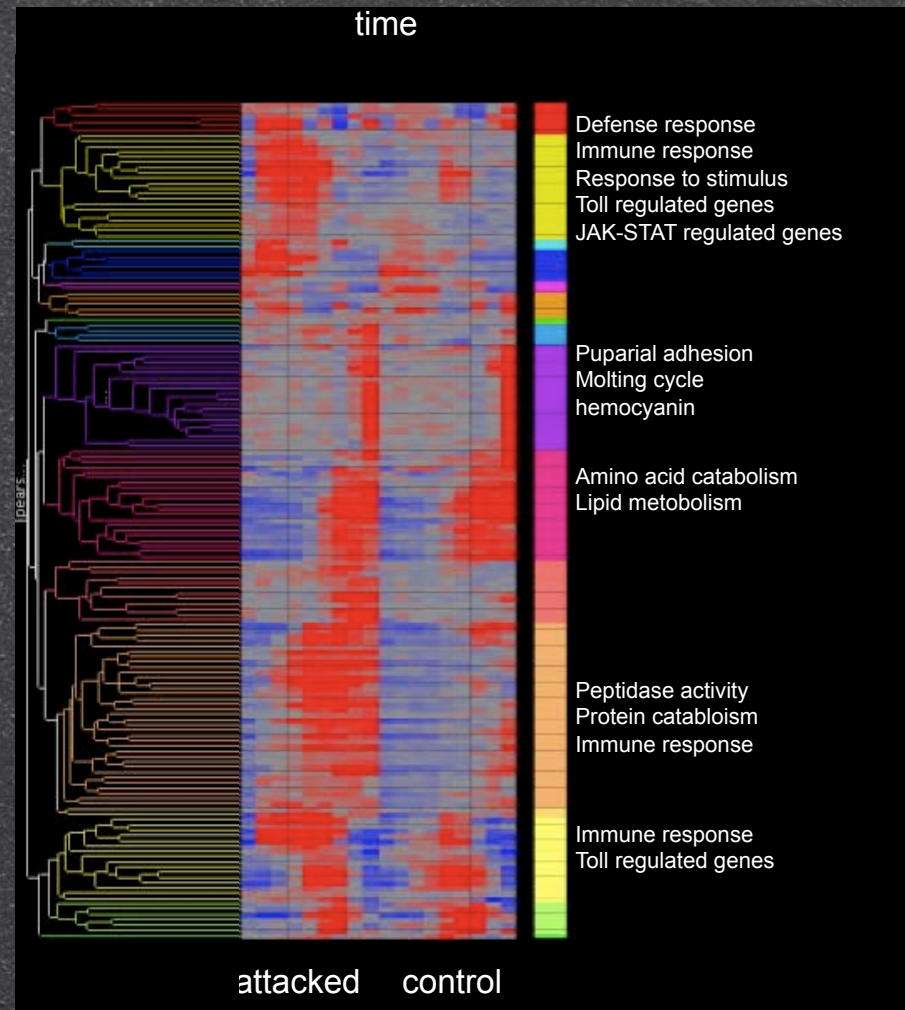
With the exceptions indicated, characters 1 to 167 are from ARRATIA (1991, 1996b, 1997) or are new characters. Because of the use of different outgroups, characters 26, 27, 28, 36, 76, 77, 78, 92, 122, 124, 125, 126, 128, 129, 130, 137, 140, and 157 changed their polarization with respect to ARRATIA (1996b, 1997), and in other cases, the presentation of some characters was slightly modified (indicated below). Characters 168 to 175 are from GRANDE & BEMIS (1998); characters 176 to 191 are taken from PINNA (1996); and characters 192 to 196 are from BRITO (1997).

1. Ethmopalatine ossification in the floor of nasal capsule articulating with autopalatine: [0] absent; [1] present. (PATTERSON & ROSEN 1977.)
2. Two paired endoskeletal ethmoidal ossifications: [0] absent; [1] present.
3. Parietal bones fused in a median element: [0] absent; [1] present.

Arratia, 1999. Zoological Journal of the Linnean Society 151:691-757.



# Ontologies can address the problems with text





Annotating data with  
ontologies:  
Gene annotation as an  
example



# Annotation objectives

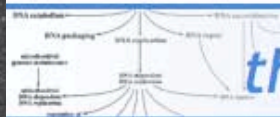
- Where and when is a particular gene product involved (cell part, cell type, body part, development stage)?
- Which functions does the gene product exert?
- With which biological processes is the gene product associated?
- In abnormal or wild-type phenotype?



# Gene Ontology (GO)

- Covers cellular component, molecular function, biological process
- Multi-species, multi-disciplinary, open-source
- Annotated gene products allow information-theoretic computation
  - E.g., quantitative assessment of commonalities among and across sets of genes





the Gene Ontology

AmiGO

[Search](#) [Browse](#) [GOOSE](#) [Other Tools](#) [Help](#)Search GO 

terms



genes or proteins



exact match

## establishment of apical/basal cell polarity

[Term information](#) [Term lineage](#) [External references](#) [17 gene product associations](#)

### Term Information

<b>Accession</b>	GO:0035089
<b>Ontology</b>	<b>biological process</b>
<b>Synonyms</b>	None
<b>Definition</b>	The specification and formation of the polarity of a cell along its apical/basal axis. [source: GOC:bf]
<b>Comment</b>	None
<b>Subset</b>	None
<b>Community</b>	There have been <a href="#">0 comments</a> for this term. If you would like to view or participate in the community annotation, please continue to the <a href="#">GONUTS page</a> .

[Back to top](#)

### Term Lineage

[Switch to viewing term parents, siblings and children](#)[Filter tree view](#) [?](#)



## Term Lineage

Switch to viewing term parents, siblings and children

### ▼ Filter tree view ?

Filter Gene Product Counts

Data source

All  
AspGD  
CGD  
dictyBase

Species

All  
Agrobacterium tum...  
Anaplasma phagocy...  
Arabidopsis thaliana

View Options

Tree view ☒ Full ☐ Compact

Set filters

Remove all filters

all : all [245476 gene products]

GO:0008150 : biological\_process [172247 gene products]

GO:0016043 : cellular component organization [12771 gene products]

GO:0032989 : cellular component morphogenesis [2548 gene products]

GO:0000902 : cell morphogenesis [2181 gene products]

GO:0007163 : establishment or maintenance of cell polarity [448 gene products]

GO:0030010 : establishment of cell polarity [183 gene products]

**GO:0035089 : establishment of apical/basal cell polarity [17 gene products]**

GO:0045198 : establishment of epithelial cell apical/basal polarity [13 gene products]

GO:0035088 : establishment or maintenance of apical/basal cell polarity [65 gene products]

**GO:0035089 : establishment of apical/basal cell polarity [17 gene products]**

GO:0045198 : establishment of epithelial cell apical/basal polarity [13 gene products]

GO:0045197 : establishment or maintenance of epithelial cell apical/basal polarity [47 gene products]

GO:0045198 : establishment of epithelial cell apical/basal polarity [13 gene products]

GO:0045199 : maintenance of epithelial cell apical/basal polarity [6 gene products]

**GO:0035090 : maintenance of apical/basal cell polarity [10 gene products]**

GO:0045199 : maintenance of epithelial cell apical/basal polarity [6 gene products]

GO:0030011 : maintenance of cell polarity [28 gene products]

**GO:0035090 : maintenance of apical/basal cell polarity [10 gene products]**

GO:0045199 : maintenance of epithelial cell apical/basal polarity [6 gene products]

Actions...

Last action: Opened

GO:0035090

Graphical View

Reset tree

View in tree browser

Download...

OBO

RDF/XML

GraphViz dot

View gene products associated with this term



## Gene Product Associations to establishment of apical/basal cell polarity ; GO:0035089 and children

Download all association information in: [gene association format](#) [RDF/XML](#)

Filter associations displayed ?

### establishment of apical/basal cell polarity ; GO:0035089 [\[show def\]](#) [\[view in tree\]](#)

Symbol, full name	Information	Qualifier	Evidence	Reference	Assigned by
<input type="checkbox"/> <a href="#">baz</a> 38 associations <a href="#">bazooka</a> <a href="#">BLAST</a>	gene from <i>Drosophila melanogaster</i>		<a href="#">IMP</a>	<a href="#">FB:FBrf0167999</a>	FlyBase
<input type="checkbox"/> <a href="#">Foxj1</a> 18 associations <a href="#">forkhead</a> <a href="#">BLAST</a> <a href="#">box J1</a>	gene from <i>Mus musculus</i>		<a href="#">IMP</a> With <a href="#">MGI:MGI:2181746</a>	<a href="#">MGI:MGI:3037476</a>	MGI
<input type="checkbox"/> <a href="#">Prkci</a> 9 associations <a href="#">protein</a> <a href="#">BLAST</a> <a href="#">kinase C, iota</a>	gene from <i>Mus musculus</i>		<a href="#">IMP</a> With <a href="#">MGI:MGI:3526850</a> With <a href="#">MGI:MGI:3000907</a>	<a href="#">MGI:MGI:3607414</a>	MGI
<input type="checkbox"/> <a href="#">Prkci</a> 16 associations <a href="#">protein</a> <a href="#">BLAST</a> <a href="#">kinase C, iota</a>	gene from <i>Rattus norvegicus</i>		<a href="#">ISO</a> With <a href="#">RGD:1331958</a>	<a href="#">RGD:1624291</a>	RGD





### establishment of epithelial cell apical/basal polarity ; GO:0045198 [\[show def\]](#) [\[view in tree\]](#)

Symbol, full name	Information	Qualifier	Evidence	Reference	Assigned by
<input type="checkbox"/> <a href="#">crb</a> 33 associations <a href="#">crumbs</a>	gene from <i>Drosophila melanogaster</i>		<a href="#">TAS</a>	<a href="#">FB:FBrf0054006</a>	FlyBase
<input type="checkbox"/> <a href="#">Crb3</a> 4 associations <a href="#">crumbs</a> <a href="#">BLAST</a> <a href="#">homolog 3 (Drosophila)</a>	gene from <i>Mus musculus</i>		<a href="#">ISA</a> With <a href="#">EMBL:AY103469</a>	<a href="#">MGI:MGI:2668769</a>	MGI
<input type="checkbox"/> <a href="#">ds</a> 24 associations <a href="#">BLAST</a>	gene from <i>Drosophila melanogaster</i>		<a href="#">IMP</a>	<a href="#">FB:FBrf0155499</a>	FlyBase



**Prkci<sup>tm1Kido</sup> Targeted Allele Detail**

Your Input Welcome

Allele	Symbol: <b>Prkci<sup>tm1Kido</sup></b> Name: targeted mutation 1, Yoshiaki Kido ID: MGI:3526850																										
Synonyms	PRClambda <sup>flox</sup>																										
Allele details	Allele Type: Targeted (Floxed/Frt) ES Cell Line: Not Specified Mutation: Insertion LoxP sites flanked exon 5 by homologous recombination. ( <a href="#">J:95378</a> ) International Mouse Strain Resource: ( <a href="#">Search for IMSR strains</a> with Prkci mutations) References and Additional Notes: ( <a href="#">See Below</a> )																										
Gene information	Symbol: <a href="#">Prkci</a>   Human Ortholog: <a href="#">PRKCI</a> Name: protein kinase C, iota Chromosome: 3:30894669-30951663 bp, + strand   Genetic Position: 13.8 cM																										
Phenotype summary 	Phenotype Summary by Mammalian Phenotype terms      Key: <table><tr><td>hm</td><td>homozygous</td><td>ht</td><td>heterozygous</td></tr><tr><td>cn</td><td>conditional genotype</td><td>cx</td><td>complex: &gt; 1 genome feature</td></tr><tr><td>tg</td><td>involves transgenes</td><td>ot</td><td>other: hemizygous, indeterminate,...</td></tr><tr><td>N</td><td>normal phenotype</td><td></td><td>expected model not found</td></tr></table> ( <a href="#">show</a> or <a href="#">hide</a> all annotated terms) Genotypes are listed in the next section.			hm	homozygous	ht	heterozygous	cn	conditional genotype	cx	complex: > 1 genome feature	tg	involves transgenes	ot	other: hemizygous, indeterminate,...	N	normal phenotype		expected model not found								
	hm	homozygous	ht	heterozygous																							
cn	conditional genotype	cx	complex: > 1 genome feature																								
tg	involves transgenes	ot	other: hemizygous, indeterminate,...																								
N	normal phenotype		expected model not found																								
<table><tr><td>Affected Systems</td><td>Genotypes:</td><td><a href="#">cn1</a></td><td><a href="#">cn2</a></td></tr><tr><td>digestive/alimentary system</td><td>▶</td><td>✓</td><td></td></tr><tr><td>endocrine/exocrine glands</td><td>▶</td><td>✓</td><td></td></tr><tr><td>homeostasis/metabolism</td><td>▶</td><td>✓</td><td></td></tr><tr><td>nervous system</td><td>▶</td><td></td><td>✓</td></tr><tr><td>vision/eye</td><td>▶</td><td></td><td>✓</td></tr></table>				Affected Systems	Genotypes:	<a href="#">cn1</a>	<a href="#">cn2</a>	digestive/alimentary system	▶	✓		endocrine/exocrine glands	▶	✓		homeostasis/metabolism	▶	✓		nervous system	▶		✓	vision/eye	▶		✓
Affected Systems	Genotypes:	<a href="#">cn1</a>	<a href="#">cn2</a>																								
digestive/alimentary system	▶	✓																									
endocrine/exocrine glands	▶	✓																									
homeostasis/metabolism	▶	✓																									
nervous system	▶		✓																								
vision/eye	▶		✓																								
Phenotypic data by genotype	Phenotypic Data by Genotype ( <a href="#">show</a> or <a href="#">hide</a> all phenotypic details)																										
	<table><tr><th>Genotype</th><th>Allelic Composition</th><th>Genetic Background</th></tr><tr><td>▶ <a href="#">cn1</a></td><td><a href="#">Prkci<sup>tm1Kido</sup>/Prkci<sup>tm1Kido</sup></a> <a href="#">Tg(Ins2-cre)25Mqn/0</a></td><td>involves: 129/Sv * C57BL/6 * DBA/2</td></tr><tr><td>▶ <a href="#">cn2</a></td><td><a href="#">Prkci<sup>tm1Kido</sup>/Prkci<sup>tm1Kido</sup></a> <a href="#">Tg(Crx-cre)1Tfur/0</a></td><td>Not Specified</td></tr></table>			Genotype	Allelic Composition	Genetic Background	▶ <a href="#">cn1</a>	<a href="#">Prkci<sup>tm1Kido</sup>/Prkci<sup>tm1Kido</sup></a> <a href="#">Tg(Ins2-cre)25Mqn/0</a>	involves: 129/Sv * C57BL/6 * DBA/2	▶ <a href="#">cn2</a>	<a href="#">Prkci<sup>tm1Kido</sup>/Prkci<sup>tm1Kido</sup></a> <a href="#">Tg(Crx-cre)1Tfur/0</a>	Not Specified															
Genotype	Allelic Composition	Genetic Background																									
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▶ <a href="#">cn2</a>	<a href="#">Prkci<sup>tm1Kido</sup>/Prkci<sup>tm1Kido</sup></a> <a href="#">Tg(Crx-cre)1Tfur/0</a>	Not Specified																									



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OBO Foundry



NCBO Bioportal: Welcome to the NCBO Bioportal

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Search resources  [Advanced Resource Search](#)

Most Active Ontologies

Ontology	Version	Notes	Mappings
<a href="#">Human disease</a>	1.50	0	17732
<a href="#">Mouse adult gross anatomy</a>	1.194	0	3905
<a href="#">NCI Thesaurus</a>	08.12d	9	3798
<a href="#">Foundational Model of Anatomy</a>	3.0	0	1997
<a href="#">Zebrafish anatomy and development</a>	1.21	0	791

Statistics

Ontologies	145
Concepts	723,806
Resources Indexed	11

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The National Center for Biomedical Ontology

http://bioontology.org/ ncbo

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Current News

- October 17, 2009 - Call for Paper/Exhibition Submissions - [BME09-CISP'09](#)
- August 10, 2009 - Call for Papers - SWESE2009
- August 7, 2009 - Call for papers - [Workshop on Semantic Web Applications in Scientific Discourse](#)
- July 31, 2009 - Call for papers - [The First International Workshop on Role of Semantic Web in Provenance Management \(SWPM 2009\)](#)
- July 24, 2009 - [ICBO](#): program [now available](#).
- July 20, 2009 - Call for papers - [8th Terminology and Artificial Intelligence Conference \(TIA 2009\)](#)

The National Center for Biomedical Ontology is a consortium of leading biologists, clinicians, informaticians.



ONTOLOGY NAME	FORMAT	VERSION	AUTHOR	UPLOADED ON	GROUP	STATUS
<a href="#">Amphibian gross anatomy (AAO)</a>	OBO Format	1.8	AmphiAnat list	07/30/2008	OBOFoundry	<a href="#">Explore</a>
<a href="#">C. elegans gross anatomy (WBbt)</a>	OBO Format	1.21	Http://brachy Administrators	07/22/2008	OBOFoundry	<a href="#">Explore</a>
<a href="#">Dictyostelium discoideum anatomy (DDANAT)</a>	OBO Format	1.9	Rex			
<a href="#">Drosophila gross anatomy (FBbt)</a>	OBO Format	1.30	Http://fbbt Administrators	07/30/2008	OBOFoundry	<a href="#">Explore</a>
<a href="#">Fungal gross anatomy (FAO)</a>	OBO Format	1.3	Fun			
<a href="#">Human developmental anatomy, abstract version (EHDA)</a>	OBO Format	1.3	EMA			
<a href="#">Human developmental anatomy, timed version (EHDA)</a>	OBO Format	1.3	EMA			
<a href="#">Medaka fish anatomy and development (MFO)</a>	OBO Format	1.1	Med			
<a href="#">Mosquito gross anatomy (TGMA)</a>	OBO Format	1.10	C. L			
<a href="#">Mouse adult gross anatomy (MA)</a>	OBO Format	1.194	Ana			
<a href="#">Mouse gross anatomy and development (EMAP)</a>	OBO Format	1.2	EMA	07/30/2008	OBOFoundry	<a href="#">Explore</a>
<a href="#">Plant structure (PO)</a>	OBO Format	1.63	Po_anatomy Administrators	04/03/2009	OBOFoundry	<a href="#">Explore</a>
<a href="#">Spider Ontology (SPD)</a>	OBO Format	1.11	Martin Ramirez	07/24/2009	OBOFoundry	<a href="#">Explore</a>
<a href="#">Teleost anatomy and development (TAO)</a>	OBO Format	1.117	Wasila Dahdul	07/22/2009	OBOFoundry	<a href="#">Explore</a>
<a href="#">Tick gross anatomy (TADS)</a>	OBO Format	1.2	Http://www Administrators	07/30/2008	OBOFoundry	<a href="#">Explore</a>
<a href="#">Xenopus anatomy and development (XAO)</a>	OBO Format	1.17	Erik Segerdell	07/22/2009	OBOFoundry	<a href="#">Explore</a>
<a href="#">Zebrafish anatomy and development (ZFA)</a>	OBO Format	1.21	ZFIN administrators	06/23/2009	OBOFoundry	<a href="#">Explore</a>

ONTOLOGY NAME	FORMAT	VERSION	AUTHOR
<a href="#">C. elegans phenotype (WBPhenotype)</a>	OBO Format	1.82	Http://tazendra Administrators
<a href="#">Cereal plant trait (TO)</a>	OBO Format	1.61	Po_anatomy Administrators
<a href="#">Human disease (DOID)</a>	OBO Format	1.50	Human_disease Administrators
<a href="#">Infectious disease (IDO)</a>	OBO Format	1.3	Lindsay Cowell
<a href="#">Mammalian phenotype (MP)</a>	OBO Format	1.274	JAX list
<a href="#">Mosquito insecticide resistance (MIRO)</a>	OBO Format	1.521	C. Louis
<a href="#">Mouse pathology (MPATH)</a>	OBO Format	1.3	Mouse_pathology Administrators
<a href="#">Pathogen transmission (TRANS)</a>	OBO Format	1.11	Human_disease Administrators
<a href="#">Phenotypic quality (PATO)</a>	OBO Format	1.163	Quality Administrators
<a href="#">Units of measurement (UO)</a>	OBO Format	1.21	Unit Administrators
<a href="#">Yeast phenotypes (YPO)</a>	OBO Format	1.14	Mike Cherry



# OBO Foundry coverage

RELATION TO TIME	CONTINUANT			OCCURRENT
	GRANULARITY	INDEPENDENT	DEPENDENT	
ORGAN AND ORGANISM		Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO) Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)
CELL AND CELLULAR COMPONENT		Cell (CL)	<b>Cellular Component (FMA, GO)</b>	<b>Biological Process (GO)</b>
MOLECULE		Molecule (ChEBI, SO, RnaO, PrO)	<b>Molecular Function (GO)</b>	Molecular Process (GO)



# OBO Foundry as a community resource

- Provides ontology building know-how:
  - Tested ontology building and community best-practices
  - Incremental, modular, bottoms-up approach to evidence-based terminology
  - Principles for developing definitions and relationships
- Provides peer-review and community feedback



# Consistent definitions for terms

- Genus-differentia: A is a type of <parent of A> that <differentia>
- Example: premaxilla is-a dermal bone
  - def: "Dermal bone that forms the anteriormost element of the upper jaw. It articulates with the maxilla posterodorsally and its antimeres medially. The premaxilla is paired."



# Interoperability: Reuse of relations

## OBO Relations Ontology (R0)

### Foundational

*is\_a*  
*part\_of*

### Spatial

*located\_in*  
*contained\_in*  
*adjacent\_to*

### Temporal

*transformation\_of*  
*derives\_from*  
*preceded\_by*

### Participation

*has\_participant*  
*has\_agent*

“Relations in Biomedical Ontologies”, Genome Biology, April 2005



# Ontological needs for computable phenotypes

“Meckel’s cartilage greatly reduced”



Meckel’s cartilage  
*Entity (E)*

decreased size  
*Quality (Q)*

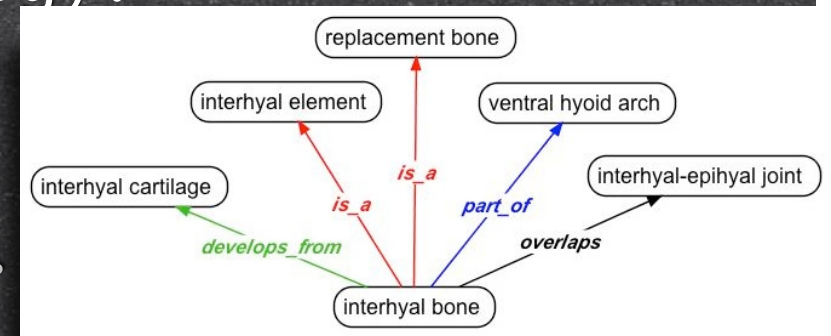
Meckel’s cartilage <i><u>Entity</u></i>	size: <i><u>Attri- bute</u></i>	decreased <i><u>Value</u></i>
<i>Character</i>		<i>Character State</i>

- In addition, need ontologies for
- who exhibits the phenotype (taxonomy)
- evidence



# Teleost Anatomy Ontology (TA0)

- Initiated in 2007 as a clone of the zebrafish anatomy ontology.
- 2371 terms, 395 new.
- Multi-species: teleosts.
- Terms are defined based on structure, homology is not implied.
- Focus is on the skeletal system as the ontology is driven by annotation.





# Taxonomically variable relationships

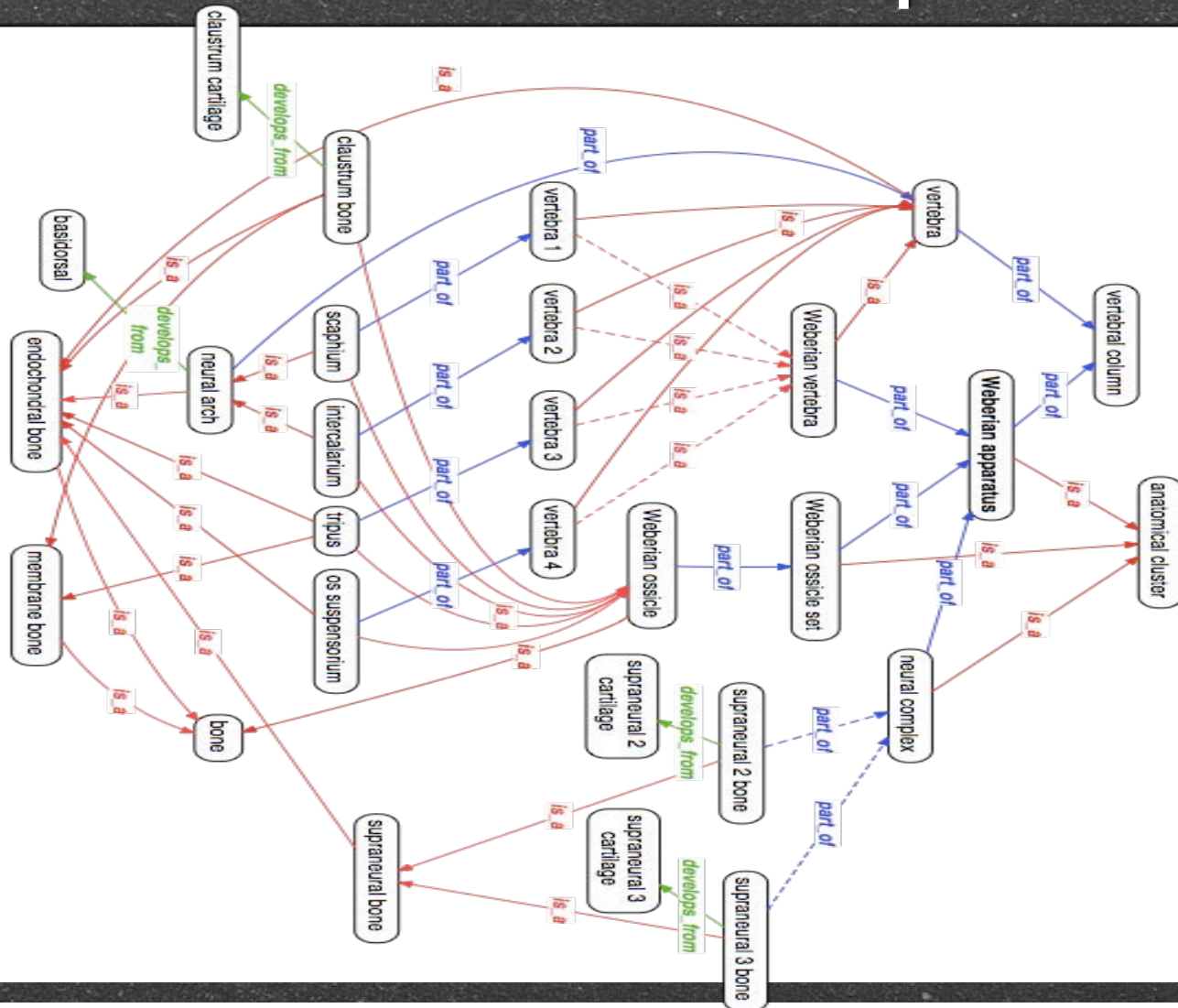




Image Record: [460945] *Chromobotia macracanthus*

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

CToL Date Submitted: 14-Jun-11

CToL Submitted By: 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

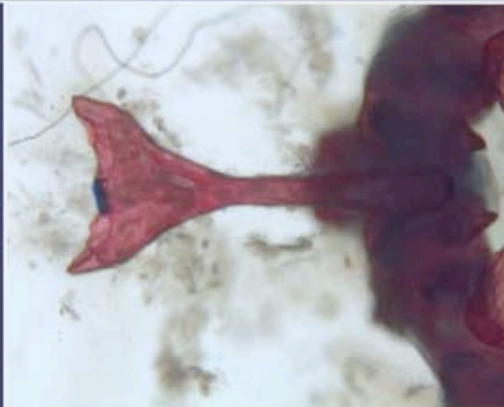
Technique: Digital Camera

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



FSI Viewer

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:

Determination

Class: Actinopterygii

Order: Cypriniformes

Family: Cobitidae

Genus: Chromobotia

Species: *Chromobotia macracanthus*

(Add Annotation...)

Basihyal  
bone

External links/identifiers

External identification: CToL-S:0001165

External identification: CToL-I:0000501

Ontology:

Teleost Anatomy TAO:0000316

Other Annotations (Add Annotation...)

TAO is already  
being used to  
link data



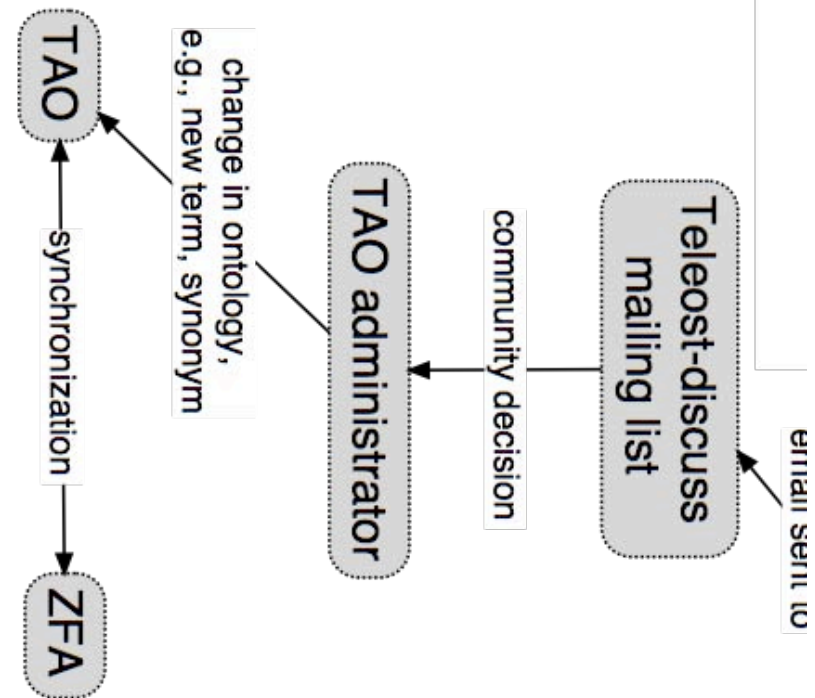
# Teleost Taxonomy Ontology (TT0)

- Based on and regularly updated from Bill Eschmeyer's Catalog of Fishes
- Includes custom additions, such as fossil taxa and higher order taxa.
- 36,060 terms (taxa), 38,000 synonyms



# Community ownership of ontology development

- OBO ontologies have a request tracker
- Participation is open
- Subject (biology) experts are key to ensure usefulness
- Ontology building is a collaborative effort





## Open Biomedical Ontologies

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### Tracker: Teleost Anatomy (TAO) term requests

Search: 
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ID	Summary	Status	Opened	Assignee	Submitter	Resolution	Priority
Assignee: <input type="text" value="Any"/> Status: <input type="text" value="Any"/> Category: <input type="text" value="Any"/> Group: <input type="text" value="Any"/> Submitter: <input type="text"/> Keyword: <input type="text"/>							
Artifact ID: <input type="text"/> <a href="#">Filter</a> <a href="#">Reset</a> <a href="#">Permalink</a>							
2812852	<a href="#">Pharyngobranchial series</a>	Open	2009-06-26	nobody	nobody	None	5
2808769	<a href="#">syn: operculare</a>	Open	2009-06-19	nobody	<a href="#">pmabee</a>	None	5
2808159	<a href="#">Parurohyal</a>	Open	2009-06-18	nobody	<a href="#">pmabee</a>	None	5
2808155	<a href="#">unbranched anal fin ray</a>	Open	2009-06-18	nobody	<a href="#">pmabee</a>	None	5
2808135	<a href="#">dorsal fin pterygiophore 1</a>	Open	2009-06-18	<a href="#">wdahdul</a>	<a href="#">wdahdul</a>	None	5
2808115	<a href="#">palate</a>	Open	2009-06-18	nobody	<a href="#">pmabee</a>	None	5
2808111	<a href="#">gill arch preferred term</a>	Open	2009-06-18	<a href="#">wdahdul</a>	<a href="#">wdahdul</a>	None	5
2807822	<a href="#">surangular</a>	Open	2009-06-17	nobody	<a href="#">tgrande</a>	None	5
2807821	<a href="#">craniotemporal musce</a>	Open	2009-06-17	nobody	<a href="#">tgrande</a>	None	5
2807819	<a href="#">diplospondyly</a>	Open	2009-06-17	nobody	<a href="#">tgrande</a>	None	5

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On Mar 25, 2008, at 5:11 PM, Vari, Richard wrote:

Hi,

Brian has pointed out some important points in noting the variation in the infraorbitals. The relative size of IO1 is often a function of head shape and it is rare that it is the largest element among characiforms. At least in some gymnotiforms it is reduced to a tubular canal not notably larger than the other elements in the series. In general it lies between the anteroventral margin of eye and the upper jaw.

IO3 and 4 are standardly separate with fusion of IO4 being with IO5 when present.

Comments about canals should be qualified with "when present" since canals are absent in some taxa, usually of smaller body sizes.

IO5 canal forms are highly variable, with canal absent in some taxa.

I did not follow the argument for the need for the dermosphenotic rather than IO6. IO6 seems to be readily identified elements even when there are less as a consequence of fusion between elements or loss.

Rich

Date: Tue, 25 Mar 2008 10:36:37 -04  
From: Brian Sidlauskas <bls16@du...>  
Subject: Re: [Obo-teleost-discuss] [ obo-Zebrafish Anatomy (ZFA) term requests-1924376 ] infraorbital definition  
To: Wasila Dahdul <dahdul@an...>

A few comments on these definitions:

- > Infraorbital 1 [synonym=lachrymal]
- > Dermal bone that covers the region
- > eye and the upper jaw. Infraorbital 1 (the lachrymal) is the first
- > and
- > largest bone in the infraorbital series. Bears the anterior most part
- > of the infraorbital canal.

IO1 is definitely not always the largest. In many characiforms IO3 is the biggest by far.

- > infraorbital 2
- > Plate like dermal bone, variable in size, though usually one of the
- > smallest in the series. Bears 2-3 neuromast pores enclosed within the
- > infraorbital canal (Nelson, 1969).
- >
- > infraorbital 3
- > Plate like dermal bone, variable in size, but usually largest in the
- > series. Usually fused with IO4 (eg. Opsariichthys, Nelson (1969)).
- > Usually bears 3 neuromast pores enclosed within the infraorbital
- > canal
- > (Nelson, 1969).

0 and 1 are possible pore numbers for these bones.

Date: Tue, 25 Mar 2008 11:13:48 -0500

From: "Arratia, Gloria" <garratia@ku.edu>

Subject: Re: [Obo-teleost-discuss] [ obo-Zebrafish Anatomy (ZFA) term requests-1924376 ] infraorbital definition

To: <bls16@du...>, "Wasila Dahdul" <dahdul@an...>

Dear Colleagues,

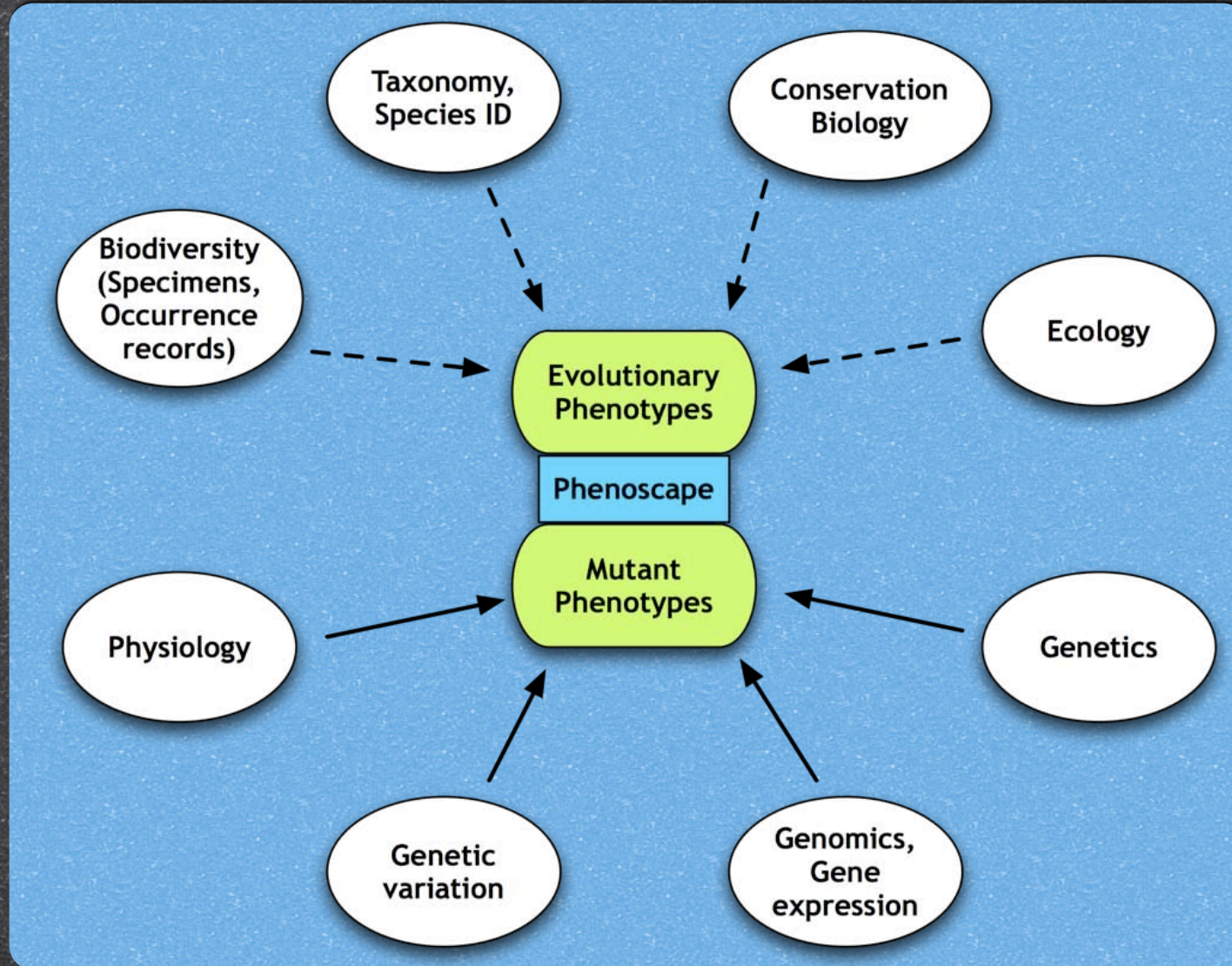
A few questions: At what level are you trying to define the infraorbital series of bones? At the teleostean basal level or at the ostariophysan level? The possible definitions presented in the last messages show a combination of alternatives that result from the fact that different teleostean levels are being used.

Probably, it may help if I define for you the conditions at the basal teleostean level:

The series of infraorbital bones at the basal teleostean level may have > the 7th or the 6th bone (the most dorsoposterior element). At this level, the common condition for the bone named dermosphenotic is the division of the canal in two branched: one that it may be associated to the supraorbital canal (or end blind in the bone) and other that is the infraorbital canal that joins the otic canal. From this pattern, there are many evolutionary transformation so that infraorbital 7 and 6 are lost, so that the dermosphenotic becomes the 5th bone of the series. Also the connections of the infraorbital canal with the supraorbital and or otic canals may be lost or modified in different teleostean subgroups.



# Systematic morphology can be part of data-rich biology





# Acknowledgements

- Phenoscape Personnel & PIs:  
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