

PHENOTYPED

Paula Mabee

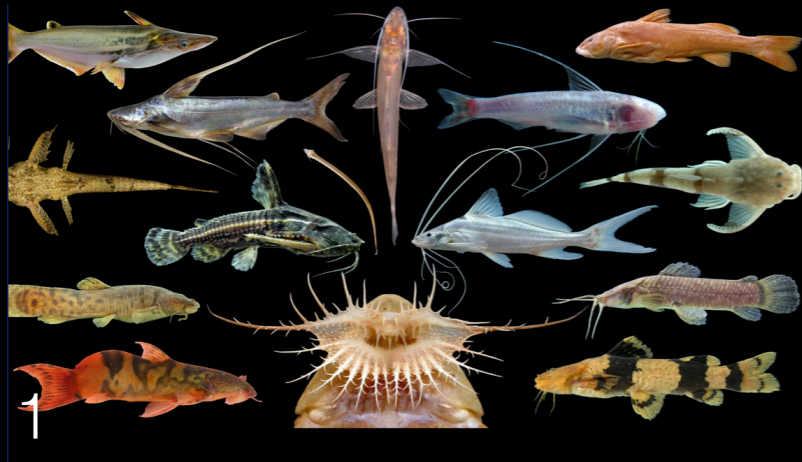
June 4, 2018

*Second Annual Digital Data in Biodiversity Research
Conference: Emerging Innovations for Biodiversity Data*

University of California, Berkeley



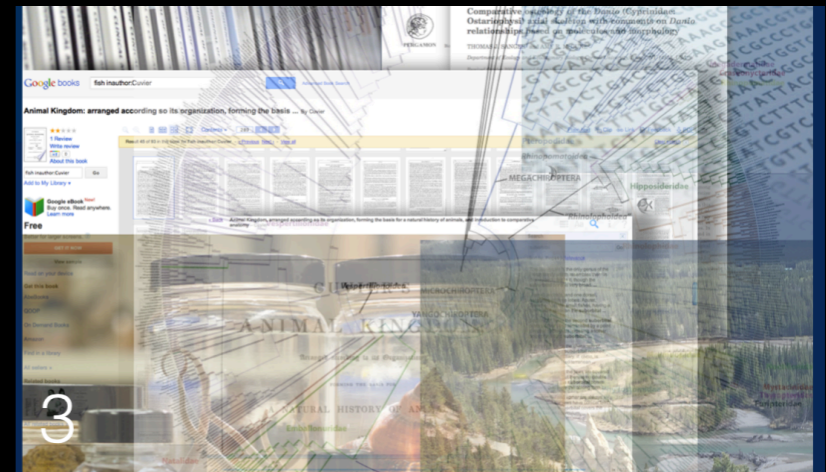
PHENOTYPED



1
The Promise 7 min
Mabee and her colleagues at NESCent initiate a bootcamp with the MOD World. They fall for semantic phenotypes but are unaware of the perils ahead.



2
Missing Link 8 min
Moving beyond fundamentals, the scientists begin to use the System's logic on the Phenotyped. Dark secrets about the genetic bases of phenotypes come to light and others have ideas to reverse the logic.



3
Beyond the Observed 10 min
When the scientists attempt to make a synthetic supermatrix of phenotypes, they realize that they can dramatically expand knowledge.



4
The Inter
Worried h
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phenotyp
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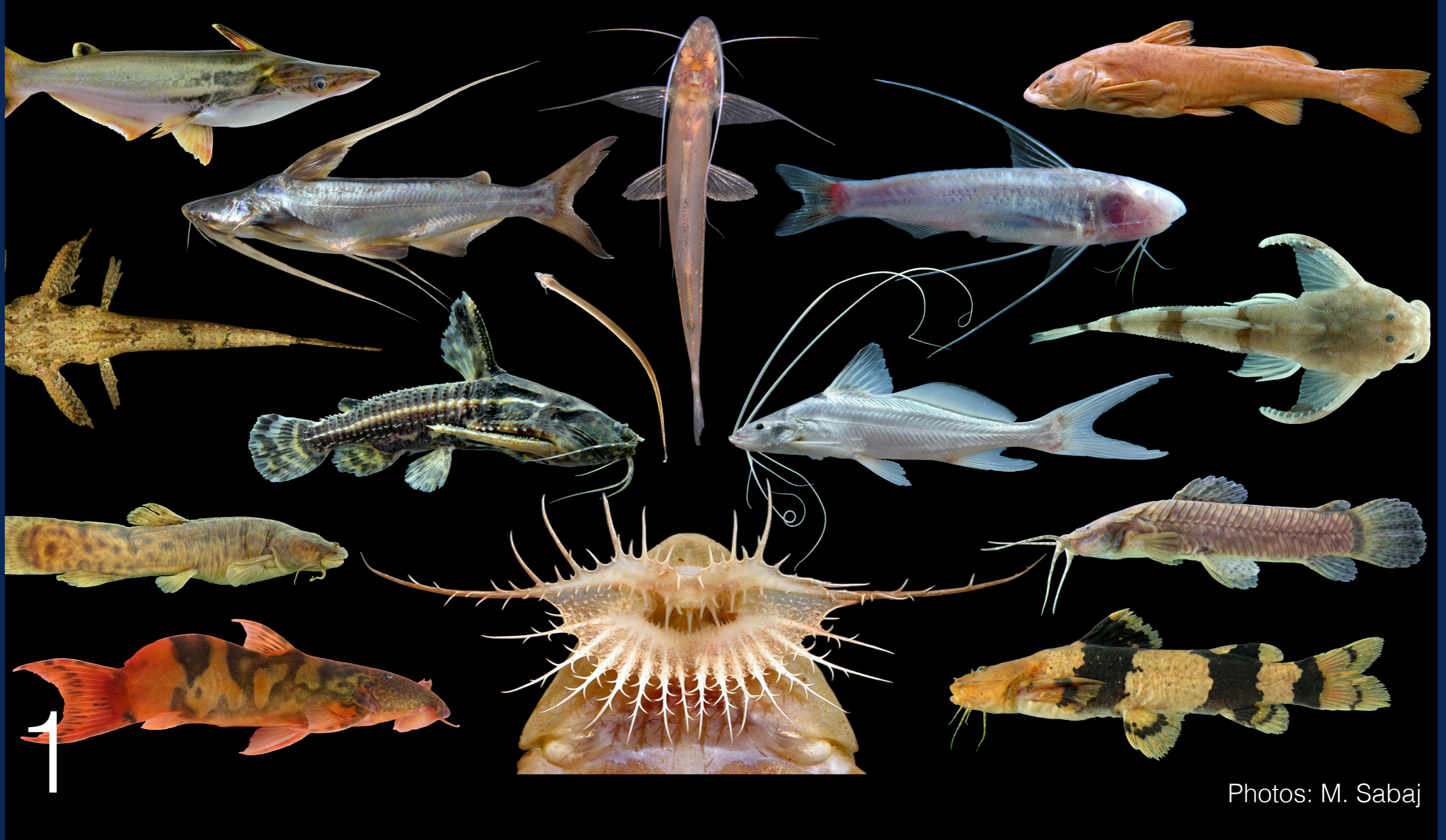
OVERVIEW

EPISODES

TRAILERS & MORE

MORE LIKE THIS

DETAILS



The Promise

7 min

Mabee and her colleagues at NESCent initiate a bootcamp with the MOD World. They fall for semantic phenotypes but are unaware of the perils ahead.



PERGAMON

Comparative osteology of the *Danio* (Cyprinidae: Ostariophysi) axial skeleton with comments on *Danio* relationships based on molecules and morphology

THOMAS J. SANGER¹ and AMY R. McCUNE*

Department of Ecology and Evolutionary Biology, Cornell University, Ithaca, NY 14853, USA

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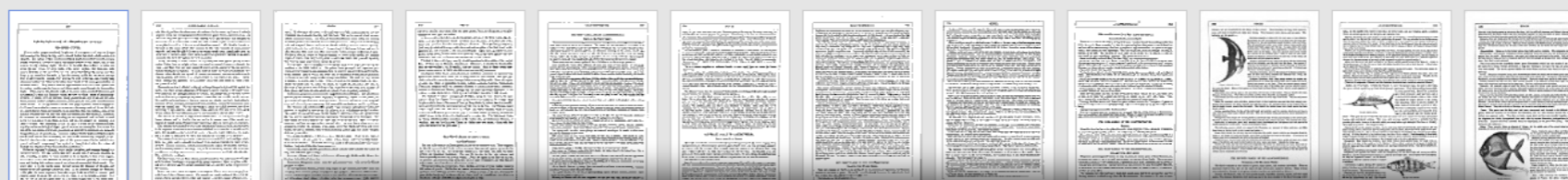
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Contents 289

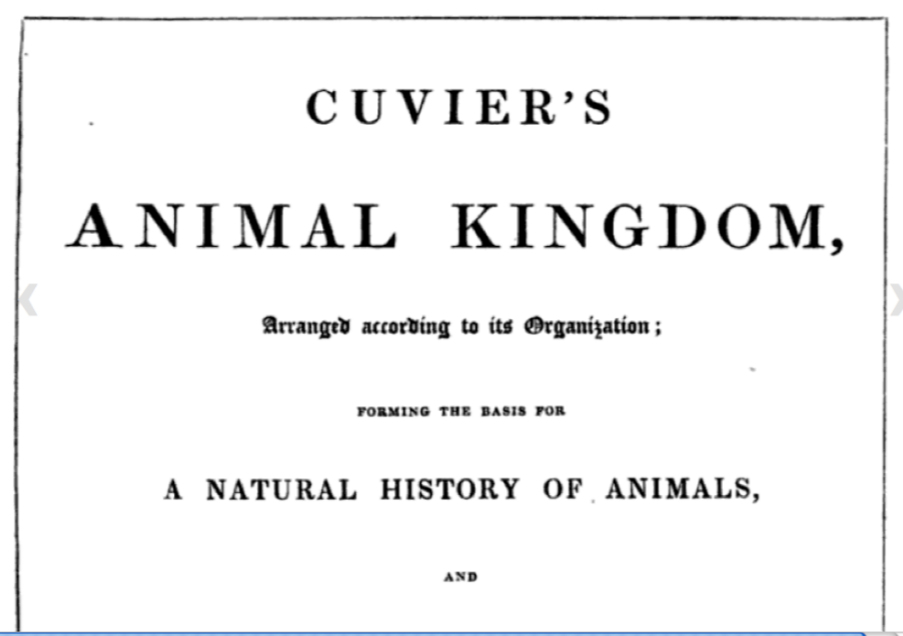
Plain text Clip Link Feedback PDF

Result 45 of 93 in this book for fish inauthor:Cuvier - Previous Next - View all

Clear search



Animal Kingdom, arranged according so its organization, forming the basis for a natural history of animals, and introduction to comparative anatomy - Cuvier



Search suborbital Go

Sort by: Pages | Relevance

The Star-gazer is the only genus of the Perch family which resembles them in this respect; but in it, though the suborbital bone is very broad, ...	294
... small ventrals, and one dorsal, consisting of three lobes. Aputet, Treacherous, are small fishes, having a formidable spine on the suborbital ...	295
ScolojuUa, have the second suborbital plate toothed, and terminated by a point directed backwards, crossing another point of the third suborbital, ...	296
The second, with the characters of Capito, have the suborbital notched, showing the maxillary. if. chclo, is common in the Mediterranean ...	305
In the living state, the jaws are covered with fleshy lips, but there is no double lip adhering to the suborbital bones. These fishes have the oblong form ...	311
... the tongue and vomer are always smooth, but the jaws have conical teeth, and the large suborbital covers the cheek like an operculum. ...	319

ix / 676

Rich legacy, digital, but not computable

Difficult for machines.....

<u>OMIM Query</u>	<u># of records</u>
"large bone"	785
"enlarged bone"	156
"big bones"	16
"huge bones"	4
"massive bones"	28
"hyperplastic bones"	12
"hyperplastic bone"	40
"bone hyperplasia"	134
"increased bone growth"	612

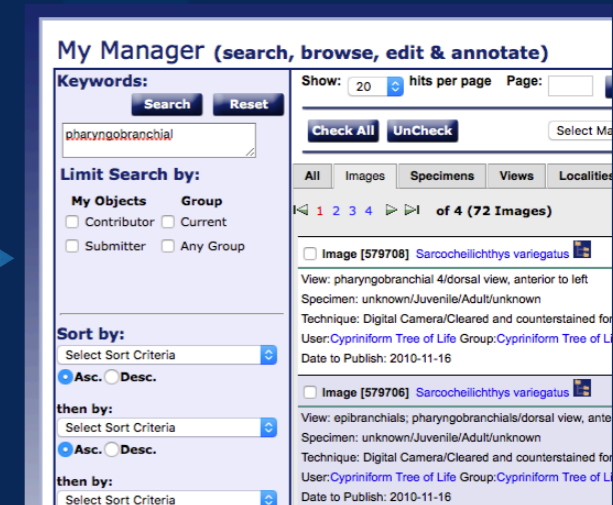
Discovery impeded

*“Find all images of keywords =
gill arch or gill arch skeleton”*

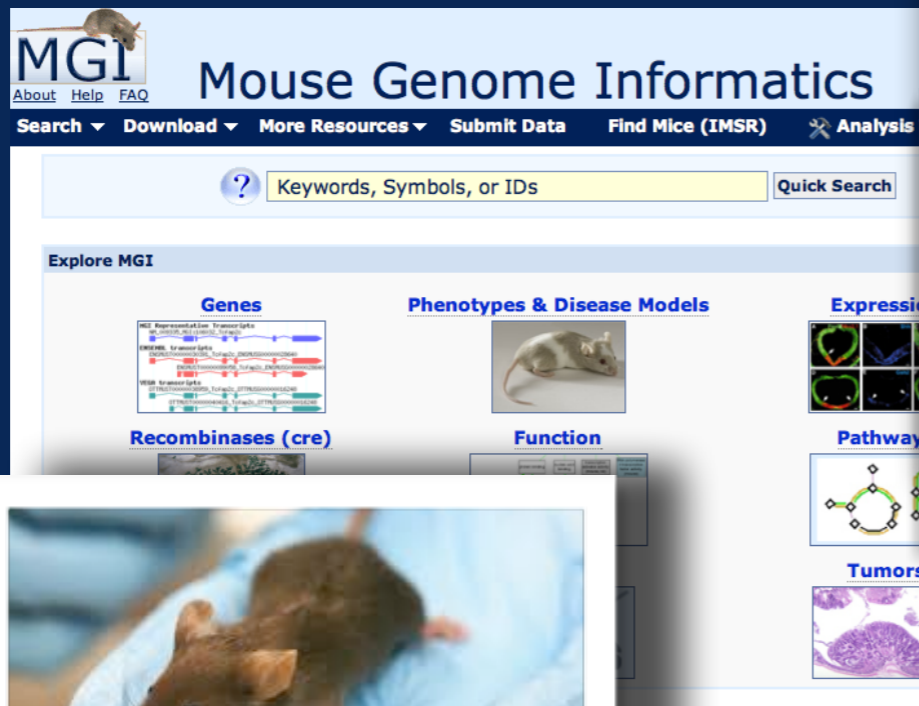
Results = 0



But there are many
images of parts of
gill arches



MOD world: using ontologies, model organism databases link phenotypes—genes

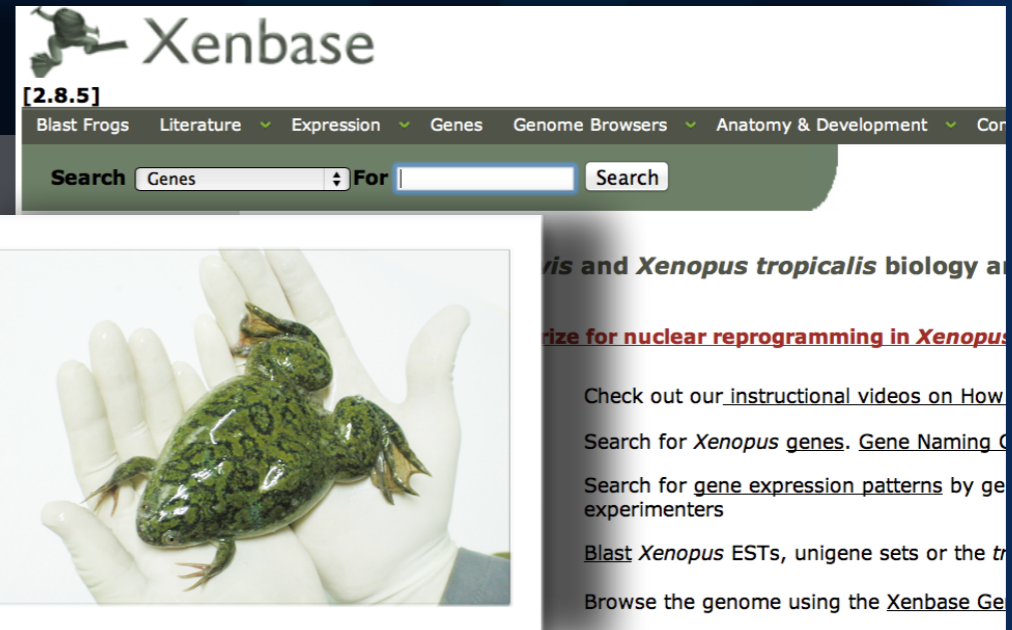


MGI Mouse Genome Informatics

Search Keywords, Symbols, or IDs

Explore MGI


- Genes
- Phenotypes & Disease Models
- Expression
- Recombinases (cre)
- Function
- Pathway
- Tumors



Xenbase [2.8.5]

Blast Frogs Literature Expression Genes Genome Browsers Anatomy & Development

Search Genes For



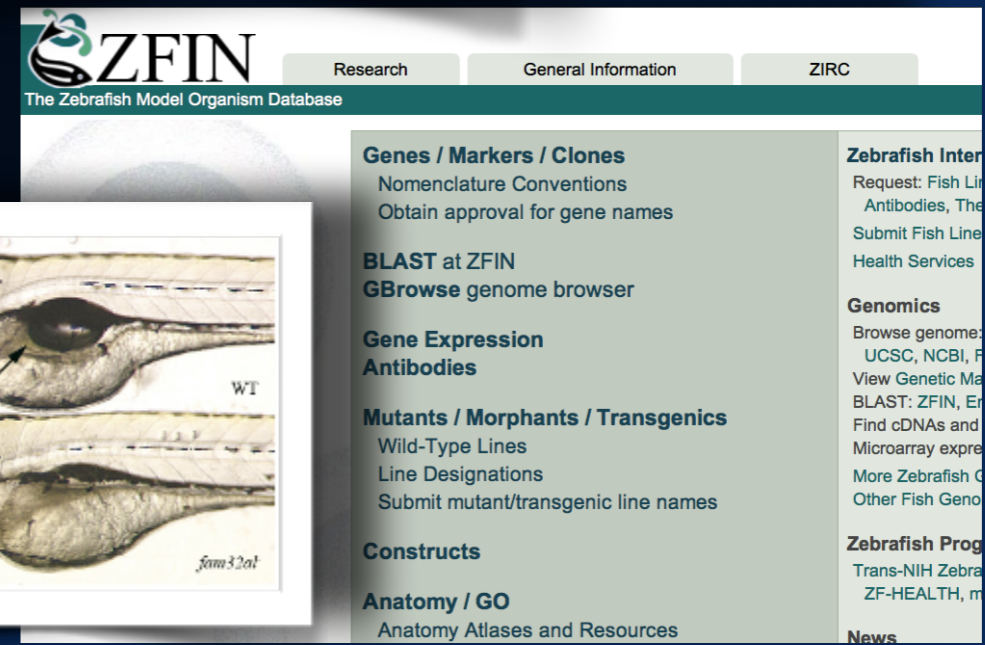
Check out our [instructional videos on How](#)

Search for *Xenopus* genes. [Gene Naming C](#)

Search for gene expression patterns by ge
experimenters

Blast *Xenopus* ESTs, unigene sets or the tr

Browse the genome using the [Xenbase Ge](#)



ZFIN The Zebrafish Model Organism Database

Research General Information ZIRC

- Genes / Markers / Clones
 - Nomenclature Conventions
 - Obtain approval for gene names
- BLAST at ZFIN
- GBrowse genome browser
- Gene Expression
- Antibodies
- Mutants / Morphants / Transgenics
 - Wild-Type Lines
 - Line Designations
 - Submit mutant/transgenic line names
- Constructs
- Anatomy / GO
 - Anatomy Atlases and Resources

Zebrafish Inter

- Request: Fish Lin
- Antibodies, The
- Submit Fish Line
- Health Services

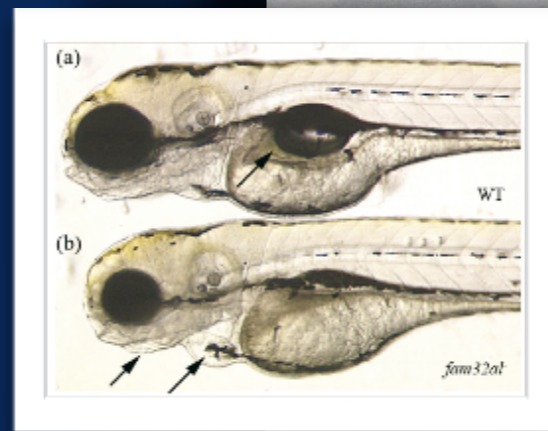
Genomics

- Browse genome:
- UCSC, NCBI, F
- View Genetic Ma
- BLAST: ZFIN, Er
- Find cDNAs and
- Microarray expre
- More Zebrafish C
- Other Fish Geno

Zebrafish Prog

- Trans-NIH Zebra
- ZF-HEALTH, m

News



Home News Tools Downloads Citing HPO Documentation HPO people

Home

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 - Defining HPO Terms
 - Annotation guide
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- HPO people

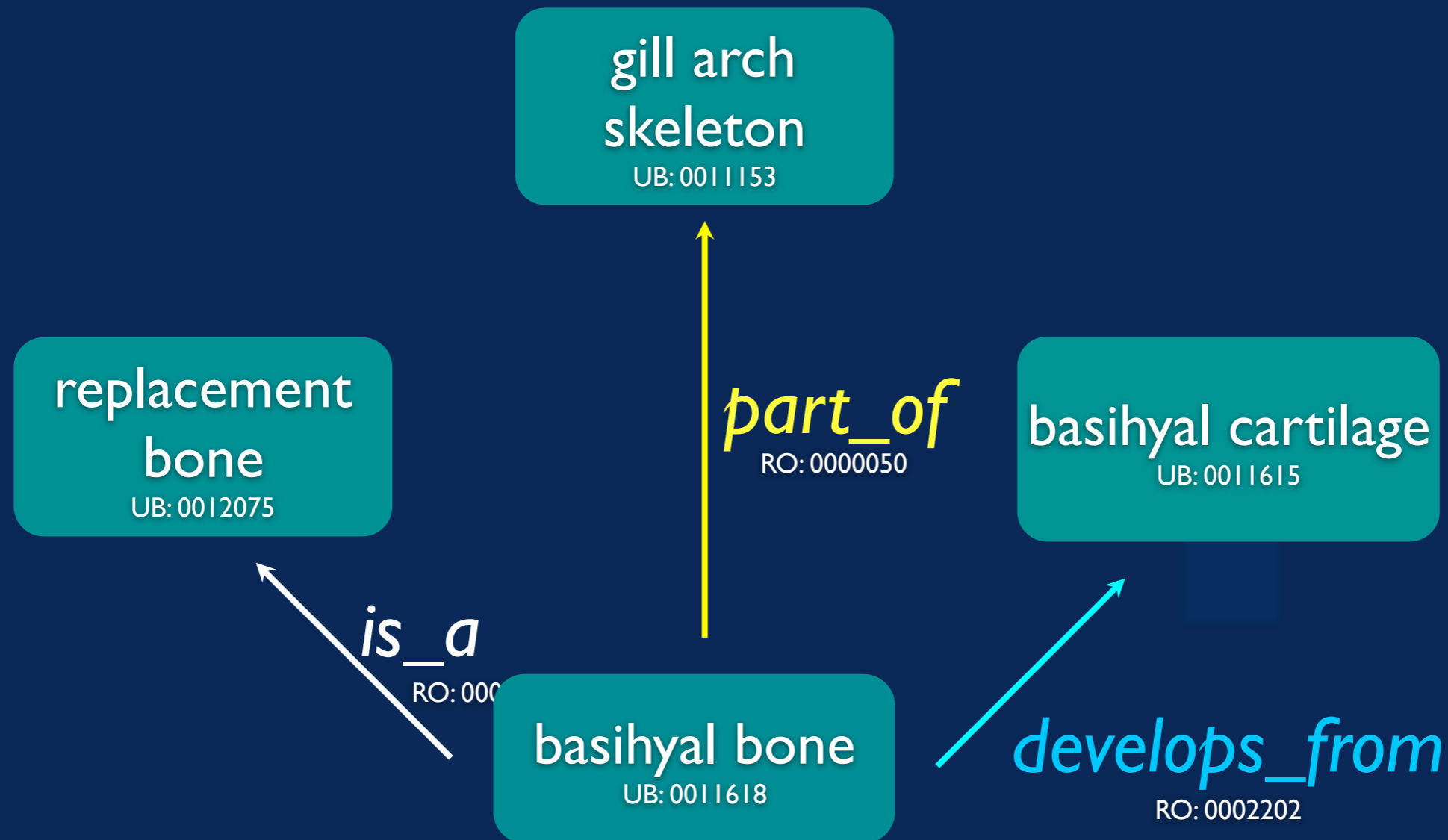


An ontology
vocabulary
(HPO) aims
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The HPO w
hugely imp
using inform
annotations

The HPO is
id Biomedical
Ontologies),
logical defini
for HPO terms
are being deve
using PATO a
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other ontologies
including the
PMA, GO, CHEBI,
and MPATH. The
HPO can be used
for clinical diagnostics
in human genetics
(Phenomizer),
bioinformatics
research on the
relationships be
tween human
phenotypic ab
normalities and
cellular and
biochemical n
etworks, for m
apping between
human and m
odel organism
phenotypes, and
for providing
a standardized
vocabulary for
clinical datab
ases, among
many other things.
There exists a
webpage for
every HPO-
term.

The HPO project
encourages input
from the medical
and genetics
community with
regards to the
ontology itself
and to clinical
annotations.

Semantics provide meaning



part_of: http://purl.obolibrary.org/obo/BFO_0000050
develops_from: http://purl.obolibrary.org/obo/RO_0002202

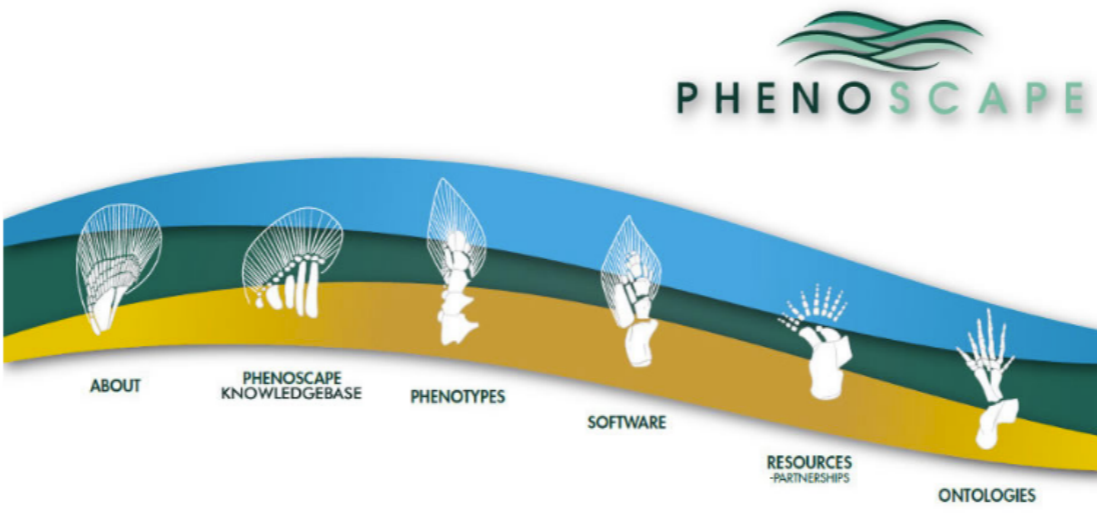
A (semantic) word is worth a thousand images...

gill arch skeleton
UB:0011153

Gill arch part	Number images
Basihyal bone	147
Basibranchial	236
Ceratobranchial	224
Hypobranchial	110
Epibranchial	220
Pharyngobranchial	92
Copula	70
TOTAL	1099

Phenoscape (2007)

Promise: Find, compare, integrate computable phenotypes

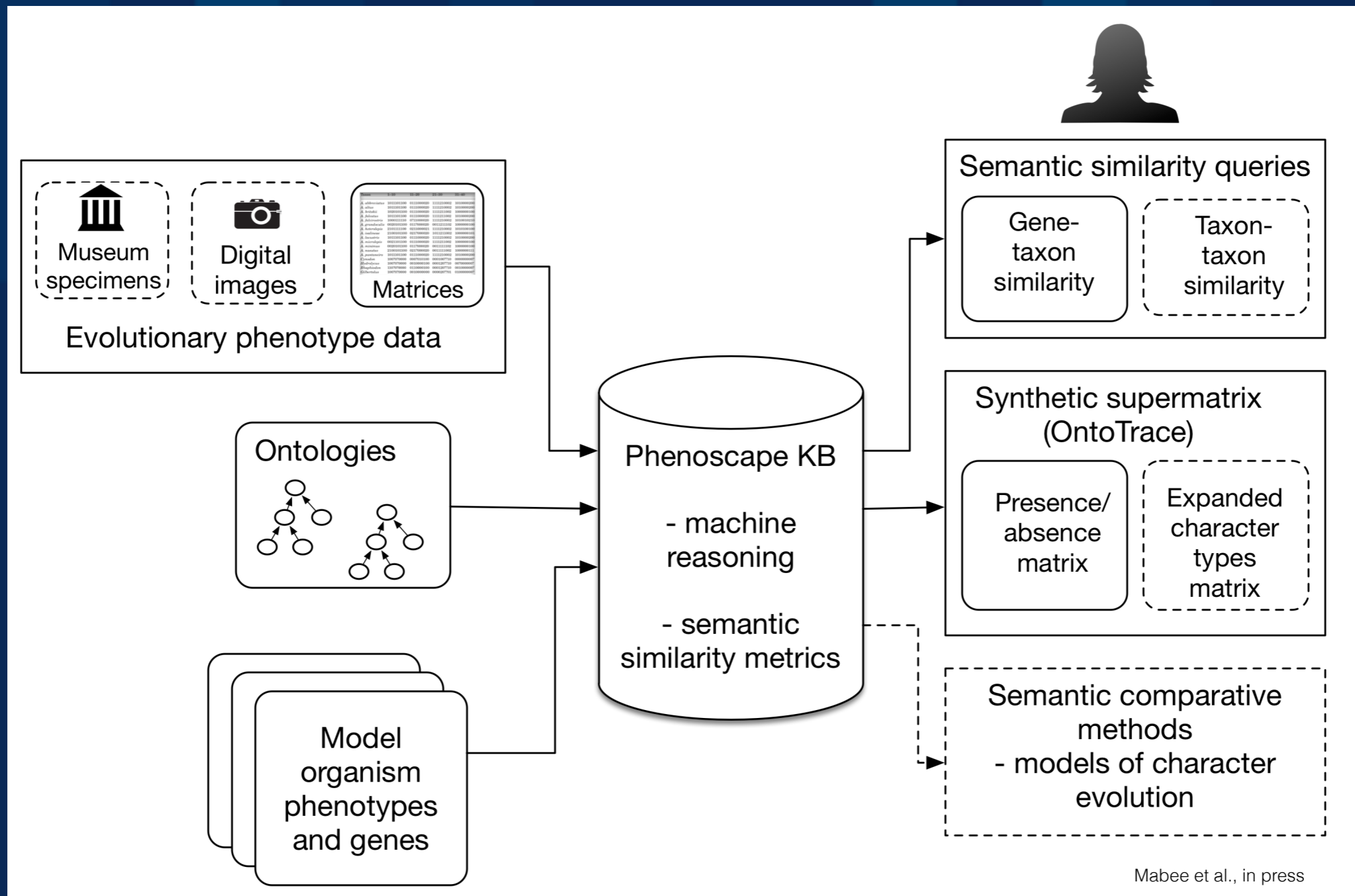


The logo features a stylized landscape with a blue sky, green hills, and a yellow ground. Six white icons representing different phenotypes are placed along the horizon. Below the icons are labels: ABOUT, PHENOSCAPE KNOWLEDGEBASE, PHENOTYPES, SOFTWARE, RESOURCES PARTNERSHIPS, and ONTOLOGIES. The word PHENOSCAPE is written in green capital letters at the top right of the logo.

Phenoscape project team

- University of North Carolina
 - [Todd Vision](#)
- National Evolutionary Synthesis Center (NESCent)
 - [Hilmar Lapp](#)
 - [Jim Balhoff](#)
 - [Prashanti Manda](#)
- University of South Dakota
 - [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 Havamizu](#)
- Cincinnati Children's Hospital Medical Center
 - [Aaron Zorn \(Xenbase\)](#)
 - [Christina James-Zorn \(Xenbase\)](#)
 - [Virgilio Ponferadda](#)
- California Academy of Sciences
 - [David Blackburn](#)
- University of Arizona
 - [Hong Cui](#)
- Lawrence Berkeley National Labs
 - [Chris Mungall](#)





Mabee et al., in press

Perils

Lots of resource & software development: phenotype ontologies, annotation software, Knowledgebase (KB), tools

Phenotyped: 4,260 species but >1 billion biological specimens in U.S. natural history collections!

TABLE 1. CHARACTER STATE MATRIX USED FOR PHYLOGENETIC ANALYSIS OF THE PLACEMENT OF *B. capapretum* WITHIN PIMELODIDAE AND *Brachyplatystoma*. Character states described in Appendix 1 and text.

	12345	67890	1	11111	11112	22222	22223	33333
<i>Steindachnerion</i>	11110	00000	00000	21000	00000	00000	00000	00011
<i>Phractocephalus-Leiarius</i> group	11110	00000	00000	00000	00000	00000	00000	00001
<i>Pimelodus</i> group	11111	11111	00000	00000	00000	00000	00000	00000
<i>Calophysus</i> group	11111	11111	00000	00000	00000	00000	00000	00000
<i>Zungaro</i>	11111	10000	00000	01001	00000	00000	00000	00010
<i>Sorubim</i> group	11111	10000	00000	20001	00000	00000	00000	00011
<i>Platynemichthys</i>	11111	10000	11000	00000	00000	00000	00000	00000
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000	00000	00000	00000	00000
<i>B. tigrinum</i>	11111	10000	11121	00111	00000	01110	00000	01201
<i>B. platyneumon</i>	11111	10000	11120	11111	00000	01001	20000	00010
<i>B. filax</i>	11111	10000	11111	11111	00000	01001	20000	00011
<i>B. capapretum</i>	11111	10000	11111	11111	00000	01001	20000	00011
<i>B. rousseauxi</i>	11111	10000	11111	11111	00000	01001	20000	00011
Heppteriidae	00000	00000	00000	00001	00000	00000	00000	00000
Pseudopimelodidae	00000	00000	00000	00000	00000	00000	00000	00000
Bagridae	00100	00000	00000	01000	00000	00000	00000	00000
Ictaluridae	00000	00000	00000	00001	00000	00000	00000	00000

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<i>Pimelodus</i> group	11111	11111	00000	00000	00000	00000	00000	00000
<i>Calophysus</i> group	11111	11111	00000	00000	00000	00000	00000	00000
<i>Zungaro</i>	11111	10000	00000	01001	00000	00000	00000	00010
<i>Sorubim</i> group	11111	10000	00000	20001	00000	00000	00000	00011
<i>Platynemichthys</i>	11111	10000	11000	00000	00000	00000	00000	00000
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000	00000	00000	00000	00000
<i>B. tigrinum</i>	11111	10000	11121	00111	00000	01110	00000	01201
<i>B. platyneumon</i>	11111	10000	11120	11111	00000	01001	20000	00010
<i>B. filax</i>	11111	10000	11111	11111	00000	01001	20000	00011
<i>B. capapretum</i>	11111	10000	11111	11111	00000	01001	20000	00011
<i>B. rousseauxi</i>	11111	10000	11111	11111	00000	01001	20000	00011
Heppteriidae	00000	00000	00000	00001	00000	00000	00000	00000
Pseudopimelodidae	00000	00000	00000	00000	00000	00000	00000	00000
Bagridae	00100	00000	00000	01000	00000	00000	00000	00000
Ictaluridae	00000	00000	00000	00001	00000	00000	00000	00000

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<i>Pimelodus</i> group	11111	11111	00000	00000	00000	00000	00000	00000
<i>Calophysus</i> group	11111	11111	00000	00000	00000	00000	00000	00000
<i>Zungaro</i>	11111	10000	00000	01001	00000	00000	00000	00010
<i>Sorubim</i> group	11111	10000	00000	20001	00000	00000	00000	00011
<i>Platynemichthys</i>	11111	10000	11000	00000	00000	00000	00000	00000
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000	00000	00000	00000	00000
<i>B. tigrinum</i>	11111	10000	11121	00111	00000	01110	00000	01201
<i>B. platyneumon</i>	11111	10000	11120	11111	00000	01001	20000	00010
<i>B. filax</i>	11111	10000	11111	11111	00000	01001	20000	00011
<i>B. capapretum</i>	11111	10000	11111	11111	00000	01001	20000	00011
<i>B. rousseauxi</i>	11111	10000	11111	11111	00000	01001	20000	00011
Heppteriidae	00000	00000	00000	00001	00000	00000	00000	00000
Pseudopimelodidae	00000	00000	00000	00000	00000	00000	00000	00000
Bagridae	00100	00000	00000	01000	00000	00000	00000	00000
Ictaluridae	00000	00000	00000	00001	00000	00000	00000	00000



Phenoscape Knowledgebase

Anatomy studies

- 651,660 phenotypes
- >5,000 taxa
- 161 studies
- >20,000 character states

Model organism datasets

- 309,383 phenotypes
- 15,447 genes
- +
- 732,658 anatomical expression
- 34,731 genes

Phenotyped specimen data

- from legacy literature
- from images
- 'born semantic' phenotypes
- predicted phenotypes

2

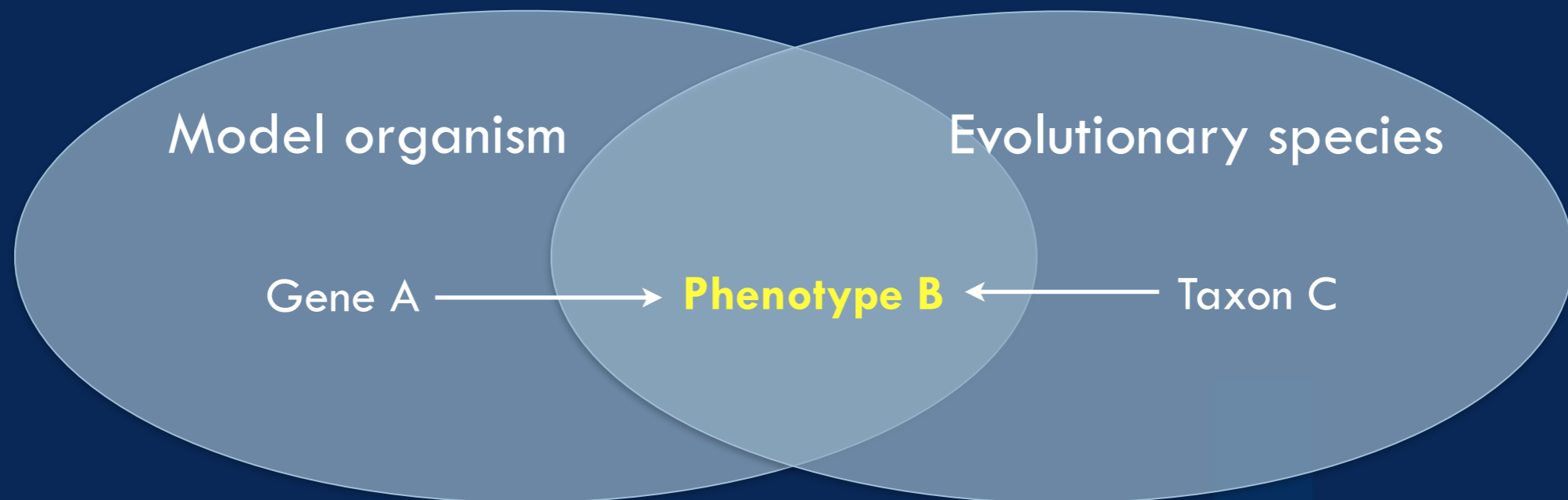


Missing Link

8 min

Moving beyond fundamentals, the scientists begin to use the System's logic on the Phenotyped. Dark secrets about the genetic bases of phenotypes come to light and others have ideas to reverse the logic.

Semantic phenotypes link species & genes



Gene-Phenotype connection

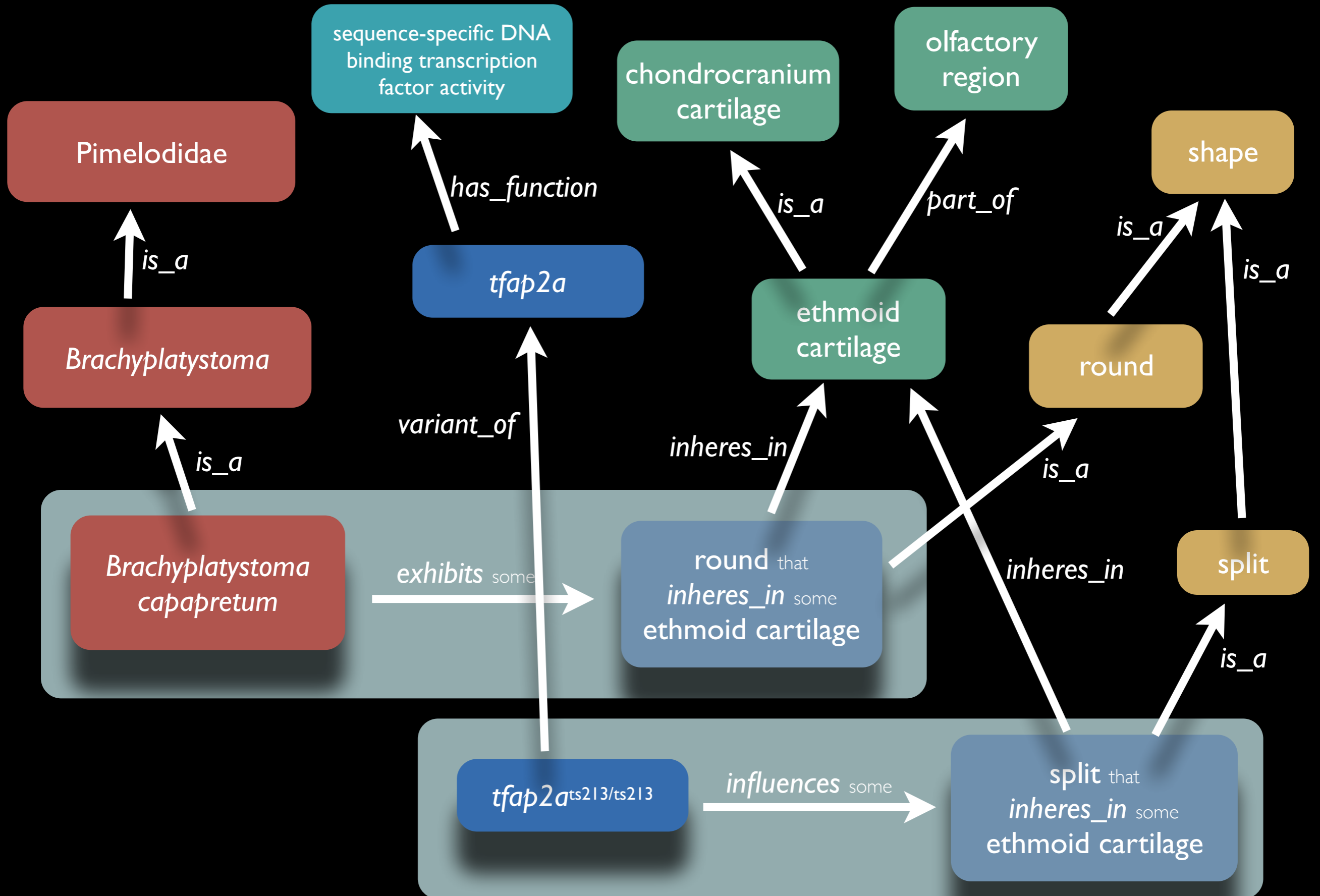




image: <http://www.ustacroport.com/buried-treasure>

KB is a Hypothesis generator
Interoperability yields 100,000's of
Gene-Phenotype hypotheses

E.g., How did the catfish lose its tongue?



Flathead Catfish; Photo by USFWS, used under Creative Commons License

Find the genes with a similar phenotype

Candidate genes

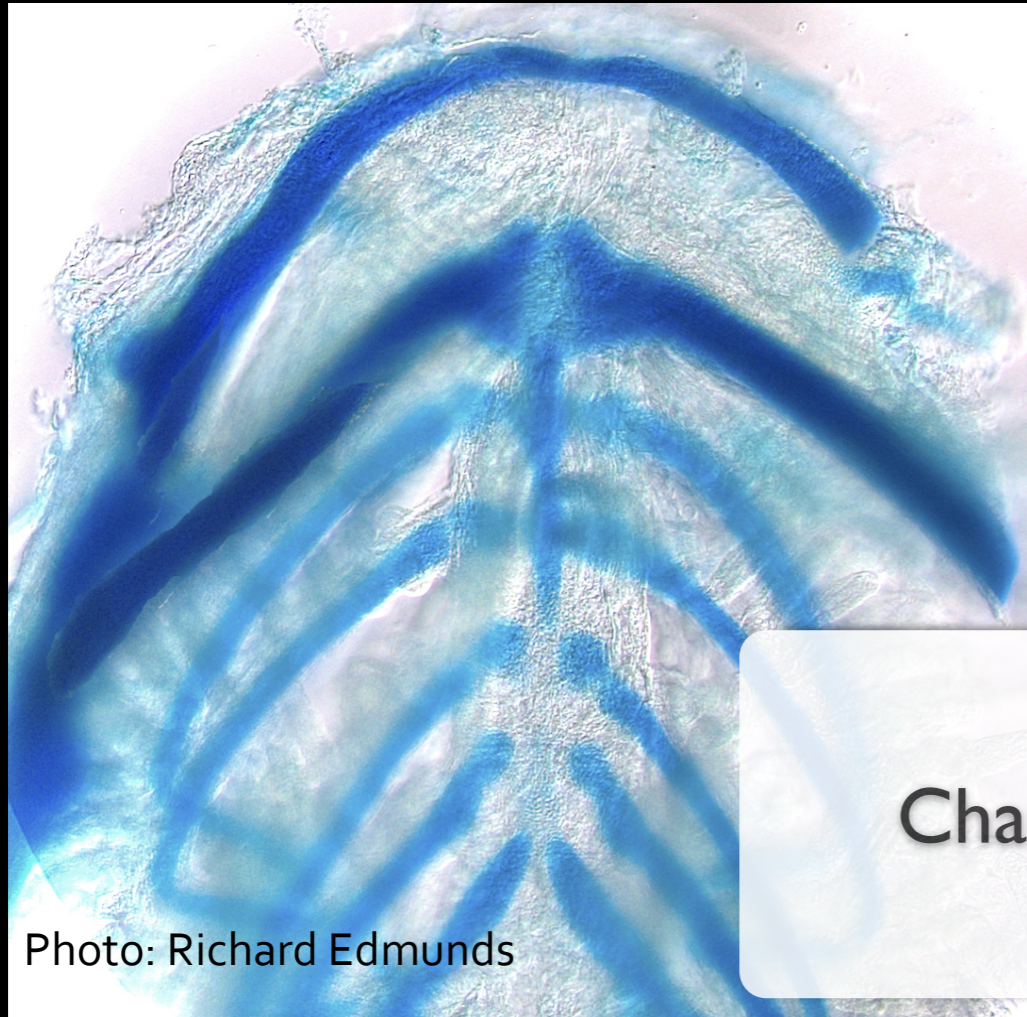
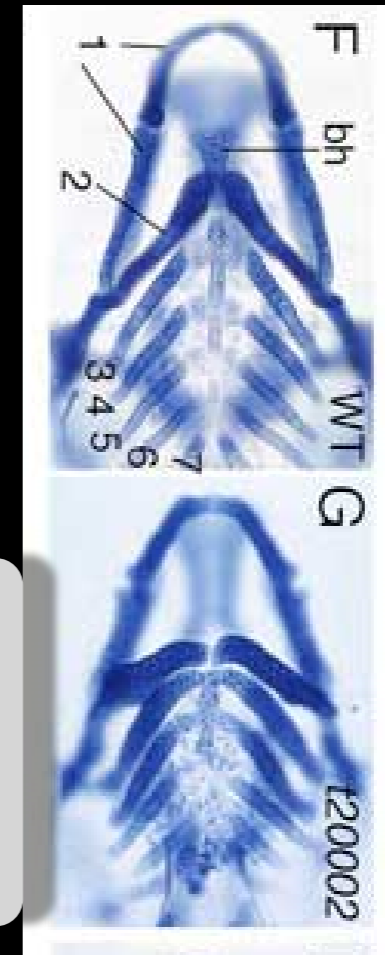


Photo: Richard Edmunds

Ictalurus punctatus

Changes in *brpf1*?



Laue et al., 2008

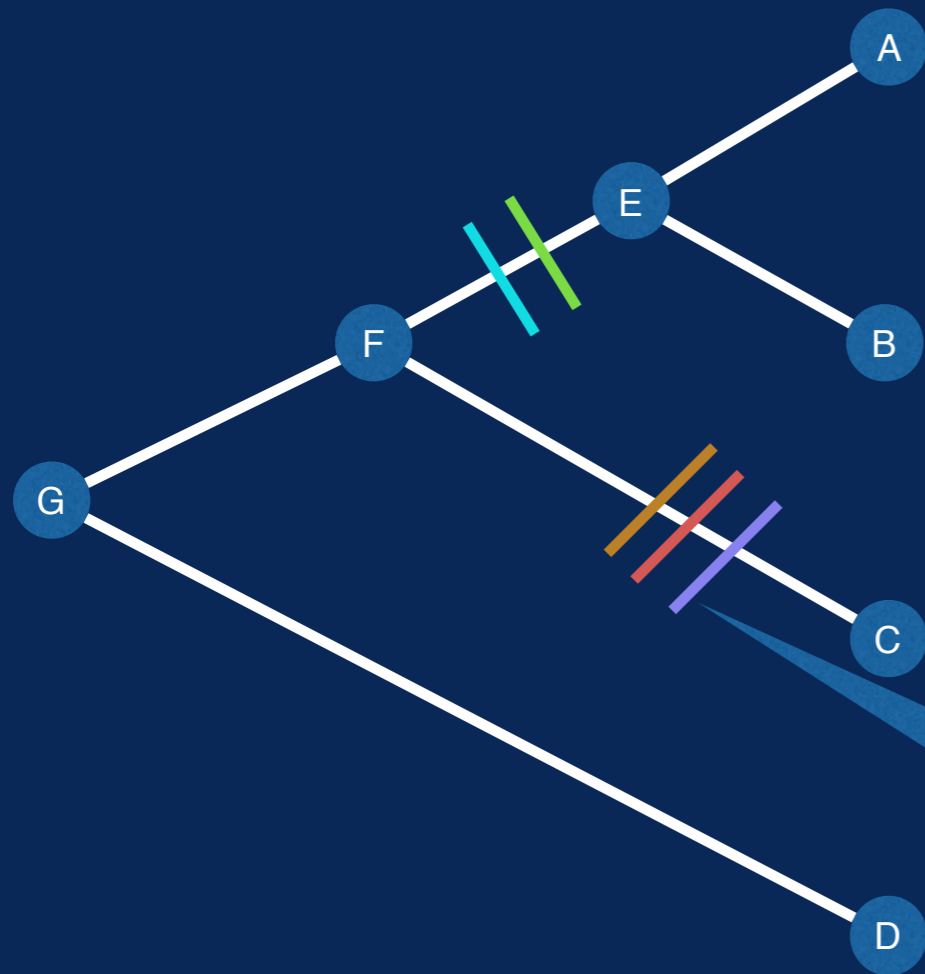
- brpf1*
- disc1*
- disp1*
- fac*
- foxd3*
- hand2*
- myst3*
- sox9a*
- unm_th9*
- unm_tn20c*
- unm_ty5*

taxa: **Siluriformes**

genes: (11)

Edmunds et al., 2016,
Mol. Biol Evol.

Phenoblast: find the genes with a similar set of phenotypes to a taxon

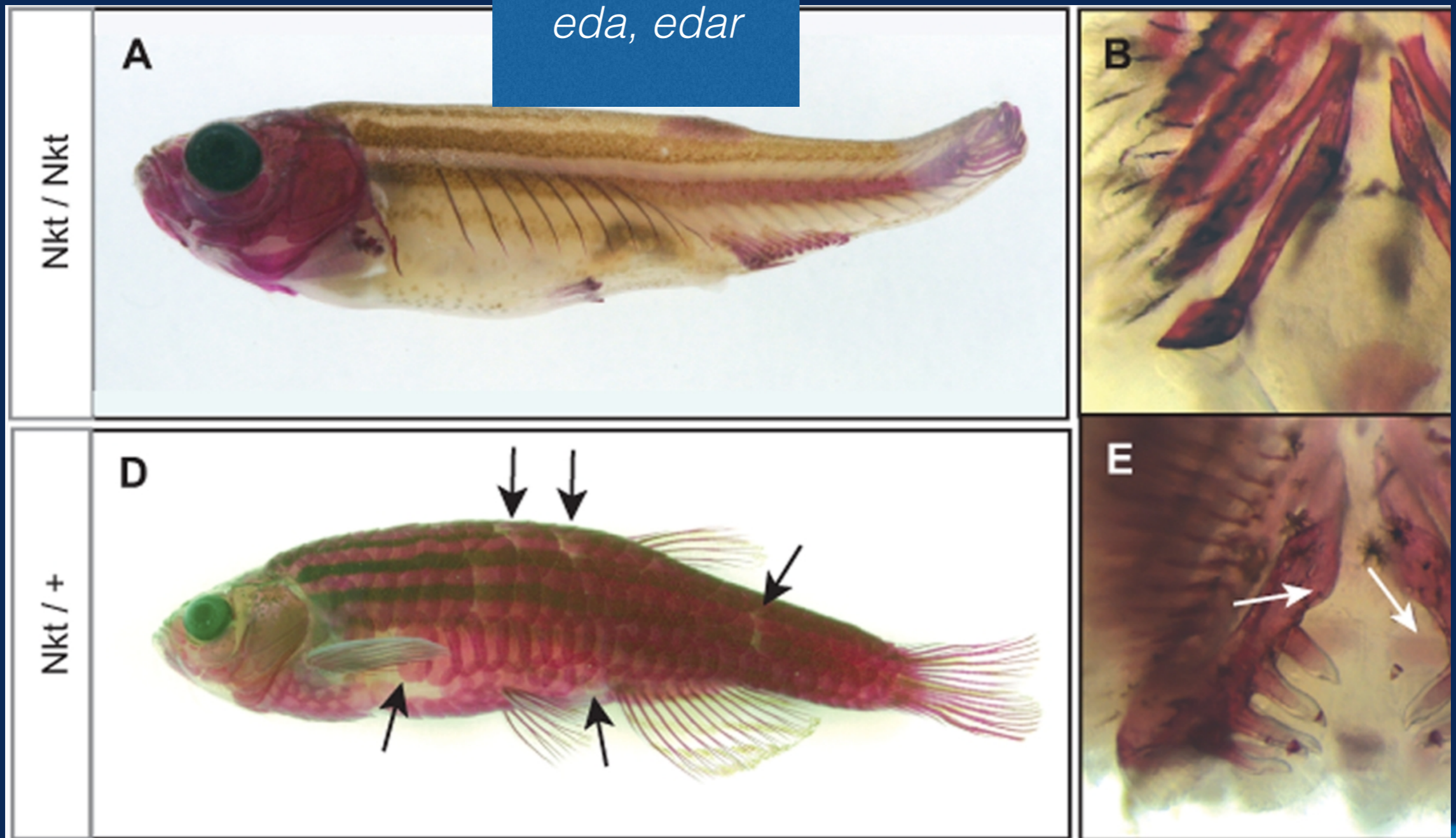


Set of phenotypes for a species



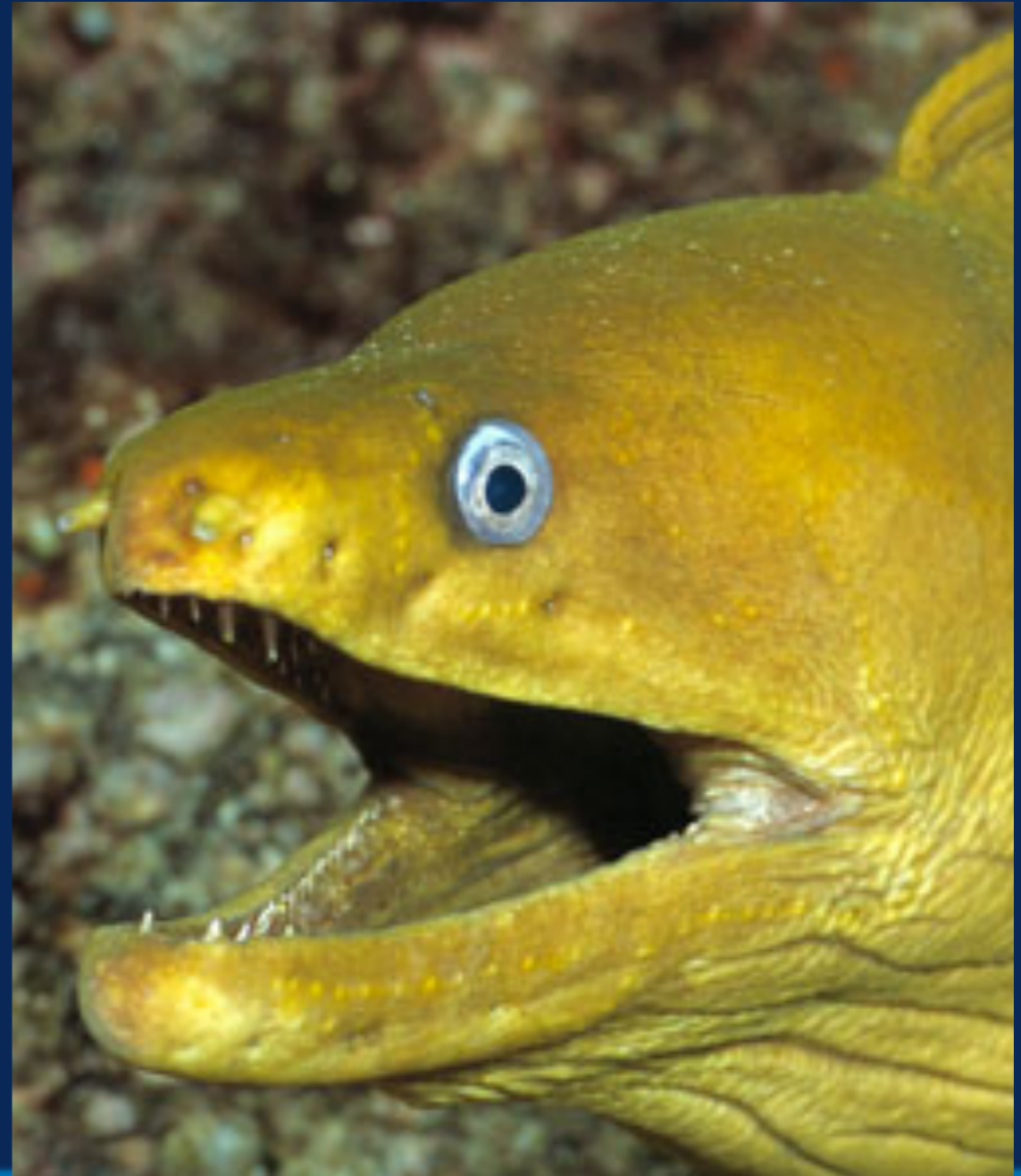
Reverse: Find the species with a phenotype similar to a gene...

eda, edar



'Candidate species' (195)

- including all eels, some catfish, etc.
- voucher specimens are in natural history collections



Phenotyped natural history specimens automatically linked with genetics



<https://celebrating200years.noaa.gov/datasets/fishcollection/>

Zoological Journal of the Linnean Society, 2006, 158, 1–14. With 14 figures

Comparative osteology of the *Danio* (Cyprinidae: Ostariophysii) axial skeleton with comments on *Danio* relationships based on molecules and morphology

THOMAS D. SANGER¹ AND AMY B. MCGRAW²

¹Department of Ecology and Evolutionary Biology, Cornell University, Ithaca, NY 14853, USA

²Department of Biology, Cornell University, Ithaca, NY 14853, USA

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3

MEGACHIROPTERA

MICROCHIROPTERA

YANGOCHIROPTERA

Rhinolophidae

Myristacidae

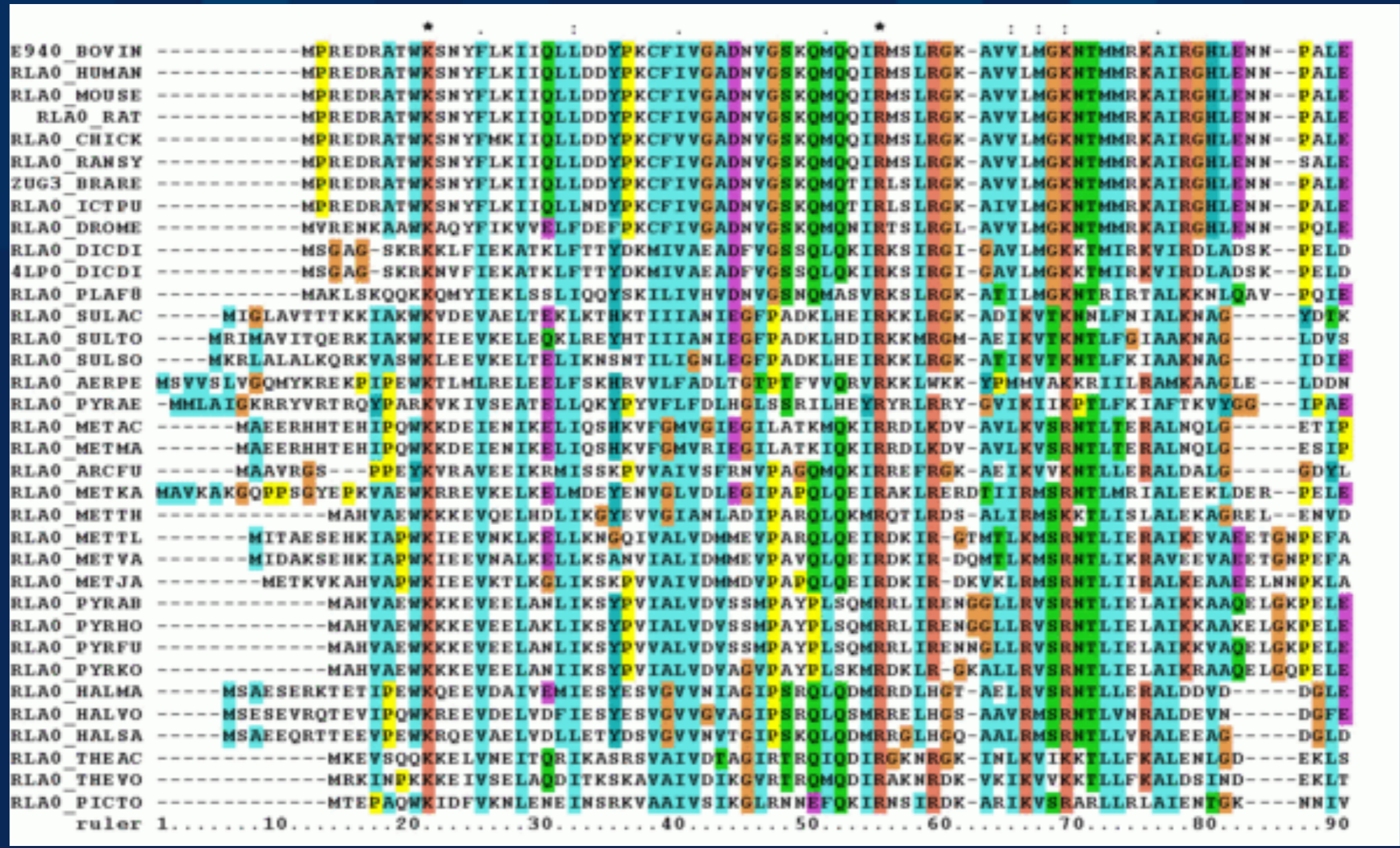
Thysanoptera

Furpteridae

Beyond the Observed

10 min

When the scientists attempt to make a synthetic supermatrix of phenotypes, they realize that they can dramatically expand knowledge.



By Miguel Andrade at English Wikipedia - Transferred from en.wikipedia to Commons.,
 CC BY-SA 3.0, <https://commons.wikimedia.org/w/index.php?curid=3930704>

Genetic supermatrix

TABLE 1. CHARACTER STATE MATRIX USED FOR PHYLOGENETIC ANALYSIS OF THE PLACEMENT OF *B. capapretum* WITHIN PIMELODIDAE AND *Brachyplatystoma*. Character states described in Appendix 1 and text.

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<i>Pimelodus</i> group	11111	11111	00000	00000			
<i>Calophysus</i> group	11111	11111	00000	00000			
<i>Zungaro</i>	11111	10000	00000	01001			
<i>Sorubim</i> group	11111	10000	00000	20001			
<i>Platynematichthys</i>	11111	10000	11000	00000			
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000			
<i>B. tigrinum</i>	11111	10000	11121	00111			
<i>B. platyneumum</i>	11111	10000	11120	11111			
<i>B. filax</i>	11111	10000	11111	11111			
<i>B. cape</i>	11111	10000	11111	11111			
<i>B. rous</i>	11111	10000	11111	11111			
Heptapteridae	11111	10000	11111	11111			
Pseudopimelodidae	11111	10000	11111	11111			
Bagridae	11111	10000	11111	11111			
Ictaluridae	11111	10000	11111	11111			

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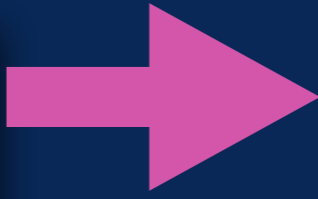
	12345	67890	11111 12345	11112 67890	22222 12345	22223 67890	33333 12345
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<i>Pimelodus</i> group	11111	11111	00000	00000	00000	00000	00000
<i>Calophysus</i> group	11111	11111	00000	00000	01110	00000	01201
<i>Zungaro</i>	11111	10000	00000	01001	20000	00000	00010
<i>Sorubim</i> group	11111	10000	00000	20001	20000	00000	00011
<i>Platynematichthys</i>	11111	10000	11000	00000	00000	00000	00000
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000	00000	00000	10010
<i>B. tigrinum</i>	11111	10000	11120	11111	11110	00000	11100
<i>B. platyneumum</i>	11111	10000	11111	11111	21101	11111	01101
<i>B. filax</i>	11111	10000	11111	11111	21101	11111	01101

TABLE 1. CHARACTER STATE MATRIX USED FOR PHYLOGENETIC ANALYSIS OF THE PLACEMENT OF *B. capapretum* WITHIN PIMELODIDAE AND *Brachyplatystoma*. Character states described in Appendix 1 and text.

	12345	67890	11111 12345	11112 67890	22222 12345	22223 67890	33333 12345
<i>Steindachnerion</i>	11110	00000	00000	21000	00000	00000	00011
<i>Phractocephalus-Leiarius</i> group	11110	00000	00000	00000	00000	00000	00001
<i>Pimelodus</i> group	11111	11111	00000	00000	00000	00000	00000
<i>Calophysus</i> group	11111	11111	00000	00000	01110	00000	01201
<i>Zungaro</i>	11111	10000	00000	01001	20000	00000	00010
<i>Sorubim</i> group	11111	10000	00000	20001	20000	00000	00011
<i>Platynematichthys</i>	11111	10000	11000	00000	00000	00000	00000
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000	00000	00000	10010
<i>B. tigrinum</i>	11111	10000	11120	11111	11110	00000	11100
<i>B. platyneumum</i>	11111	10000	11111	11111	21101	11111	01101
<i>B. filax</i>	11111	10000	11111	11111	21101	11111	01101

TABLE 1. CHARACTER STATE MATRIX USED FOR PHYLOGENETIC ANALYSIS OF THE PLACEMENT OF *B. capapretum* WITHIN PIMELODIDAE AND *Brachyplatystoma*. Character states described in Appendix 1 and text.

	12345	67890	11111 12345	11112 67890	22222 12345	22223 67890	33333 12345
<i>Steindachnerion</i>	11110	00000	00000	21000	00000	00000	00011
<i>Phractocephalus-Leiarius</i> group	11110	00000	00000	00000	00000	00000	00001
<i>Pimelodus</i> group	11111	11111	00000	00000	00000	00000	00000
<i>Calophysus</i> group	11111	11111	00000	00000	01110	00000	01201
<i>Zungaro</i>	11111	10000	00000	01001	20000	00000	00010
<i>Sorubim</i> group	11111	10000	00000	20001	20000	00000	00011
<i>Platynematichthys</i>	11111	10000	11000	00000	00000	00000	00000
<i>Brachyplatystoma vaillantii</i>	11111	10000	11111	11000	00000	00000	10010
<i>B. tigrinum</i>	11111	10000	11121	00111	11000	00000	10010
<i>B. platyneumum</i>	11111	10000	11120	11111	11110	00000	11100
<i>B. filax</i>	11111	10000	11111	11111	21101	11111	01101
<i>B. capapretum</i>	11111	10000	11111	11111	21101	11111	01101
<i>B. rousseauxii</i>	11111	10000	11111	11111	21101	11100	01101
Heptapteridae	00000	00000	00000	00001	00000	00000	00001
Pseudopimelodidae	00000	00000	00000	00000	01000	00000	10010
Bagridae	00100	00000	00000	01000	00000	00000	00000
Ictaluridae	00000	00000	00000	00001	00000	00000	00000



	0	1	0	1	0	0	1	0	1	0	1	1	0	1	0	1	0	0	1	0	1
Alligator	0	1	0	1	0	0	1	0	1	0	1	1	0	1	0	1	0	0	1	0	1
Alligator	1	0	0	0	1	0	1	1	0	1	1	0	1	0	0	0	1	0	1	1	0
Alligator	0	1	0	1	0	1	0	1	0	0	1	0	0	1	0	1	0	1	0	1	0
Alligator	1	0	1	1	1	0	1	1	0	0	1	0	1	0	1	1	1	0	1	1	0
Alligator	0	0	1	1	1	0	1	0	1	1	0	1	0	0	1	1	1	0	1	0	1
Alligator	1	0	0	0	1	0	0	1	0	0	1	0	1	0	0	0	1	0	0	1	0
Alligator	1	0	1	1	1	0	1	0	1	1	0	1	0	1	1	1	0	1	0	1	1
Alligator	1	0	1	1	1	0	1	1	1	1	0	1	0	1	1	1	0	1	1	1	1
Alligator	1	0	1	1	1	0	1	1	1	1	0	1	0	1	1	1	0	1	1	1	0
Alligator	0	0	1	1	0	1	1	1	1	0	1	1	1	1	0	1	1	0	1	1	0
Alligator	1	0	1	1	1	0	1	1	1	0	0	1	0	1	0	1	1	0	1	1	0
Alligator	1	0	1	1	0	1	0	1	0	1	1	0	1	0	1	1	0	1	0	1	0
Alligator	1	0	1	1	1	1	1	1	1	1	1	1	1	1	0	1	1	1	1	1	1
Alligator	1	0	1	0	0	0	1	1	0	0	1	0	1	0	1	0	0	0	1	1	0
Alligator	1	0	1	1	0	1	0	1	0	1	1	0	1	0	1	1	0	1	0	1	0
Alligator	0	0	1	1	1	1	0	1	1	1	0	0	0	1	1	1	0	0	1	1	0
Alligator	1	0	1	1	1	1	1	1	1	1	1	0	1	0	1	1	1	1	1	1	1
Alligator	0	0	1	0	0	1	1	0	1	1	0	1	0	0	1	0	0	1	1	0	1
Alligator	0	0	1	0	0	0	1	0	1	1	0	0	0	1	0	0	0	1	0	0	1
Alligator	1	0	1	1	1	1	1	0	1	1	0	0	1	0	1	1	1	1	1	0	1
Alligator	0	0	1	1	1	1	1	0	1	1	0	1	0	0	1	1	1	1	1	0	1

Phenotype supermatrix (+/-)

❖ *OntoTrace*: Software tool that returns +/- synthetic matrices for desired taxa and characters

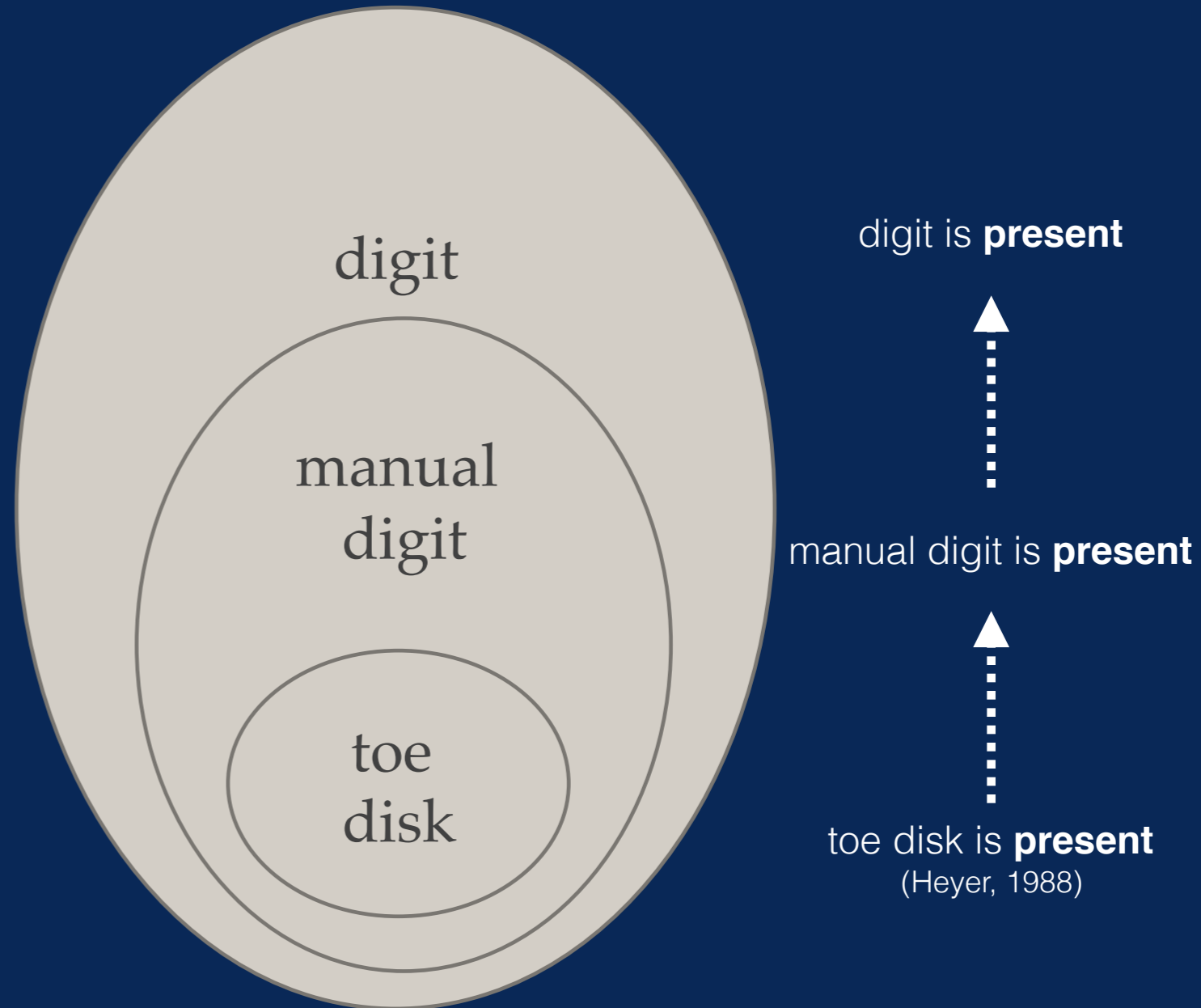
Dececchi, Balhoff, Lapp, & Mabee (2015) *Systematic Biology* 64(6), 936-952.

Infer presence, from presence of part

- “Bottom up”
- If part of an entity is present, then the entity is present
- If a species has toe disks, it has digits

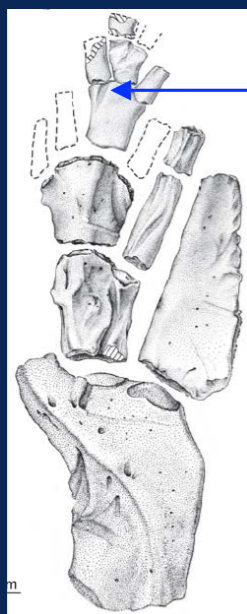


toe disks



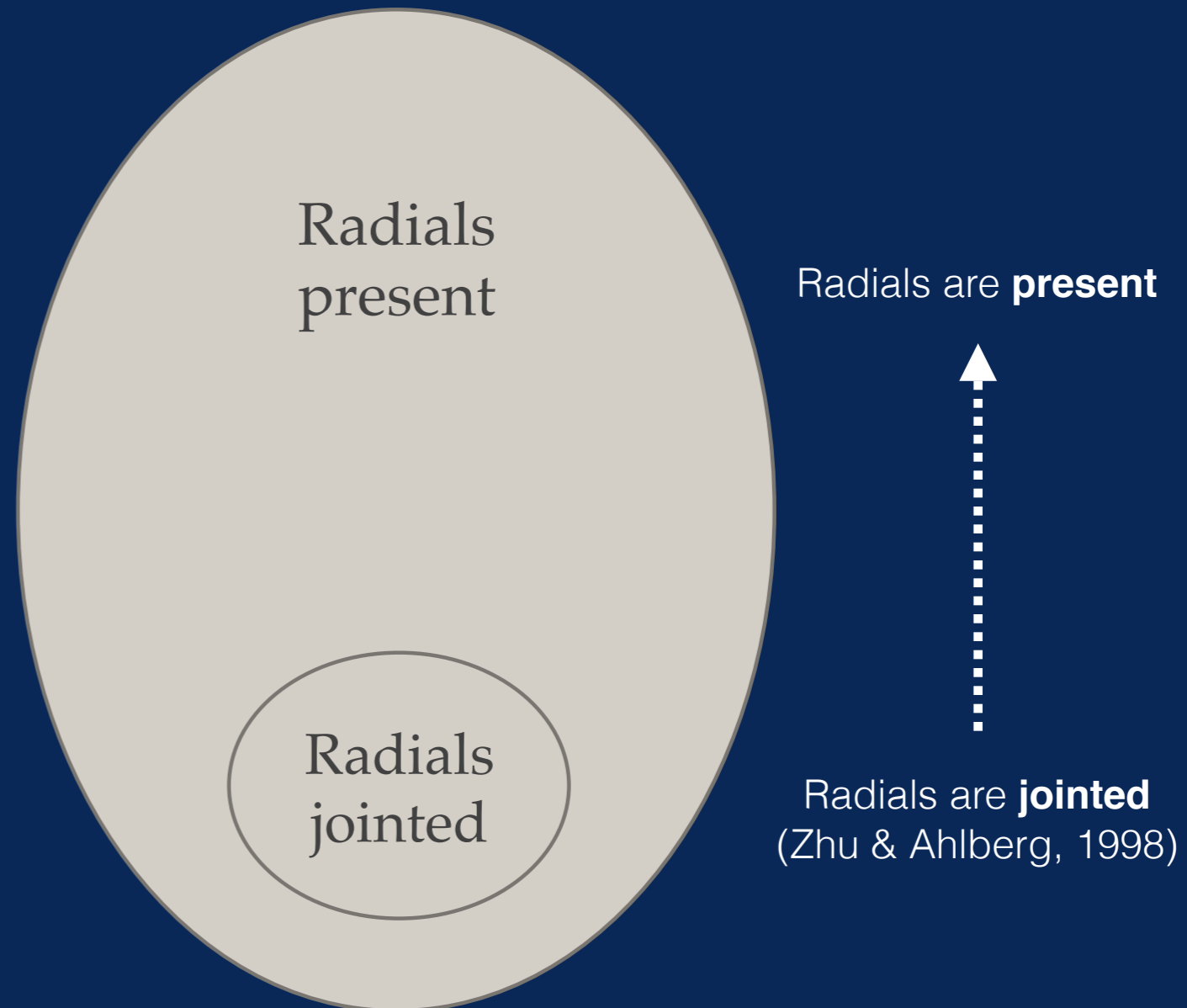
Infer presence, from quality of entity

- If an entity is asserted to have a quality other than absence, it is present
- E.g., If radials are jointed, they are present



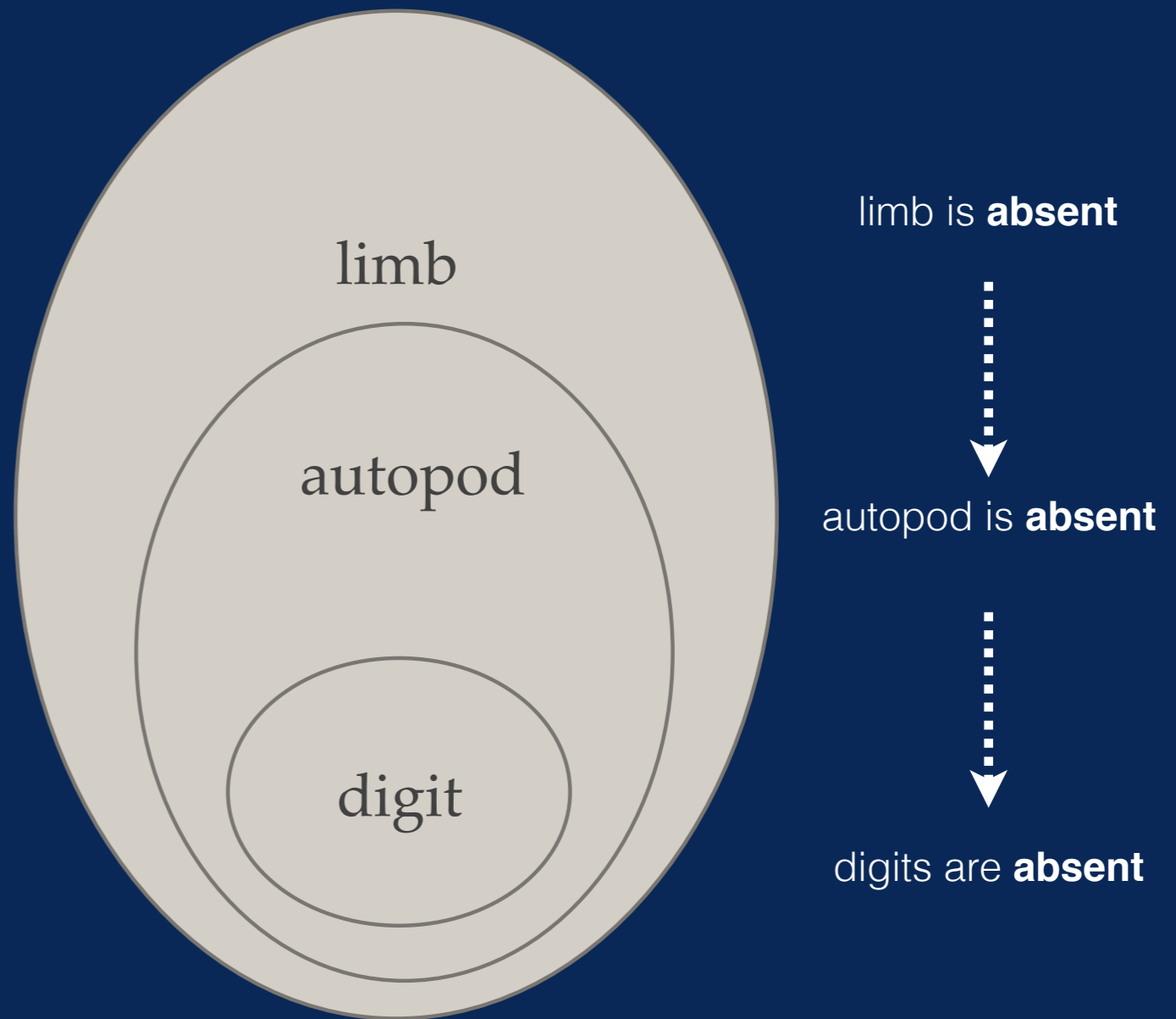
radials jointed

Modified from Shubin et al. 2006



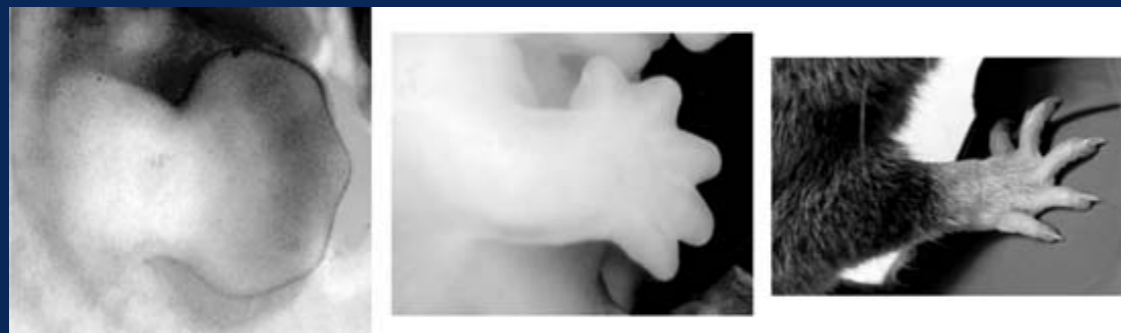
Infer absence, from absence of parent

- “Top down”
- If parent is absent, all children and parts are absent
- If snakes lack limbs, they lack digits

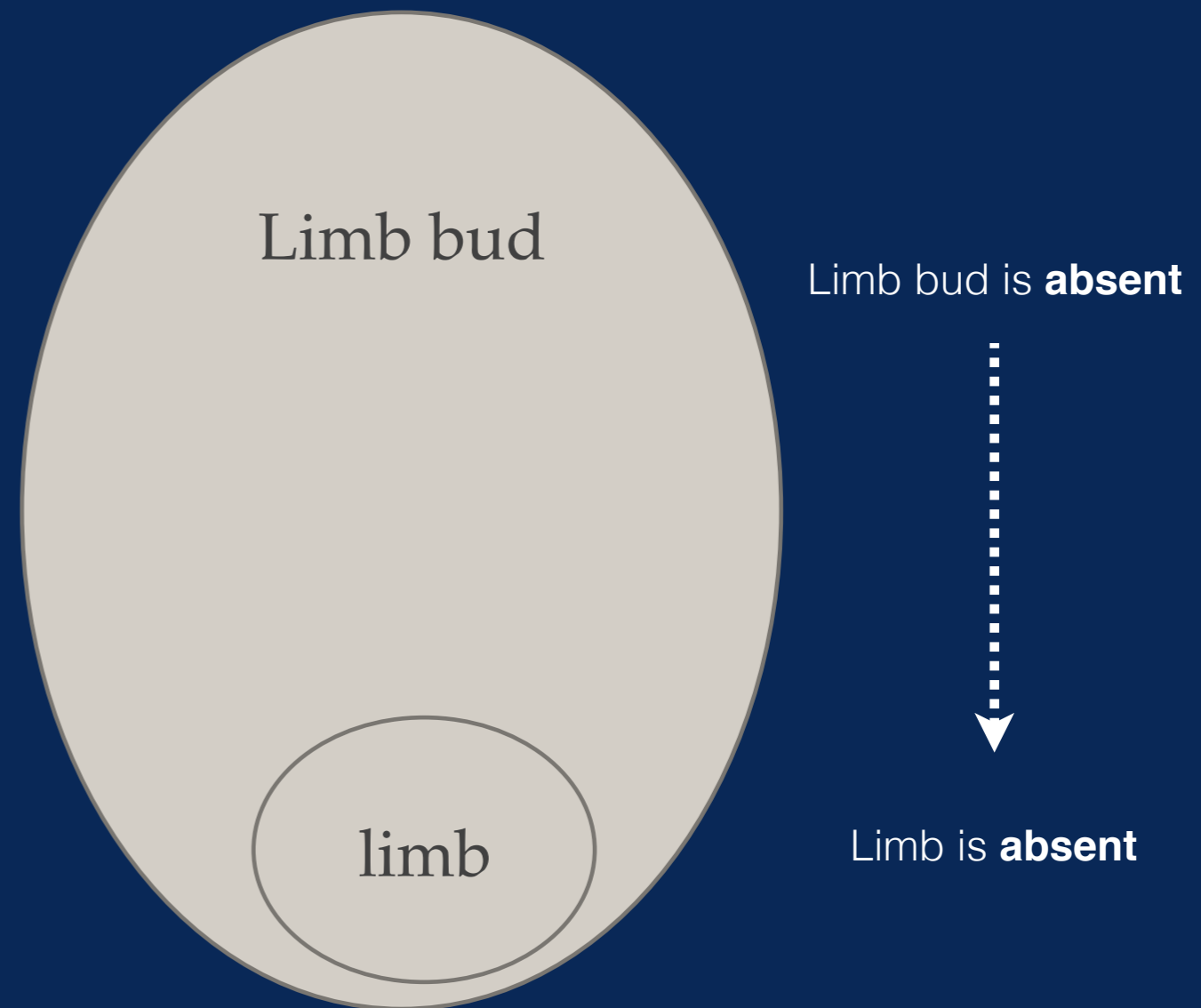


Infer absence, from absence of developmental precursor

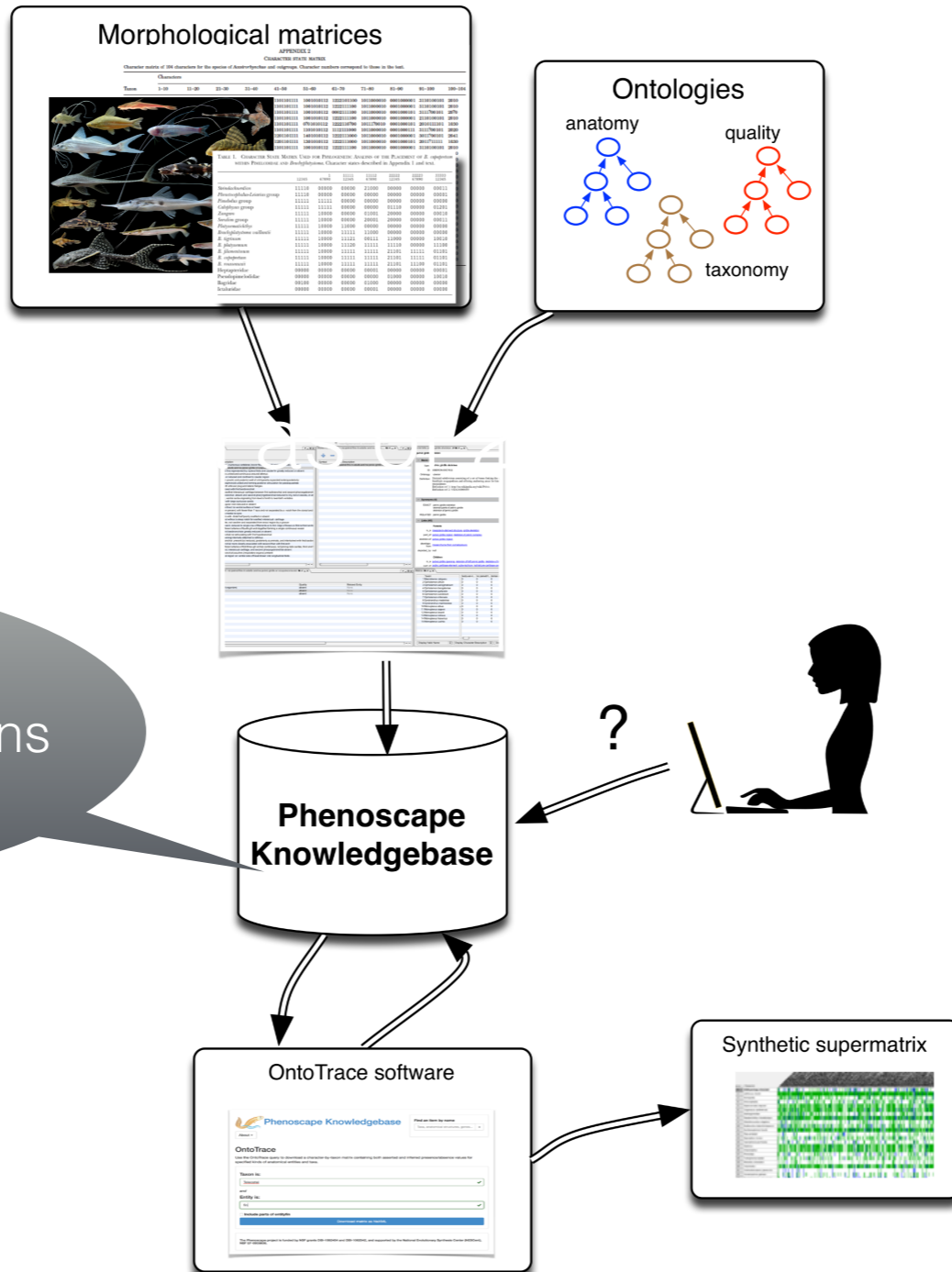
- If developmental precursor is absent, entity is absent
- If species lacks limb buds, they lack limbs



from Sears, 2011



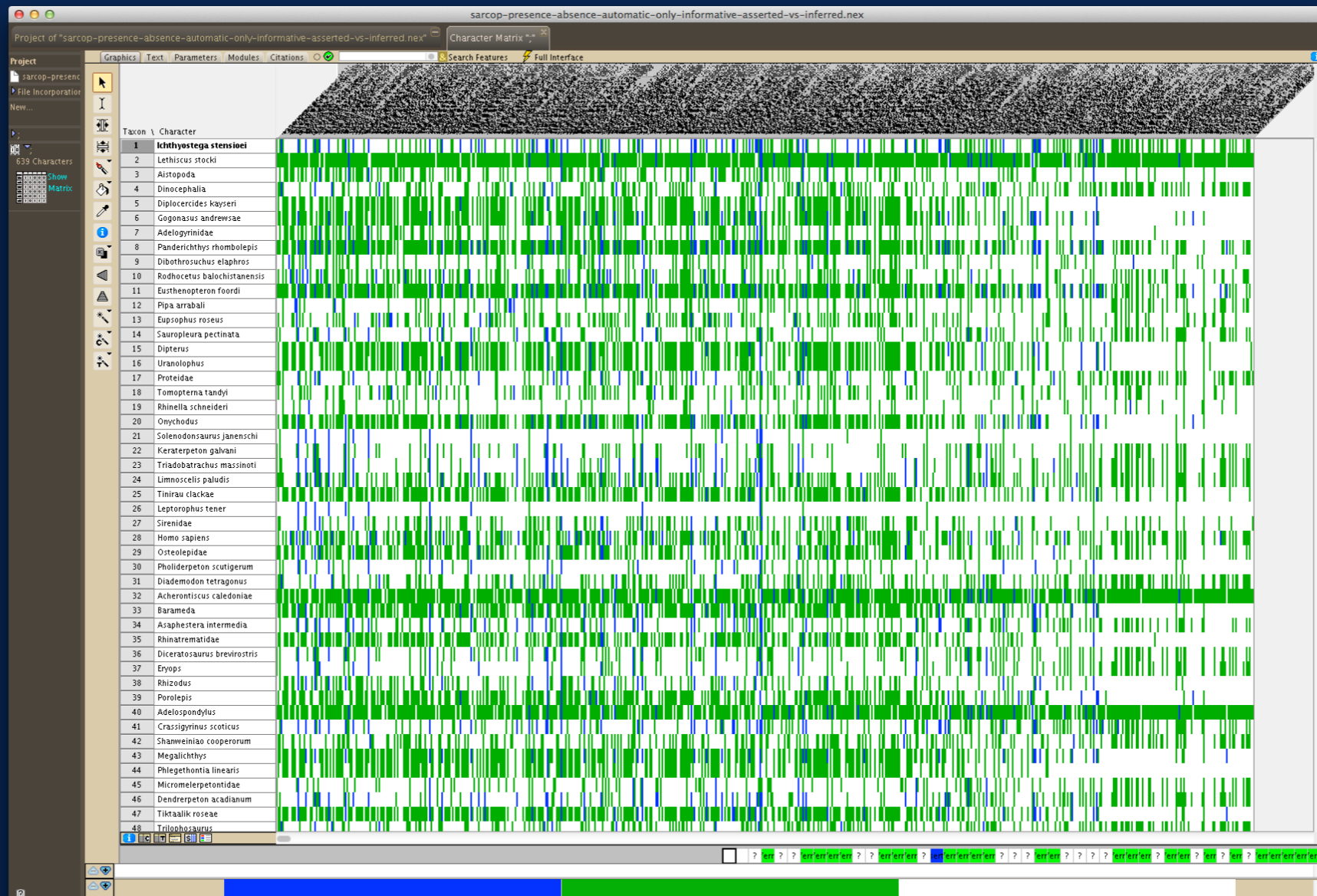
Query: “fin, limb, girdle & parts” “Sarcopterygii”



(Data as of 2018-05-11)

Dececchi, Balhoff, Lapp, & Mabee
(2015) *Systematic Biology* 64(6),
936-952.

'Synthetic morphological supermatrix'

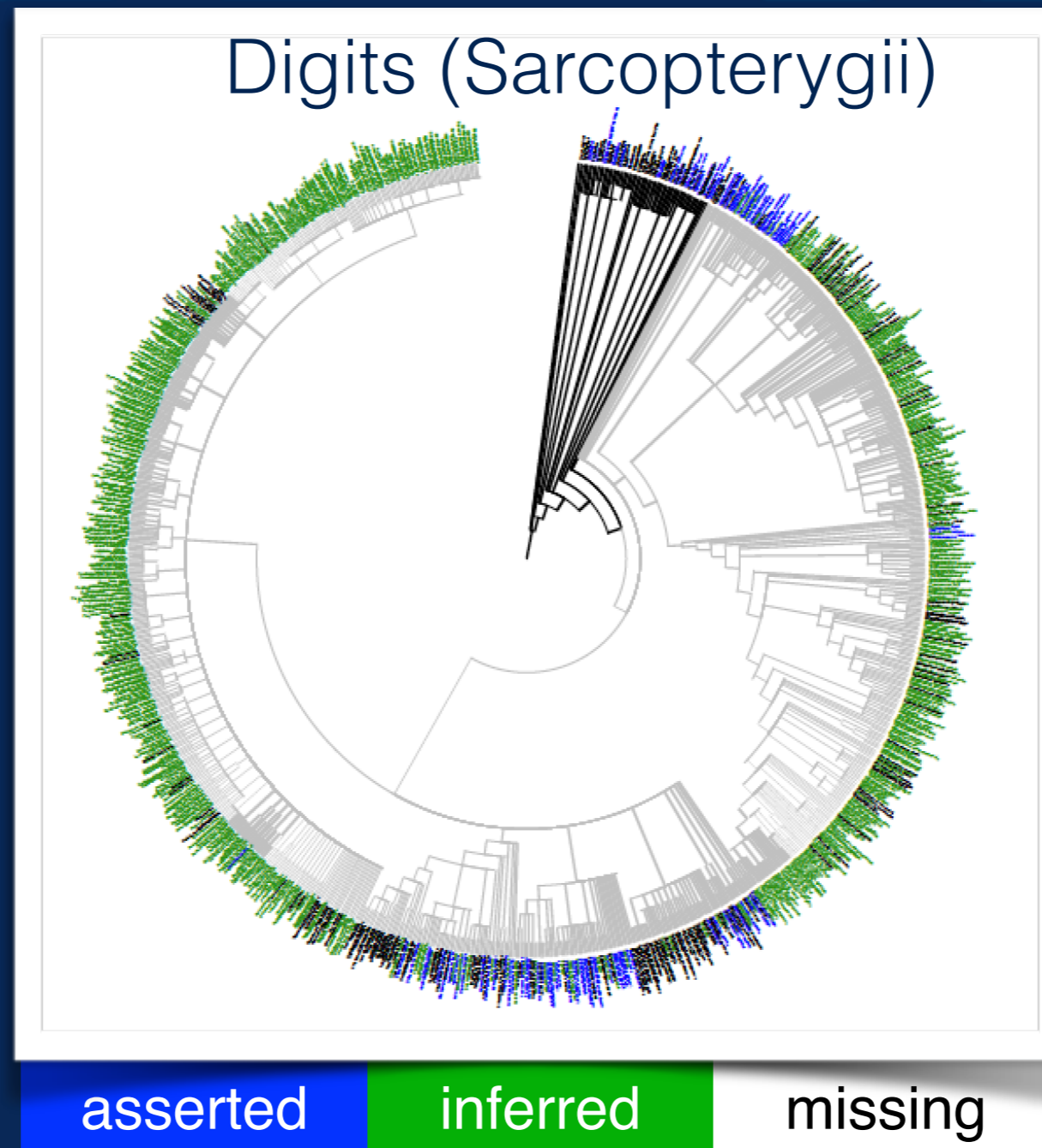


asserted

inferred

missing

Inference provides most of the data for this phenotype



Enabled: Isolation, quantification of data conflicts

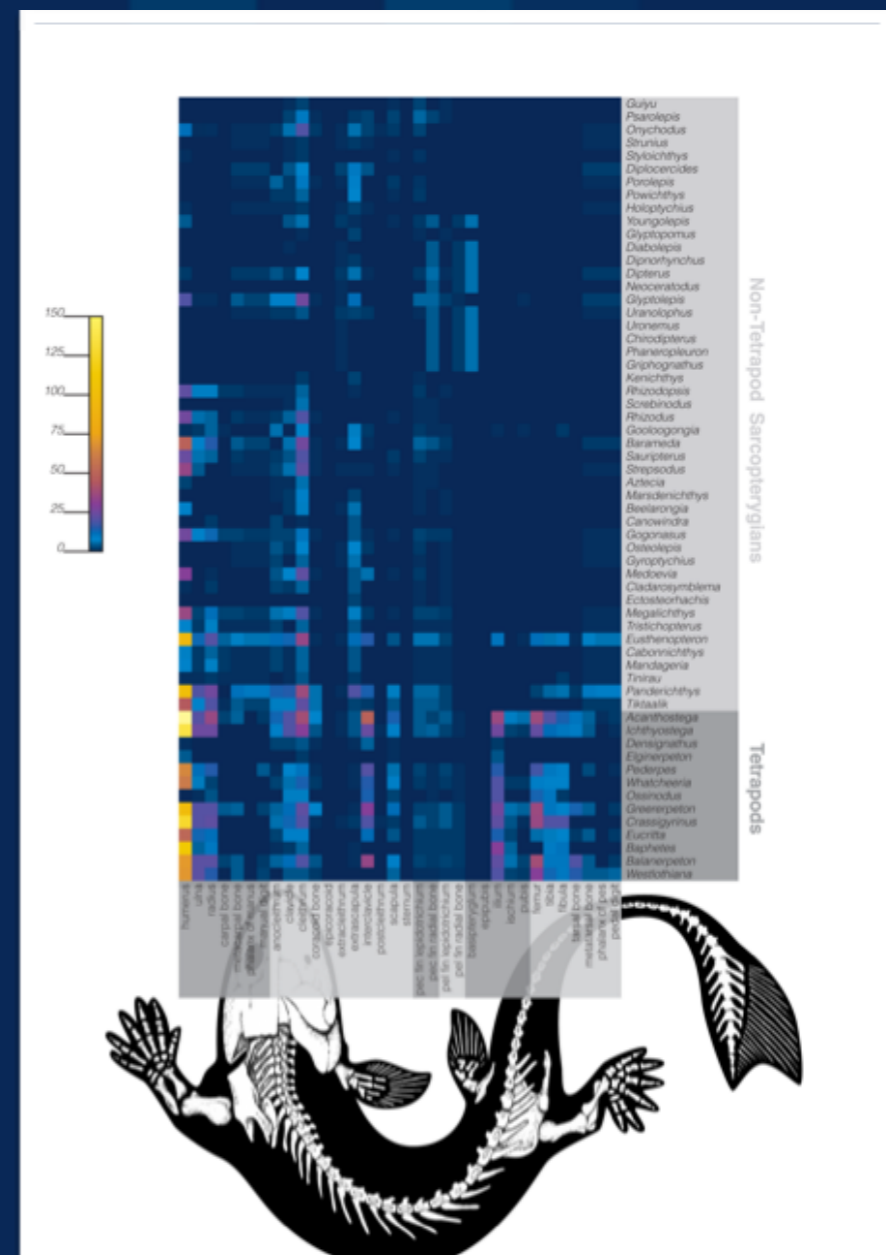
The screenshot shows a window titled "Matrix" containing a data table. The table has 15 columns numbered 1 to 15 and 24 rows of taxon names. The cells contain values 0, 1, or "?". A red arrow points to the cell at row 190, column 4, which contains "0 and 1". The text "presence & absence" is written in white on the red arrow.

Taxon	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
164 Astylosternus di...	?	1	?	?	1	?	?	?	?	?	1	?	?	1	?
165 Ptyonius marshii	?	?	1	?	?	?	?	?	?	?	?	?	?	?	0
166 Gooloogongia	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?
167 Pipidae	0	1	1	?	1	?	?	?	?	?	?	?	?	?	0
168 Phrynomantis	?	1	?	?	1	?	?	?	?	?	?	?	?	1	?
169 Baptonis adven...	?	1	?	?	1	?	?	?	?	?	?	?	?	?	?
170 Boophis madag...	?	1	?	?	1	?	?	?	?	?	1	1	?	0	?
171 Archeria	0	?	1	1	1	?	?	?	?	?	?	?	?	?	1
172 Phonerpeton pri...	?	?	1	?	?	?	?	?	?	?	?	?	?	?	?
173 Osornophryne b...	?	?	?	?	1	?	?	?	?	?	?	1	?	?	?
174 Petropedetes ca...	?	1	?	?	1	?	?	?	?	?	1	1	?	0 and 1	?
175 Tropidosuchus r...	0	1	?	1	1	?	?	?	?	?	?	?	?	?	?
176 Morganucodonti...	?	1	1	0	1	?	?	?	?	?	?	?	?	?	?
177 Hemiphractus	?	1	?	?	1	?	?	?	?	?	?	?	?	1	?
178 Sterropterygion ...	?	?	1	?	?	0	0	0	?	?	0	0	?	?	?
179 Euphlyctis cyan...	?	1	?	?	1	?	?	?	?	?	1	?	?	1	?
180 Afrixalus fulvovit...	?	1	?	?	1	?	1	?	?	?	?	?	?	1	?
181 Amietophrynus ...	?	?	?	?	1	?	?	?	?	?	?	1	?	?	?
182 Archeria crassid...	?	?	1	1	?	?	?	?	?	?	?	?	?	?	1
183 Litargosuchus le...	?	?	?	?	1	?	?	?	0	?	?	?	?	?	?
184 Kotlassia prima	0	?	1	1	?	?	?	?	?	?	?	?	?	?	0
185 Captorhinidae	?	1	1	0	1	?	?	?	?	?	?	?	?	?	1
186 Neldasaurus wri...	?	?	1	?	?	?	?	?	?	?	?	?	?	?	0
187 Melanophrynisc...	?	1	?	?	1	?	?	?	?	?	?	?	?	1	?
188 Archaeotherium ...	?	1	?	?	1	?	1	?	?	?	1	?	?	1	?
189 Proceratophrys ...	?	?	?	?	?	?	?	?	?	?	?	1	?	?	?
190 Acheloma cum...	0	?	1	0 and 1	?	?	?	?	?	?	?	?	?	?	0
191 Pederpes finney...	?	?	1	0 and 1	?	?	?	?	?	?	?	?	?	?	1
192 Rhynchoskos st...	0	?	1	1	?	?	?	?	?	?	?	1	?	?	0
193 Thrinaxodon lior...	?	1	1	0	1	?	?	?	?	?	?	?	?	?	?
194 Lemur catta	?	1	?	?	1	?	1	?	?	0	1	1	?	1	?
195 Anhydrophryne ...	?	1	?	?	1	?	?	?	?	?	1	?	?	1	?
196 Phrynobatrachu...	?	1	?	?	1	?	?	?	?	?	1	1	?	0 and 1	?
197 Hyopsodus paul...	?	1	?	?	1	?	1	?	?	0	1	1	?	1	?
198 Cycloramphus f...	?	1	?	?	1	?	?	?	?	?	1	1	?	0	?
199 Pelobates fuscus	?	1	?	?	1	?	?	?	?	?	1	1	0	1	?
200 Herrerasaurus i...	0	1	?	1	1	?	1	?	0	?	1	1	?	?	?
201 Notharctus tene...	?	1	?	?	1	?	1	?	?	?	1	1	?	1	?
202 Allobates talam...	?	1	?	?	1	?	?	?	?	?	1	1	?	0	?
203 Apheliscus insid...	?	?	?	?	?	?	?	?	?	0	?	?	?	?	?
204 Manis pentadact...	?	1	?	?	1	?	1	?	?	1	1	1	?	1	?

774 conflicted cells (0.5%) of 146,451 populated cells

Enabled: Phenotype enrichment

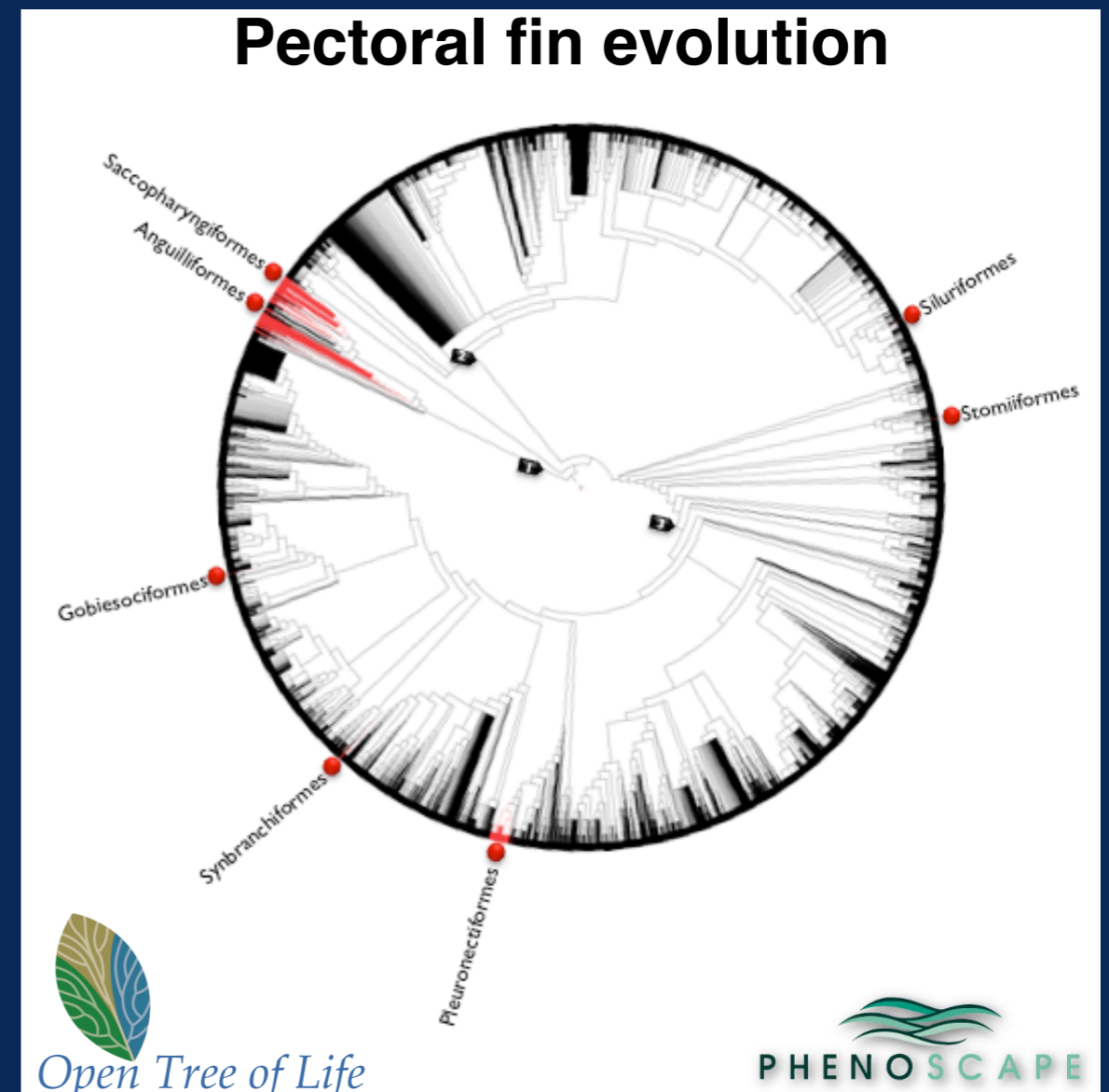
- Heat map:
 - Cell color reflects number of character states:
 - for each anatomical entity (column)
 - for each taxon (row)
 - Dark blue = empty cells



Sarcopterygii, fins, limbs, girdles

Enabled: Large-scale ancestral state reconstruction

Enhancement to sparse data



Potential: scale up phenotypic data to drive genomic discoveries

Cell Reports
Report

Cell
PRESS
Open
ACCESS

A “Forward Genomics” Approach Links Genotype to Phenotype using Independent Phenotypic Losses among Related Species

Michael Hiller,^{1,5,*} Bruce T. Schaar,¹ Vahan B. Indjeian,^{1,3} David M. Kingsley,^{1,3} Lee R. Hagey,⁴ and Gill Bejerano^{1,2,*}

¹Department of Developmental Biology

²Department of

Stanford Univ

³Howard Hugl

⁴Department of

⁵Present addr

01307 Dresde

*Corresponde

http://dx.doi.

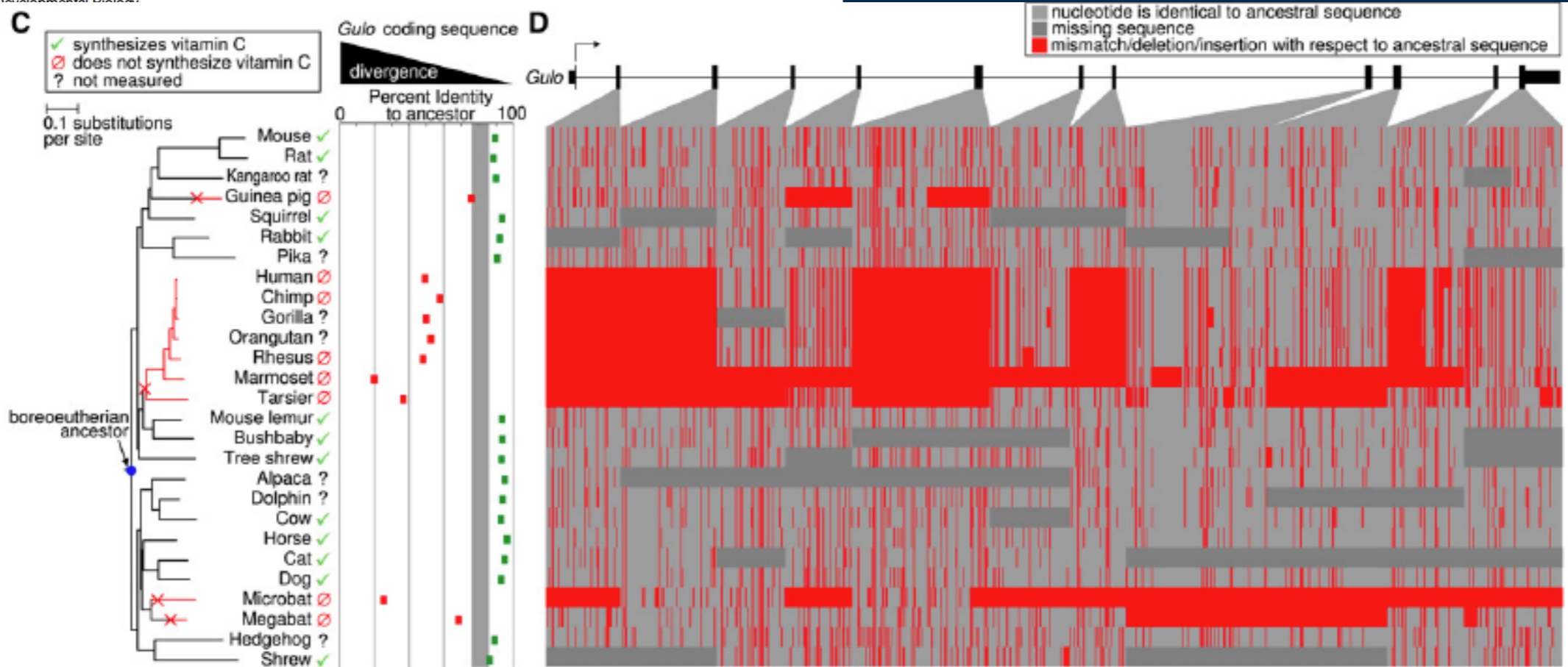


Figure 2. A Forward Genomics Screen to Match an Ancestral Presence/Absence Trait Pinpoints *Gulo* Inactivation in Vitamin C-Non-synthesizing Species



https://canadianmuseumofnature.files.wordpress.com/2014/10/cmm_clct_276.jpg?w=640



Geography,
Environment

The Intermediary

3 min

Worried humans reached out to the Machine for help in predicting climate change. Cleverly connecting phenotype with collection data, It told the humans which species would be likely to win the competition.

PHENOTYPED

■ Help me...

- * Related taxa with similar phenotypes from dissimilar environments
- * Unrelated taxa with similar phenotypes from similar environments
- * Understand the direction of phenotypic change in a particular species over time
- * Predict the likely changes in phenotype and species' interactions based on collections data

Reasoning across phenotype & environment

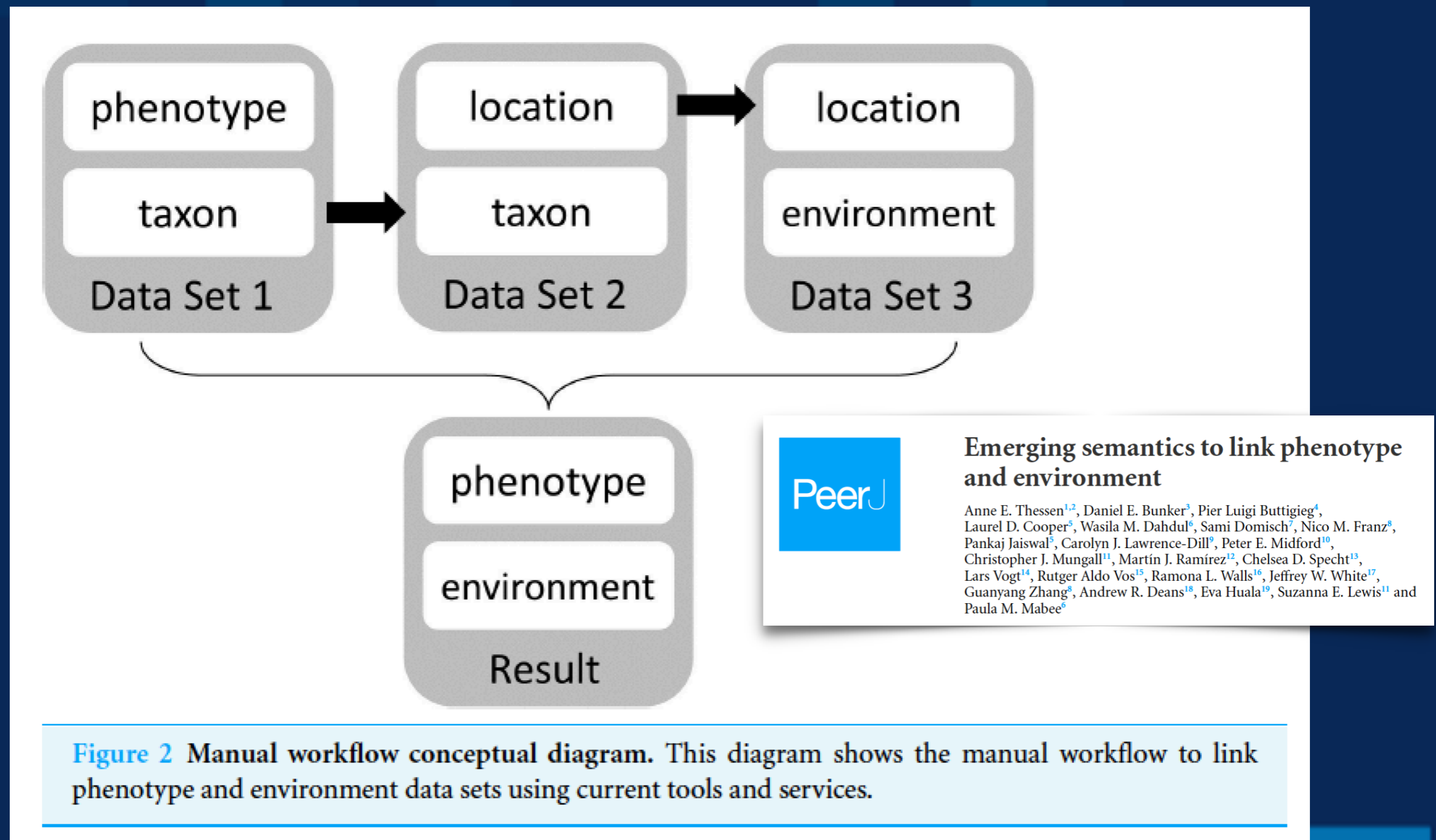
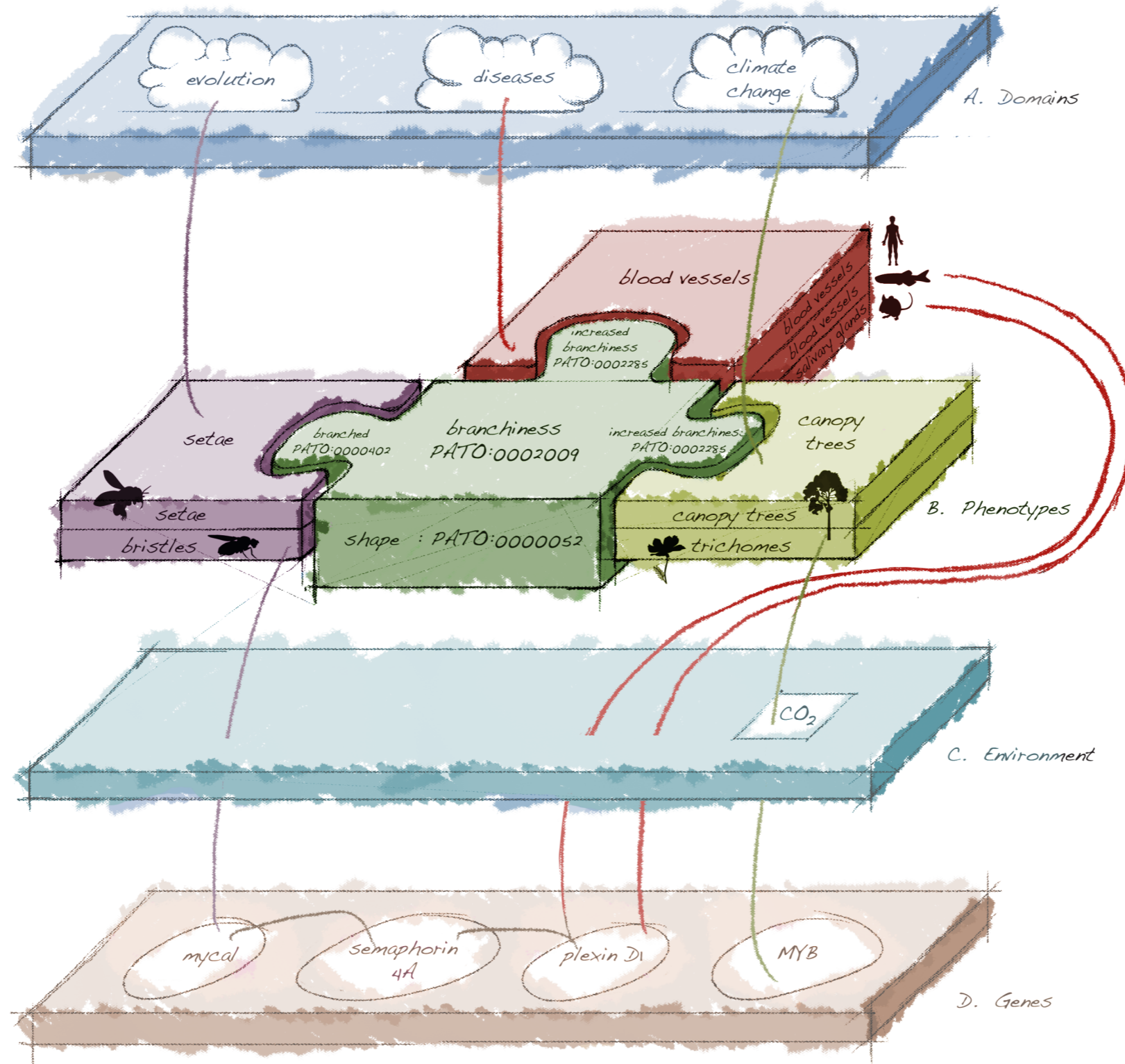


Figure 2 Manual workflow conceptual diagram. This diagram shows the manual workflow to link phenotype and environment data sets using current tools and services.



2015 Deans et al. *PLOS Biology*. January 5, 2015. doi: 10.1371/journal.pbio.1002033

Phenotypes a portal to understanding the integration between organism and environment

Acknowledgements

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- NSF Phenotype Research Coordination Network (phenotypercn.org), DBI-0956049
- Phenoscope contributors, Advisory Board, Data sources (see: <http://phenoscope.org/wiki/Acknowledgments>)
- Phenoscope collaborators

