Towards ubiquitous OWL computing: Simplifying programmatic authoring of and querying with OWL axioms

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Did all the work:

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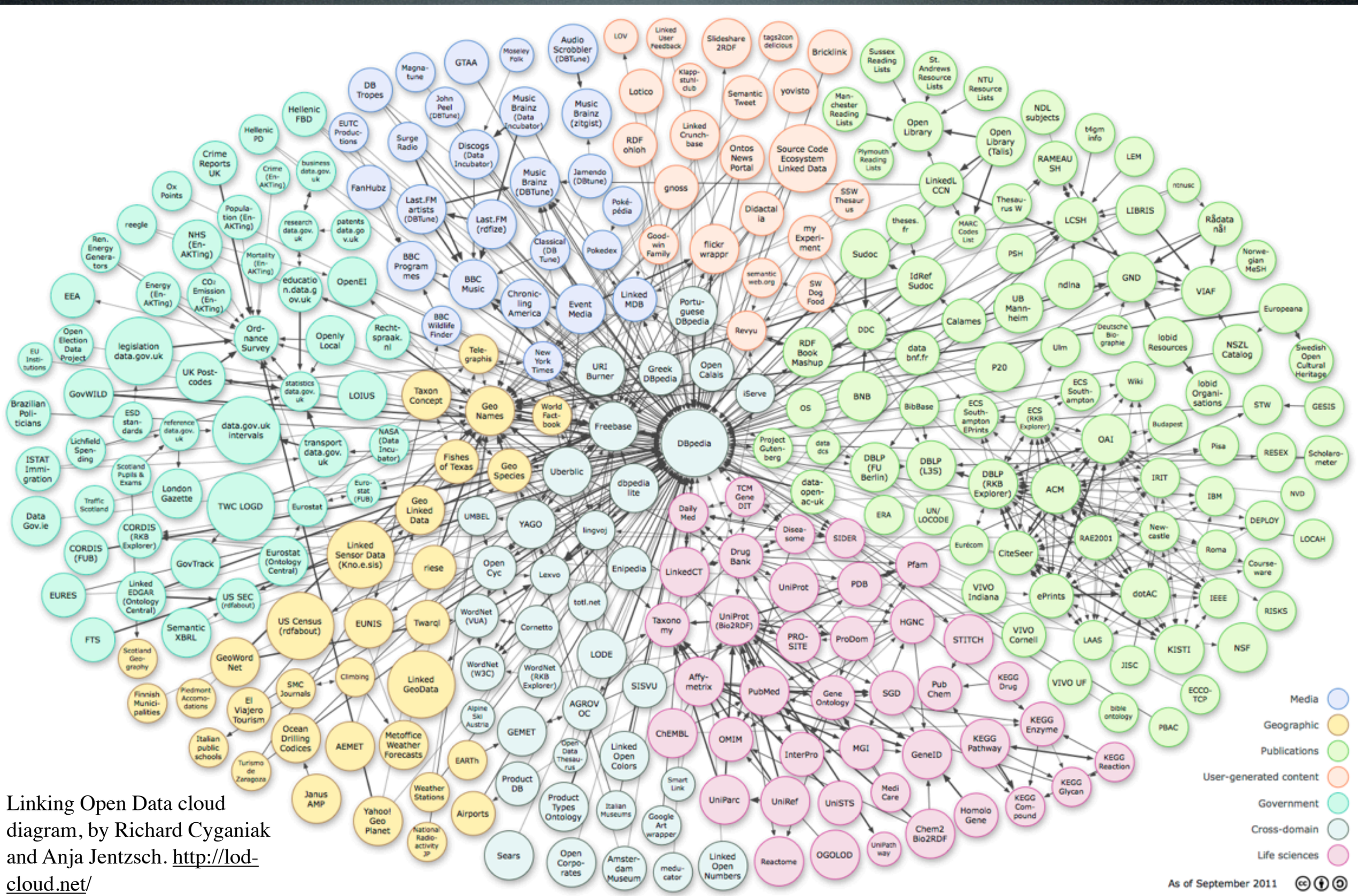
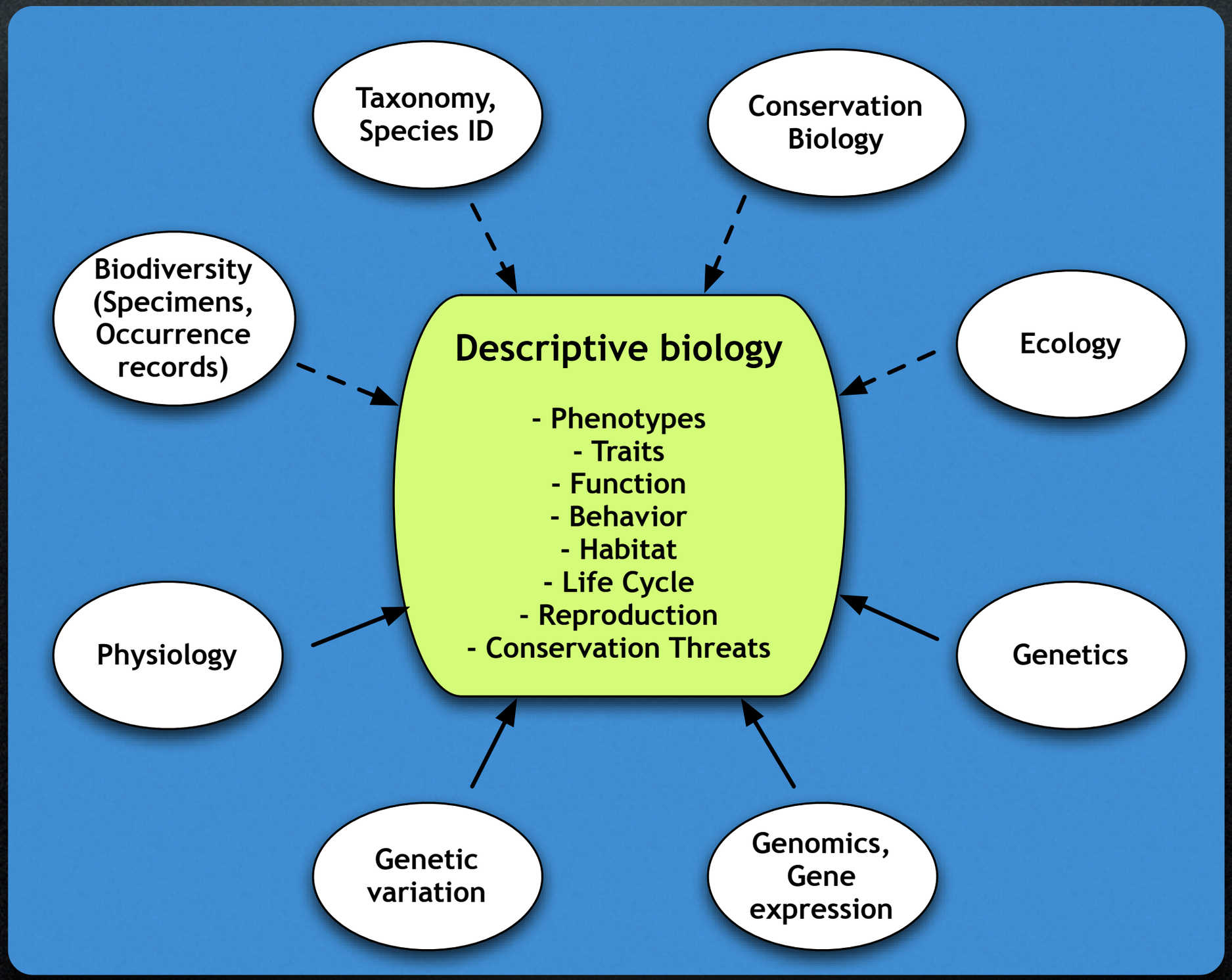
http://github.com/balhoff

RDF is a powerful data integrator



Linking Open Data cloud diagram, by Richard Cyganiak and Anja Jentzsch. http://lod-cloud.net/

Ontologies allow integrating across descriptive biology



Translational bioinformatics

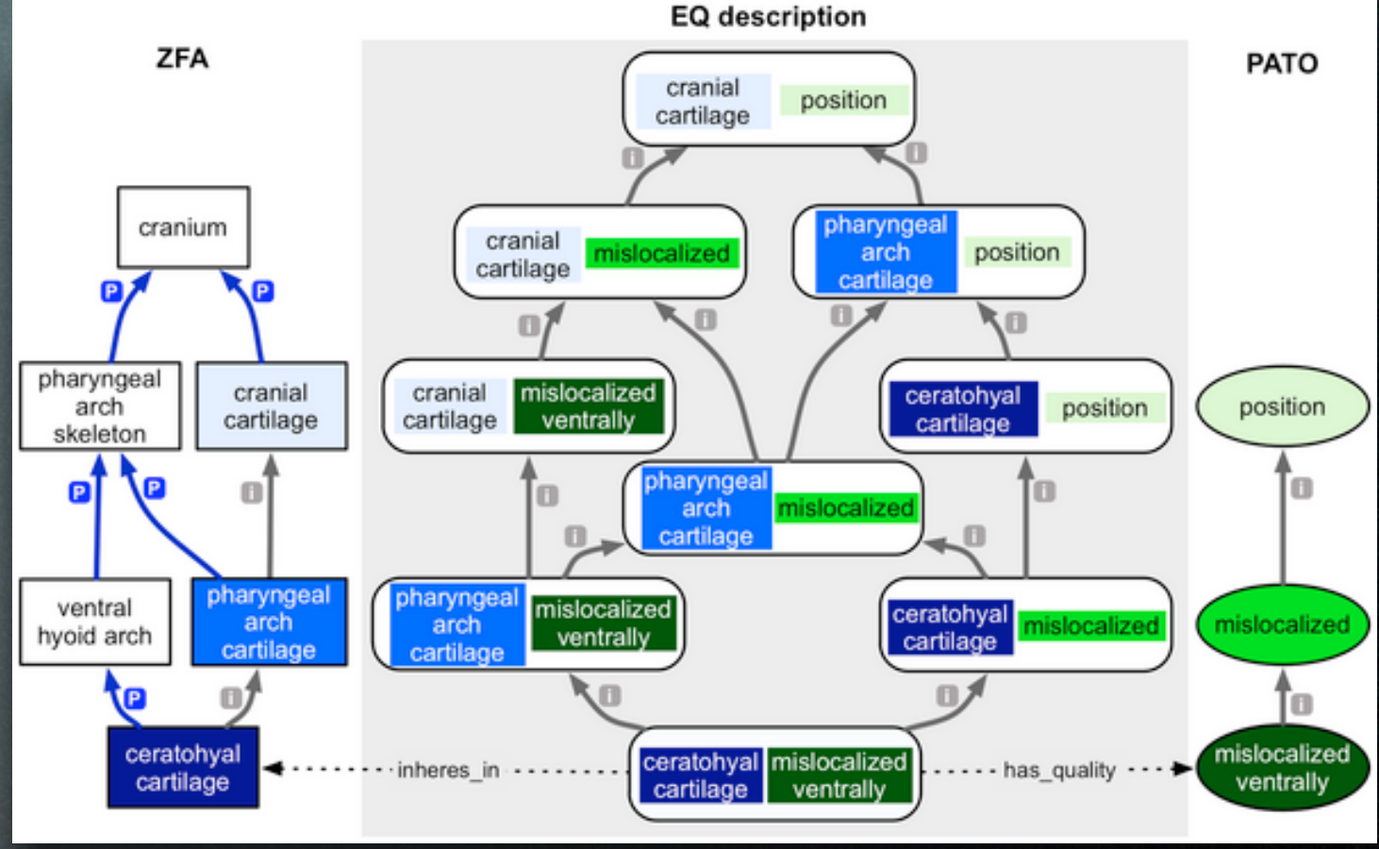


Fig. 3, Washington et al (2009)

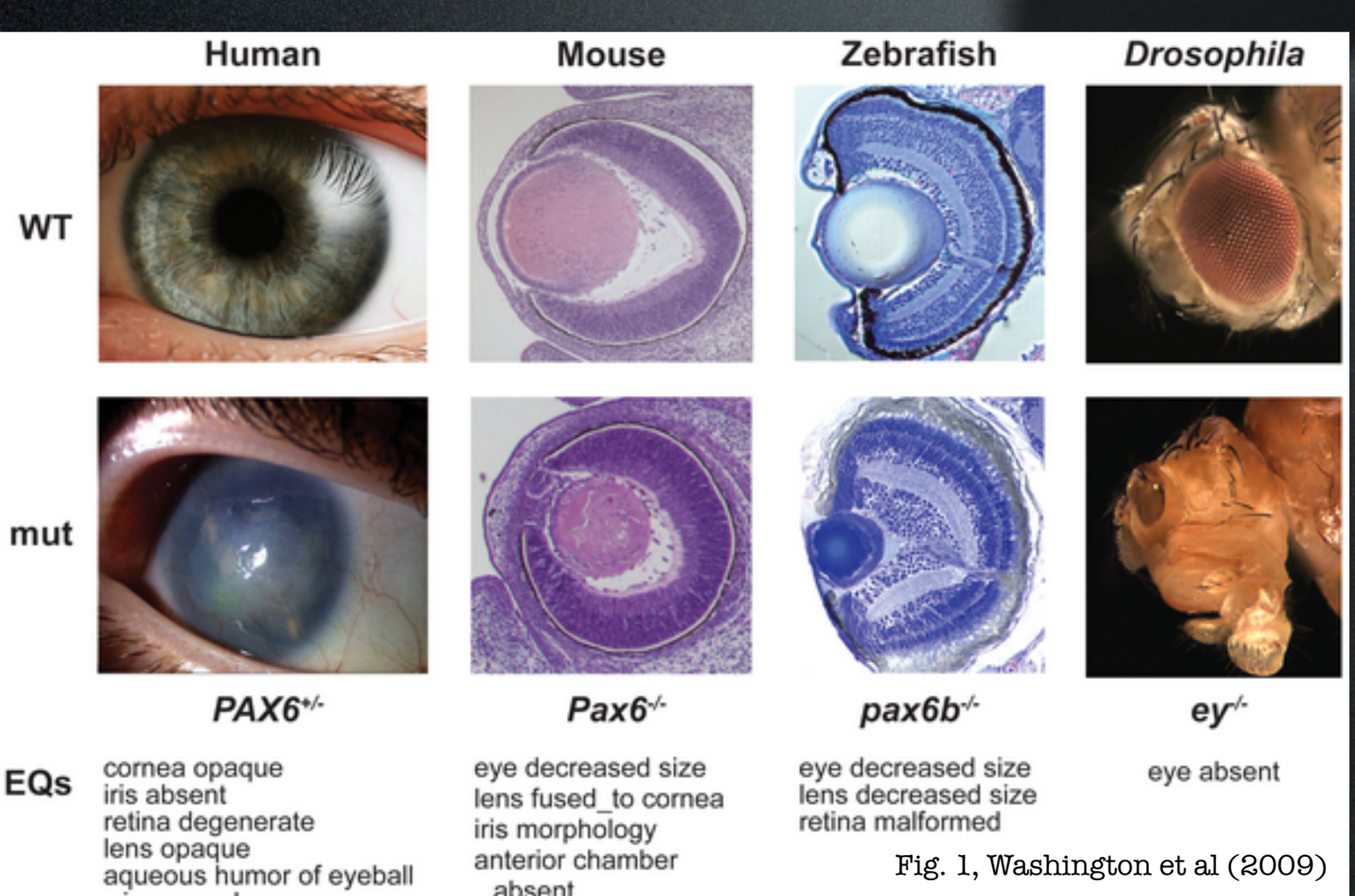


Fig. 1, Washington et al (2009)

Model organism -> Human

Translational biodiversity informatics

PHENOSCAPE

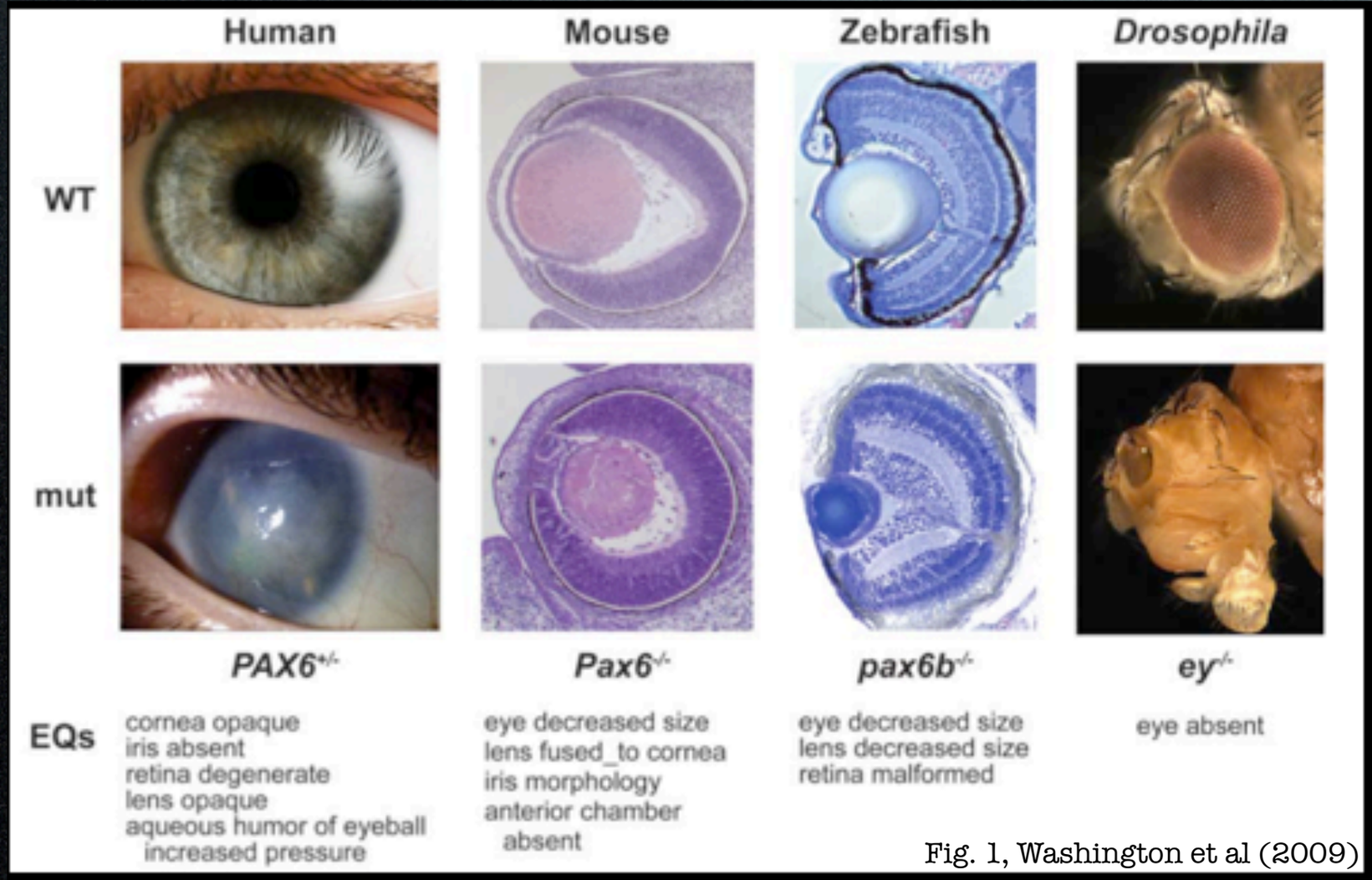
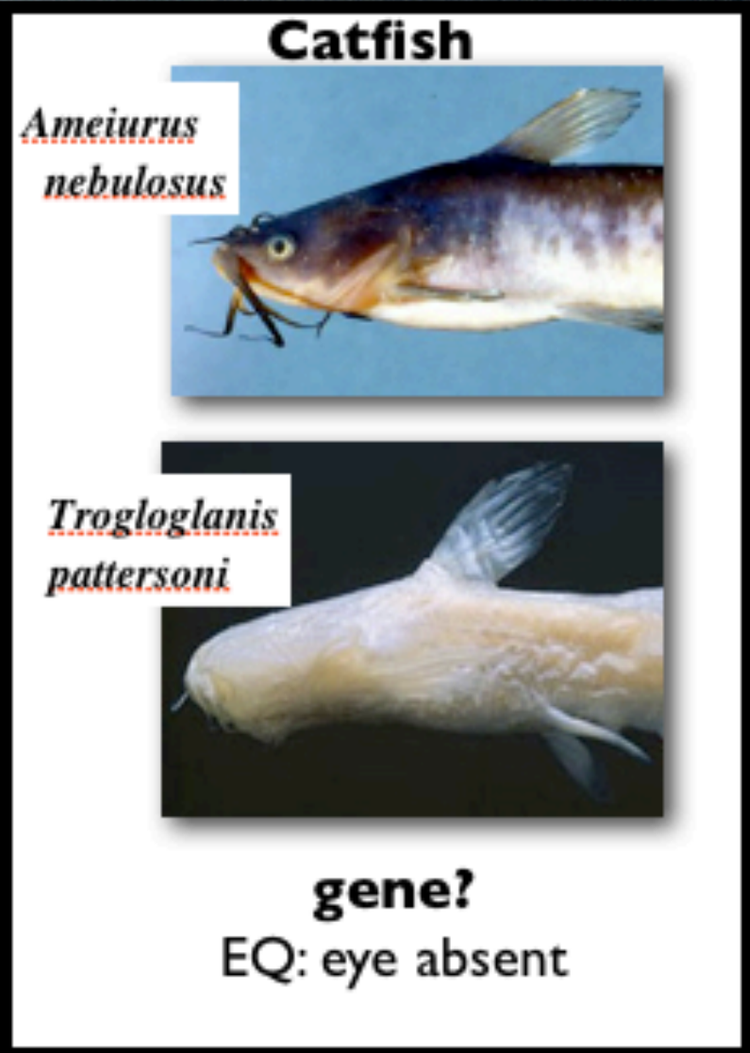


Fig. 1, Washington et al (2009)



Model organism genes -> Evolutionary diversity by semantic similarity of phenotypes

Integrate across studies & fields by virtue of ontologies

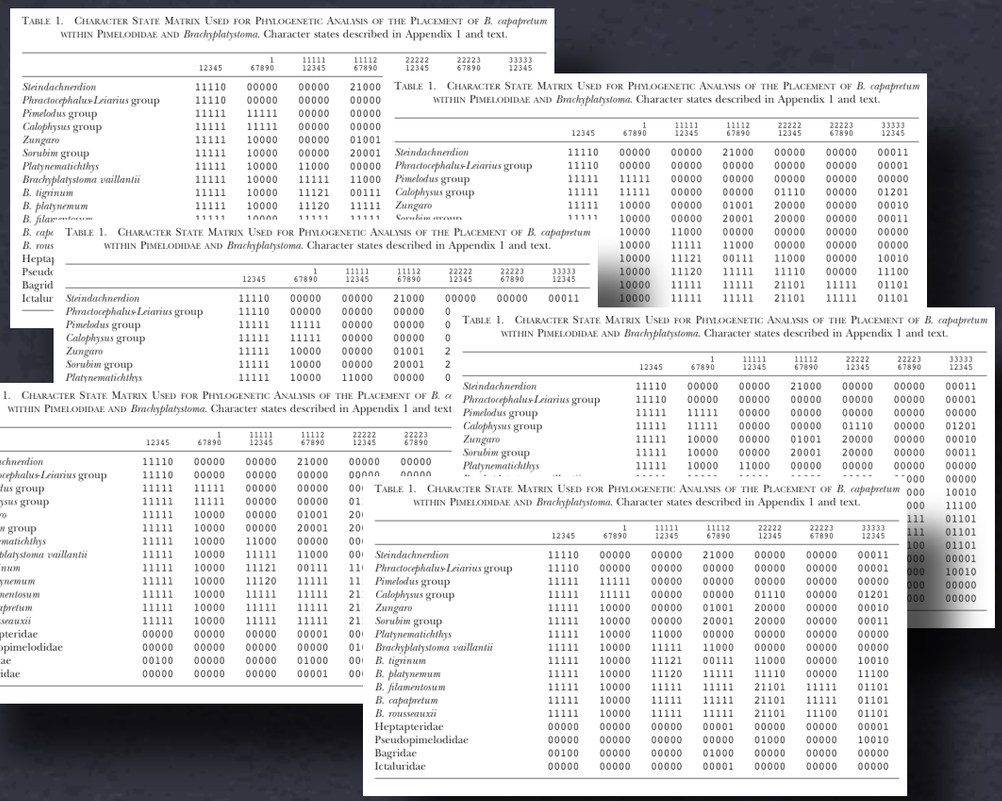
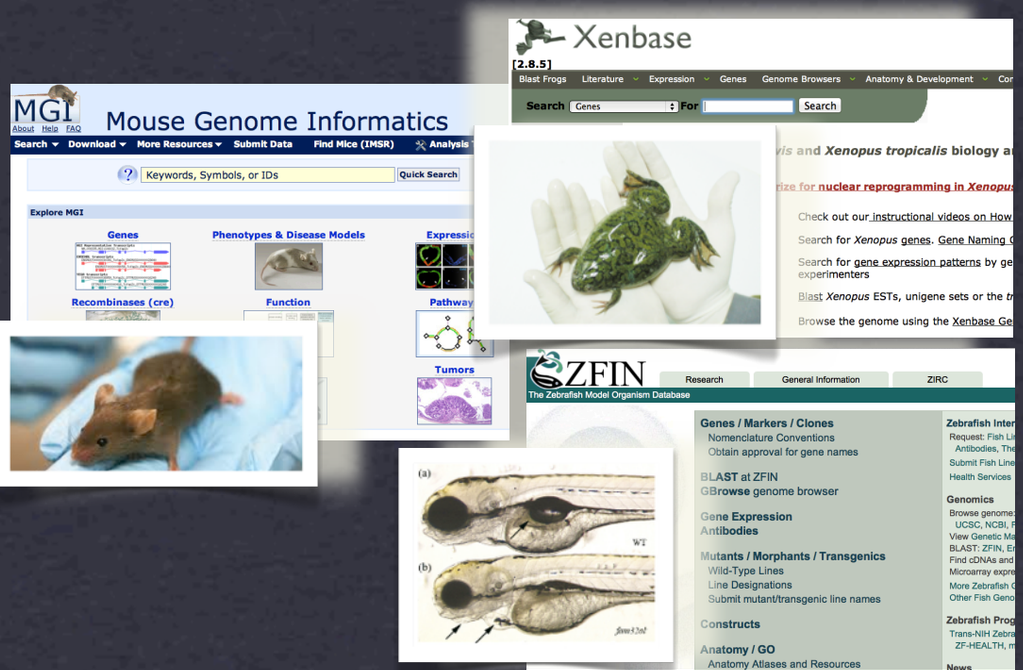


TABLE 1. CHARACTER STATE MATRIX USED FOR PHYLOGENETIC ANALYSIS OF THE PLACEMENT OF *B. capapretum* WITHIN PIMELODIDAE AND *Brachyplatystoma*. Character states described in Appendix 1 and text.

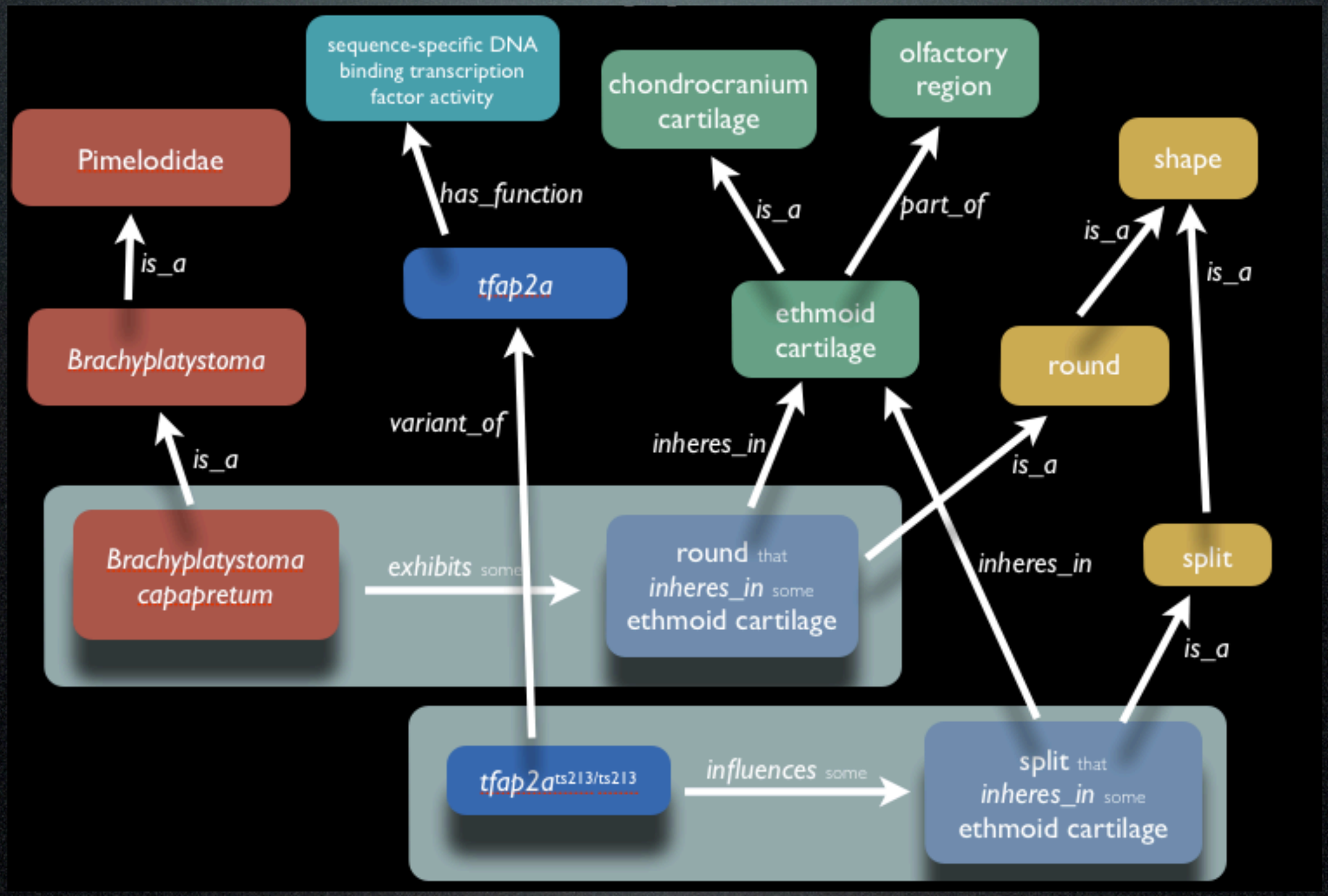
	12345	1 67890	11111 12345	11112 67890	22222 12345	22223 67890	33333 12345
Steindachnerdion	11110	00000	00000	21000	00000	00000	00011
Phractocephalus-Leiarius group	11110	00000	00000	00000	00000	00000	00001
Pimelodus group	11111	11111	00000	00000	00000	00000	00000
Calophysus group	11111	11111	00000	00000	01110	00000	01201
Zungaro	11111	10000	00000	01001	20000	00000	00010
Sorubim group	11111	10000	00000	20001	20000	00000	00011
Platynematichthys	11111	10000	11000	00000	00000	00000	00000
Brachyplatystoma vaillantii	11111	10000	11111	11000	00000	00000	00000
B. tigrinum	11111	10000	11121	00111	11000	00000	10010
B. platynemum	11111	10000	11120	11111	11110	00000	11100
B. filamentosum	11111	10000	11111	11111	21101	11111	01101
B. capapretum	11111	10000	11111	11111	21101	11111	01101
B. rousseauxii	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

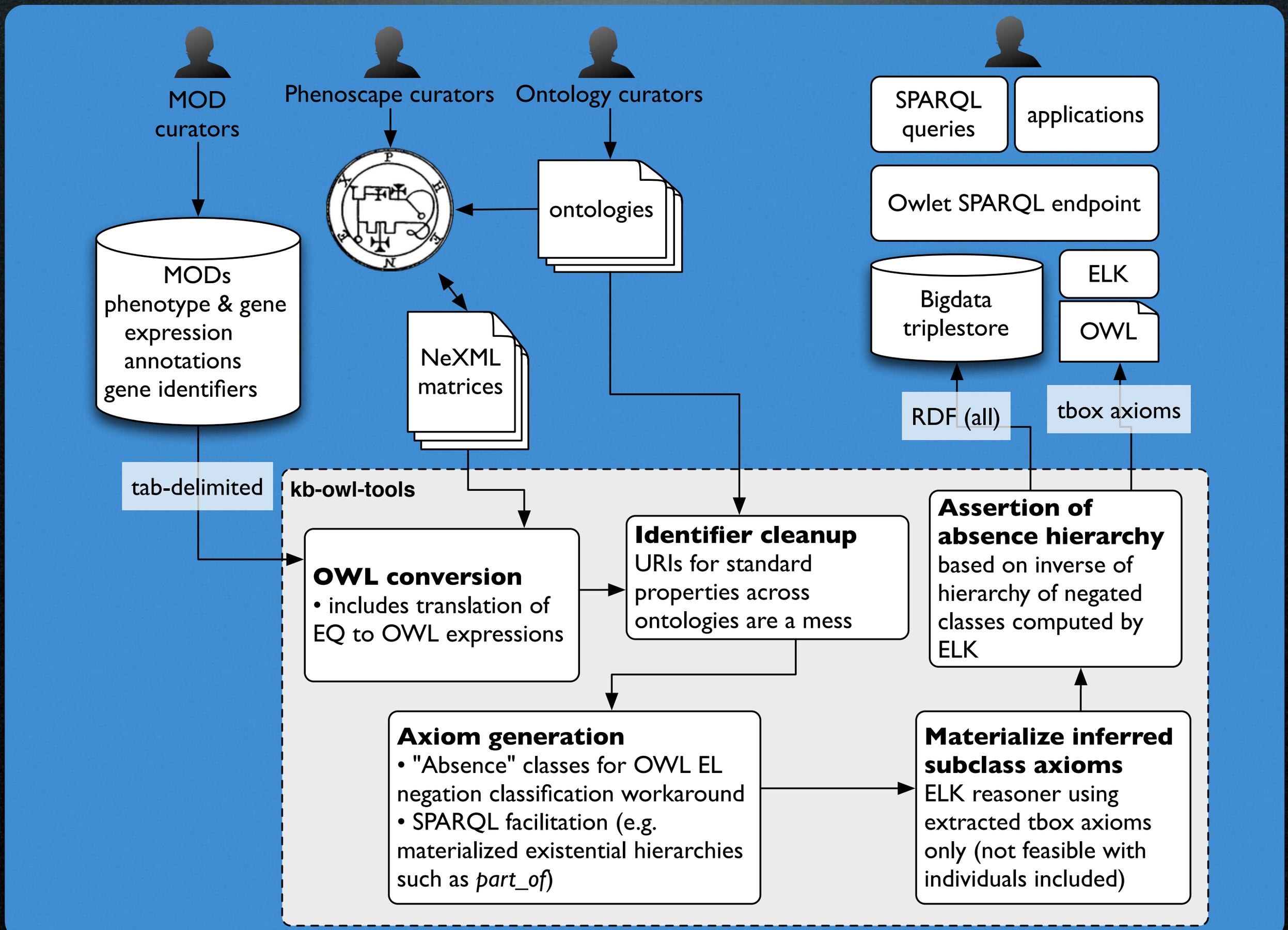


Comparative studies + Model organism datasets

= Phenoscape Knowledgebase

Computable via shared ontologies, rich semantics, OWL reasoning




MOD curators
Phenoscape curators
Ontology curators
SPARQL queries
applications
Owlet SPARQL endpoint
ontologies
MODs phenotype & gene expression annotations gene identifiers
NeXML matrices
Bigdata triplestore
ELK
OWL
RDF (all)
tbox axioms
tab-delimited
kb-owl-tools
OWL conversion
• includes translation of EQ to OWL expressions
Identifier cleanup
URIs for standard properties across ontologies are a mess
Assertion of absence hierarchy
based on inverse of hierarchy of negated classes computed by ELK
Axiom generation
• "Absence" classes for OWL EL negation classification workaround
• SPARQL facilitation (e.g. materialized existential hierarchies such as part_of)
Materialize inferred subclass axioms
ELK reasoner using extracted tbox axioms only (not feasible with individuals included)

Ontology axiom authoring at scale is cumbersome

- If a developmental precursor is absent, the structure is absent.

"For every anatomical structure, assert that the class of things not having the structure as a part is a subclass of the class of things not having the structure's developmental precursor structure as a part."

(See Balhoff et al, Phenotype Day, http://phenoday2014.bio-lark.org/pdf/11.pdf)

Scowl: marrying programming to ontology language

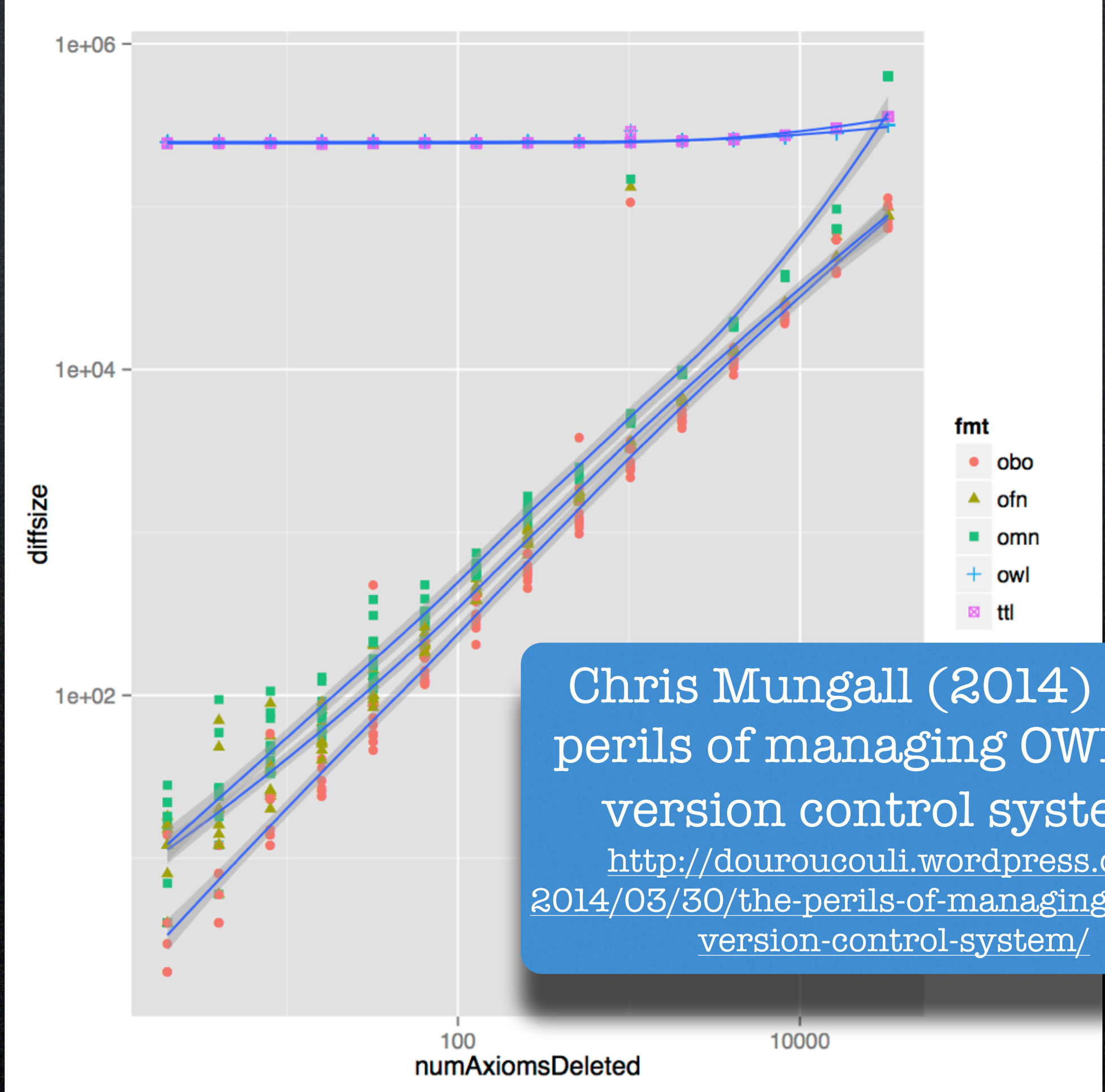
"For every anatomical structure, assert that the class of things not having the structure as part is a subclass of the class of things not having the structure's developmental precursor as part."

```
for {
    term <- reasoner.getSubclasses(anatomical_structure)
} yield
(not (hasPart some term)) SubClassOf (not (hasPart some (developsFrom
some term)))
```

Scowl

- Allows declarative approach to composing OWL expressions and axioms using the OWL API.
- Implemented as a library in Scala
- Exploits ‘`implicit class`’ construct in Scala
  - Compiler turns declarative expressions into JVM instructions
- Class, Annotation, Property axioms

```
val hasFather = ObjectProperty("http://example.org/hasFather")
val hasBrother = ObjectProperty("http://example.org/hasBrother")
val hasUncle = ObjectProperty("http://example.org/hasUncle")
val axiom = hasUncle SubPropertyChain (hasFather o hasBrother)
```



Chris Mungall (2014) "The perils of managing OWL in a version control system"

http://douroucouli.wordpress.com/2014/03/30/the-perils-of-managing-owl-in-a-version-control-system/

```
object AnatomyOntology extends App {

  val factory = OWLManager.getOWLDataFactory
  val ns = "http://example.org/anatomy.owl#"
  val head = Class(ns + "001")
  val body = Class(ns + "002")
  val hand = Class(ns + "003")
  val arm = Class(ns + "004")
  val anatomical_structure = Class(ns + "005")
  val part_of = ObjectProperty(ns + "006")
  val label = factory.getRDFSLabel

  val ontology = Ontology("http://example.org/anatomy.owl", Set(
    head Annotation (label, "head"),
    head SubClassOf anatomical_structure,
    head SubClassOf (part_of some body),
    head SubClassOf (not(part_of some arm)),

    body Annotation (label, "body"),
    body SubClassOf anatomical_structure,

    arm Annotation (label, "arm"),
    arm SubClassOf anatomical_structure,
    arm SubClassOf (part_of some body),

    hand Annotation (label, "hand"),
    hand SubClassOf anatomical_structure,
    hand SubClassOf (part_of some arm)))

  ontology.getOWLOntologyManager.saveOntology(
                                ontology,
                                IRI.create(new File(args(0))))
}
```

Similarly motivated effort: Tawny-OWL

- By Phil Lord
- Allows construction of OWL ontologies in Clojure.
- “the ontology engineering equivalent of R”
- https://github.com/phillord/tawny-owl

Scowl

- Home:
  http://github.com/phenoscape/scowl
- MIT-licensed

Ontology-driven querying in SPARQL is not pretty

SPARQL: genes expressed in head muscles

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX ao: <http://purl.obolibrary.org/obo/my-anatomy-ontology/>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
SELECT DISTINCT ?gene
WHERE
{
?gene ao:expressed_in ?structure .
?structure rdf:type ?structure_class .
# Triple pattern selecting structure:
?structure_class rdfs:subClassOf "ao:muscle” .
?structure_class rdfs:subClassOf ?restriction
?restriction owl:onProperty ao:part_of .
?restriction owl:someValuesFrom "ao:head" .
}
```

- Awkward, lengthy, complicated
- Error-prone
- Slow

Owlet: bridging from SPARQL to DL queries

- Want to allow arbitrary selection of structures of interest, using rich semantics:

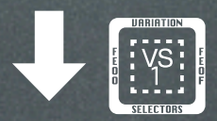
  (part_of some (limb/fin or girdle skeleton))
  or (connected_to some girdle skeleton)

- RDF triplestores provide very limited reasoning expressivity, and scale poorly with large ontologies.
- However, ELK can answer class expression queries within seconds.

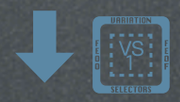
owlet: A little OWL in SPARQL

- **owlet** interprets OWL class expressions embedded within SPARQL queries
- Uses any OWL API-based reasoner to preprocess query.
  - We use ELK that holds terminology in memory.
- Replaces OWL expression with FILTER statement listing matching terms

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX ao: <http://purl.obolibrary.org/obo/my-anatomy-ontology/>
PREFIX ow: <http://purl.org/phenoscape/owlet/syntax#>
SELECT DISTINCT ?gene
WHERE
{
?gene ao:expressed_in ?structure .
?structure rdf:type ?structure_class .
# Triple pattern containing an OWL expression:
?structure_class rdfs:subClassOf "ao:muscle and (ao:part_of some ao:head)"^^ow:omn .
}
```



owlet




```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX ao: <http://purl.obolibrary.org/obo/my-anatomy-ontology/>
PREFIX ow: <http://purl.org/phenoscape/owlet/syntax#>
SELECT DISTINCT ?gene
WHERE
{
?gene ao:expressed_in ?structure .
?structure rdf:type ?structure_class .
# Filter constraining ?structure_class to the terms returned by the OWL query:
FILTER(?structure_class IN (ao:adductor_mandibulae, ao:constrictor_dorsalis, ...))
}
```

owlet: A little OWL in SPARQL

- Implemented in Scala, can be used in Java
- Home: http://github.com/phenoscape/owlet
- MIT-licensed
- Unique to owlet, compared to some related efforts (e.g., SPARQL-DL, Terp in Pellet):
  - Any SPARQL endpoint / OWL API-based reasoner combination
  - No non-standard syntax

owlery: REST web services for owlet

- Allows federation via SERVICE keyword in SPARQL
- http://github.com/phenoscape/owlery

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX ao: <http://purl.obolibrary.org/obo/my-anatomy-ontology/>
PREFIX ow: <http://purl.org/phenoscape/owlet/syntax#>
SELECT DISTINCT ?gene
WHERE
{
?gene ao:expressed_in ?structure .
?structure rdf:type ?structure_class .
  # Triple pattern containing an OWL expression, handled by federated query:
  SERVICE <http://owlery.example.org/sparql> {
  ?structure_class rdfs:subClassOf
         "ao:muscle and (ao:part_of some ao:head)"^^ow:omn .
  }
}
```

Summary

- Programming and computing with OWL is more complicated than need be
- Need a thriving ecosystem of small tools that fill common gaps
- Scowl, Owlet, and Owlery are small steps in that direction
- Vision: programming with ontologies as easy and ubiquitous as with alignments

Acknowledgements

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- Chris Mungall (LBL)
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