

# The Ontology of Craniofacial Development and Malformation

James Brinkley (PI)

Onard Mejino

Todd Detwiler

Structural Informatics Group

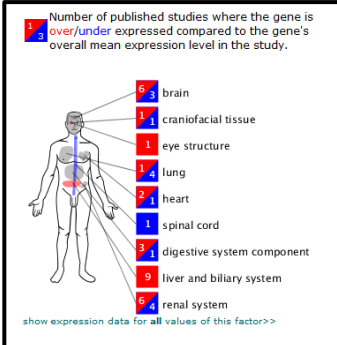
University of Washington

Seattle

# Goals

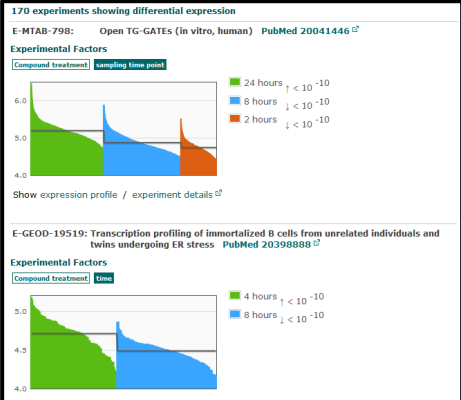
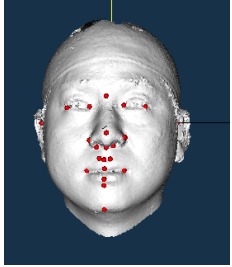
- Create an ontology for use by FaceBase
- Standardized terms
  - for annotation
  - retrieval by keyword search
- Relations
  - representation of knowledge
  - to allow semantic integration

# FaceBase



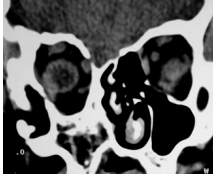
Genomic studies

Medical imaging

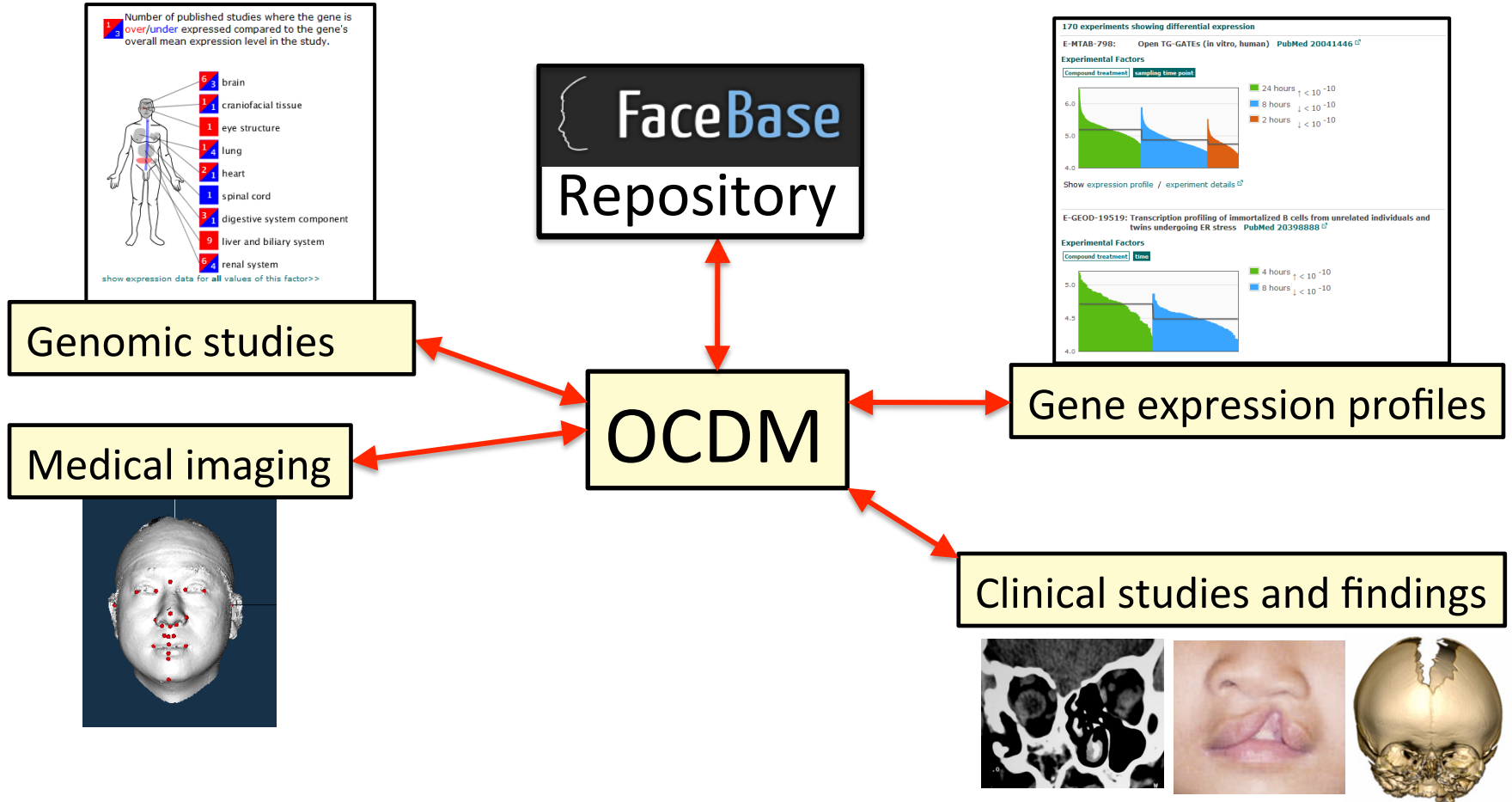


Gene expression profiles

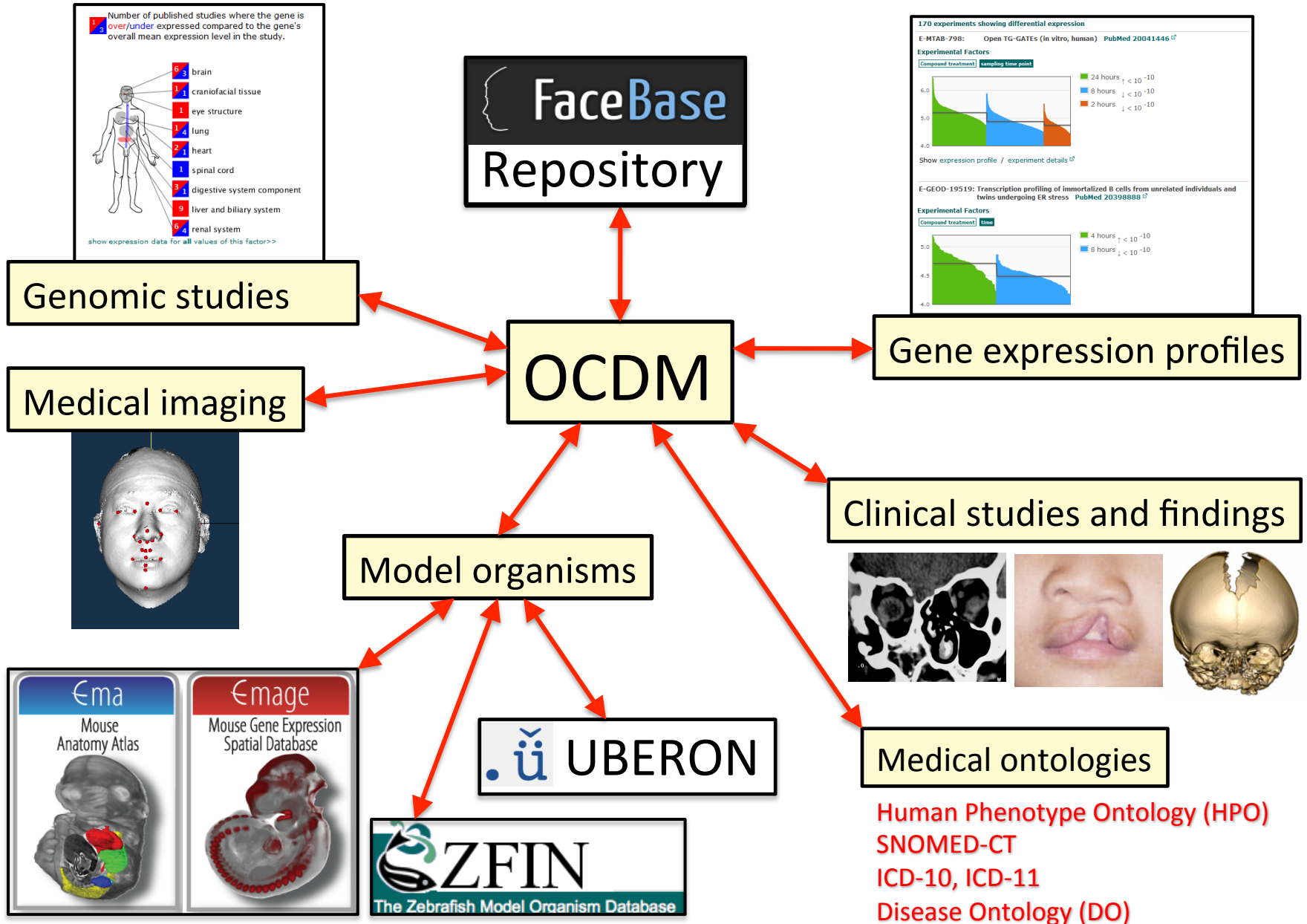
Clinical studies and findings



# OCDM



# Importing Other Ontologies





# Principles of the OCDM

- Based on human anatomy and development
  - Foundational Model of Anatomy (FMA)
- Utilize existing ontologies as much as possible
- Modular
- Ontology best practices

# FaceBase 1

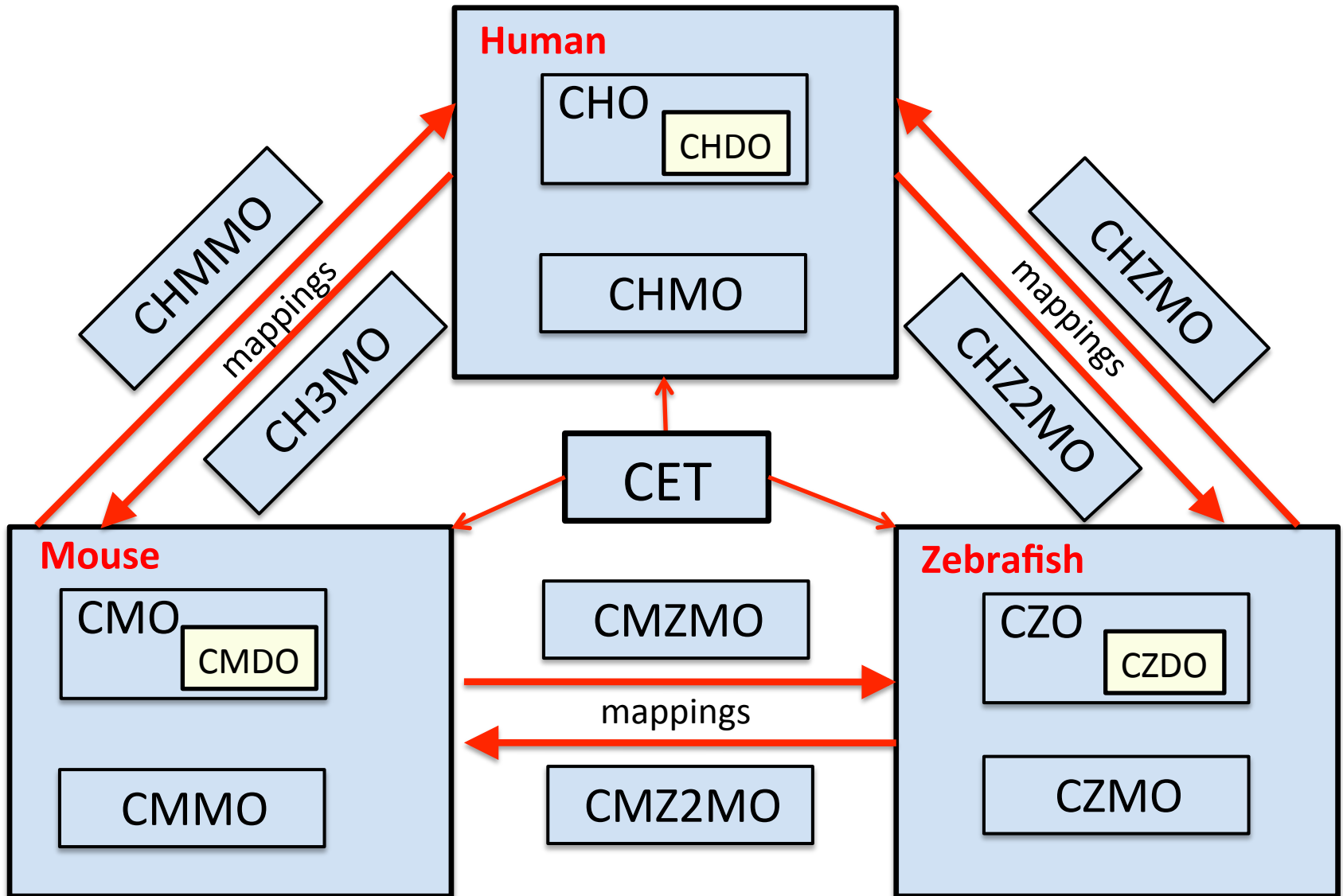
- Use Case
  - Cleft lip and/or palate
- Species
  - Mouse, human
- Developed overall framework
- Anatomy
  - Mouth and nose
  - Additional anatomy “for free”



# FaceBase 2

- Filling in the framework created in FaceBase 1
- Use Cases
  - Craniosynostosis
  - Midface hypoplasia
  - Interactive atlas of normal skull development
  - Mandible development
  - 3-D craniofacial morphometrics
- Species: human, mouse, zebrafish
- Anatomy: Musculoskeletal system of head, additional “for free”

# OCDM



# Facebase 2 Milestones

Year	Task	Dates
1	1. Conversion to OWL	
	a. CHO to OWL	May 1, 2014 – Aug 1, 2014
	b. OCDDM to OWL	June 1, 2014 – July 1, 2014
	c. Review and Testing	July 1, 2014 – Aug 1, 2014
	d. Ongoing methodology	Aug 1, 2014 – Ongoing
2	2. Canonical musculoskeletal system (MS) of head	
	a. CHO (Human)	May 1, 2014 – Aug 31, 2014
	b. CMO (Mouse)	Sept 1, 2014 – Dec, 31, 2014
	c. CZO (Zebrafish)	Jan 1, 2015 – July 31, 2015
	d. Termlists for Hub	Feb 15, 2015 -- Ongoing
3	3. Embryonic development of MS of head	
	a. CHDO (Human)	Aug 1, 2015 – Nov 30, 2015
	b. CMDO (Mouse)	Dec 1, 2015 – March 31, 2016
	c. CZDO (Zebrafish)	April 1, 2016 – Sept 30, 2016
3	4. Anatomy mappings	
	a. CHO with CMO	Oct 1, 2016 – Jan 31, 2017
	b. CMO with CZO	Feb 1, 2017 – June 30, 2017

# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications

# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications

# Craniofacial Zebrafish Ontology (CZO)

---

- same ontological framework as **CHO** (human) and **CMO** (mouse)

- content derived from

*Zebrafish anatomical ontology (ZFIN: <https://zfin.org/>)*

*FaceBase domain experts (Harris and Fischer)*

*Publication - Cubbage and Mabee (J. Morphol.229: 121-160)*

- unique source ID retained

E.g.

cranial vault (**ZAO**) [http://purl.obolibrary.org/obo/ZFA\\_0005606](http://purl.obolibrary.org/obo/ZFA_0005606)

suture (**CZO**) <http://purl.org/sig/ont/czo/czo0000043>



# Craniofacial Zebrafish Ontology (CZO)

- 'Organ (Danio rerio)'
  - ▼ ● 'Cavitated organ (Danio rerio)'
    - ▼ ● 'Organ with cavitated organ parts (Danio rerio)'
      - ▼ ● 'Bone organ (Danio rerio)'
        - ▼ ● 'Cartilage bone (Danio rerio)'
          - ▼ ● 'Endochondral bone (Danio rerio)'
            - ▶ ● 'Autopalatine (Danio rerio)'  
[http://purl.obolibrary.org/obo/ZFA\\_0000620](http://purl.obolibrary.org/obo/ZFA_0000620)
            - ▶ ● 'Basioccipital bone (Danio rerio)'
            - ▶ ● 'Ceratobranchial bone (Danio rerio)'
            - ▶ ● 'Epihyal bone (Danio rerio)'
            - ▶ ● 'Epiotic bone (Danio rerio)'
            - ▶ ● 'Exoccipital bone (Danio rerio)'
            - ▶ ● 'Hyomandibula (Danio rerio)'
            - ▶ ● 'Hypobranchial bone (Danio rerio)'
            - ▶ ● 'Hypohyal bone (Danio rerio)'
            - ▶ ● 'Kinethmoid bone (Danio rerio)'
            - ▶ ● 'Metapterygoid bone (Danio rerio)'
            - ▶ ● 'Orbitosphenoid bone (Danio rerio)'
            - ▶ ● 'Pre-ethmoid bone (Danio rerio)'
            - ▶ ● 'Pro-otic bone (Danio rerio)'
            - ▶ ● 'Quadrate bone (Danio rerio)'
            - ▶ ● 'Retro-articular bone (Danio rerio)'
            - ▶ ● 'Sclerotic bone (Danio rerio)'

Description: 'Autopalatine (Danio rerio)'

Equivalent To +

SubClass Of +

- 'Endochondral bone (Danio rerio)'
- articulates\_with some 'Maxilla (Danio rerio)'
- articulates\_with some 'Pre-ethmoid bone (Danio rerio)'
- regional\_part some 'Anterior dorsomedial process of autopalatine (Danio rerio)'
- regional\_part some 'Anterior lateral process of autopalatine (Danio rerio)'
- regional\_part some 'Anterior ventromedial process of autopalatine (Danio rerio)'

SubClass Of (Anonymous Ancestor)

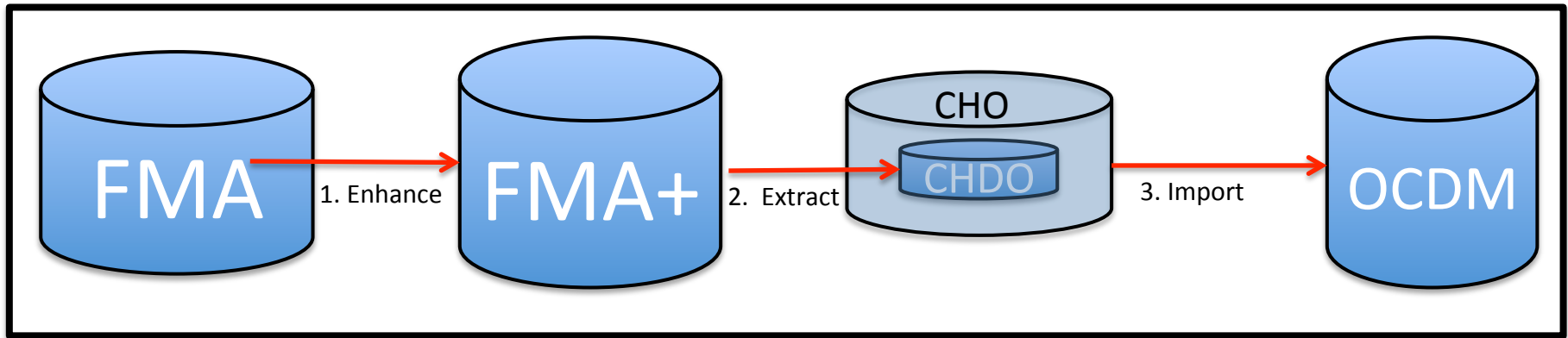
- has\_dimension value "true"
- species value "zebrafish"^^string
- has\_mass value "true"



# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications

# Development of human craniofacial musculoskeletal system



1. **Enhance** the FMA (Foundational Model of Anatomy) with embryology content
2. **Extract** as CHDO (Craniofacial Human Development Ontology) component of CHO (Craniofacial Human Ontology)
3. **Import** CHO/CHDO into OCDM

# Development of craniofacial musculoskeletal system

## Class hierarchy: 'First pharyngeal arch'

- 'Developmental structure'
- ▶ ● 'Cardinal subdivision of developmental organ system'
- ▼ ● 'Cardinal subdivision of developmental organism'
- ▶ ● 'Cardinal body part of embryo'
- ▶ ● 'Cardinal body part of fetus'
- ▶ ● 'Subdivision of blastocyst'
- ▼ ● 'Subdivision of cardinal body part of embryo'
- 'Anterior limb bud'
- 'Aortopulmonary septum'
- 'Cardiogenic area'
- 'Caudal rim of embryonic disc'
- ▶ ● 'Embryonic body fold'
- ▶ ● 'Foot plate'
- 'Genital tubercle'
- ▶ ● 'Hand plate'
- 'Left aorticopulmonary cushion'
- ▶ ● 'Pharyngeal apparatus'
- 'Posterior limb bud'
- 'Primitive middle ear'
- ▼ ● 'Region of pharyngeal apparatus'
- ▼ ● 'Pharyngeal arch'
- 'Fifth pharyngeal arch'
- ▶ ● 'First pharyngeal arch'
- ▶ ● 'Fourth pharyngeal arch'
- ▶ ● 'Second pharyngeal arch'
- 'Sixth pharyngeal arch'
- ▶ ● 'Third pharyngeal arch'
- ▶ ● 'Pharyngeal groove'
- ▶ ● 'Pharyngeal membrane'
- ▶ ● 'Pharyngeal pouch'
- ▶ ● 'Region of pharyngeal arch'
- 'Right aorticopulmonary cushion'
- 'Seessel's pocket'
- ▶ ● 'Subdivision of body proper of embryo'
- ▼ ● 'Subdivision of head of embryo'
- ▶ ● 'Cardinal subdivision of head of embryo'
- 'Cranial rim of embryonic disc'
- ▶ ● 'Embryonic part of head of embryo'

## Annotations: 'First pharyngeal arch'

CHDO

- Annotations +
- label [language: en]  
First pharyngeal arch
  - FMAID [type: string]  
293019
  - 'preferred name' [type: string]  
First pharyngeal arch
  - synonym [type: string]  
First branchial arch
  - synonym [type: string]  
Mandibular arch

## Description: 'First pharyngeal arch'

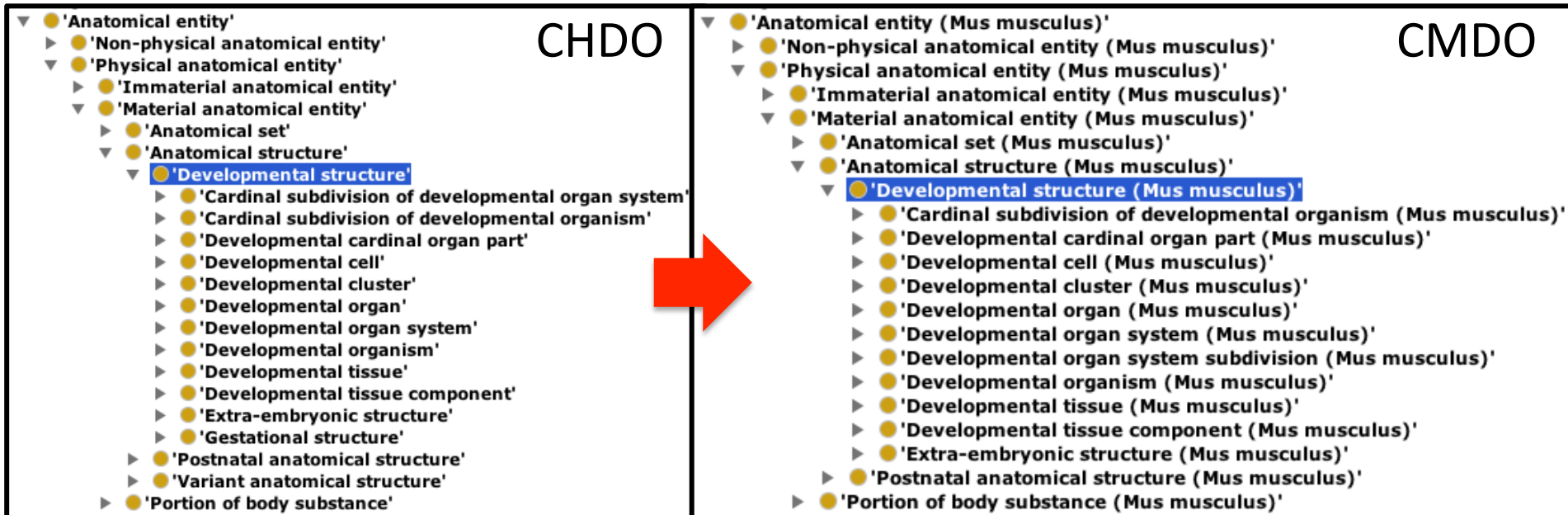
- Equivalent To +
- SubClass Of +
- 'constitutional part' some 'Ectoderm of first pharyngeal arch'
  - 'constitutional part' some 'Endoderm of first pharyngeal arch'
  - 'constitutional part' some 'Mesenchyme of first pharyngeal arch'
  - 'has developmental stage' some 'Carnegie stage 12'
  - 'has developmental stage' some 'Carnegie stage 13'
  - 'has developmental stage' some 'Carnegie stage 14'
  - 'has developmental stage' some 'Carnegie stage 15'
  - 'Pharyngeal arch'
  - 'regional part of' some 'Pharyngeal apparatus'
  - 'derives' some 'Maxillary process'
- SubClass Of (Anonymous Ancestor)
- 'has dimension' value true

# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications


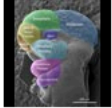
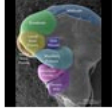
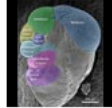


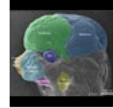


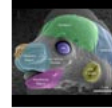

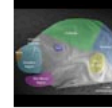




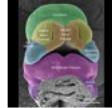




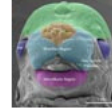

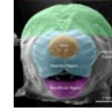







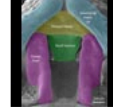
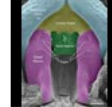



# Human as initial template for mouse

## CHDO template for CMDO

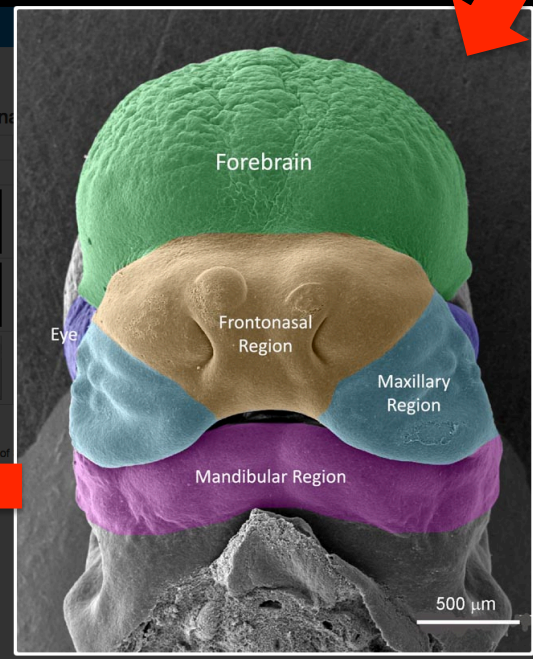
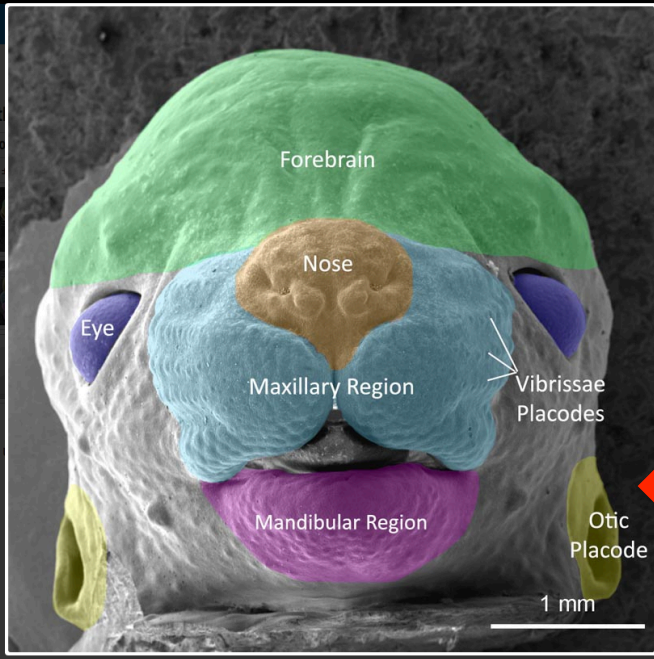
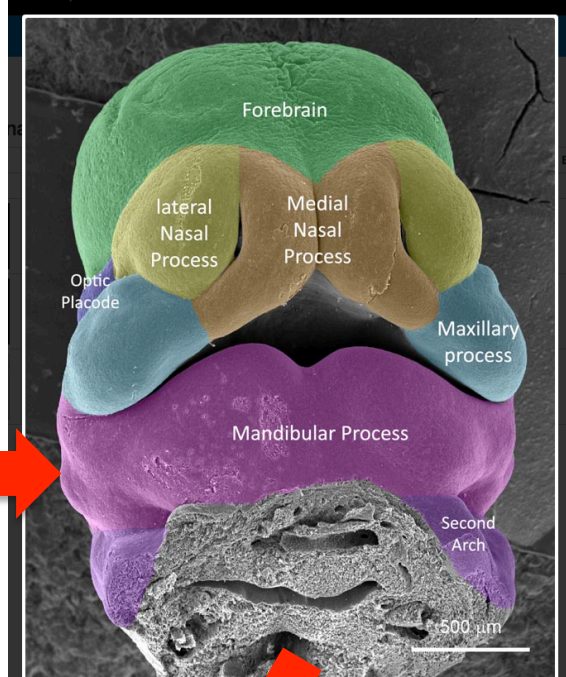
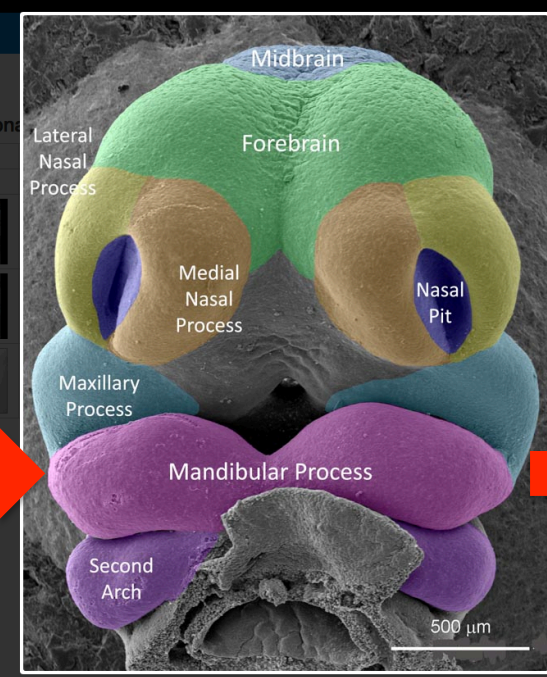
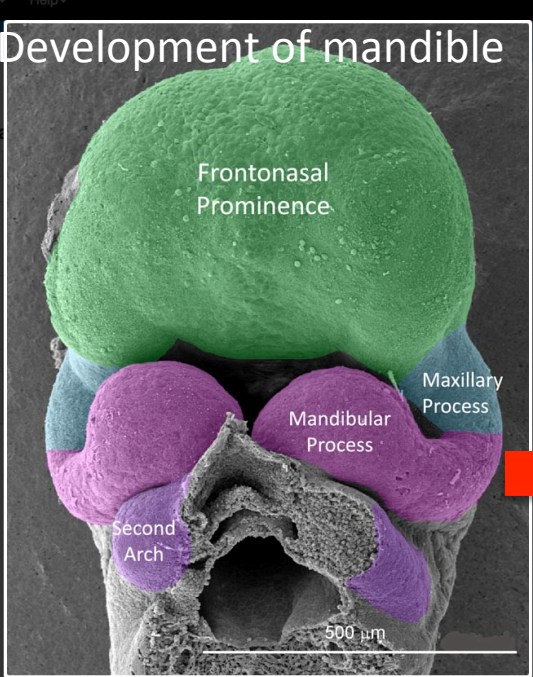


# Mouse Anatomy

C57BL/6 mouse embryo (click thumbnails to enlarge) -- Anatomy Marked

	E9.5	E10	E10.5	E11	E11.5	E12	E12.5	E13	E13.5	E14	E14.5	E15
# of Somites	23 ± 2	32 ± 2	37 ± 2	42 ± 2	45 ± 2							
Side view												
Frontal view												
Palate												

# Development of mandible











# Craniofacial Mouse Developmental Ontology (CMDO)

Class hierarchy: 'Mandibular part of first pharyngeal arch (Mus musculus)'

- ▶ ● 'Pharyngeal apparatus (Mus musculus)'
  - ▼ ● 'Region of pharyngeal apparatus (Mus musculus)'
    - ▶ ● 'Pharyngeal arch (Mus musculus)'
    - ▶ ● 'Pharyngeal groove (Mus musculus)'
    - ▶ ● 'Pharyngeal membrane (Mus musculus)'
    - ▶ ● 'Pharyngeal pouch (Mus musculus)'
    - ▼ ● 'Region of pharyngeal arch (Mus musculus)'
      - ▼ ● 'Region of first pharyngeal arch (Mus musculus)'
        - ▶ ● 'Lateral lingual swelling (mus musculus)'
        - ▶ ● 'Mandibular part of first pharyngeal arch (Mus musculus)'
        - ▶ ● 'Maxillary process (Mus musculus)'
        - ▶ ● 'Maxillary-mandibular groove (Mus musculus)'
        - ▶ ● 'Medial lingual swelling of first pharyngeal arch (Mus musculus)'
    - ▶ ● 'Subdivision of body proper of embryo (Mus musculus)'
    - ▶ ● 'Subdivision of head of embryo (Mus musculus)'
      - ▶ ● 'Wall of notochordal process (Mus musculus)'

- ▶ ● 'Developmental cardinal organ part (Mus musculus)'
- ▶ ● 'Developmental cell (Mus musculus)'
- ▶ ● 'Developmental cluster (Mus musculus)'
- ▼ ● 'Developmental organ (Mus musculus)'
- ▶ ● 'Bone primordium (Mus musculus)'
- ▶ ● 'Dental bud (Mus musculus)'
- ▼ ● 'Embryonic bone organ (Mus musculus)'
- ▶ ● 'Embryonic auditory ossicle (Mus musculus)'
- ▶ ● 'Embryonic basioccipital bone (Mus musculus)'

Annotations: 'Mandibular part of first pharyngeal arch (Mus musculus)'

preferred\_name [type: string] @ X O  
Mandibular part of first pharyngeal arch

synonym [type: string] @ X O  
Mandibular process

verified [type: boolean] @ X O

Description: 'Mandibular part of first pharyngeal arch (Mus musculus)'

Equivalent To +

SubClass Of +

- 'Region of first pharyngeal arch (Mus musculus)'
- constitutional\_part some 'Ectoderm of mandibular part of first pharyngeal arch (Mus musculus)'
- constitutional\_part some 'Endoderm of mandibular part of first pharyngeal arch (Mus musculus)'
- constitutional\_part some 'Mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
- regional\_part\_of some 'First pharyngeal arch (Mus musculus)'

# Craniofacial Mouse Developmental Ontology (CMDO)

- ▶ ● 'Mesenchyme of embryonic vomeronasal organ (Mus musculus)'
- ▶ ● 'Mesenchyme of frontonasal process (Mus musculus)'
- ▶ ● 'Mesenchyme of lateral lingual swelling (Mus musculus)'
- ▶ ● 'Mesenchyme of left nasal region of frontonasal process (Mus musculus)'
- ▼ ● 'Mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'Mesenchyme of mandibular part of left first pharyngeal arch (Mus musculus)'
  - 'Mesenchyme of mandibular part of right first pharyngeal arch (Mus musculus)'
  - 'TS14 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS15 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS16 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS17 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS18 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS19 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS20 mesenchyme of mandibular process (Mus musculus)'
- ▶ ● 'Mesenchyme of maxillary process (Mus musculus)'
- ▶ ● 'Mesenchyme of medial lingual swelling (Mus musculus)'
- ▶ ● 'Mesenchyme of nasal process (Mus musculus)'
- ▶ ● 'Mesenchyme of nasal region of frontonasal process (Mus musculus)'
- ▶ ● 'Mesenchyme of nasal septum (Mus musculus)'

Description: 'TS17 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'

Equivalent To +

SubClass Of +

● 'Mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'

● has\_embryonic\_day some 'E10.5 (Mus musculus)'

● has\_Theiler\_stage some 'TS17 (Mus musculus)'

SubClass Of (Anonymous Ancestor)

● constitutional\_part\_of some 'Mandibular part of first pharyngeal arch (Mus musculus)'

● species value "mouse"^^string

● has\_dimension value true

● has\_boundaries true

# Craniofacial Mouse Developmental Ontology (CMDO)

## E10.5 mandibular part of first pharyngeal arch, mesenchyme

- ▶ ● 'Mesenchyme of embryonic vomeronasal organ (Mus musculus)'
- ▶ ● 'Mesenchyme of frontonasal process (Mus musculus)'
- ▶ ● 'Mesenchyme of lateral lingual swelling (Mus musculus)'
- ▶ ● 'Mesenchyme of left nasal region of frontonasal process (Mus musculus)'
- ▼ ● 'Mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'Mesenchyme of mandibular part of left first pharyngeal arch (Mus musculus)'
  - 'Mesenchyme of mandibular part of right first pharyngeal arch (Mus musculus)'
  - 'TS14 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS15 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS16 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS17 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS18 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS19 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'
  - 'TS20 mesenchyme of mandibular process (Mus musculus)'
- ▶ ● 'Mesenchyme of maxillary process (Mus musculus)'
- ▶ ● 'Mesenchyme of medial lingual swelling (Mus musculus)'
- ▶ ● 'Mesenchyme of nasal process (Mus musculus)'
- ▶ ● 'Mesenchyme of nasal region of frontonasal process (Mus musculus)'
- ▶ ● 'Mesenchyme of nasal septum (Mus musculus)'

Description: 'TS17 mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'

Equivalent To +

SubClass Of +

● 'Mesenchyme of mandibular part of first pharyngeal arch (Mus musculus)'

● has\_embryonic\_day some 'E10.5 (Mus musculus)'

● has\_Theiler\_stage some 'TS17 (Mus musculus)'

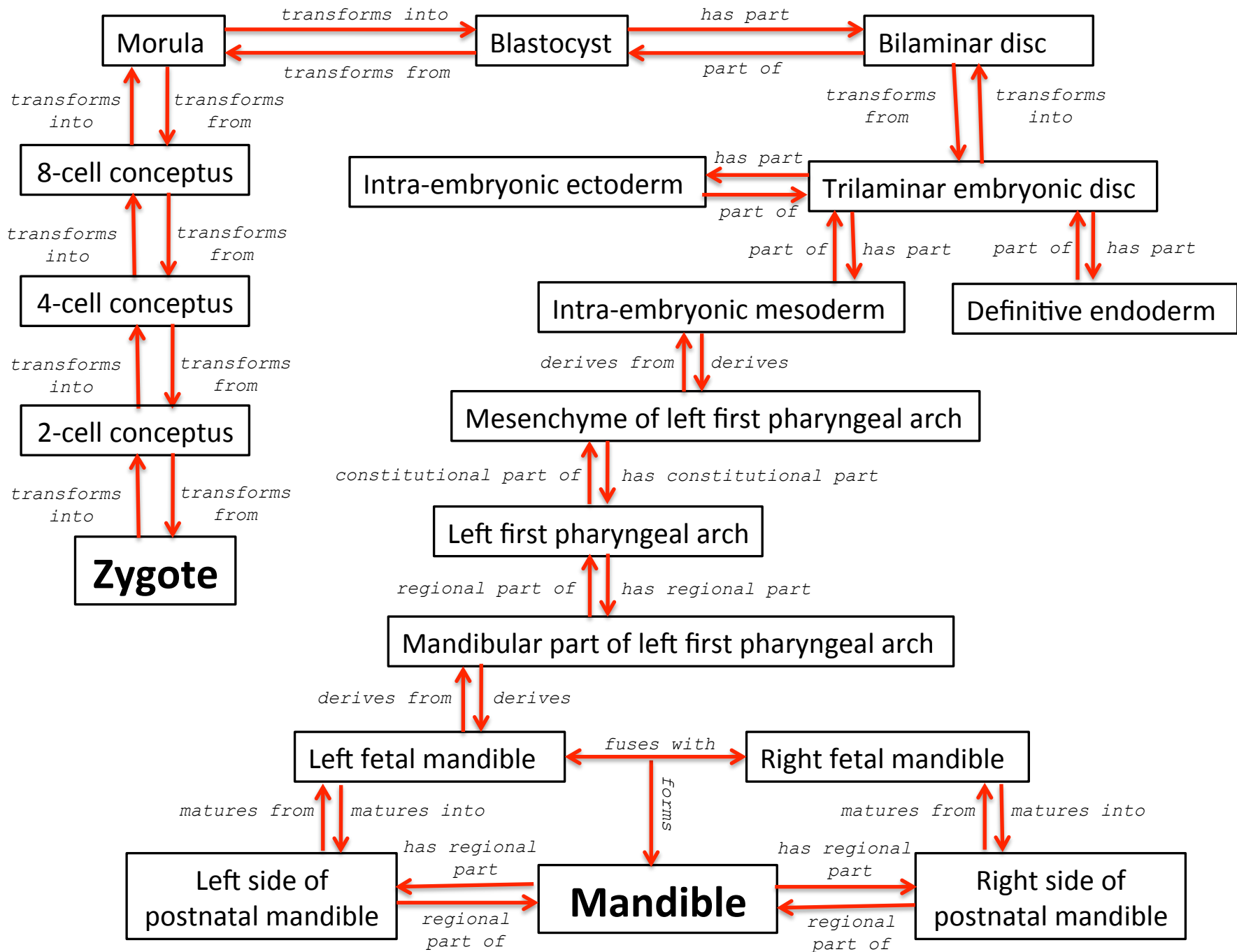
SubClass Of (Anonymous Ancestor)

● constitutional\_part\_of some 'Mandibular part of first pharyngeal arch (Mus musculus)'

● species value "mouse"^^string

● has\_dimension value true

● has\_boundaries true



# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications

## HUB metadata domains

Species

Mouse strain

Specimen type

Anatomy

*Human*

*Mouse*

*Zebrafish*

Malformation phenotype

*Human*

*Mammalian*

Developmental stage

*Human*

*Mouse*

*Zebrafish*

Developmental day

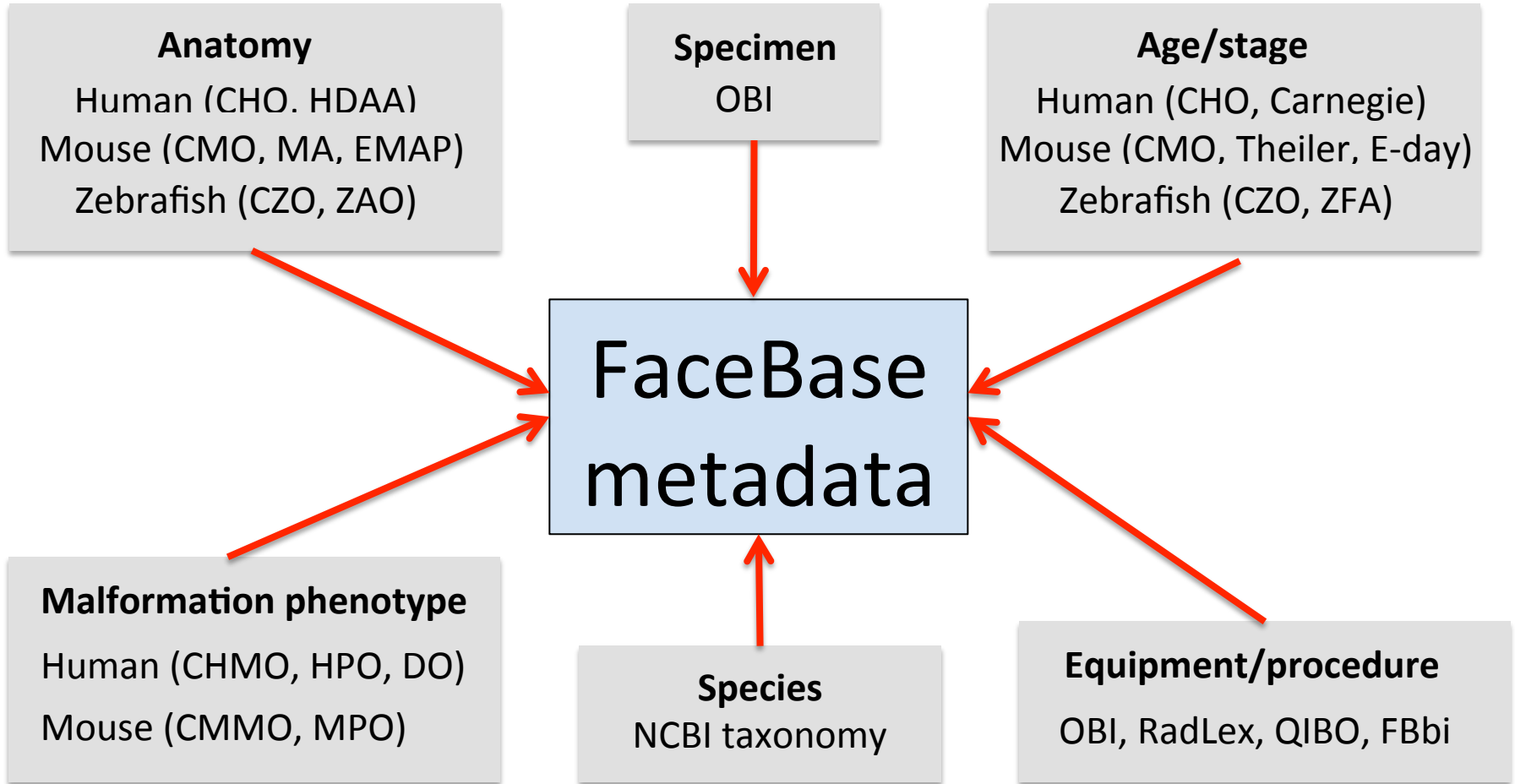
*Human*

*Mouse*

Imaging method/device



# Metadata Annotation List



### **OCDM components:**

<b>CHO</b>	Craniofacial Human Ontology
<b>CMO</b>	Craniofacial Mouse Ontology
<b>CZO</b>	Craniofacial Zebrafish Ontology
<b>CHMO</b>	Craniofacial Human Malformation Ontology
<b>CMMO</b>	Craniofacial Mouse Malformation Ontology

### **External ontology sources:**

<b>EMAP</b>	Mouse Gross Anatomy and Development Ontology
<b>HPO</b>	Human Phenotype Ontology
<b>MA</b>	Mouse Adult Gross Anatomy Ontology
<b>MPO</b>	Mammalian Phenotype Ontology
<b>HDAA</b>	Human Developmental Anatomy Ontology
<b>DO</b>	Disease Ontology
<b>ZFA</b>	Zebrafish Anatomy and Development Ontology
<b>OBI</b>	Ontology of Biomedical Investigation
<b>RadLex</b>	Radiology Lexicon
<b>QIBO</b>	Quantitative Imaging Biomarker Ontology
<b>Fbbi</b>	Biological Imaging Methods Ontology

# HUB metadata domains

## Species

## Source: NCBI taxonomy

<b>Danio rerio</b>	Zebrafish	<a href="http://purl.bioontology.org/ontology/NCBITAXON/7955">http://purl.bioontology.org/ontology/NCBITAXON/7955</a>
<b>Mus musculus</b>	Mouse	<a href="http://purl.bioontology.org/ontology/NCBITAXON/10090">http://purl.bioontology.org/ontology/NCBITAXON/10090</a>
<b>Homo sapiens</b>	Human	<a href="http://purl.bioontology.org/ontology/NCBITAXON/9606">http://purl.bioontology.org/ontology/NCBITAXON/9606</a>
<b>Pan troglodytes</b>	Chimpanzee	<a href="http://purl.bioontology.org/ontology/NCBITAXON/9598">http://purl.bioontology.org/ontology/NCBITAXON/9598</a>

# HUB metadata domains

## **Specimen**

## **Source**

### **Ontology of Biological Investigation (OBI)**

**Tissue specimen**

[http://purl.obolibrary.org/obo/OBI\\_0001479](http://purl.obolibrary.org/obo/OBI_0001479)

**Whole mount tissue**

[http://purl.obolibrary.org/obo/OBI\\_1000049](http://purl.obolibrary.org/obo/OBI_1000049)

### **Biological Imaging Methods Ontology**

**Cryofixed tissue**

[http://purl.obolibrary.org/obo/FBbi\\_00000013](http://purl.obolibrary.org/obo/FBbi_00000013)

**Embedded tissue**

[http://purl.obolibrary.org/obo/FBbi\\_00000015](http://purl.obolibrary.org/obo/FBbi_00000015)

### **FaceBase member**

**Cultured explant**

Tomek/Wysocka/Selleri

**Cryostat slice sample**

Aho/Wysocka/Selleri

# HUB metadata domains

## Human anatomy

### Source

#### Foundational Model of Anatomy (FMA)

Mandible

<http://purl.org/sig/ont/fma/fma52748>

Frontal suture

<http://purl.org/sig/ont/fma/fma52989>

#### Human Development Anatomy Ontology (EHDA2)

Frontonasal process

[http://purl.obolibrary.org/obo/EHDA2\\_0000578](http://purl.obolibrary.org/obo/EHDA2_0000578)

Head neural crest

[http://purl.obolibrary.org/obo/EHDA2\\_0004420](http://purl.obolibrary.org/obo/EHDA2_0004420)

#### Cell Ontology (CL)

Embryonic stem cell

[http://purl.obolibrary.org/obo/CL\\_0002322](http://purl.obolibrary.org/obo/CL_0002322)

# HUB metadata domains

## **Mouse anatomy**

### **Source**

**Craniofacial Mouse Ontology (CMO of OCDM)**

**Nasal placode**

<http://purl.org/sig/ont/cmo/cmo0010281>

**Neural crest**

<http://purl.org/sig/ont/cmo/cmo0008902>

**Mouse Gross Anatomy and Development Ontology (EMAP)**

**Fronto-nasal process**

[http://purl.obolibrary.org/obo/EMAPA\\_16681](http://purl.obolibrary.org/obo/EMAPA_16681)

**Branchial arch**

[http://purl.obolibrary.org/obo/EMAPA\\_16117](http://purl.obolibrary.org/obo/EMAPA_16117)

**Mouse Adult Gross Anatomy Ontology (MA)**

**Frontal bone**

[http://purl.obolibrary.org/obo/MA\\_0001466](http://purl.obolibrary.org/obo/MA_0001466)

**Face**

[http://purl.obolibrary.org/obo/MA\\_0002473](http://purl.obolibrary.org/obo/MA_0002473)

# HUB metadata domains

## **Zebrafish anatomy**

## **Source**

**Zebrafish Anatomy and Development Ontology (ZFA)**

**Cranial vault**

[http://purl.obolibrary.org/obo/ZFA\\_0005606](http://purl.obolibrary.org/obo/ZFA_0005606)

**Premaxilla**

[http://purl.obolibrary.org/obo/ZFA\\_0000567](http://purl.obolibrary.org/obo/ZFA_0000567)

**Craniofacial Zebrafish Ontology (CZO of OCDM)**

**Suture**

<http://purl.org/sig/ont/czo/czo0000043>

**Gill rakers**

<http://purl.org/sig/ont/czo/czo0001198>

# HUB metadata domains

## Human malformation phenotype

### Source

#### Craniofacial Human Malformation Ontology (CHMO)

**Labiopalatal cleft**

<http://purl.org/sig/ont/chmo/chmo0000536>

**Pseudoachondroplasia**

<http://purl.org/sig/ont/chmo/chmo0001002>

#### Human Phenotype Ontology (HPO)

**Craniosynostosis**

[http://purl.obolibrary.org/obo/HP\\_0001363](http://purl.obolibrary.org/obo/HP_0001363)

**Micrognathia**

[http://purl.obolibrary.org/obo/HP\\_0000347](http://purl.obolibrary.org/obo/HP_0000347)



# HUB metadata domains

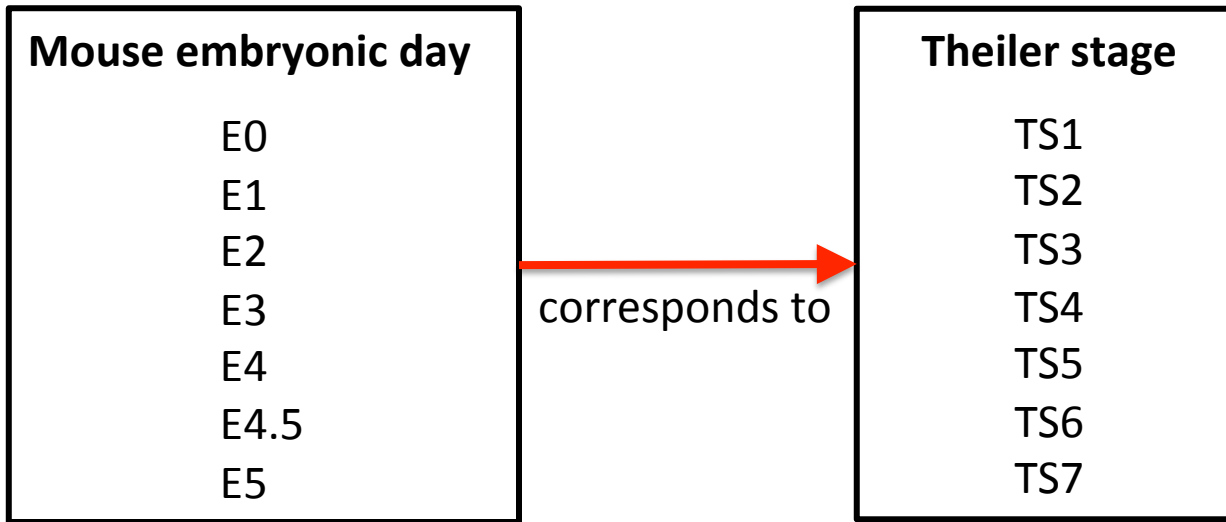
## Mouse embryo day

E0	<a href="http://purl.org/sig/ont/cmo/cmo0013382">http://purl.org/sig/ont/cmo/cmo0013382</a>
E1	<a href="http://purl.org/sig/ont/cmo/cmo0008717">http://purl.org/sig/ont/cmo/cmo0008717</a>
E3	<a href="http://purl.org/sig/ont/cmo/cmo0013384">http://purl.org/sig/ont/cmo/cmo0013384</a>
E4	<a href="http://purl.org/sig/ont/cmo/cmo0008738">http://purl.org/sig/ont/cmo/cmo0008738</a>
E4.5	<a href="http://purl.org/sig/ont/cmo/cmo0008768">http://purl.org/sig/ont/cmo/cmo0008768</a>
E5	<a href="http://purl.org/sig/ont/cmo/cmo0008771">http://purl.org/sig/ont/cmo/cmo0008771</a>
E6	<a href="http://purl.org/sig/ont/cmo/cmo0008774">http://purl.org/sig/ont/cmo/cmo0008774</a>

## Theiler stage

TS1	<a href="http://purl.org/sig/ont/cmo/cmo0013383">http://purl.org/sig/ont/cmo/cmo0013383</a>
TS2	<a href="http://purl.org/sig/ont/cmo/cmo0008718">http://purl.org/sig/ont/cmo/cmo0008718</a>
TS3	<a href="http://purl.org/sig/ont/cmo/cmo0013385">http://purl.org/sig/ont/cmo/cmo0013385</a>
TS4	<a href="http://purl.org/sig/ont/cmo/cmo0008736">http://purl.org/sig/ont/cmo/cmo0008736</a>
TS5	<a href="http://purl.org/sig/ont/cmo/cmo0008739">http://purl.org/sig/ont/cmo/cmo0008739</a>
TS6	<a href="http://purl.org/sig/ont/cmo/cmo0008769">http://purl.org/sig/ont/cmo/cmo0008769</a>
TS7	<a href="http://purl.org/sig/ont/cmo/cmo0008772">http://purl.org/sig/ont/cmo/cmo0008772</a>

# HUB metadata domains



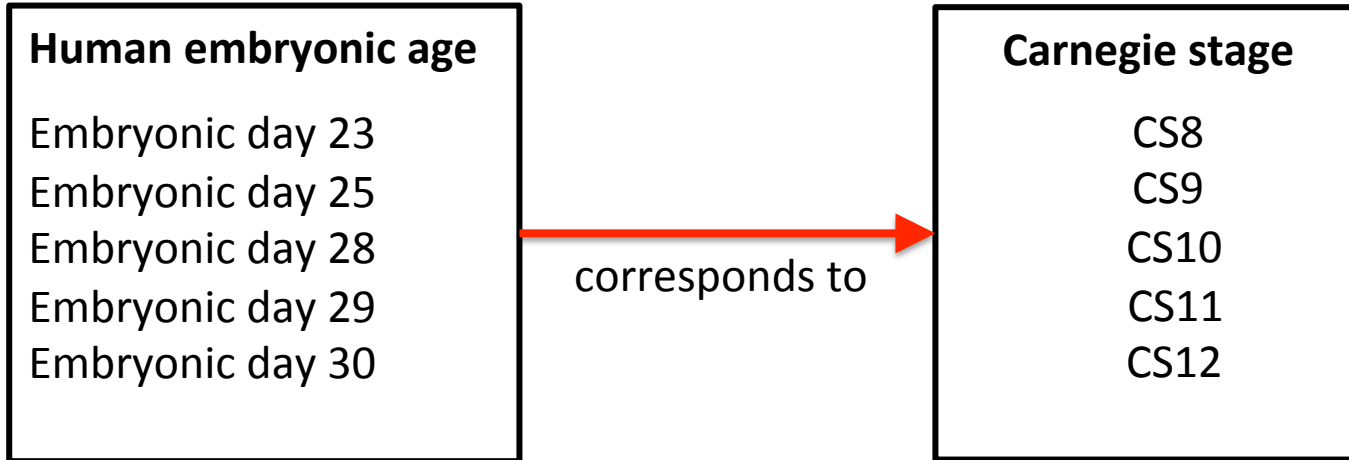
## CMO (OCDM)

The screenshot shows a web-based interface for the CMO (OCDM). On the left is a tree view of classes. The root is "Developmental age (Mus musculus)", which is expanded to show "Gestational age (Mus musculus)", which is further expanded to show "Embryonic day (Mus musculus)". Under "Embryonic day (Mus musculus)", several subclasses are listed, including "E0 (Mus musculus)", "E1 (Mus musculus)", "E10 (Mus musculus)", "E10.5 (Mus musculus)", "E11 (Mus musculus)", "E11.5 (Mus musculus)", "E12 (Mus musculus)", "E12.5 (Mus musculus)", and "E13.5 (Mus musculus)". The "E10.5 (Mus musculus)" class is highlighted in blue, and a red arrow points from it to the right-hand pane.

The right-hand pane displays the details for the "E10.5 (Mus musculus)" class. At the top, it shows the "Description: E10.5 (Mus musculus)". Below this, there are several sections:

- Equivalent To**: A plus sign (+) in a circle.
- SubClass Of**: A plus sign (+) in a circle, followed by a list of parent classes: "Embryonic day (Mus musculus)" and "corresponds\_to\_Theiler\_stage some 'TS17 (Mus musculus)'".
- SubClass Of (Anonymous Ancestor)**: A list of properties: "has\_dimension value false" and "species value 'mouse'^^string".

# HUB metadata domains



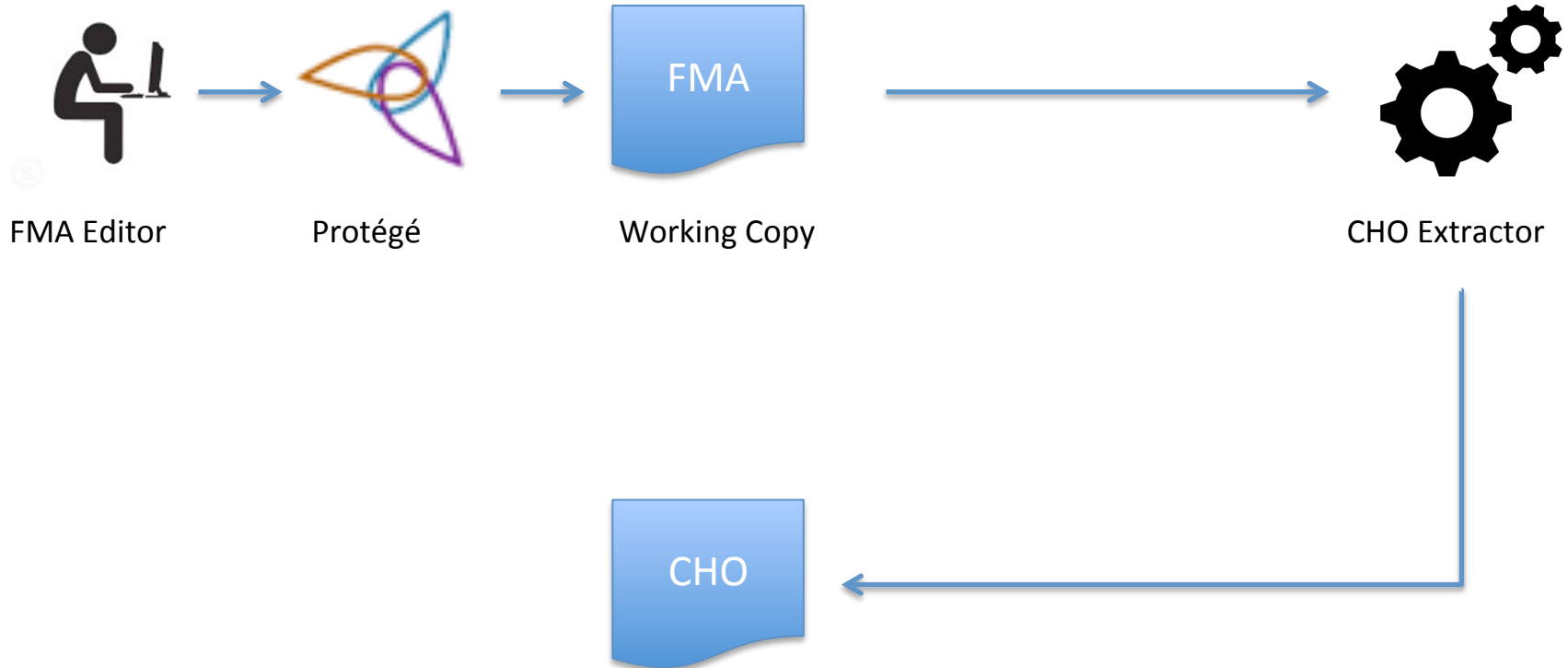
## FMA

The screenshot shows a hierarchical ontology view on the left and a detailed property view on the right. The left view shows a tree structure under "Gestational age" with "Embryonic age" expanded to show days 23, 25, 28, 29, 30, 32, and 33. The "Embryonic day 28" entry is highlighted in blue. The right view shows the "Description: 'Embryonic day 28'" header and two expandable sections: "Equivalent To" and "SubClass Of". The "SubClass Of" section is expanded, showing a relationship: "'corresponds to' some 'Carnegie stage 10'" with a yellow dot next to it, and "'Embryonic age'" below it. A red arrow points from the blue highlight in the left view to the yellow dot in the right view.

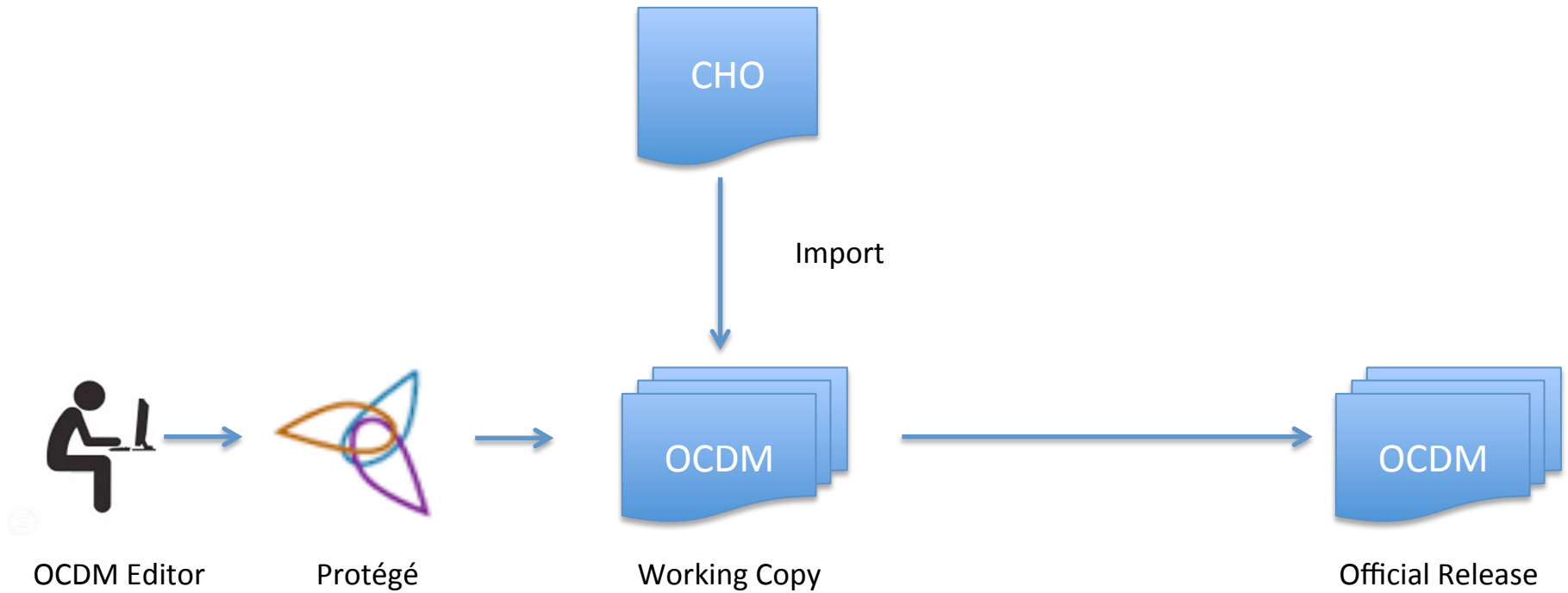
# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications

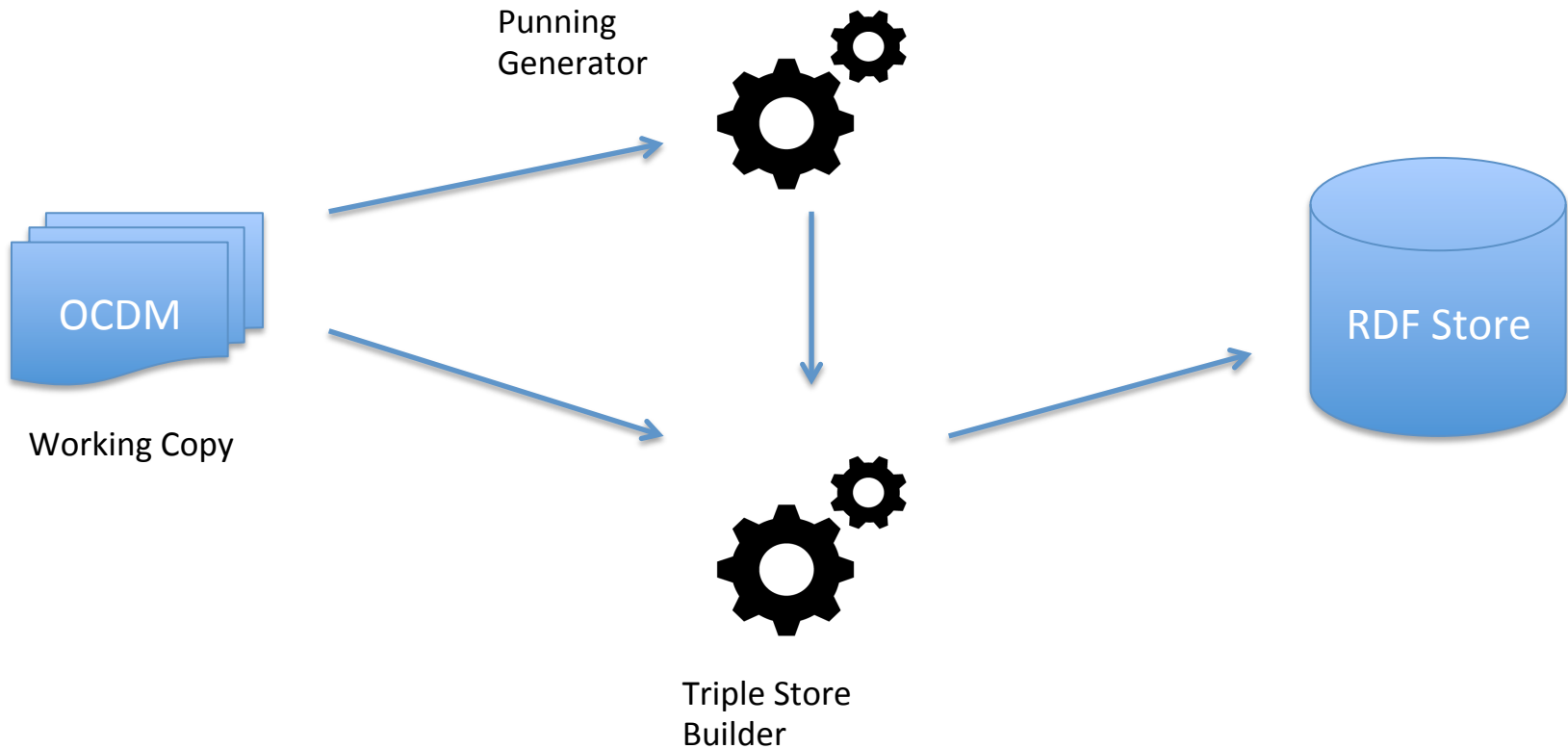
# Methods 1: FMA to CHO



# Methods 2: CHO to OCDM



# Methods 3: OCDM to Queryable Resource



# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications



# Facebase 2 Milestones

Year	Task	Dates
1	1. Conversion to OWL	
	a. CHO to OWL	May 1, 2014 – Aug 1, 2014
	b. OCDDM to OWL	June 1, 2014 – July 1, 2014
	c. Review and Testing	July 1, 2014 – Aug 1, 2014
	d. Ongoing methodology	Aug 1, 2014 – Ongoing
2	2. Canonical musculoskeletal system (MS) of head	
	a. CHO (Human)	May 1, 2014 – Aug 31, 2014
	b. CMO (Mouse)	Sept 1, 2014 – Dec, 31, 2014
	c. CZO (Zebrafish)	Jan 1, 2015 – July 31, 2015
	d. Termlists for Hub	Feb 15, 2015 -- Ongoing
3	3. Embryonic development of MS of head	
	a. CHDO (Human)	Aug 1, 2015 – Nov 30, 2015
	b. CMDO (Mouse)	Dec 1, 2015 – March 31, 2016
	c. CZDO (Zebrafish)	April 1, 2016 – Sept 30, 2016
3	4. Anatomy mappings	
	a. CHO with CMO	Oct 1, 2016 – Jan 31, 2017
	b. CMO with CZO	Feb 1, 2017 – June 30, 2017

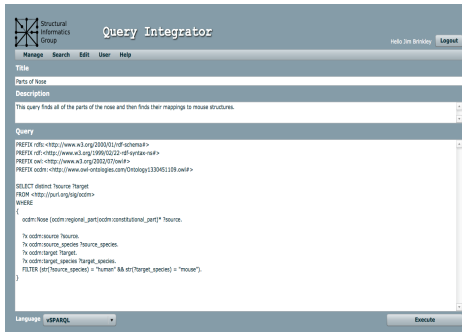
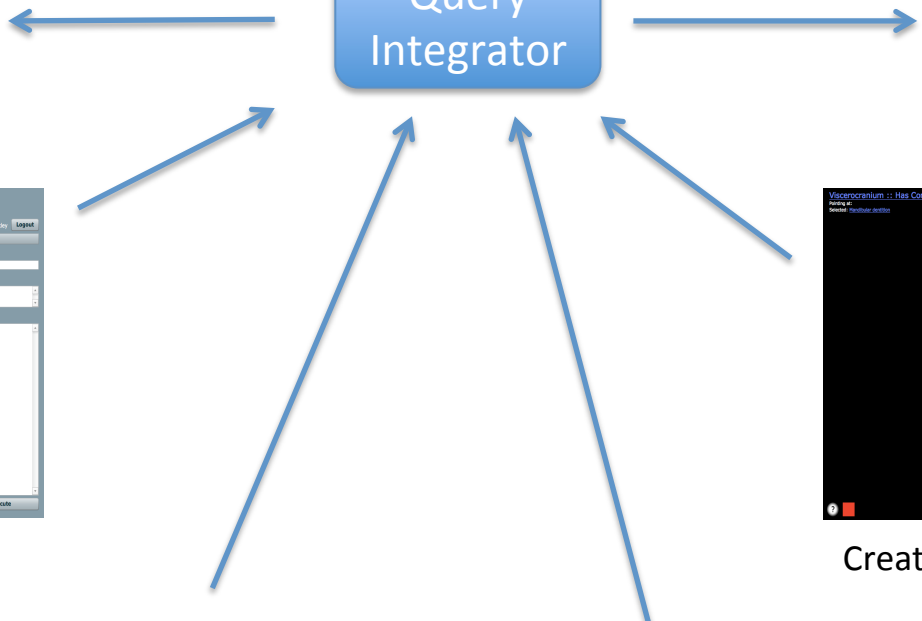
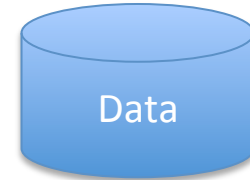
# Future Milestones

Year	Task	Date
4	5. Craniofacial malformation (facial and cranial vault dysmorphology)	
	a. CHMO (Human)	July 1, 2017 – Nov 30, 2017
	b. CMMO (Mouse)	Dec 1, 2017 – March 31, 2018
	c. CZMO (Zebrafish)	April 1, 2018 – Aug 31, 2018
5	6. Malformation mappings	
	a. CHMO with CMMO	Sept 1, 2018 – Dec 31, 2018
	b. CHMO with CZMO	Jan 1, 2019 – April 30, 2019
	7. Connect all species structure to molecular level	Ongoing

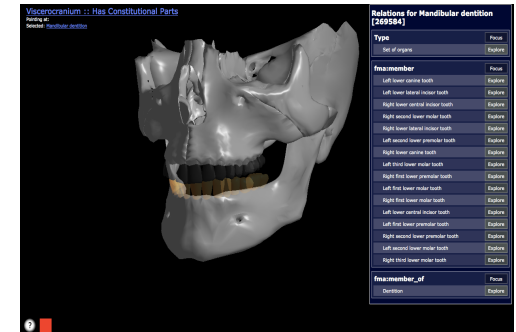
# Topics

- Craniofacial Zebrafish Ontology (CZO)
- Human craniofacial musculoskeletal system (cMSK) development
- Mouse cMSK development
- Hub metadata terms
- Ongoing Methodology
- Year 3 plans and beyond
- Potential Applications

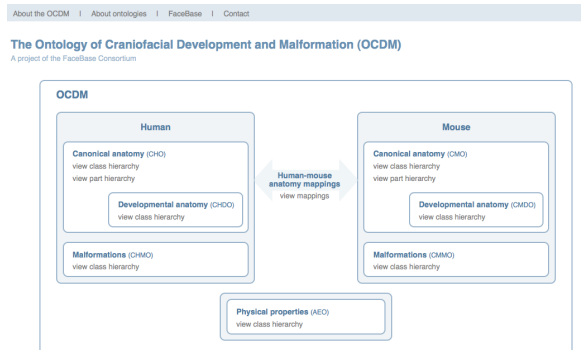
# Potential Access and Applications



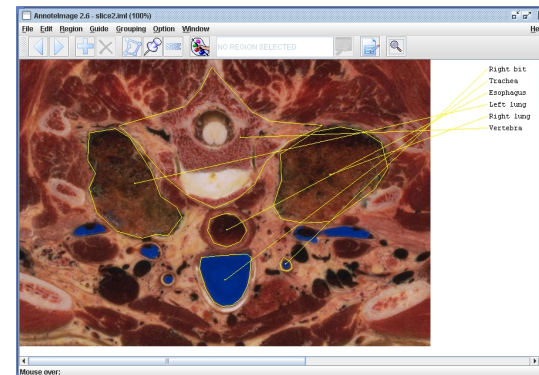
Create and save queries



Create 3-D scenes



View the OCM



Annotate Data

# Personnel

- Onard Mejino
- Todd Detwiler
- Tim Cox
- Trond Nilsen
- Melissa Clarkson
- Linda Shapiro
- Michael Cunningham

# Links

- Overview of OCDDM
  - <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4041627/>
- Foundational Model Browser
  - <http://xiphoid.biostr.washington.edu/fma/index.html>
- Obtaining the OCDDM
  - <https://www.facebase.org/ocdm/>

# Questions for Discussion

- How to make it easy to annotate FaceBase data with detailed terms from OCDDM and other ontologies
- How to utilize rich relations in OCDDM and other ontologies to facilitate “intelligent” search