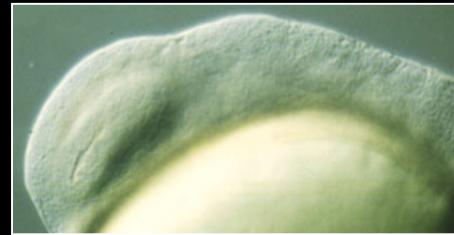




Genetic basis of animal diversity & evolution



Supported by NIH P41 HG002659 and U54 HG004028, NSF DBI-0641025,
Cambridge University, Stanford University, NESCent &
the University of Oregon



Melissa Haendel
Kevin Schaper
Erik Segerdell
Sierra Taylor

FlyBase

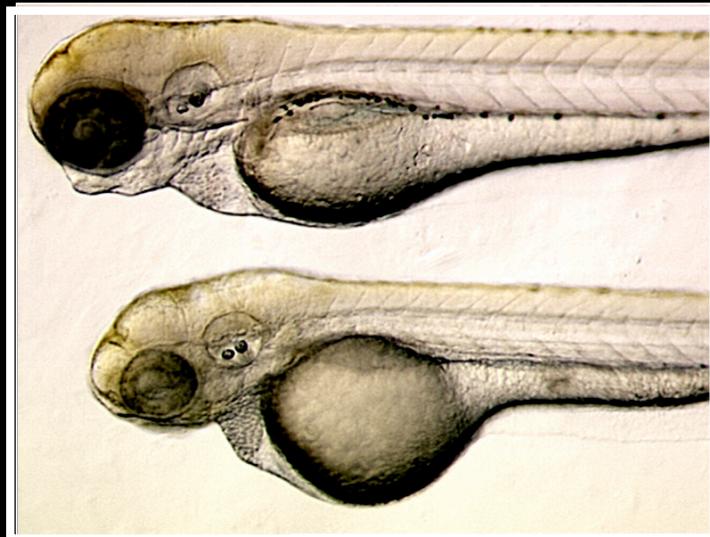
Michael Ashburner
Rachel Drysdale
George Gkoutos



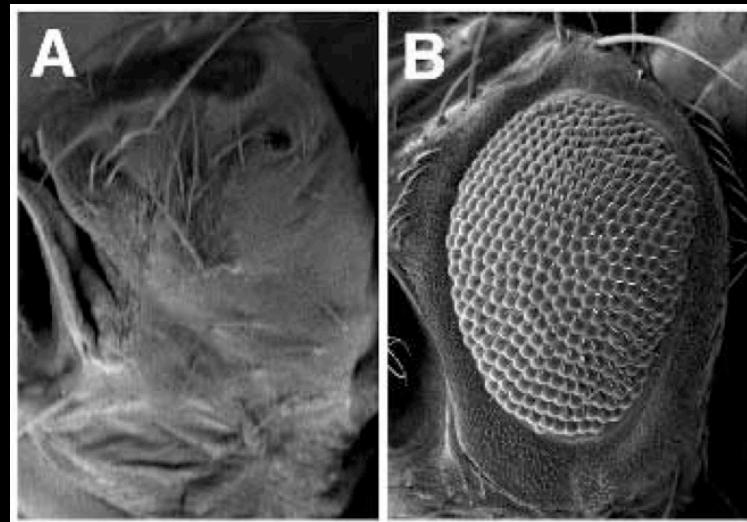
Mark Gibson
Suzi Lewis
Chris Mungall
Nicole Washington

- 1. Animal models of human disease
(diversity & evolution)**
- 2. Introduction to ontologies**
- 3. Representing mutant phenotypes**

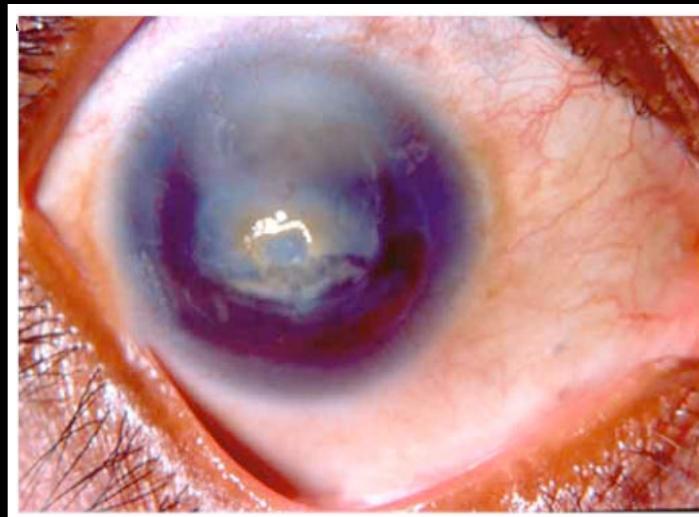
Animal models of human disease



zebrafish



fly



human

EYA gene mutants



Animal disease models:

Humans

Mutant Gene



Mutant or missing
Protein



Mutant Phenotype
(disease)

Animal models

Mutant Gene



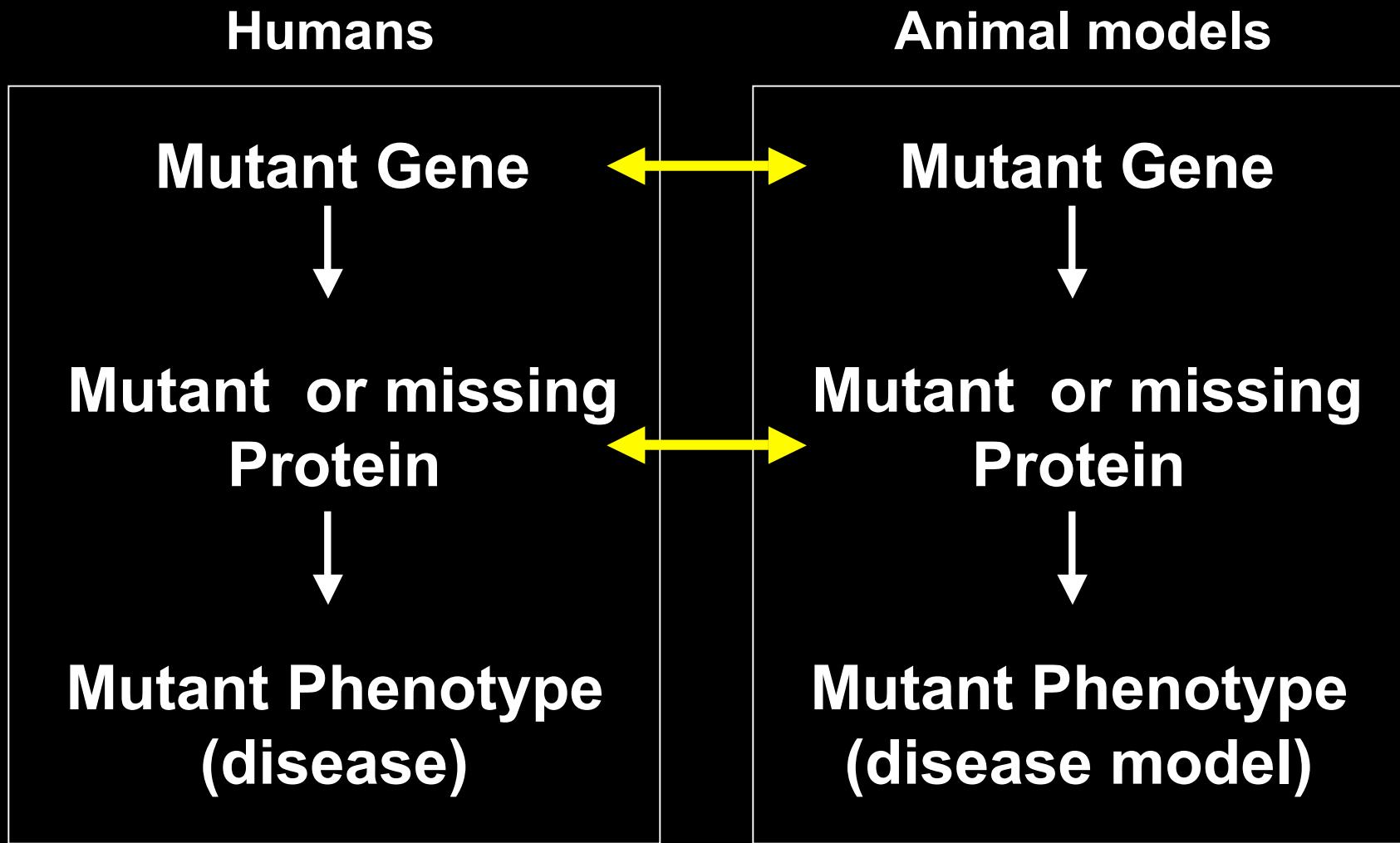
Mutant or missing
Protein



Mutant Phenotype
(disease model)

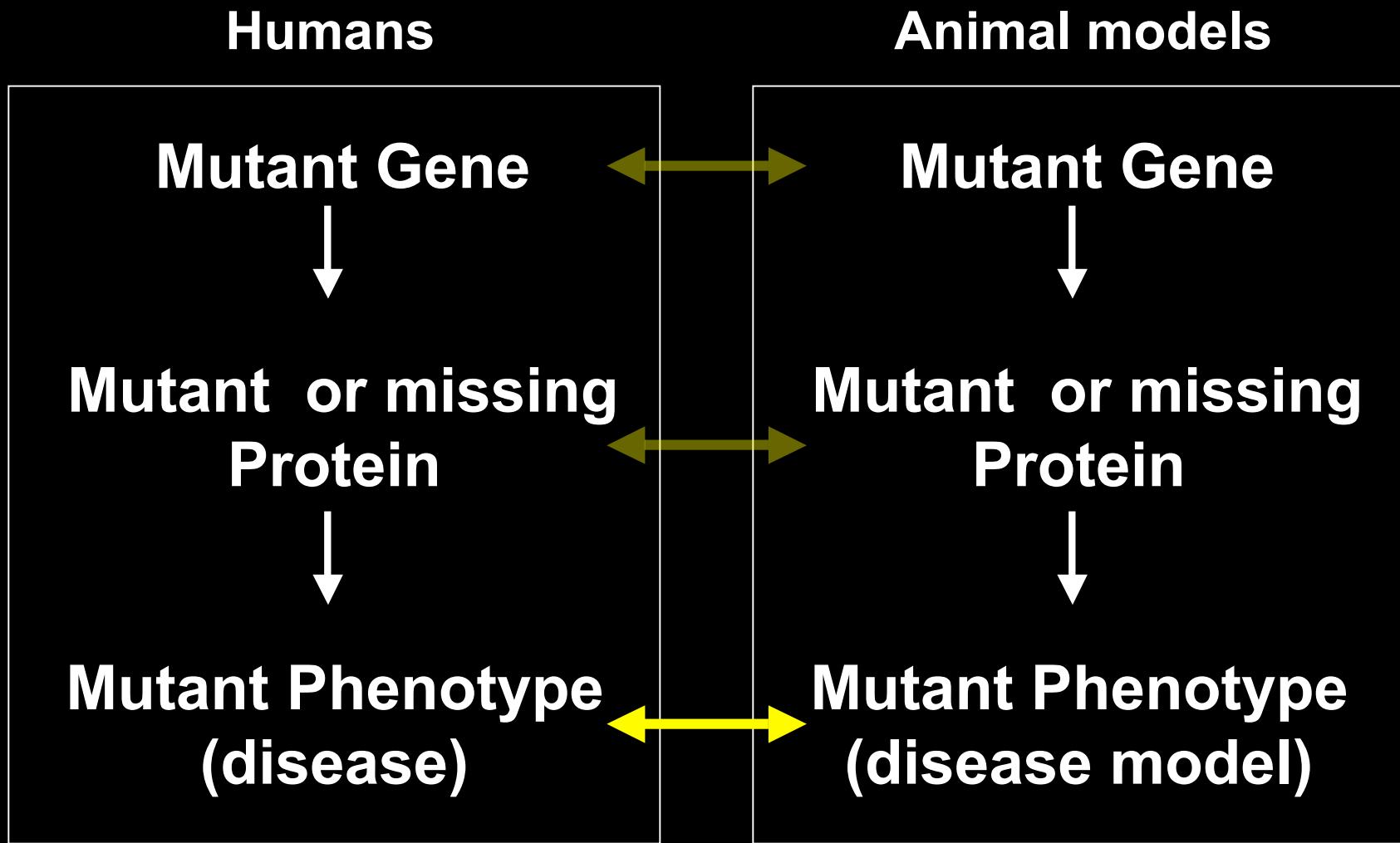


Animal disease models:



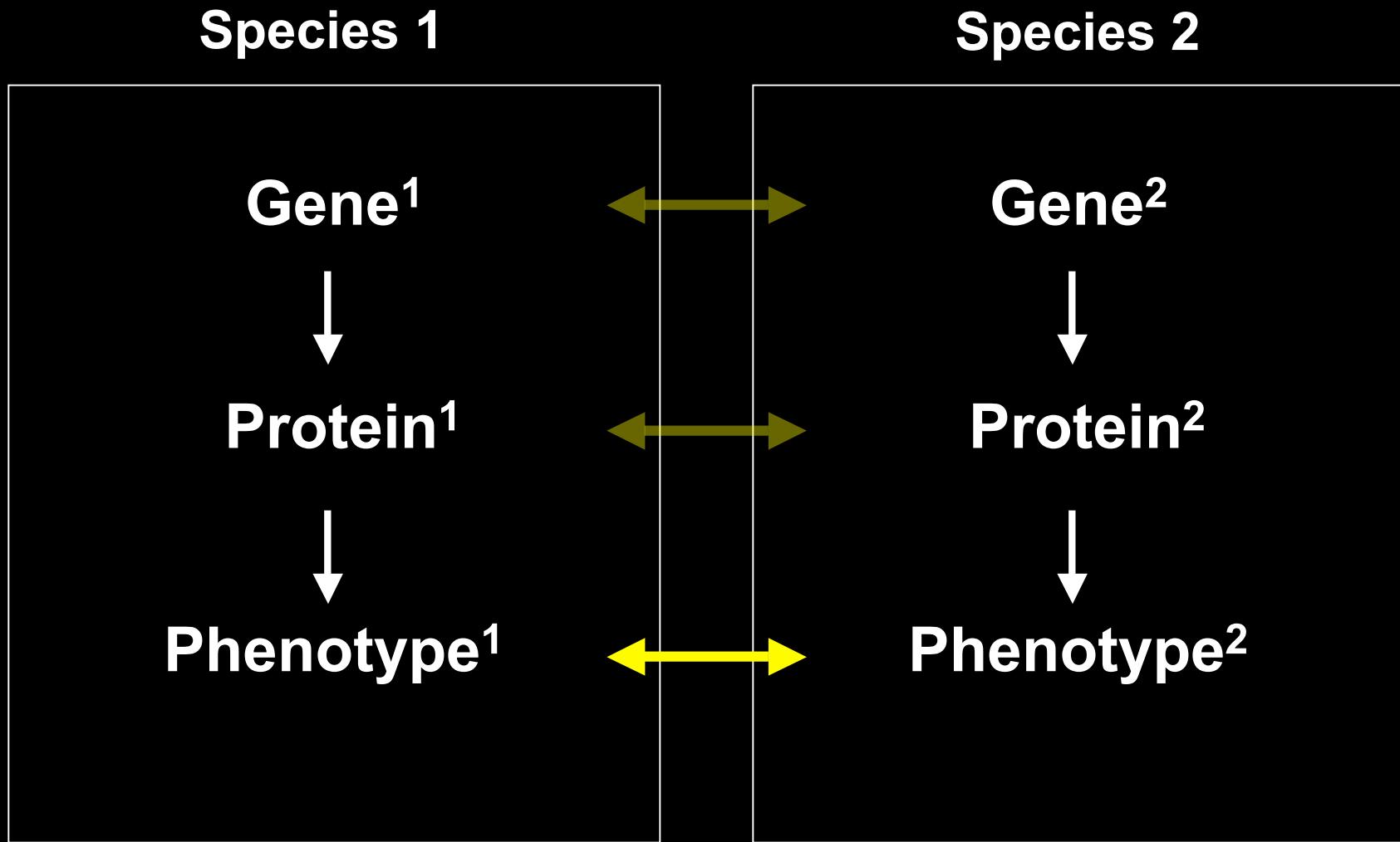


Animal disease models:



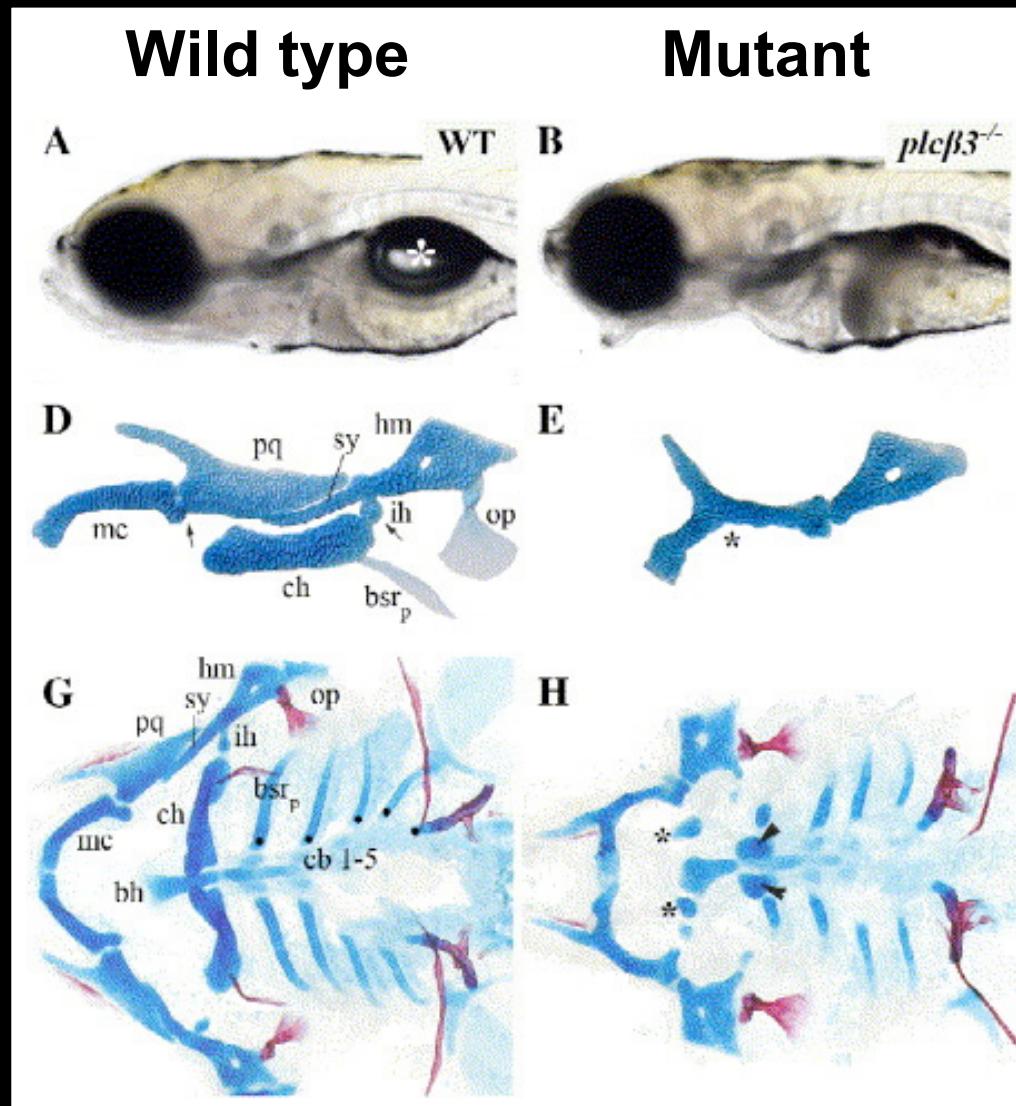


Animal diversity & evolution:



- 1. Animal models of human disease
(diversity & evolution)**
- 2. Introduction to ontologies**
- 3. Representing mutant phenotypes**

Phenotypes require complex descriptions



Vocabulary

Animal

Cranial skeleton

Dorsal mandibular arch

Head

Palatoquadrate cartilage

Pharyngeal arch

Ontology

Animal

■ Head

■ Cranial skeleton

■ Pharyngeal arch

■ Dorsal mandibular arch

■ Palatoquadrate cartilage

- 1. Animal models of human disease
(diversity & evolution)**
- 2. Introduction to ontologies**
- 3. Representing mutant phenotypes**

**1. Animal models of human disease
(diversity & evolution)**

2. Introduction to ontologies

3. Representing mutant phenotypes

- a. Ontologies**
- b. Syntax**



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Developmental Biology 304 (2007) 194–207

DEVELOPMENTAL
BIOLOGY

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phospholipase C, beta 3 is required for Endothelin1 regulation of pharyngeal arch patterning in zebrafish

Macie B. Walker^{*,1}, Craig T. Miller², Mary E. Swartz, Johann K. Eberhart, Charles B. Kimmel

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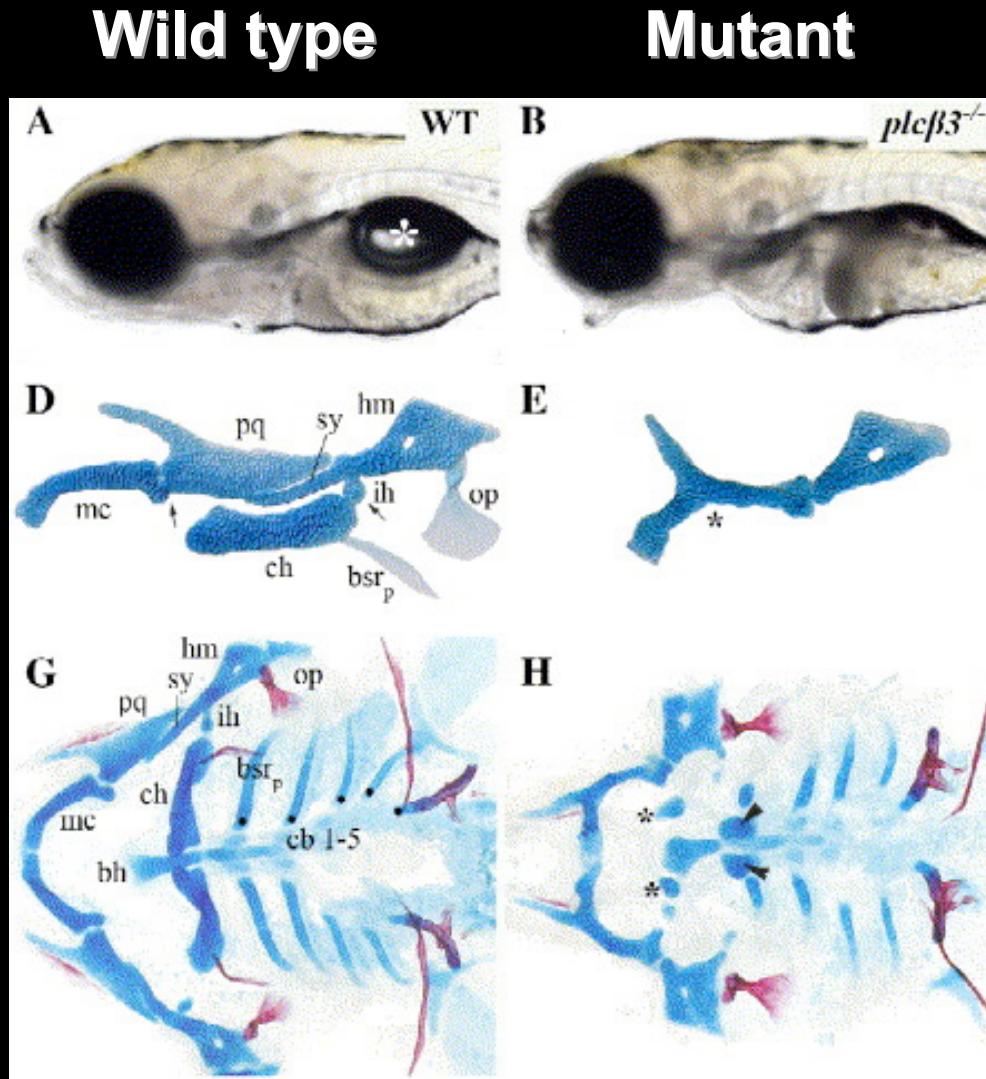
Available online 19 December 2006

Abstract

Genetic and pharmacological studies demonstrate that Endothelin1 (Edn1) is a key signaling molecule for patterning the facial skeleton in fish, chicks, and mice. When Edn1 function is reduced early in development the ventral lower jaw and supporting structures are reduced in size and often fused to their dorsal upper jaw counterparts. We show that *schmerle* (*she*) encodes a zebrafish ortholog of Phospholipase C, beta 3 (Plc β 3) required in cranial neural crest cells for Edn1 regulation of pharyngeal arch patterning. Sequencing and co-segregation demonstrates that two independent *she* (*plc β 3*) alleles have missense mutations in conserved residues within the catalytic domains of Plc β 3. Homozygous *plc β 3* mutants are phenotypically similar to *edn1* mutants and exhibit a strong arch expression defect in Edn1-dependent Distalless (Dlx) genes as well as expression defects in several Edn1-dependent intermediate and ventral arch domain transcription factors. *plc β 3* also genetically interacts with *edn1*, supporting a model in which Edn1 signals through a G protein-coupled receptor to activate Plc β 3. Mild skeletal defects occur in *plc β 3* heterozygotes, showing the *plc β 3* mutations are partially dominant. Through a morpholino-mediated deletion in the N-terminal PH domain of Plc β 3, we observe a partial rescue of facial skeletal defects in homozygous *plc β 3* mutants, supporting a hypothesis that an intact PH domain is necessary for the partial dominance we observe. In addition, through mosaic analyses, we show that wild-type neural crest cells can efficiently rescue facial skeletal defects in homozygous *plc β 3* mutants, demonstrating that Plc β 3 function is required in neural crest cells and not other cell types to pattern the facial skeleton.

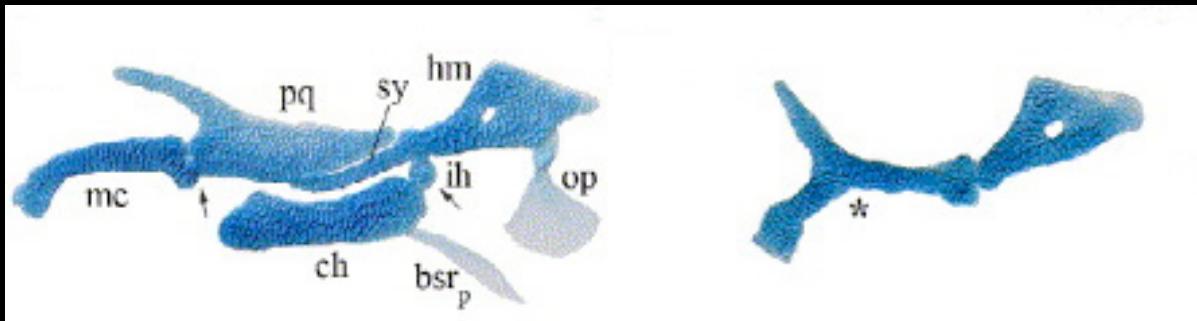
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Keywords: Dlx; Endothelin1; Neural crest; *plc β 3*; *schmerle*



“Mutations in the *plcβ3* gene result in smaller palatoquadrate and Meckel’s cartilages plus fusion of the joint”

Phenotype = entity + quality

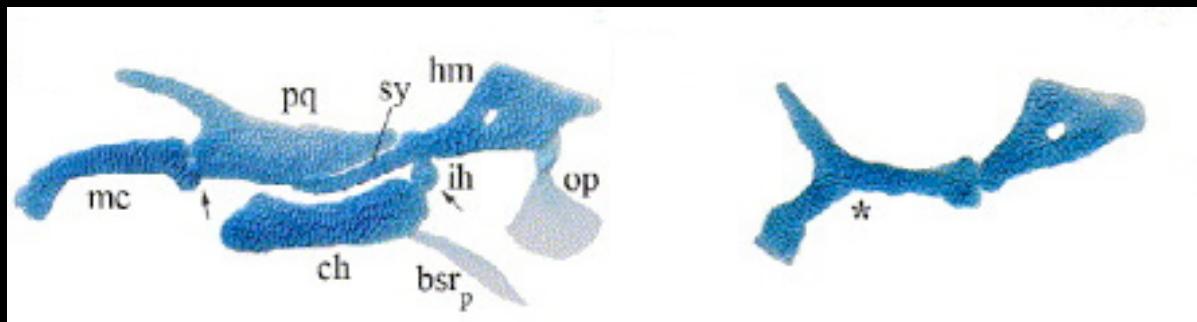


Wild type

plcβ3^{-/-}

Phenotype = entity + quality

P_1 = palatoquadrate + reduced



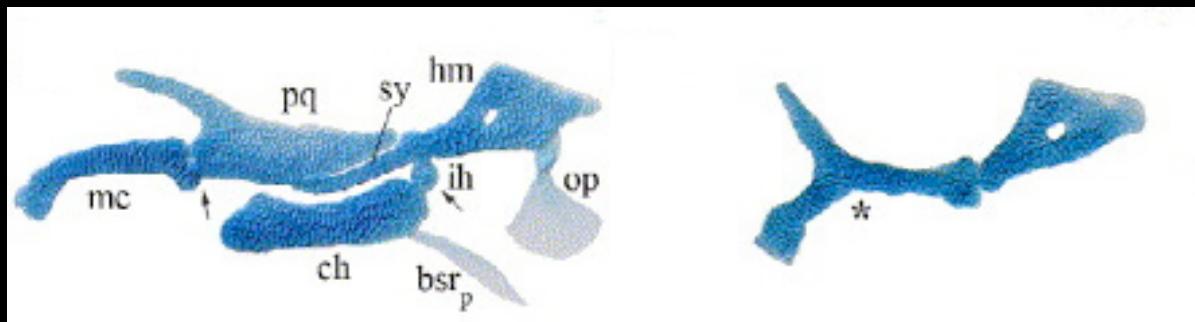
Wild type

plcβ3-/-

Phenotype = entity + quality

P_1 = palatoquadrate + reduced

P_2 = Meckel's + reduced



Wild type

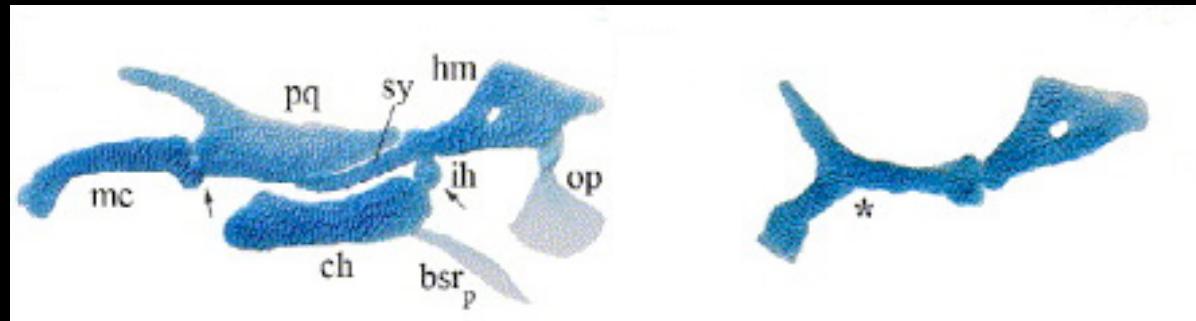
plcβ3^{-/-}

Phenotype = entity + quality

P_1 = palatoquadrate + reduced

P_2 = Meckel's + reduced

P_3 = Palatoquadrate/
Meckel's joint + fused



Wild type

plcβ3^{-/-}

Phenotype = entity + quality

P_1 = palatoquadrate + reduced

P_2 = Meckel's + reduced

P_3 = Palatoquadrate/
Meckel's joint + fused

ZAO:
palatoquadrate

Meckel's

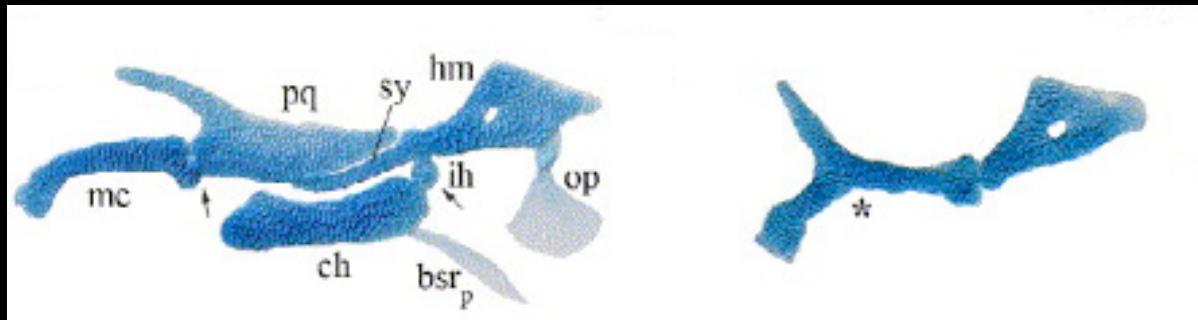
Palatoquadrate/
Meckel's joint

PATO:
reduced

reduced

fused

Wild type



Mutant



PUB=PMID:17239364

GT=plcb3[th210/th210] (AB)

E=ZAO:050711-2/***palatoquadrate***/

Q=PATO:0000638 /***reduced***/

T=during(ZFS:0000030) /***during (day 4)***/

Tag=PATO:0000460 /***abnormal***/

[Home](#)[Mutants / Transgenics](#)[BLAST](#) [Anatomy](#) [Publications](#) [People](#) [Labs](#) [Companies](#) [Acc #](#)[Wild-Types](#)[Genes / Markers / Clones](#)[Expression](#)[Maps](#)Site Search:

Search for mutant / transgenic lines

Your Input Welcome

Gene/Allele name

contains

LG: AnyMutagen: AnyMutation Type: Any

Affected Anatomy [Enter one anatomical term per line]

 Include substructures

Phenotype in:

- Every term entered
- Any term entered

Filters:

- Show only characterized mutations
- Show only transgenics
- Show all

20 results per page

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Site Search:

ZIRC

Search for mutant / transgenic lines

Your Input Welcome

Gene/Allele name

contains

LG: Any

Mutagen: Any

Mutation Type: Any

Affected Anatomy [Enter one anatomical term per line]

palat

- adductor arcus palatini
- communicating vessel palatocerebral artery
- levator arcus palatini
- median palatocerebral vein
- palatine
- palatocerebral artery
- palatocerebral artery.
- palatocerebral vein
- palatoquadrate arch
- palatoquadrate cartilage

Search by phenotype
type in:
any term entered
any term entered

Filters:

- Show only characterized mutations
- Show only transgenics
- Show all

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Site Search:

Mutant / Transgenic Lines Search Results (7 records found)

[Modify Search](#)

Your Input Welcome

| Genotype(Background) | Phenotype | Allele | Parental Zygosity | Type | Affected Gene(s) | LG |
|---|-----------------------------|----------|-------------------|----------------------|-----------------------------|----------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | 3 figure(s) | | | deficiency | sox9b, sox8 | 3 Details |
| rereb^{ru622/ru622} | 1 figure(s) | ru622 | | point mutation | rereb | 23 Details |
| sox9a^{hi1134Tg/+;sox9a^{tw37/+}} | 1 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| | | tw37 | | unknown | sox9a | 12 Details |
| sox9a^{hi1134Tg/hi1134Tg} | 1 figure(s) | hi1134Tg | ♀ +/- ♂ +/- | transgenic insertion | sox9a | 12 Details |
| sox9a^{hi1134Tg} | 3 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| sox9a^{tw37/+} | 1 figure(s) | tw37 | | unknown | sox9a | 12 Details |
| sox9a^{tw37/tw37} | 5 figure(s) | tw37 | ♀ +/- ♂ +/- | unknown | sox9a | 12 Details |

Modify your search

Your Input Welcome

Gene/Allele name contains LG: Any Mutagen: Any Mutation Type: Any

Affected Anatomy [Enter one anatomical term per line]

palatograde cartilage

 Include substructures

Phenotype in:

- Every term entered
- Any term entered

Filters:

- Show only characterized mutations
- Show only transgenics
- Show all

20 results per page



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Your Input Welcome

| Genotype(Background) | Phenotype | Allele | Parental Zygosity | Type | Affected Gene(s) | LG |
|---|-----------------------------|----------|-------------------|----------------------|-----------------------------|----------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | 3 figure(s) | | | deficiency | sox9b, sox8 | 3 Details |
| rereb^{ru622/ru622} | 1 figure(s) | ru622 | | point mutation | rereb | 23 Details |
| sox9a^{hi1134Tg/+;sox9a^{tw37/+}} | 1 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| | | tw37 | | unknown | sox9a | 12 Details |
| sox9a^{hi1134Tg/hi1134Tg} | 1 figure(s) | hi1134Tg | ♀ +/- ♂ +/- | transgenic insertion | sox9a | 12 Details |
| sox9a^{hi1134Tg} | 3 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| sox9a^{tw37/+} | 1 figure(s) | tw37 | | unknown | sox9a | 12 Details |
| sox9a^{tw37/tw37} | 5 figure(s) | tw37 | ♀ +/- ♂ +/- | unknown | sox9a | 12 Details |

Modify your search

Your Input Welcome

Gene/Allele name contains LG: Any Mutagen: Any Mutation Type: Any

Affected Anatomy [Enter one anatomical term per line]

 palatoguadrate cartilage

Filters:

- Show only characterized mutations
- Show only transgenics
- Show all

20 results per page

 Include substructures

Phenotype in:

- Every term entered
- Any term entered

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Mutant / Transgenic Lines Search Results (7 records found)

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| Genotype(Background) | Phenotype | Allele | Parental Zygosity | Type | Affected Gene(s) | LG |
|---|-----------------------------|----------|-------------------|----------------|-----------------------------|----------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | 3 figure(s) | | | deficiency | sox9b, sox8 | 3 Details |
| rereb^{ru622/ru622} | 1 figure(s) | ru622 | | point mutation | rereb | 23 Details |
| sox9a^{hi1134Tg/+;sox9a^{tw37/+}} | 1 figure(s) | hi1134Tg | | transgenic | sox9a | 12 Details |
| sox9a^{hi1134Tg/hi1134Tg} | | | | | | 2 Details |
| sox9a^{hi1134Tg} | | | | | | 2 Details |
| sox9a^{tw37/+} | | | | | | 2 Details |
| sox9a^{tw37/tw37} | | | | | | 2 Details |

[Modify your search](#)Gene/Allele name (contains)

Affected Anatomy [Enter one anatomical term per line]

palatoquadrate cartilage

 Include substructures

Phenotype in:

- Every term entered
 Any term entered

Mutation Type:

Filters:

- Show only characterized mutations
 Show only transgenics
 Show all

20 results per page

Affected Anatomy [Enter one anatomical term per line]

palatoquadrate cartilage

 Include substructures

Phenotype in:

- Every term entered
 Any term entered

Mutant / Transgenic Lines Search Results (7 records found)

[Modify Search](#)

Your Input Welcome

Genotype(Background)

[Df\(LG03:sox8,sox9b\)b971/b971](#)

Phenotype

[3 figure\(s\) !\[\]\(847172ace9f417f0ef2d71cc34021152_img.jpg\)](#)[rereb^{ru622/ru622}](#)[1 figure\(s\)](#)[sox9a^{hi1134Tg/+;sox9a^{tw37/+}}](#)[1 figure\(s\)](#)[sox9a^{hi1134Tg/hi1134Tg}](#)[1 figure\(s\)](#)[sox9a^{hi1134Tg}](#)[3 figure\(s\) !\[\]\(7ac9149c311be44e3668c36226e60d19_img.jpg\)](#)[sox9a^{tw37/+}](#)[1 figure\(s\)](#)[sox9a^{tw37/tw37}](#)[5 figure\(s\) !\[\]\(fc8805fd82493e66a74a5840b065b7ae_img.jpg\)](#)

All mutations that affect this cartilage

Affected Gene(s)

[sox9b, sox8](#)3 [Details](#)[rereb](#)23 [Details](#)[sox9a](#)12 [Details](#)[hi1134Tg](#)

transgenic insertion

[tw37](#)

unknown

[hi1134Tg](#)[♀ +/- ♂ +/-](#)[hi1134Tg](#)

transgenic insertion

[tw37](#)

unknown

[tw37](#)[♀ +/- ♂ +/-](#)[tw37](#)

unknown

[sox9a](#)12 [Details](#)[sox9a^{tw37/tw37}](#)[♀ +/- ♂ +/-](#)[tw37](#)

unknown

[sox9a](#)12 [Details](#)

Modify your search

Your Input Welcome

Gene/Allele name contains LG: Mutagen: Mutation Type:

Affected Anatomy [Enter one anatomical term per line]

palatoguadrate cartilage

 Include substructures

Phenotype in:

- Every term entered
- Any term entered

Filters:

- Show only characterized mutations
- Show only transgenics
- Show all

20 results per page

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Site Search:

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Your Input Welcome

| Genotype(Background) | Phenotype | Allele | Parental Zygosity | Type | Affected Gene(s) | LG |
|---|-----------------------------|----------|-------------------|----------------------|-----------------------------|----------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | 3 figure(s) | | | deficiency | sox9b, sox8 | 3 Details |
| rereb^{ru622/ru622} | 1 figure(s) | ru622 | | point mutation | rereb | 23 Details |
| sox9a^{hi1134Tg/+;sox9a^{tw37/+}} | 1 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| | | tw37 | | unknown | sox9a | 12 Details |
| sox9a^{hi1134Tg/hi1134Tg} | 1 figure(s) | hi1134Tg | ♀ +/- ♂ +/- | transgenic insertion | sox9a | 12 Details |
| sox9a^{hi1134Tg} | 3 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| sox9a^{tw37/+} | 1 figure(s) | tw37 | | unknown | sox9a | 12 Details |
| sox9a^{tw37/tw37} | 5 figure(s) | tw37 | ♀ +/- ♂ +/- | unknown | sox9a | 12 Details |

Modify your search

Your Input Welcome

Gene/Allele name contains LG: Any Mutagen: Any Mutation Type: Any

Affected Anatomy [Enter one anatomical term per line]

palatograde cartilage

Filters:

- Show only characterized mutations
- Show only transgenics
- Show all

20 results per page

 Include substructures

Phenotype in:

- Every term entered
- Any term entered

SEARCH

RESET

[Modify Search](#)
[Your Input Welcome](#)

Mutant / Transgenic Lines Search Results (7 records found)

| Genotype(Background) | Phenotype | Allele | Parental Zygosity | Type | Affected Gene(s) | LG |
|---|------------------------------|----------|-------------------|----------------------|-----------------------------|----------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | 3 figure(s) | | | deficiency | sox9b, sox8 | 3 Details |
| rereb^{ru622}/ru622 | 1 figure(s) | ru622 | | point mutation | rereb | 23 Details |
| sox9a^{hi1134Tg/+},sox9a^{tw37/+} | 1 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| | | tw37 | | unknown | sox9a | 12 Details |
| sox9a^{hi1134Tg/hi1134Tg} | 1 figure(s) | hi1134Tg | ♀ +/- ♂ +/- | transgenic insertion | sox9a | 12 Details |
| sox9a^{hi1134Tg} | 3 figure(s) | hi1134Tg | | transgenic insertion | sox9a | 12 Details |
| sox9a^{tw37/+} | | | | | | 12 Details |
| sox9a^{tw37/tw37} | | | | | | 12 Details |

[sox9a^{hi1134Tg}](#)

View
phenotype

[3 figure\(s\)](#)
[Modify your search](#)
[Gene/Allele name](#) contains

[Mutation Type:](#) Any

Affected Anatomy [Enter one anatomical term per line]

palatoquadrata cartilage

 Include substructures

Phenotype in:

- Every term entered
- Any term entered

Filters:

- Show only characterized mutations
- Show only transgenics
- Show all

20 results per page

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ZFIN ID: ZDB-FIG-050323-3

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Yan et al., 2005 - A pair of Sox: distinct and overlapping functions of zebrafish sox9 co-orthologs in craniofacial and pectoral fin development. Development 132(5):1069-1083 - [Full text @ Development](#)

ADDITIONAL FIGURES

PHENOTYPE:

Genotype(s): [Df\(LG03:sox8,sox9b\)b971/b971](#) ▾, [Df\(LG03:sox8,sox9b\)b971/b971;sox9a^{hi1134Tg/hi1134Tg}](#) ▾, [sox9a^{hi1134Tg}](#) ▾

Observed In: [Meckels cartilage](#), [ceratobranchial](#), [ceratohyal cartilage](#), [dentary](#) ... (all 14) ▶

Stage Range : [Day 4](#)

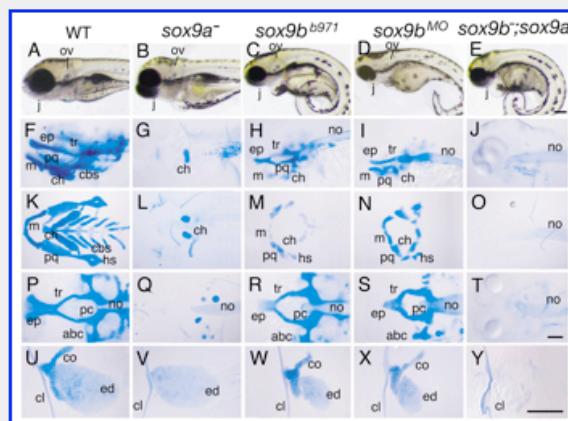


Fig. 3 Wild-type, sox9 mutants, and sox9b-morpholino-treated animals. (A-E) Live larvae; (F-J) lateral view of head of Alcian-stained larvae. (K-O) Flat mount of Alcian-stained pharyngeal arches. (P-T) Flat mount of Alcian-stained neurocranium. (U-Y) Alcian-stained pectoral fin bud. All animals at 4 dpf. (A,F,K,P,U) Wild type; (B,G,L,Q,V) sox9a mutant, showing deletion of most cranial cartilage and scapulocoracoid; (C,H,M,R,W) sox9b^{b971}; neurocranium, arches, and endochondral disc reduced; (D,I,N,S,X) sox9b MO-injected animals phenotypically similar to sox9b^{b971}; (E,J,O,T,Y) double mutant, arches, neurocranium, and fin deleted. Abbreviations: abc, anterior basicranial commissure; cbs, ceratobranchials; ch, ceratohyal, m, Meckel's cartilage; cl, cleithrum; co, scapulocoracoid; ed, endochondral disc; ep, ethmoid plate; hs, hyosympathetic; j, jaw; ov, otic vesicles; no, notochord; pc, parachordal; pq, palatoquadrate; tr, trabecula. Scale bars: in E, 100 µm for A-E; in T, 100 µm for F-T; in Y, 100 µm for U-Y.

Phenotype details

| Fish | Stage | Observed in | Phenotype |
|--|-----------------------|--|--------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | Day 4 | ceratobranchial | absent, abnormal |
| | Day 4 | Meckel's cartilage | decreased size, abnormal |
| | Day 4 | palatoquadrate cartilage | decreased size, abnormal |
| | Day 4 | ceratohyal cartilage | decreased size, abnormal |
| | Day 4 | hyosympathetic cartilage | decreased size, abnormal |

ZFIN ID: ZDB-FIG-050323-3

Your Input Welcome

[Yan et al., 2005](#) - A pair of Sox: distinct and overlapping functions of zebrafish sox9 co-orthologs in craniofacial and pectoral fin development. Development 132(5):1069-1083 - [Full text @ Development](#)

ADDITIONAL FIGURES

PHENOTYPE:

Genotype(s): [Df\(LG03:sox8,sox9b\)b971/b971](#), [Df\(LG03:sox8,sox9b\)b971/b971;sox9a^{hi1134Tg/hi1134Tg}](#), [sox9a^{hi1134Tg}](#)

Observed In: [Meckels cartilage](#), [ceratobranchial](#), [ceratohyal cartilage](#), [dentary](#) ... (all 14) ▶

Stage Range : [Day 4](#)

Description of phenotype

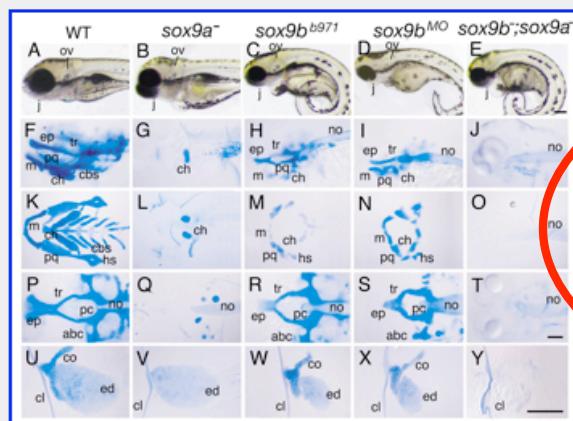


Fig. 3 Wild-type, sox9 mutants, and sox9b-morpholino-treated animals. (A-E) Live larvae; (F-J) lateral view of head of Alcian-stained larvae. (K-O) Flat mount of Alcian-stained pharyngeal arches. (P-T) Flat mount of Alcian-stained neurocranium. (U-Y) Alcian-stained pectoral fin bud. All animals at 4 dpf. (A,F,K,P,U) Wild type; (B,G,L,Q,V) sox9a mutant, showing deletion of most cranial cartilage and scapulocoracoid; (C,H,M,R,W) sox9b^{b971}; neurocranium, arches, and endochondral disc reduced; (D,I,N,S,X) sox9b MO-injected animals phenotypically similar to sox9b^{b971}; (E,J,O,T,Y) double mutant, arches, neurocranium, and fin deleted. Abbreviations: abc, anterior basicranial commissure; cbs, ceratobranchials; ch, ceratohyal, m, Meckel's cartilage; cl, cleithrum; co, scapulocoracoid; ed, endochondral disc; ep, ethmoid plate; hs, hyosympathetic; j, jaw; ov, otic vesicles; no, notochord; pc, parachordal; pq, palatoquadrate; tr, trabecula. Scale bars: in F, 100 µm for A-E; in T, 100 µm for F-T; in Y, 100 µm for U-Y.

Phenotype details

| Fish | Stage | Observed in | Phenotype |
|--|-----------------------|--|--------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | Day 4 | ceratobranchial | absent, abnormal |
| | Day 4 | Meckel's cartilage | decreased size, abnormal |
| | Day 4 | palatoquadrate cartilage | decreased size, abnormal |
| | Day 4 | ceratohyal cartilage | decreased size, abnormal |
| | Day 4 | hyosympathetic cartilage | decreased size, abnormal |

ZFIN ID: ZDB-FIG-050323-3

Your Input Welcome

[Yan et al., 2005](#) - A pair of Sox: distinct and overlapping functions of zebrafish sox9 co-orthologs in craniofacial and pectoral fin development. Development 132(5):1069-1083 - [Full text @ Development](#)

ADDITIONAL FIGURES

PHENOTYPE:

Genotype(s): [Df\(LG03:sox8,sox9b\)b971/b971](#) ▾, [Df\(LG03:sox8,sox9b\)b971/b971;sox9a^{hi1134Tg/hi1134Tg}](#) ▾, [sox9a^{hi1134Tg}](#) ▾

Observed In: Meckels cartilage, ceratobranchial, ceratohyal cartilage, dentary... (cell 14) ▾

Stage Range : Day 4

Genes involved

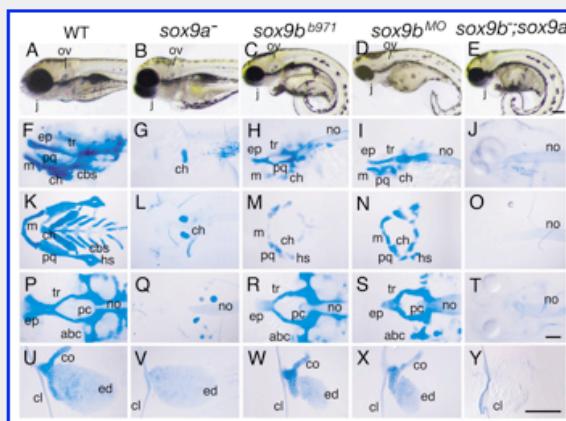


Fig. 3 Wild-type, sox9 mutants, and sox9b-morpholino-treated animals. (A-E) Live larvae; (F-J) lateral view of head of Alcian-stained larvae. (K-O) Flat mount of Alcian-stained pharyngeal arches. (P-T) Flat mount of Alcian-stained neurocranium. (U-Y) Alcian-stained pectoral fin bud. All animals at 4 dpf. (A,F,K,P,U) Wild type; (B,G,L,Q,V) sox9a mutant, showing deletion of most cranial cartilage and scapulocoracoid; (C,H,M,R,W) sox9b^{b971}; neurocranium, arches, and endochondral disc reduced; (D,I,N,S,X) sox9b MO-injected animals phenotypically similar to sox9b^{b971}; (E,J,O,T,Y) double mutant, arches, neurocranium, and fin deleted. Abbreviations: abc, anterior basicranial commissure; cbs, ceratobranchials; ch, ceratohyal, m, Meckel's cartilage; cl, cleithrum; co, scapulocoracoid; ed, endochondral disc; ep, ethmoid plate; hs, hyosympathetic; j, jaw; ov, otic vesicles; no, notochord; pc, parachordal; pq, palatoquadrate; tr, trabecula. Scale bars: in E, 100 µm for A-E; in T, 100 µm for F-T; in Y, 100 µm for U-Y.

Phenotype details

| Fish | Stage | Observed in | Phenotype |
|--|-----------------------|--|--------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | Day 4 | ceratobranchial | absent, abnormal |
| | Day 4 | Meckel's cartilage | decreased size, abnormal |
| | Day 4 | palatoquadrate cartilage | decreased size, abnormal |
| | Day 4 | ceratohyal cartilage | decreased size, abnormal |
| | Day 4 | hyosympathetic cartilage | decreased size, abnormal |

ZFIN ID: ZDB-FIG-050323-3

Your Input Welcome

Yan et al., 2005 - A pair of Sox: distinct and overlapping functions of zebrafish sox9 co-orthologs in craniofacial and pectoral fin development. Development 132(5):1069-1083 - [Full text @ Development](#)

ADDITIONAL FIGURES

PHENOTYPE:

Genotype(s): [Df\(LG03:sox8,sox9b\)b971/b971](#), [Df\(LG03:sox8,sox9b\)b971/b971;sox9a^{hi1134Tg/hi1134Tg}](#), [sox9a^{hi1134Tg}](#)

Observed In: [Meckels cartilage](#), [ceratobranchial](#), [ceratohyal cartilage](#), [dentary](#) ... (all 14) ▶

Stage Range : [Day 4](#)

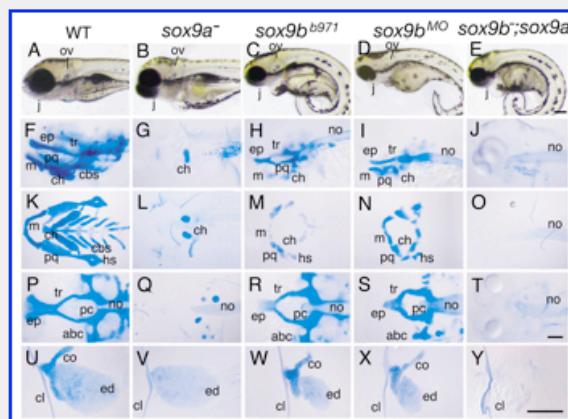


Fig. 3 Wild-type, sox9 mutants, and sox9b-morpholino-treated animals. (A-E) Live larvae; (F-J) lateral view of head of Alcian-stained larvae. (K-O) Flat mount of Alcian-stained pharyngeal arches. (P-T) Flat mount of Alcian-stained neurocranium. (U-Y) Alcian-stained pectoral fin bud. All animals at 4 dpf. (A,F,K,P,U) Wild type; (B,G,L,Q,V) sox9a mutant, showing deletion of most cranial cartilage and scapulocoracoid; (C,H,M,R,W) sox9b^{b971}; neurocranium, arches, and endochondral disc reduced; (D,I,N,S,X) sox9b MO-injected animals phenotypically similar to sox9b^{b971}; (E,J,O,T,Y) double mutant, arches, neurocranium, and fin deleted. Abbreviations: abc, anterior basicranial commissure; cbs, ceratobranchials; ch, ceratohyal, m, Meckel's cartilage; cl, cleithrum; co, scapulocoracoid; ed, endochondral disc; ep, ethmoid plate; hs, hyosympathetic; j, jaw; ov, otic vesicles; no, notochord; pc, parachordal; pq, palatoquadrate; tr, trabecula. Scale bars: in E, 100 µm for A-E; in T, 100 µm for F-T; in Y, 100 µm for U-Y.

Phenotype details

| Fish | Stage | Observed in | Phenotype |
|--|-----------------------|--|--------------------------|
| Df(LG03:sox8,sox9b)b971/b971 | Day 4 | ceratobranchial | absent, abnormal |
| | Day 4 | Meckel's cartilage | decreased size, abnormal |
| | Day 4 | palatoquadrate cartilage | decreased size, abnormal |
| | Day 4 | ceratohyal cartilage | decreased size, abnormal |
| | Day 4 | hyosympathetic cartilage | decreased size, abnormal |

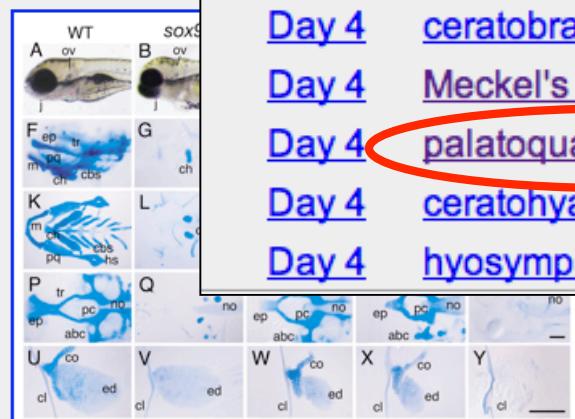
ZFIN ID: ZDB-FIG-050323-3

[Yan et al., 2005](#) - A pair of Sox: distinct and overlapping functions of zebrafish sox9 co-orthologs in craniofacial and pectoral fin development. Development 132(5):1069-1083 - [Full text @ Development](#)

Your Input Welcome

[ADDITIONAL FIGURES](#)

PHENOTYPE:

Genotype(s): [Df\(LG03:sox8,sox9b\)b971/b971](#) ▾, [Df\(LG03:](#)Observed In: [Meckel's cartilage ceratobranchial ceratohyal](#)Stage Range : [D](#)

Find everything about this cartilage

[i1134Tg](#) ▾

| Stage | Observed in | Phenotype |
|-----------------------|--|--------------------------|
| Day 4 | ceratobranchial | absent, abnormal |
| Day 4 | Meckel's cartilage | decreased size, abnormal |
| Day 4 | palatoquadrate cartilage | decreased size, abnormal |
| Day 4 | ceratohyal cartilage | decreased size, abnormal |
| Day 4 | hyosymplectic cartilage | decreased size, abnormal |

m, Meckel's cartilage; cl, cleithrum; co, scapulocoracoid; ed, endochondral disc; ep, ethmoid plate; hs, hyosymplectic; j, jaw; ov, otic vesicles; no, notochord; pc, parachordal; pq, palatoquadrate; tr, trabecula. Scale bars: in E, 100 µm for A-E; in T, 100 µm for F-T; in Y, 100 µm for U-Y.

Phenotype details

Fish

[Df\(LG03:sox8,sox9b\)b971/b971](#)

| Stage | Observed in | Phenotype |
|-----------------------|--|--------------------------|
| Day 4 | ceratobranchial | absent, abnormal |
| Day 4 | Meckel's cartilage | decreased size, abnormal |
| Day 4 | palatoquadrate cartilage | decreased size, abnormal |
| Day 4 | ceratohyal cartilage | decreased size, abnormal |
| Day 4 | hyosymplectic cartilage | decreased size, abnormal |



Research

General Information

Site Search:

ZIRC

Home Genes / Markers / Clones Expression BLAST Mutants / Tg Anatomy Maps Publications

ZFIN ID: ZDB-ANAT-060331-24

Your Input Welcome

Name: *palatoquadrate cartilage***Synonyms:** quadrate cartilage , pterygoquadrate**Appears at****Evident until**

Unknown

[Adult](#) (90d-730d, breeding adult)**Relationships**Contained by: [dorsal mandibular arch](#)

Contains:

Develops from:

Develops into: [autopalatine](#) [metapterygoid](#) [quadrate](#)**All Expressed Genes**[col10a1](#) , [sox9a](#)**All Mutants / Transgenic lines**[Df\(LG03:sox8,sox9b\)b971/b971](#) , [rereb^{ru622/ru622}](#) , [sox9a^{hi1134Tg/+;sox9a^{tw37/+}}](#) ,
[sox9a^{hi1134Tg/hi1134Tg}](#) , [sox9a^{hi1134Tg}](#) , [sox9a^{tw37/+} ... 7 total \(7 if substructures included\)](#)

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Research

General Information

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ZIRC

Home Genes / Markers / Clones Expression BLAST Mutants / Tg Anatomy Maps Publications

ZFIN ID: ZDB-ANAT-060331-24

Your Input Welcome

Name: *palatoquadrate cartilage***Synonyms:** quadrate cartilage , pterygoquadrate**Appears at****Evident until**

Unknown

[Adult](#) (90d-730d, breeding adult)**Relationships**Contained by: [dorsal mandibular arch](#)

Contains:

Develops from:

Develops into: [autopalatine](#) [metapterygoid](#) [quadrata](#)

All mutations that
affect this cartilage

All Expressed Genes[col10a1](#) , [sox9a](#)**All Mutants / Transgenic lines**

[Df\(LG03:sox8.sox9b\)b971/b971](#) , [rereb^{ru622/ru622}](#) , [sox9a^{hi1134Tg/+;sox9a^{tw37/+}}](#) ,
[sox9a^{hi1134Tg/hi1134Tg}](#) , [sox9a^{hi1134Tg}](#) , [sox9a^{tw37/+} ... 7 total](#) (7 if substructures included)

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Research General Information ZIRC

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Site Search:

ZFIN ID: ZDB-ANAT-060331-24

Your Input Welcome

Name: *palatoquadrate cartilage*

Synonyms: quadrate cartilage , pterygoquadrate

Appears at

Unknown

Evident until

[Adult](#) (90d-730d, breeding adult)

Relationships

Contained by: [dorsal mandibular arch](#)

Contains:

Develops from:

Develops into: [autopalatine](#) [metapterygoid](#) [quadrate](#)

Ontological
relationships

All Expressed Genes

[col10a1](#) , [sox9a](#)

All Mutants / Transgenic lines

[Df\(LG03:sox8.sox9b\)b971/b971](#) , [rereb^{ru622/ru622}](#) , [sox9a^{hi1134Tg/+;sox9a^{tw37/+}}](#) ,
[sox9a^{hi1134Tg/hi1134Tg}](#) , [sox9a^{hi1134Tg}](#) , [sox9a^{tw37/+}](#) ... [7 total](#) ([7](#) if substructures included)

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Phenotype
(clinical sign) = entity + quality

Anatomical ontology (CARO)

Cell & tissue ontology

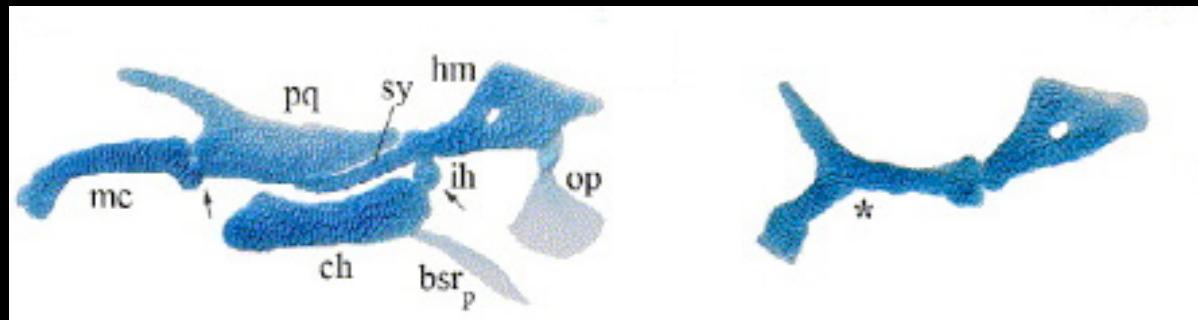
Developmental ontology + **PATO**

Gene ontology

biological process

molecular function

cellular component





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Research

General Information

Site Search:

ZIRC

ZFIN ID: ZDB-GENE-030421-3

Gene Name: *phospholipase C, gamma 1*

Your Input Welcome

Gene Symbol: *plcg1*

Previous Names: fj46f04, ded, wu:fj57a05, wu:fj46f04(1), dead beat

[Nomenclature History](#)

GENE EXPRESSION: ([current status](#))

All expression data: [5 figure\(s\)](#) from 3 publications

Wild Type Stages, Structures: [Zygote:1-cell](#) (0.00h-0.75h) to [Adult](#) (90d-730d, breeding adult)
[brain](#), [heart](#) ... ([all 7](#)) ▶

MUTANTS AND TARGETED KNOCKDOWNS:

Mutant line(s): [7 genotypes \(5 alleles\)](#)

Phenotype: ([current status](#))

Data: [5 figure\(s\)](#) from 2 publications

Observed in: [anatomical system](#), [circulation](#), [eye](#), [heart](#), [nucleate erythrocyte](#) ... ([all 7](#)) ▶

Knockdown reagents: [MO1-plcg1 \(1\)](#), [MO2-plcg1 \(1\)](#), [MO3-plcg1 \(1\)](#)

GENE PRODUCTS:

Gene Ontology

[Ontology](#)

[GO Term](#)

Molecular Function [calcium ion binding](#) ([more](#))

Biological Process [fin regeneration](#) ([more](#))

SEQUENCE INFORMATION:

| Type | Accession # | Length | Analysis |
|---|--------------------------------------|---------|-------------------|
| cDNA: | RefSeq:NM_194407 (2) | 4103 bp | - Select Tool - ▾ |
| Genomic: | GenBank:AY163169 (1) | 618 bp | - Select Tool - ▾ |
| Polypeptide: | UniProt:Q804J6 (1) | 1312 aa | - Select Tool - ▾ |
| Sequence Clusters: | UniGene:120304 (1) | | |
| All Sequence Information (16) | | | |

OTHER *p1cg1* GENE / MARKER PAGES:

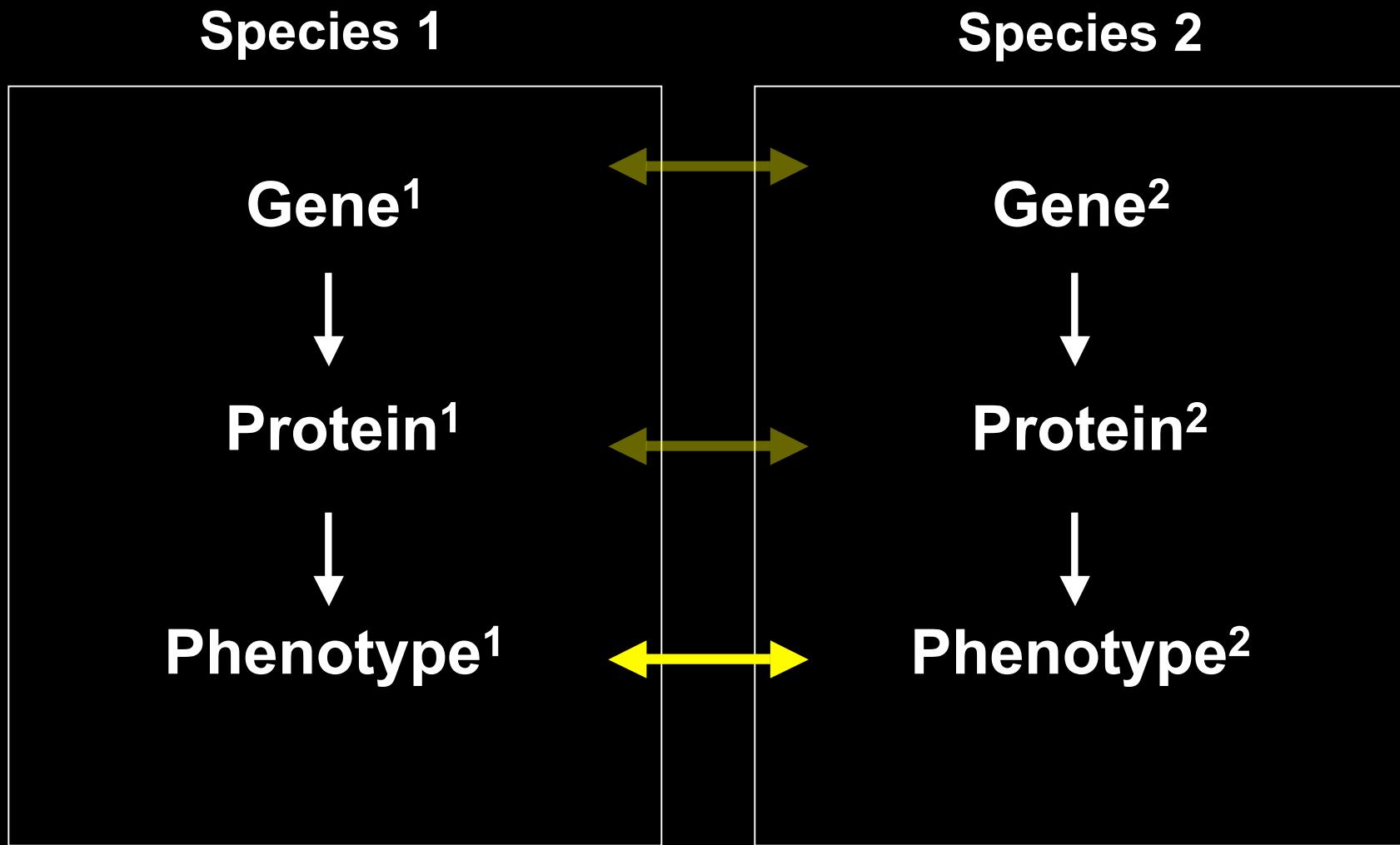
- [Entrez Gene:373867 \(1\)](#)
- [Ensembl\(Zv7\):ENSDARG00000038442 \(1\)](#)

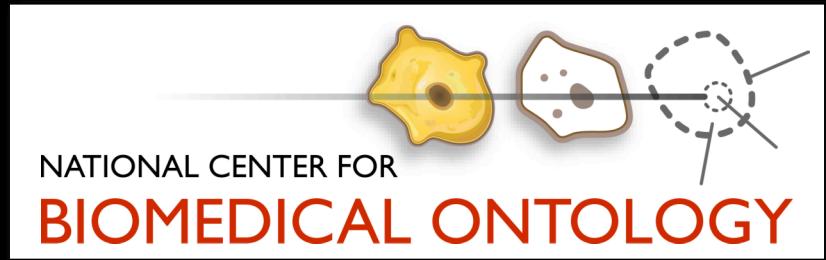
MAPPING INFORMATION:LG: 23 [Details](#)**ORTHOLOGY:**

| Species | Symbol | Chromosome (Position) | Accession # | Evidence |
|-----------------------------------|--------|-----------------------|---|----------|
| Zebrafish | plcg1 | 23 | | CL |
| Human | PLCG1 | 20 (q12-q13.1) | • OMIM:172420 • Entrez Gene:5335 | CL |
| Orthology Details | | | | |

CITATIONS (19)

Animal diversity & evolution:





**Supported by NIH P41 HG002659 and U54 HG004028, NSF DBI-0641025,
Cambridge University, Stanford University, NESCent &
the University of Oregon**