



Linking Evolution to Genomics: Motivation, Approach, Project Goals

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Workshop outcome:

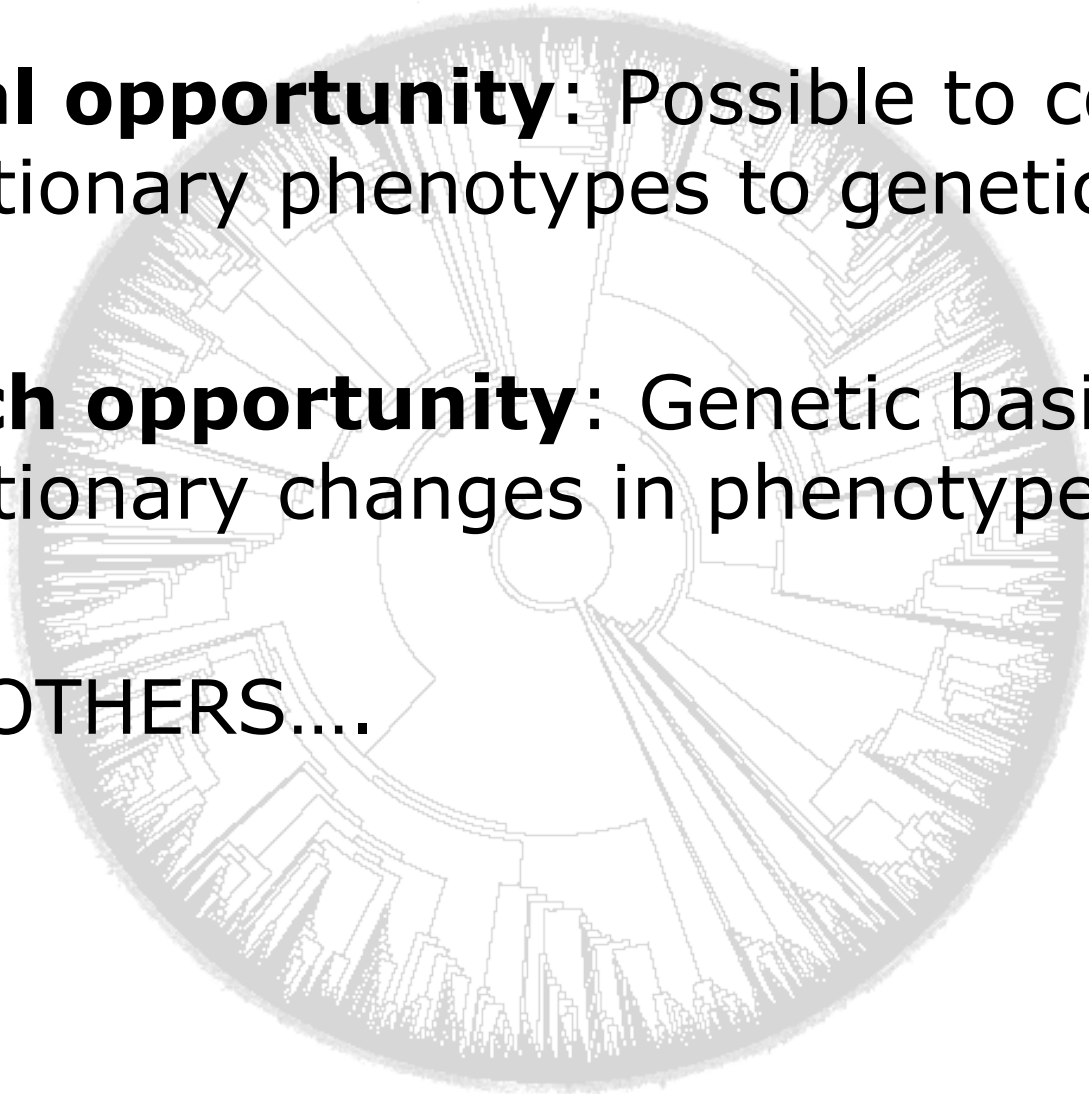
- ❑ What research questions would you like us to enable?
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Motivation & Background:

Practical opportunity: Possible to connect evolutionary phenotypes to genetics

Research opportunity: Genetic basis for evolutionary changes in phenotype?

AND OTHERS....



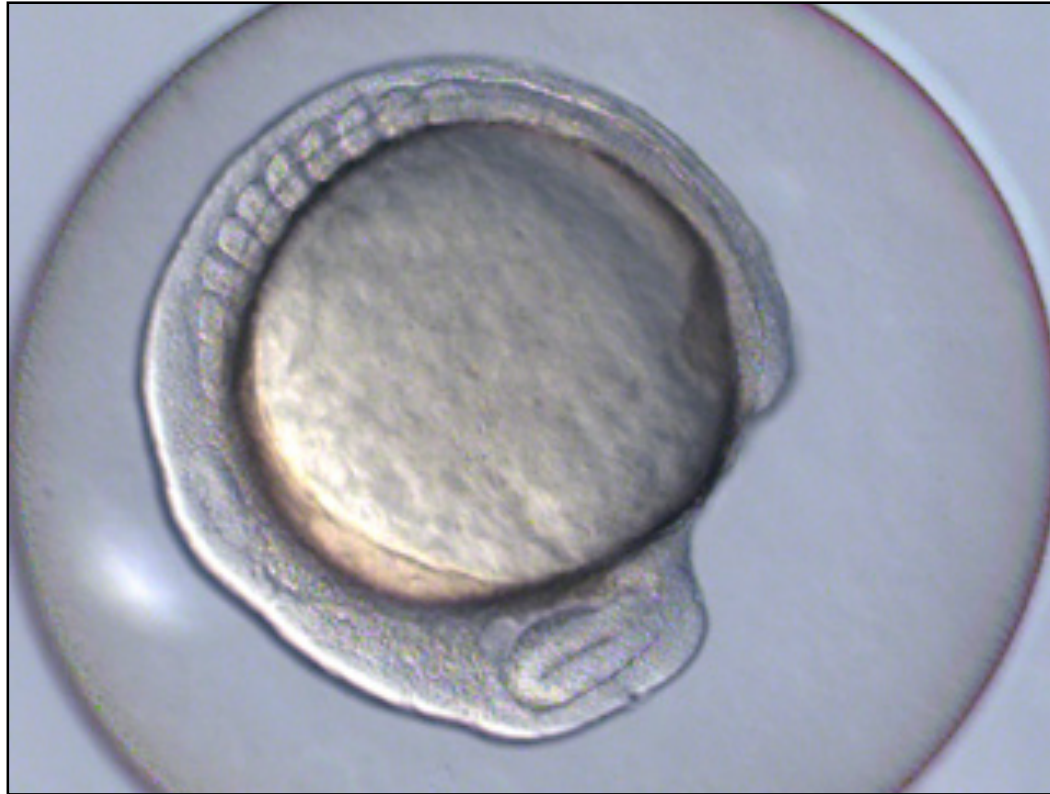


Door opener

Model organism databases developed methods to computerize and compute on phenotype

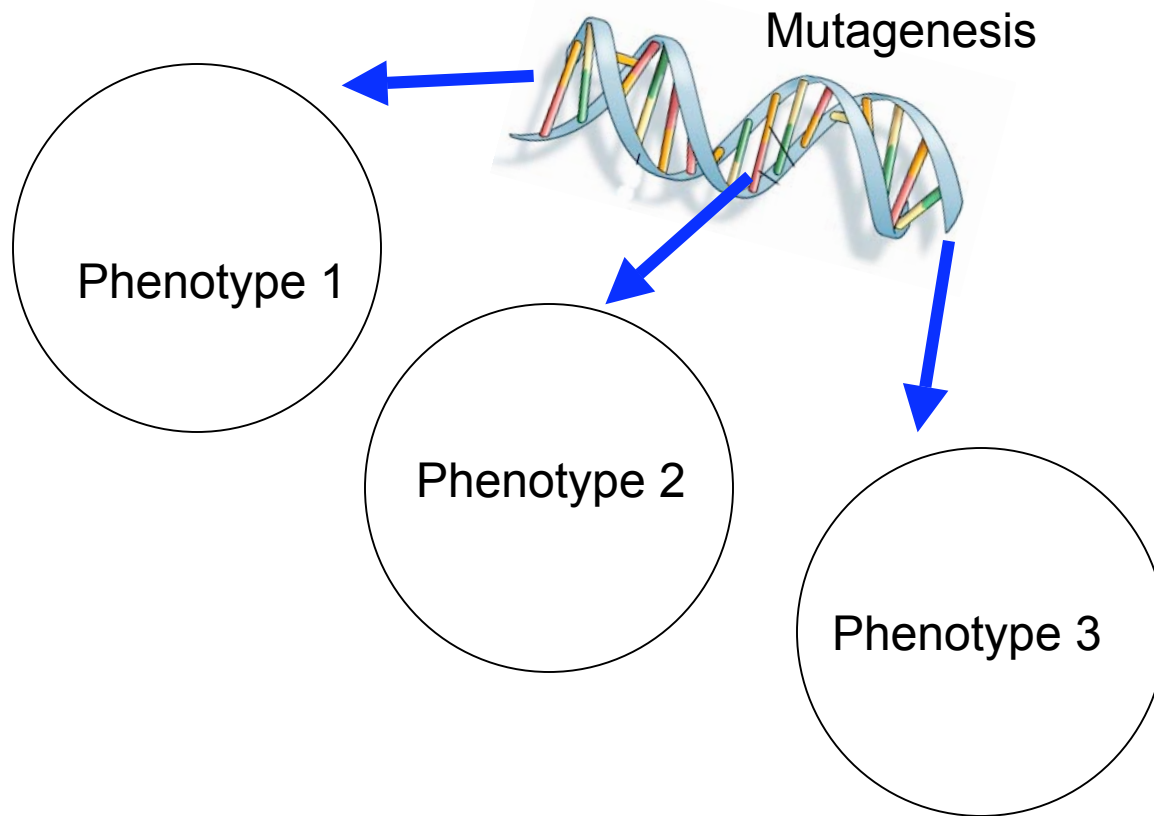
- Zebrafish genetic database (zfin.org)

Zebrafish is a model organism: Genetic approach



Model organism (zebrafish):

Mutagenesis produces phenotypes



Zebrafish phenotypes (relative to wt):

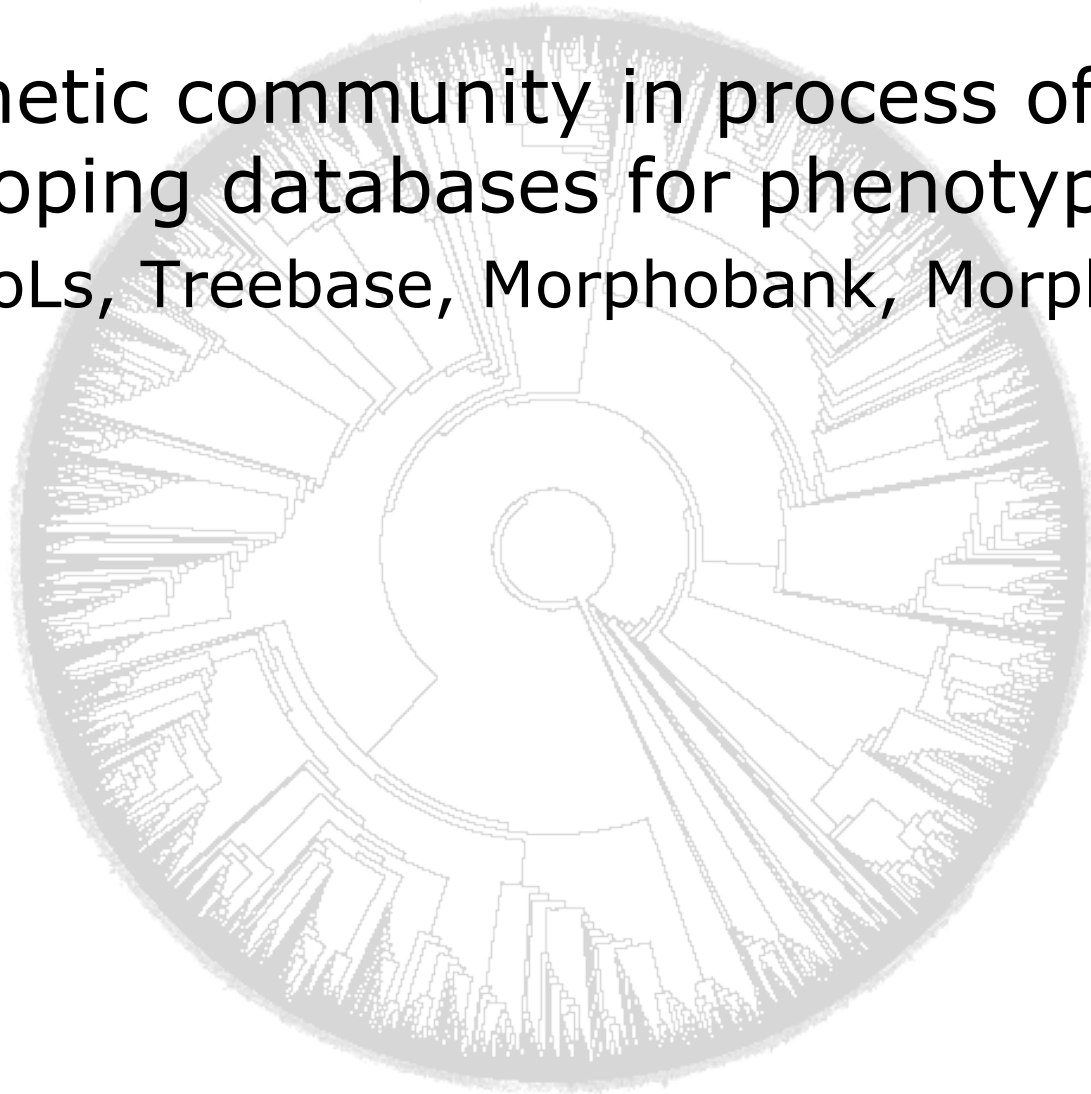
Morphology

Mutant/gene

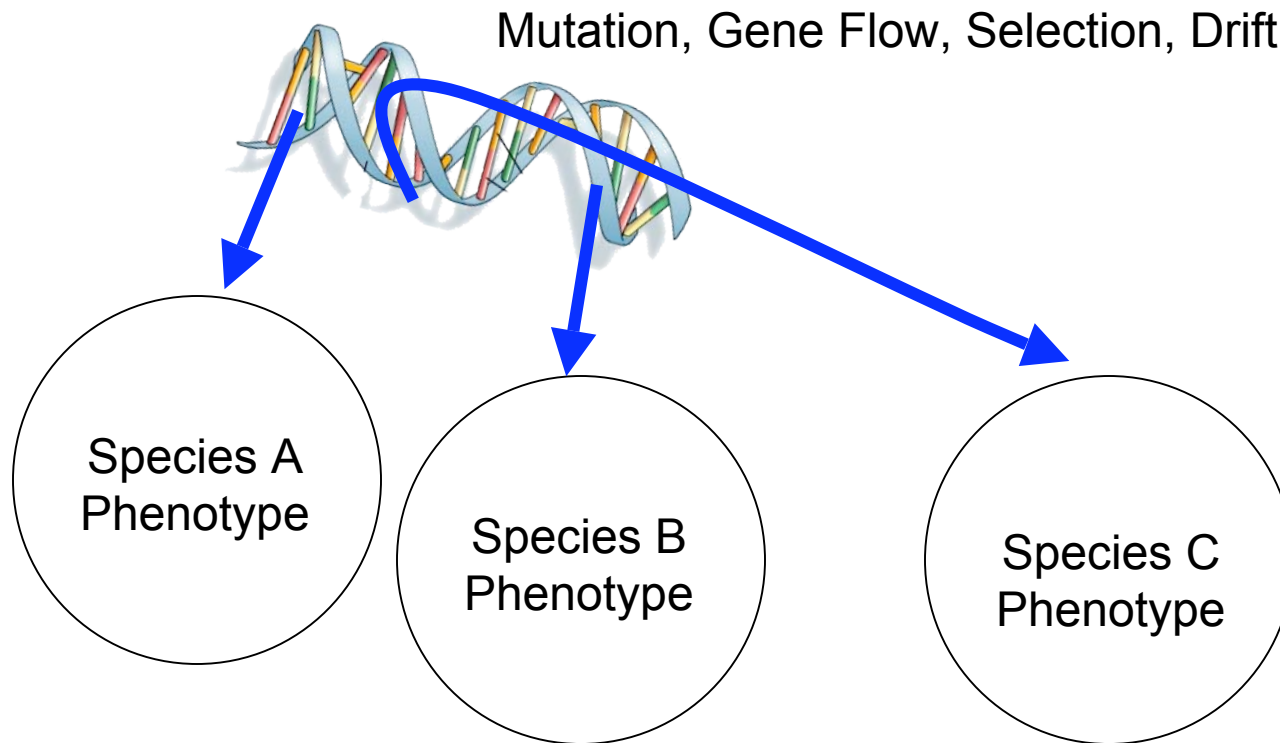
□ Maxilla : size reduction		<i>sox9ahi1134</i>
□ Dentary : size reduction		<i>sox9ahi1134</i>
□ Retroarticular : loss		<i>edn1</i>
□ Opercle : size reduction; loss	←	<i>sox9ahi1134; lockjaw</i>
□ Ceratohyal : shape change		<i>val</i>
□ Branchiostegals : number decrease		<i>edn1</i>
□ Branchiostegals : shape change		<i>she, stu, edn1-MO</i>
□ Opercle : lost		<i>edn1, lockjaw</i>
□ Opercle : size increase	←	<i>edn1</i>
□ Hypobranchials : loss		<i>val</i>
□ Ceratobranchial 5 : size reduction		<i>sox9ahi1134</i>
□ Arches 2–5 : reduced or absence		<i>lockjaw</i>
□ Arches 4–6 : loss		<i>duckbill, flathead</i>
□ Ethmoid : loss	←	<i>chameleon, cyclops, detour, etc.</i>
□ Trabeculae : fused		<i>chameleon, cyclops, detour, etc.</i>
□ Pectoral fin : loss		<i>Fgf24-MO</i>
□ Median fins : loss lepidotrichia		<i>finless</i>
□ Scapulocoracoid : loss		<i>sox9a loss</i>
□ Neural and hemal spines : alignment		<i>chordin</i>

Simultaneously...

Phylogenetic community in process of developing databases for phenotype (e.g. ToLs, Treebase, Morphobank, Morphbank)



Evolution also produces phenotypes



But, the genetic bases of morphology unknown

Mutation, Gene Flow, Selection, Drift



Species A
Morphology

Species B
Morphology

Species C
Morphology

Cypriniform fish species

Morphology

- ❑ **Branchiostegal rays:** number
- ❑ **Basibranchial 2:** T-shaped
- ❑ **Basibranchial 4:** development
- ❑ **Basibranchial 4:** shape keeled
- ❑ **Basihyal:** shape
- ❑ **Basihyal tooth plate**
- ❑ **Copula 3**
- ❑ **Hypobranchial 3:** development
- ❑ **Hypobranchial 4**
- ❑ **Ceratobranchial 5:** size
- ❑ **Ceratobranchial 5:** teeth
- ❑ **Infrapharyngeals;** number
- ❑ **Infrapharyngeal 1:** presence
- ❑ **Epibranchial 1:** uncinata process
- ❑ **Pharyngeal** teeth: rows
- ❑ **Interhyal:** size
- ❑ **Ceratohyal:** shape
- ❑ **Anterior ceratohyal:** shape

Genes
?

Interdisciplinary Fish Working Group (NESCent):

□ Zebrafish (zfin.org)

- Researchers
- Informaticians



ZFIN

□ Fish evolution

- Cypriniform Tree of Life
- Other fish morphologists



Goals:

- Communication
- Will model organism methods work for evolutionary biologists?
- Changes required for use by evolutionary biologists?



Results:

1. Phenotype is common ground for model organism & evolutionary biologists.
2. If evolutionary phenotypes are databased using ontologies, they can be connected to zebrafish phenotypic & genomic data.

Conservation of gene sequence & function

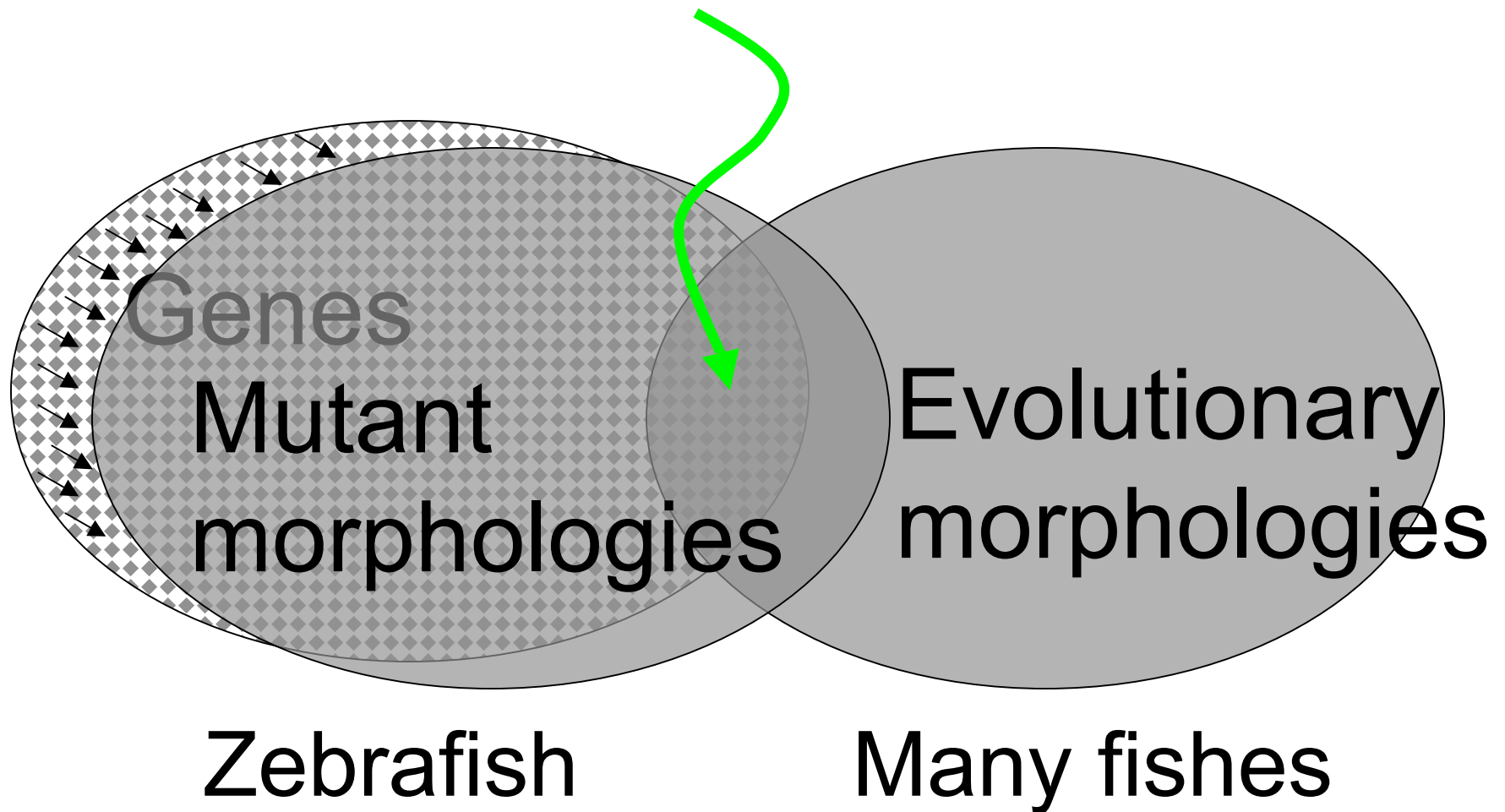


(Lamason et al., 2005)

Zebrafish Human

Conservation of gene sequence & function 16 December 2005: The lightly pigmented *golden* zebrafish show a striking resemblance to lighter skinned humans. The zebrafish pigment gene

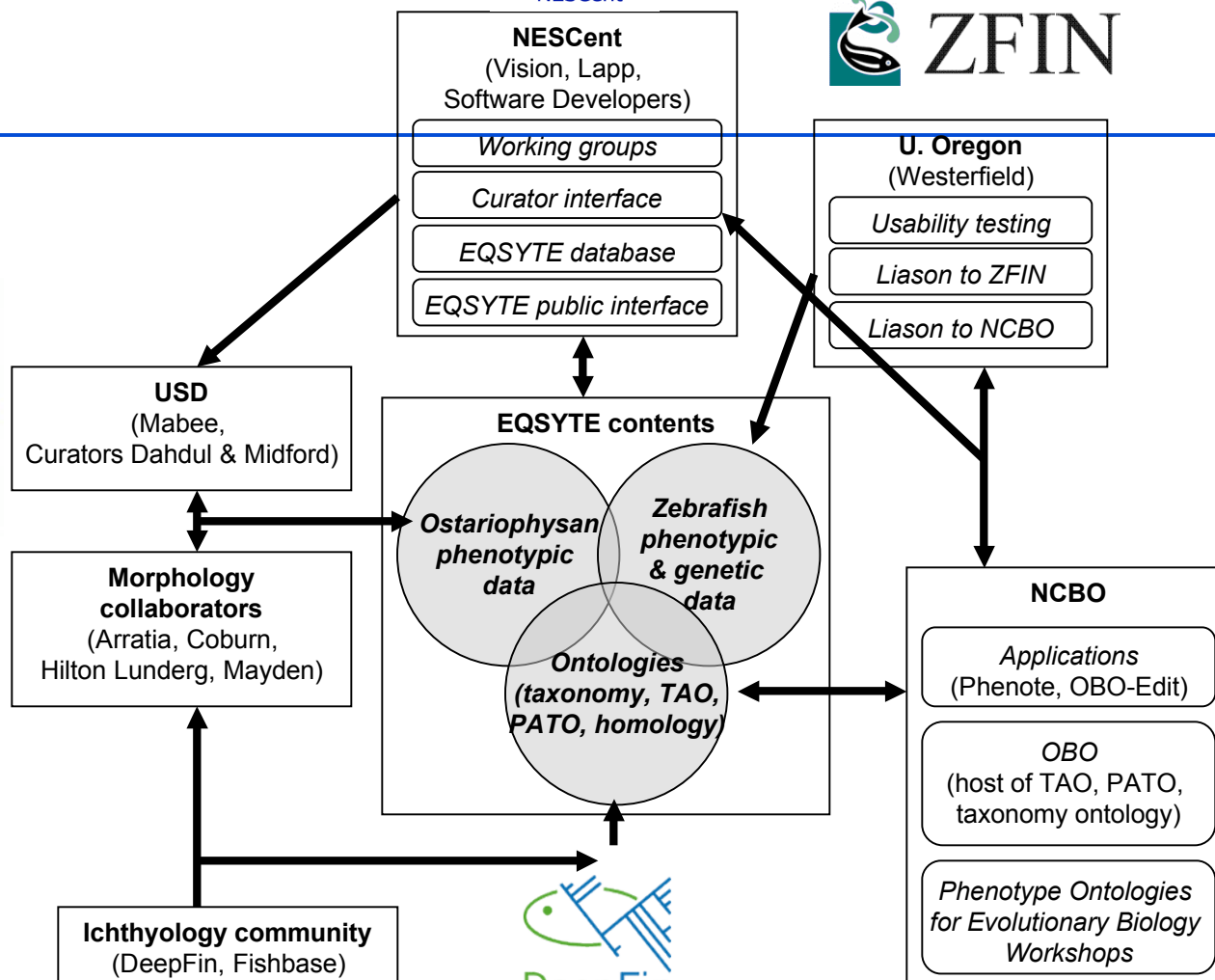
Uses: co-query data



Informatics approach:



- ☐ Prototype using fishes/zebrafish
- ☐ Develop database of phenotypic characters (skeletal) for fishes
- ☐ Connect to database of mutant phenotypes (skeletal) for zebrafish
- ☐ Access genetic and associated molecular data via shared phenotypes





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