Genetic and genomic basis of repeated evolution

Katie Peichel

University of Bern

Evolution repeats itself!
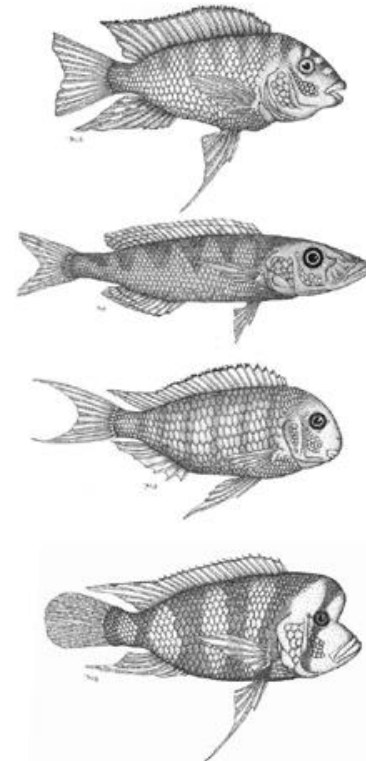
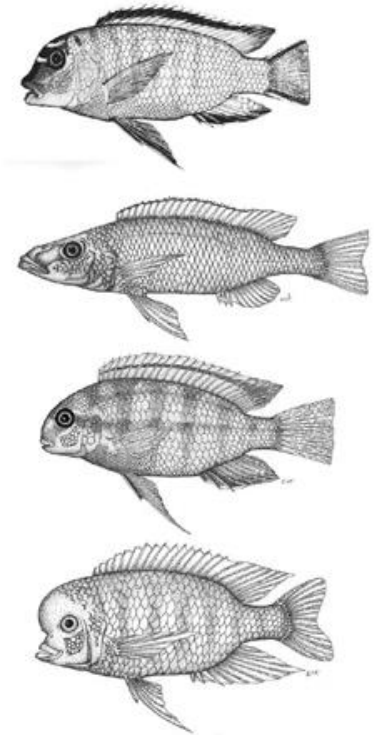Placentals

Marsupials

Lake Tanganyika   Lake Malawi

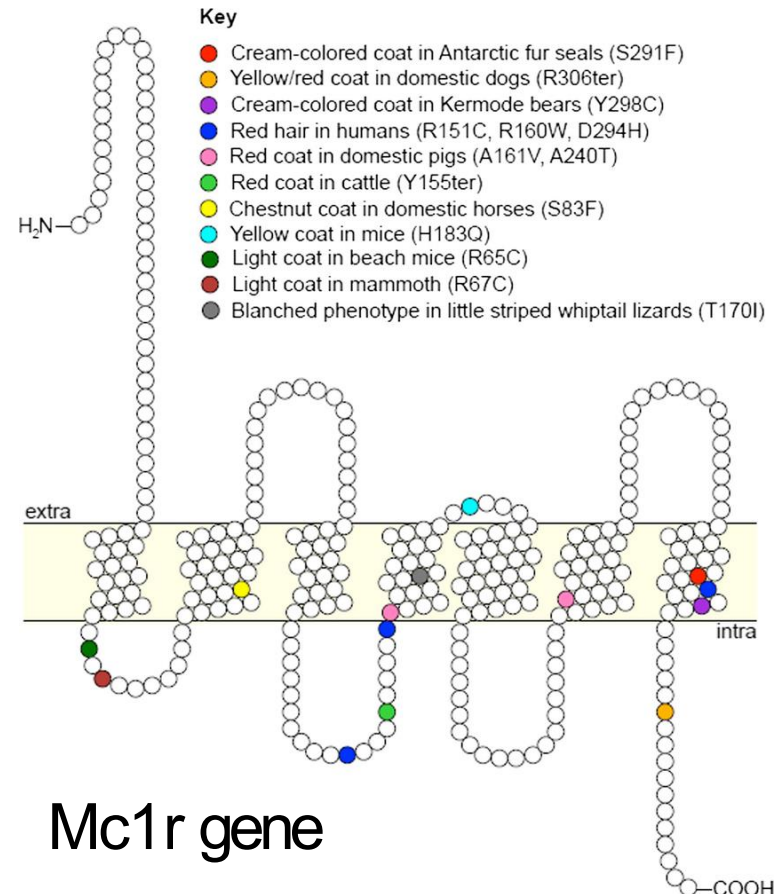# Evolution also repeats itself at the genetic level!

fence lizards | pocket mice

Key
- 🔴 Cream-colored coat in Antarctic fur seals (S291F)
- 🟠 Yellow/red coat in domestic dogs (R306ter)
- 🟣 Cream-colored coat in Kermode bears (Y298C)
- 🔵 Red hair in humans (R151C, R160W, D294H)
- 🩷 Red coat in domestic pigs (A161V, A240T)
- 🟢 Red coat in cattle (Y155ter)
- 🟡 Chestnut coat in domestic horses (S83F)
- 🩵 Yellow coat in mice (H183Q)
- 🟢 Light coat in beach mice (R65C)
- 🔴 Light coat in mammoth (R67C)
- ⚫ Blanched phenotype in little striped whiptail lizards (T170I)

Mc1r gene

Hoekstra (2006) *Heredity*

Peters et al (2016) *Ecology & Evolution*

How common is repeated phenotypic evolution at the genetic level?

Dolph Schluter

Matt Arnegard

Gina Conte

How common is repeated phenotypic evolution at the genetic level?

- Objective literature search revealed 25 case studies of two types:
  - genetic mapping studies
  - candidate gene studies
- Diverse taxa
  - fungi, plants, invertebrates, vertebrates
- Diverse traits
  - morphology, life history, toxins and toxin resistance, ability to utilize specific food sources
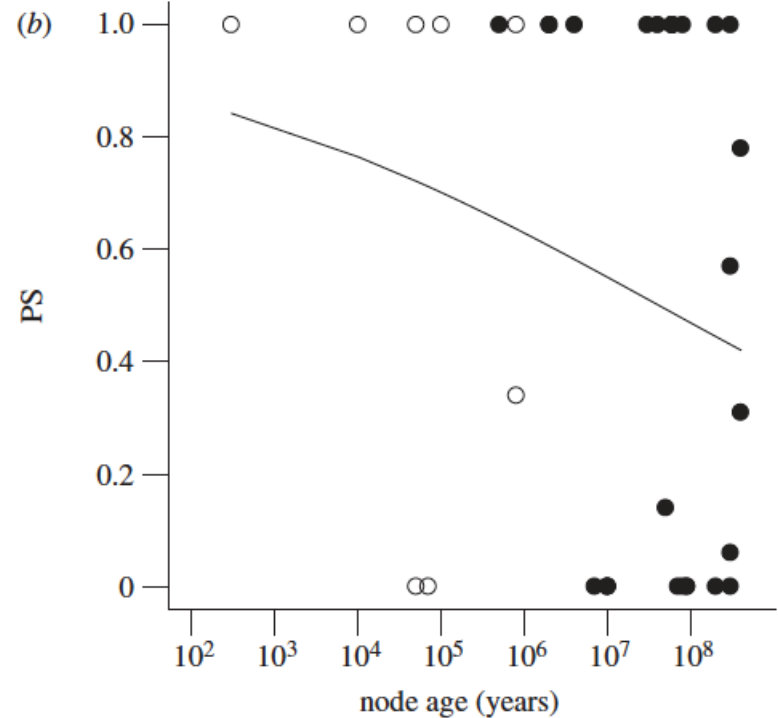
Probability of gene reuse is high!
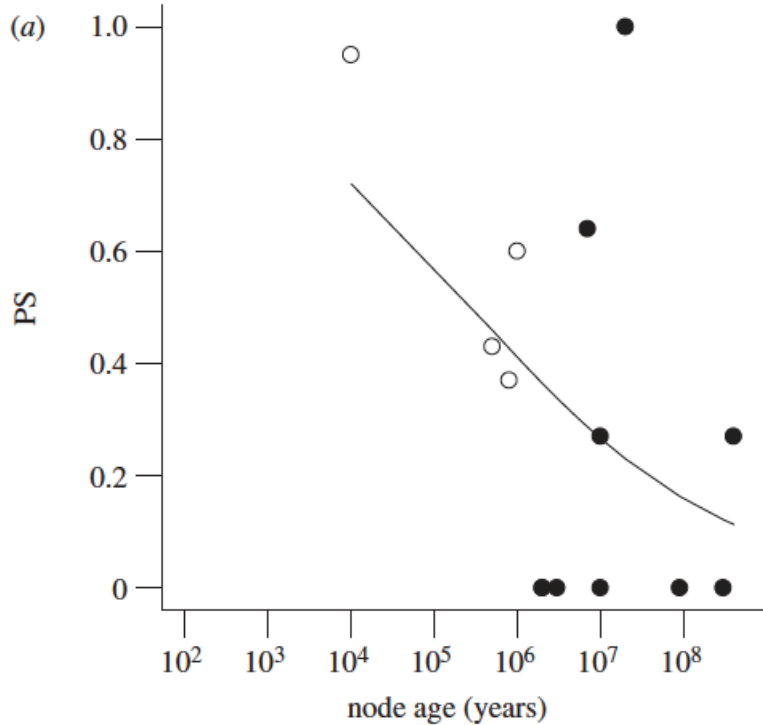
(a) Genetic mapping
PS = 0.32 ± 0.10 s.e.
○ PS = 0.47 ± 0.15 s.e.
● PS = 0.24 ± 0.12 s.e.

(b) Candidate gene
PS = 0.55 ± 0.08 s.e.
○ PS = 0.67 ± 0.17 s.e.
● PS = 0.51 ± 0.09 s.e.

Conte et al (2012) *Proceedings of the Royal Soc*

Caveats

- Publication bias

- Small number of traits

- Detecting genes of small effect

- Different studies were done at different times in different ways

- Planning to revisit this meta-analyses with Magdalena Bohutínská and Sam Yeaman

Questions

- How often do the same genetic changes underlie repeated phenotypic evolution?

- Why might some genetic changes be used more frequently when similar phenotypes evolve?

Threespine stickleback

Small teleost fish
Lives in ocean, lakes, and streams
Extensive phenotypic variation
Replicate evolutionary events
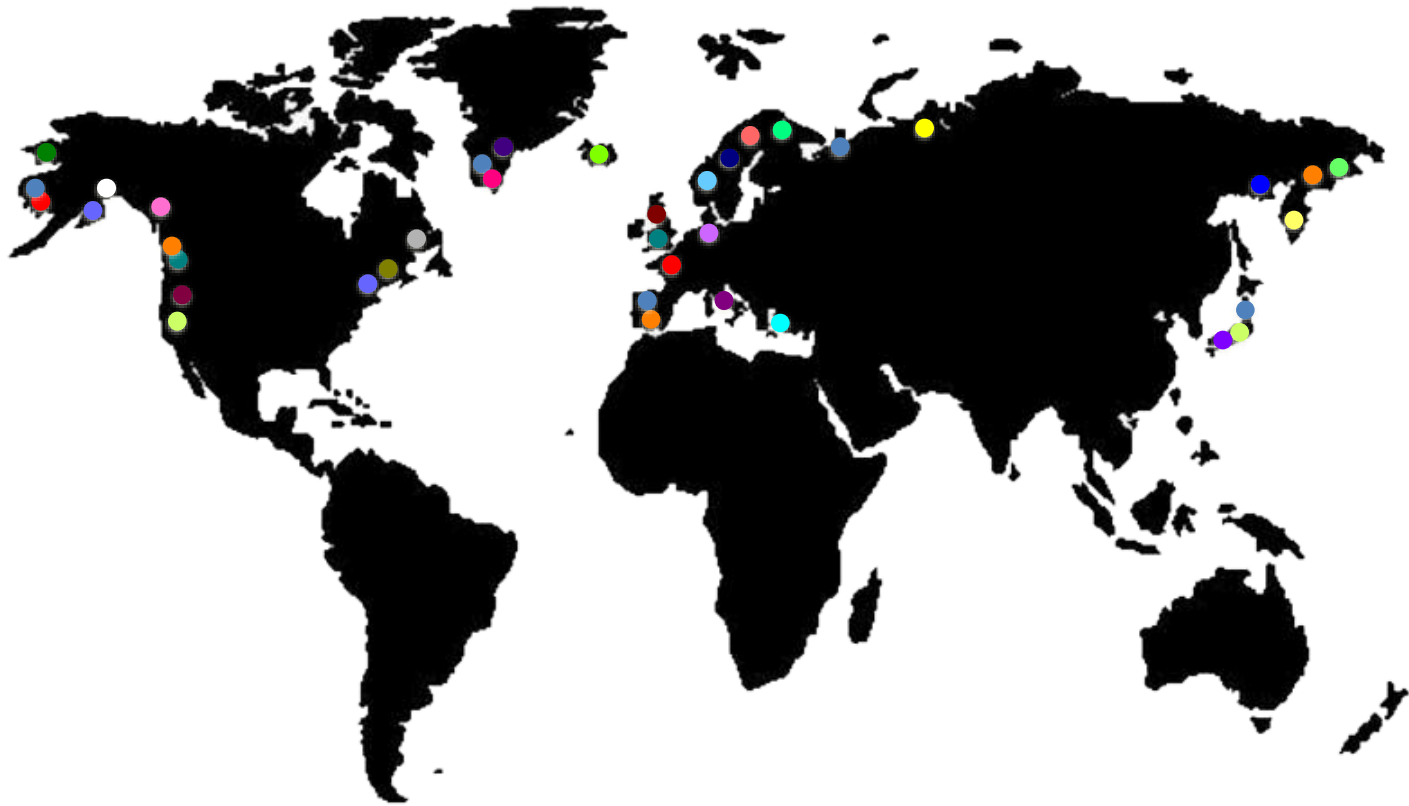Divergent populations can be crossed
Genetic tools
Genome sequence(s)

Ancestral marine populations

Derived freshwater populations

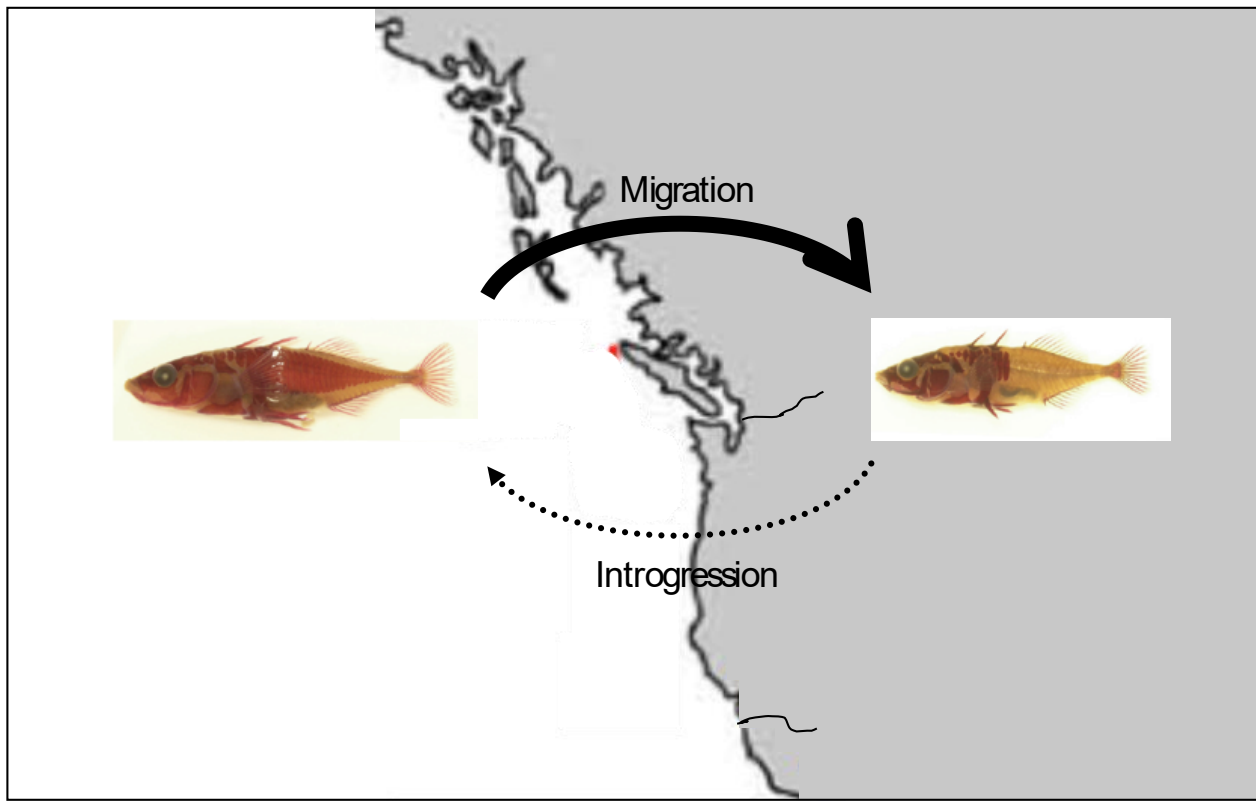# Marine *vs* freshwater sticklebacks

marine

freshwater

Photos by Seiichi Mori and Jun Kitano

Marine *vs* freshwater sticklebacks

Large, silvery, plated
Migratory, schooling
Saltwater & freshwater tolerant
Lives 2 years

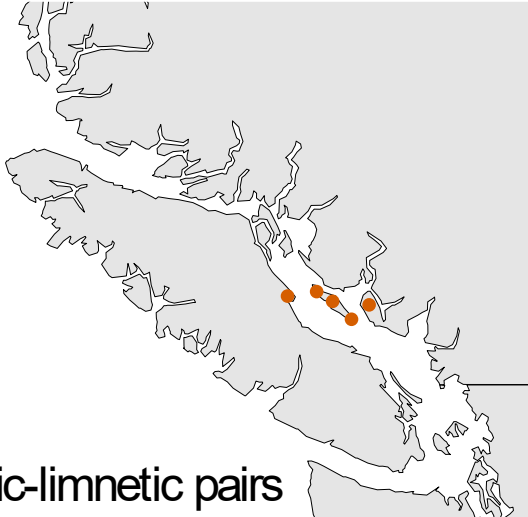Small, striped, unplated
Resident, non-schooling
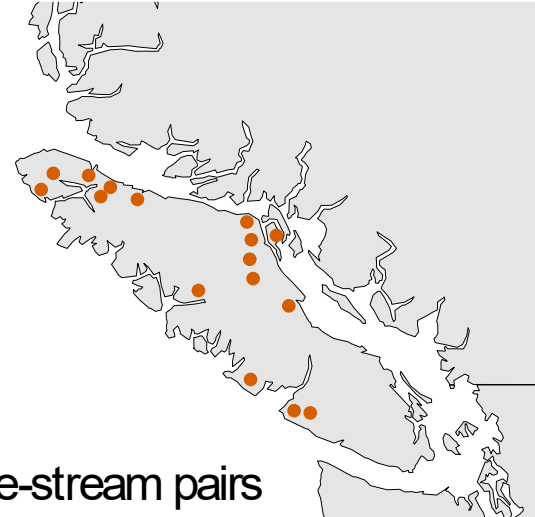Saltwater intolerant
Lives 1 year

Photos by Seiichi Mori and Jun Kitano

Repeated gene flow between marine and freshwater populations

Freshwater stickleback species pairs
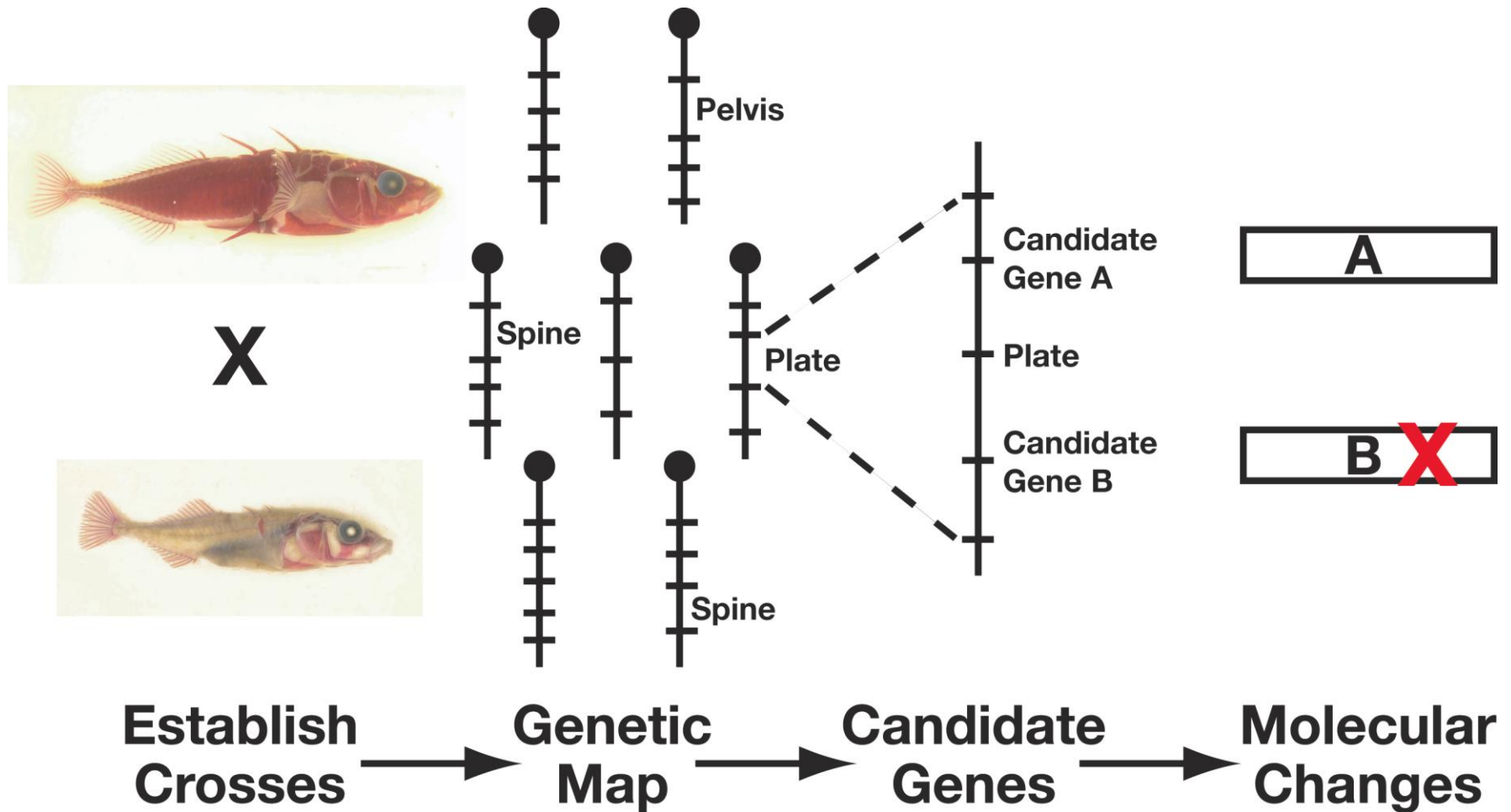

Benthic-limnetic pairs

Lake-stream pairs

Questions

- How often do the same genetic changes underlie repeated phenotypic evolution?

- Why might some genetic changes be used more frequently when similar phenotypes evolve?

Quantitative trait locus (QTL) mapping: which genotypes underlie parallel phenotypes?
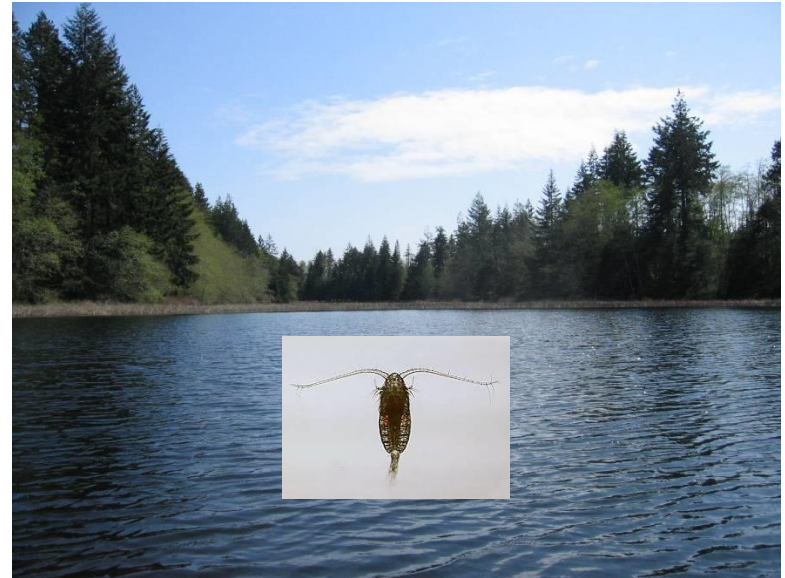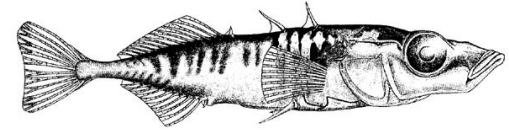

Establish Crosses → Genetic Map → Candidate Genes → Molecular Changes

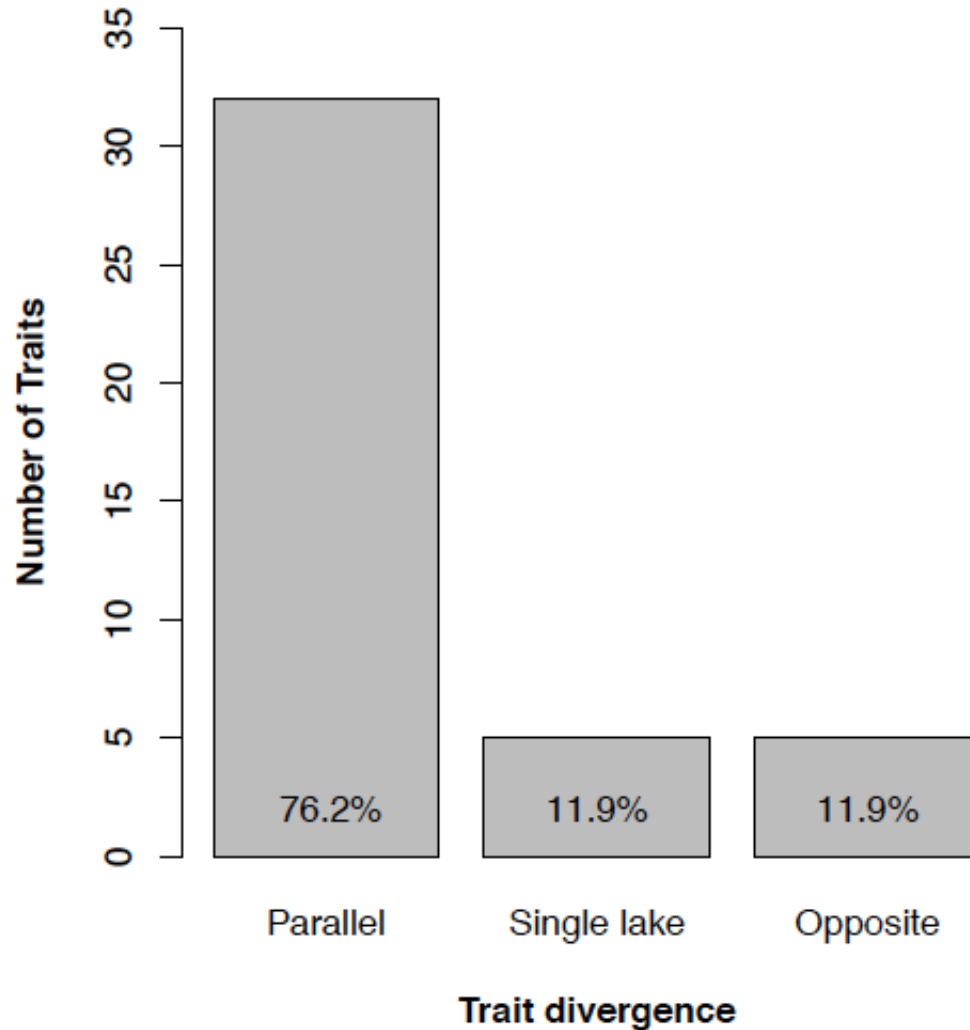# Benthic-limnetic pairs

benthic
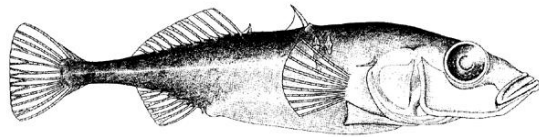
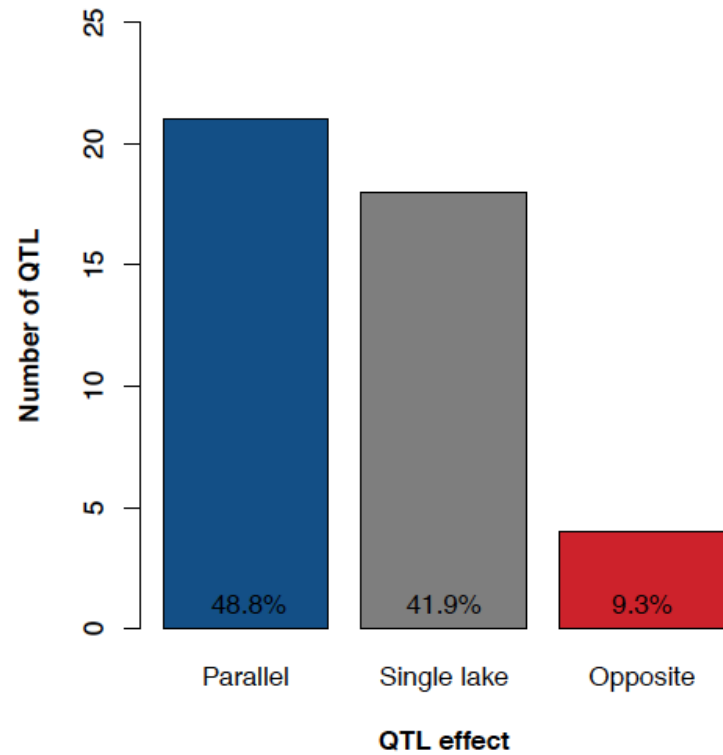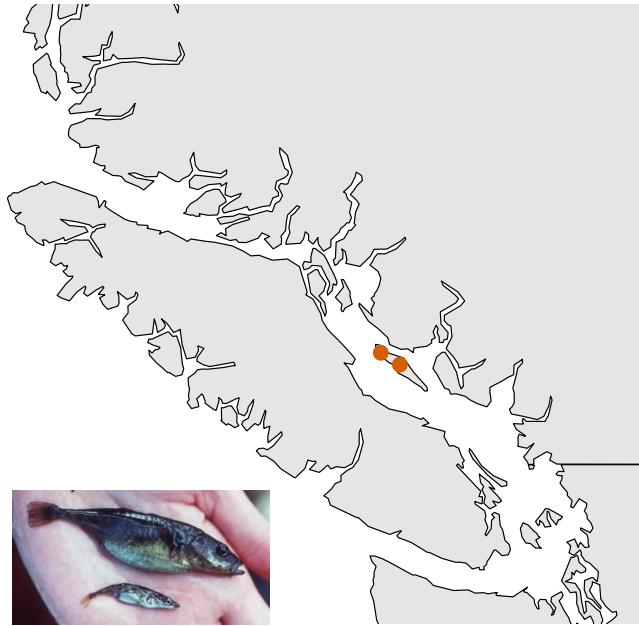limnetic

QTL mapping in two benthic-limnetic pairs

- One Paxton cross: 407 F2s
- One Priest cross: 328 F2s
- 58 morphological traits

Conte et al (2015)
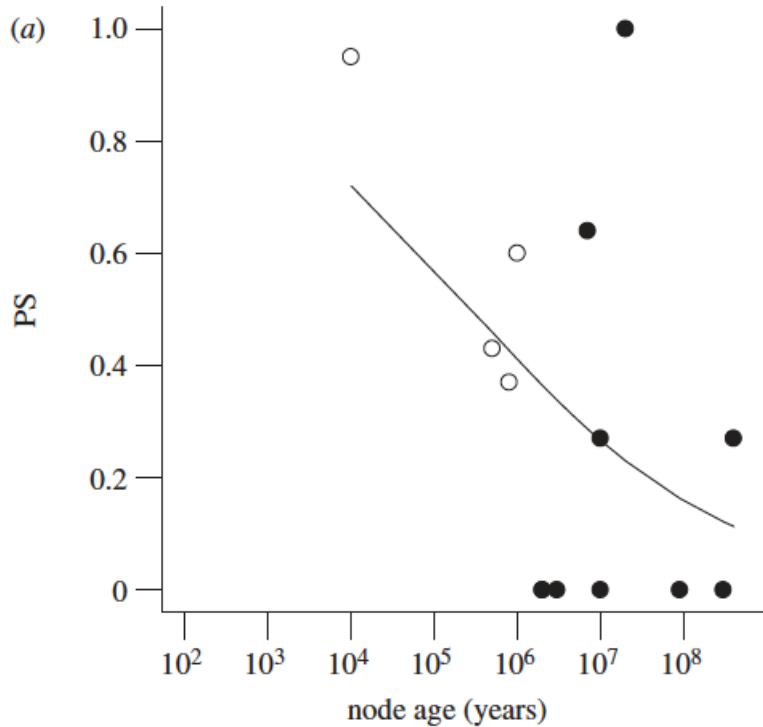
High parallelism in trait divergence in parental populations

Conte et al (2015)

Half of benthic-limnetic QTL are shared

Conte et al (2015)

Probability of gene reuse
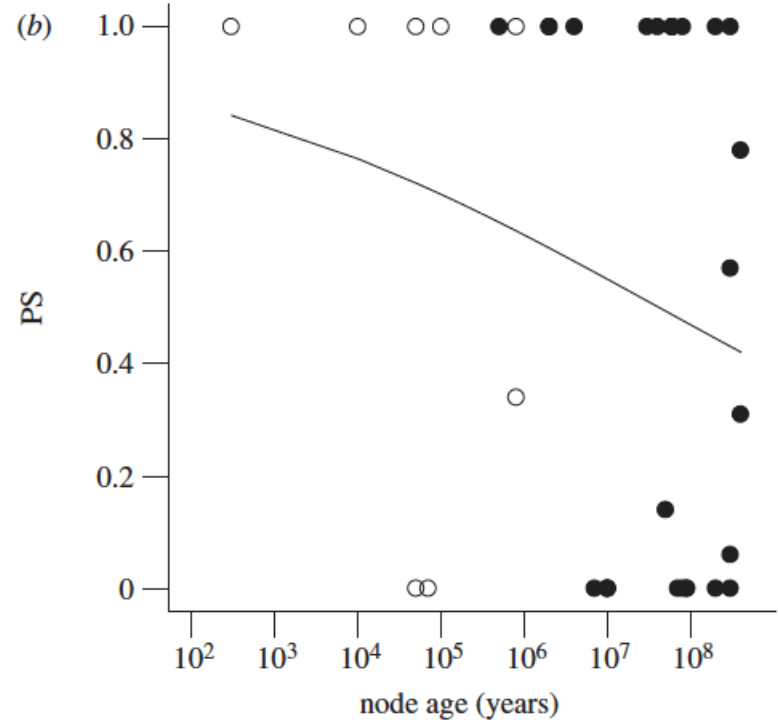

(a) Genetic mapping
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0.38

(b) Candidate gene
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Conte et al (2012) *Proceedings of the Royal Soc*

Lake-stream pairs

Stream habitat

Lake habitat

Collaboration with Yoel Stuart, Dan Bolnick, Andrew

QTL mapping in four lake-stream pairs

Watershed	F1 Family	# Individuals	# Markers	% Genotyped	Significant QTL alpha = 0.05
Boot	1	259	592	87.6	86
Boot	2	274	359	88.2	37
Misty	1	198	230	85.8	19
Misty	2	214	353	82.2	32
Pye	1	72	420	84.2	7
Pye	2	91	715	89.6	24
Pye	4	166	526	89.3	34
Roberts	3	70	635	89.2	6
Roberts	5	141	440	87.9	12

63 morphological traits

Hilary Poore

Poore et al *submitted*

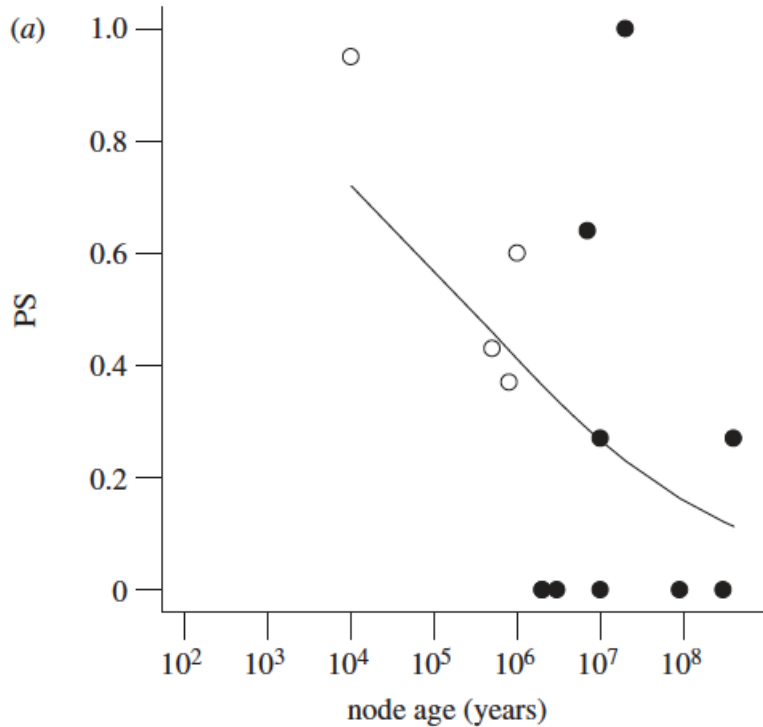# Low parallelism in trait divergence in parental populations
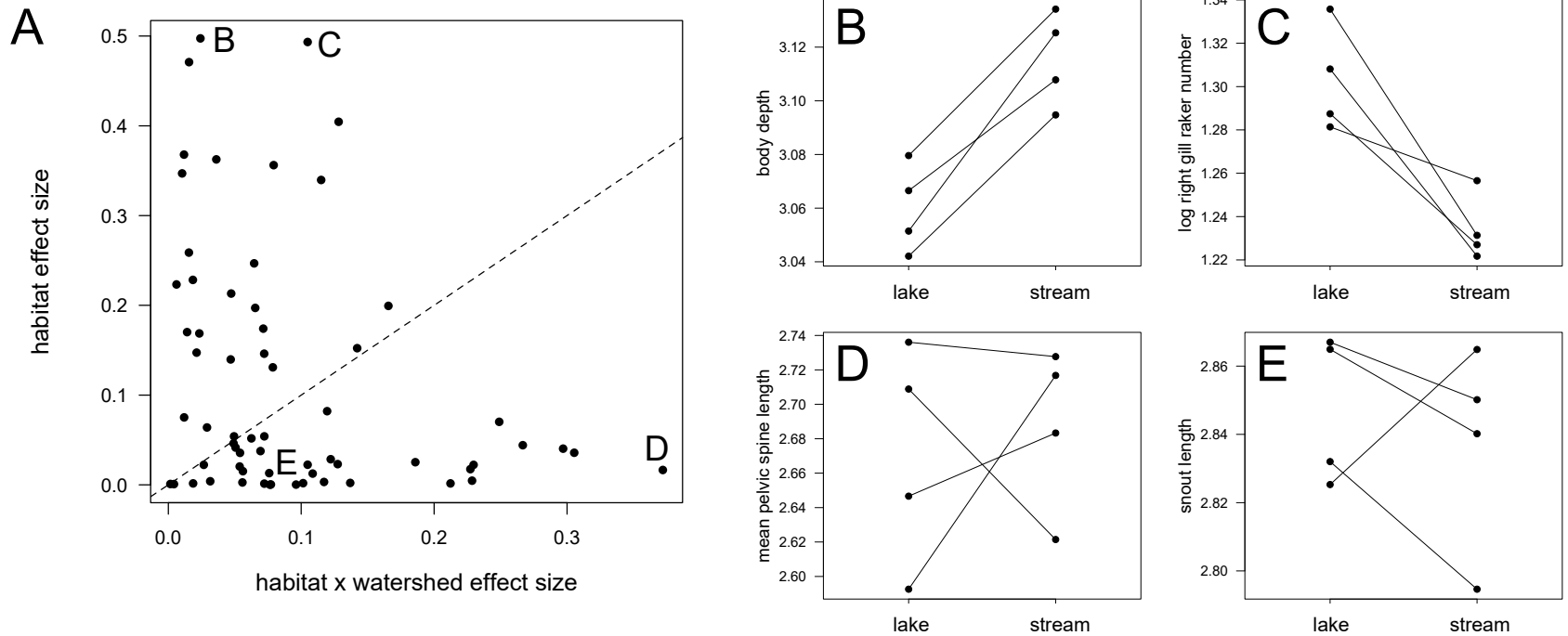
Few lake-stream QTL are shared
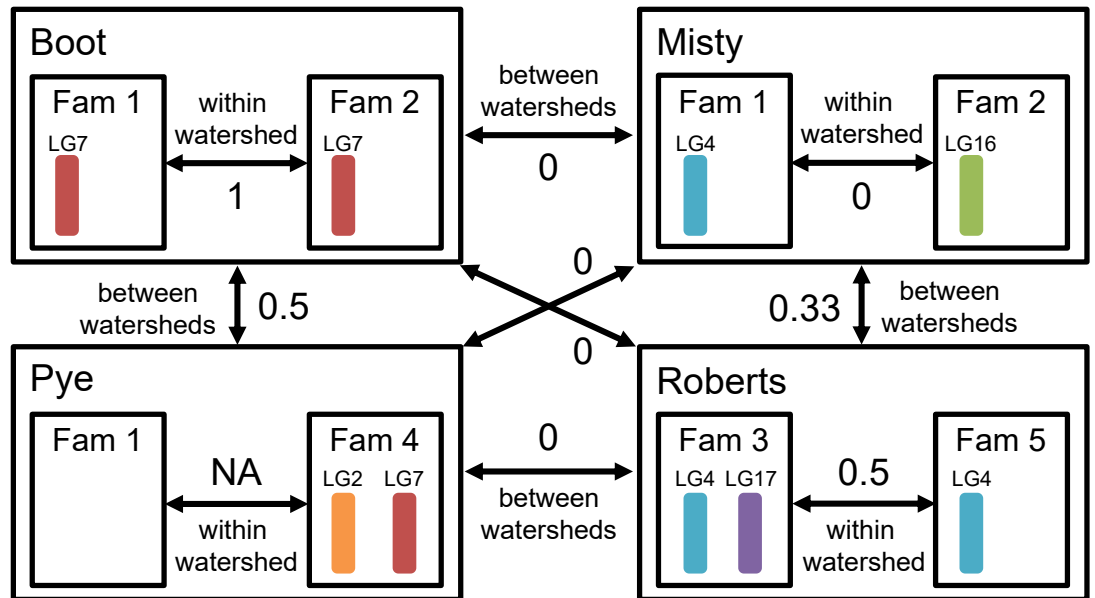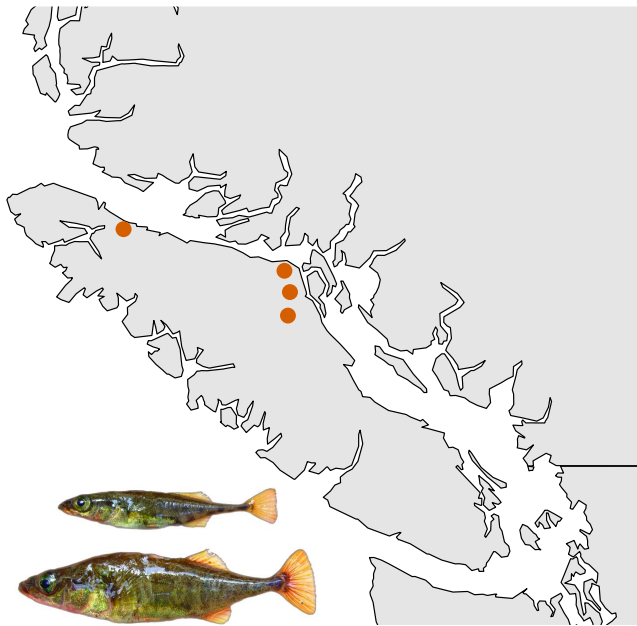
among watershed proportion shared QTL = 0.4 for plate number

Among watershed proportion shared QTL = 0.154 across traits

Poore et al *submitted*

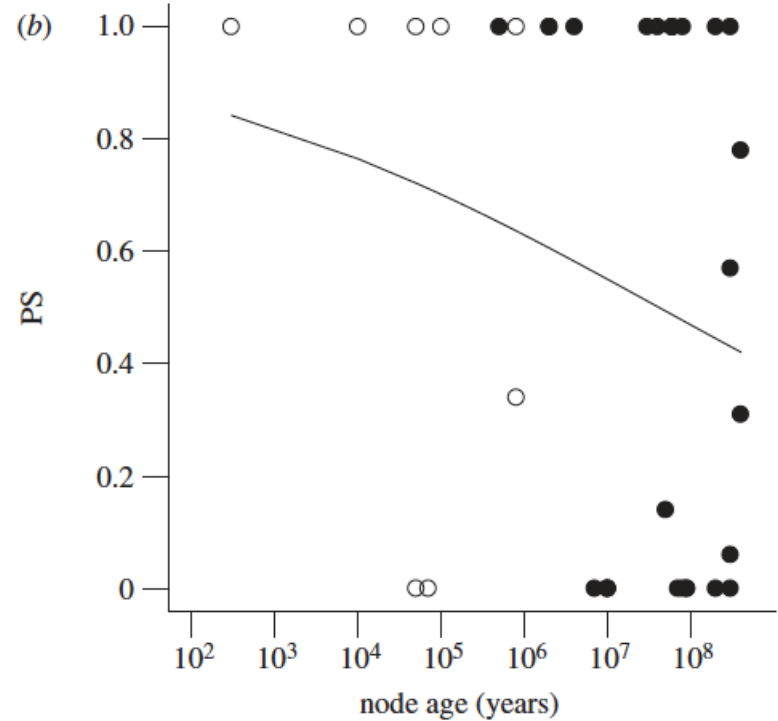# Probability of gene reuse

(a) Genetic mapping
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0.154 → ○ PS = 0.47 ± 0.15 s.e.
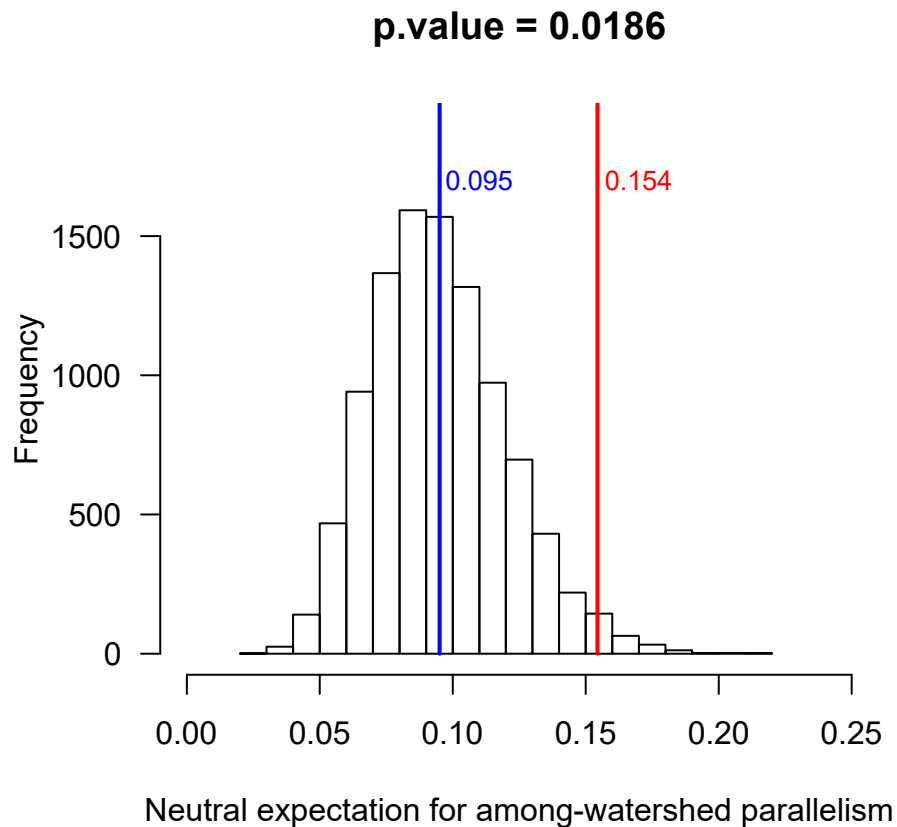● PS = 0.24 ± 0.12 s.e.

(b) Candidate gene
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Conte et al (2012) *Proceedings of the Royal Soc*

Proportion of shared QTL is still higher than expected

p.value = 0.0186

Frequency

Neutral expectation for among-watershed parallelism

Population genomics: which genotypes are associated with parallel adaptation?
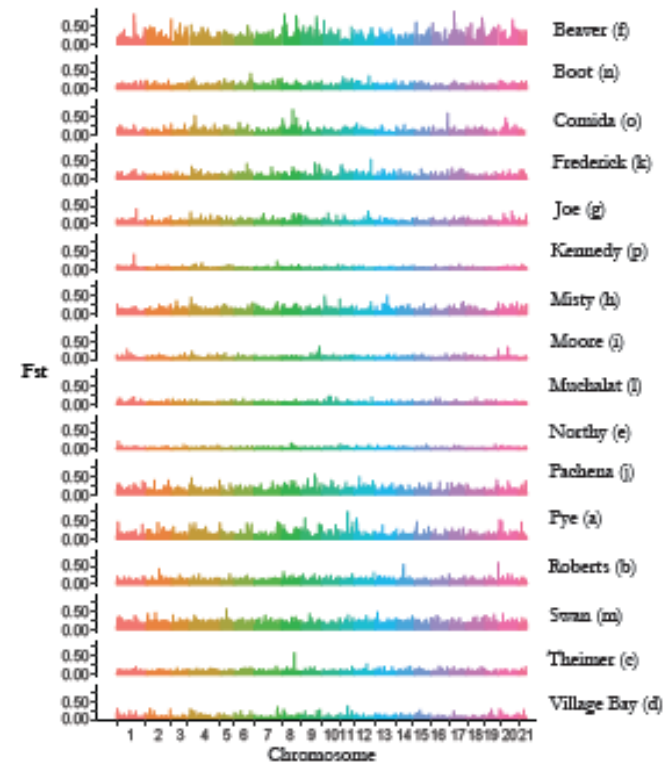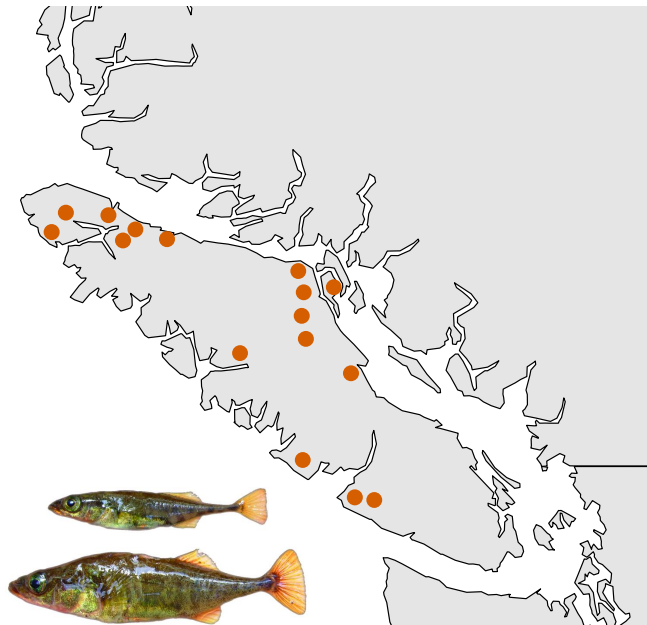

Diana Rennison
UC San Diego

- Lake-stream data
  - 16 lake-stream pairs (32 populations)
  - Sequencing for 24 individuals/population

- Benthic-limnetic data
  - 3 benthic-limnetic pairs (6 populations)
  - Sequencing for 20 individuals/population

- Fst between each lake-stream or benthic-limnetic pair was calculated in 50 kbp windows
  - 2513 windows in 16 lake-stream pairs
  - 5733 windows in 3 benthic-limnetic pairs

Highly repeatable genomic differentiation in benthic-limnetic pairs


Rennison & Peichel (2022) *Molecular Ecolo*
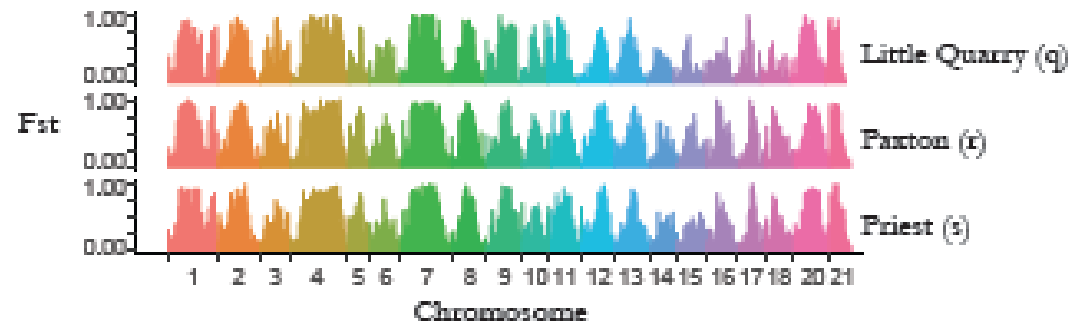
Lower repeatability of genomic differentiation in lake-stream pairs

Rennison et al (2019) *Philosophical Transactions of the Royal*

Questions

- How often do the same genetic changes underlie repeated phenotypic evolution?
  - Sometimes, but not always

- Why might some genetic changes be used more frequently when similar phenotypes evolve?
  - Pleiotropy? See Rennison and Peichel (2022) *Molecular Ecology* 31: 1476-1486

# Why do the benthic-limnetic and lake-stream pairs differ?
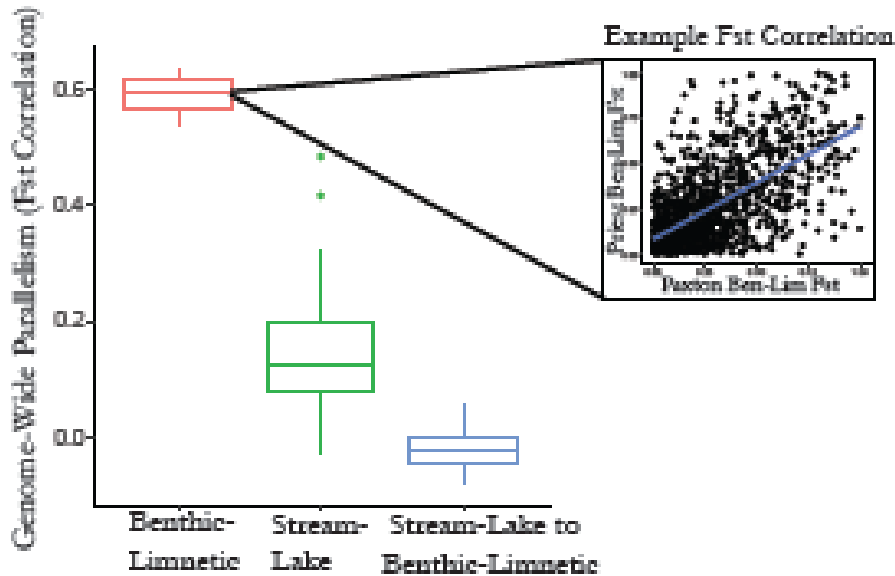
- Source of standing genetic variation?
- Evolutionary history?
- Extent of gene flow?
- Strength of parallel selection?

Problem!
We are only examining extant populations and lack information on the founding ancestral populations and ecosystem changes over time
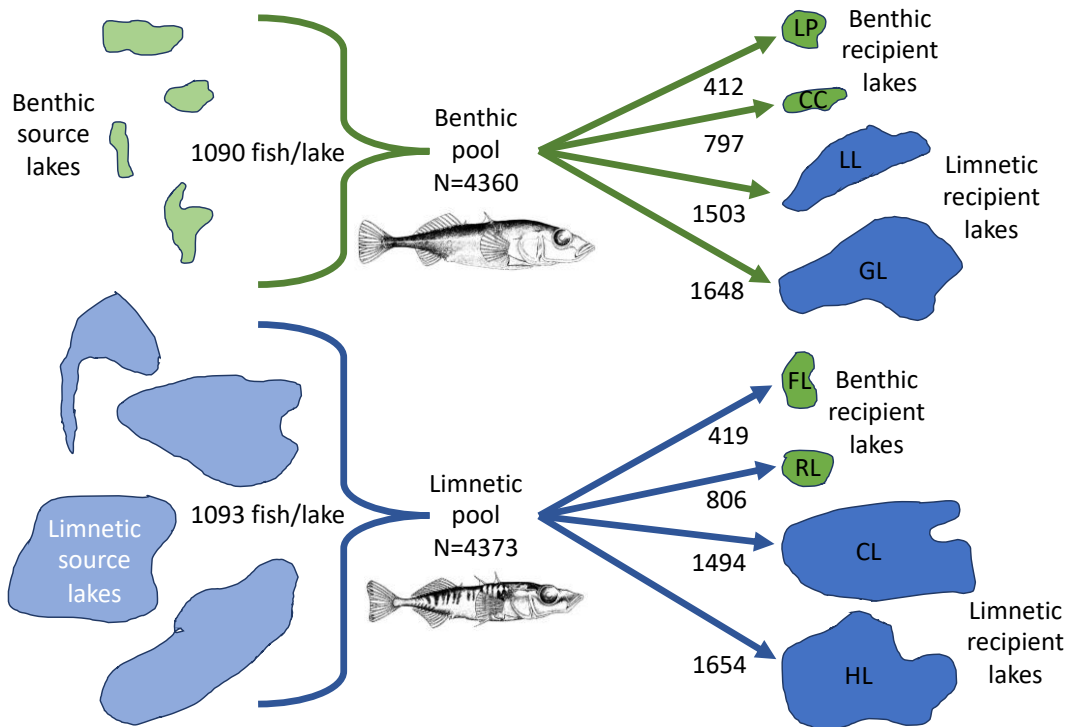
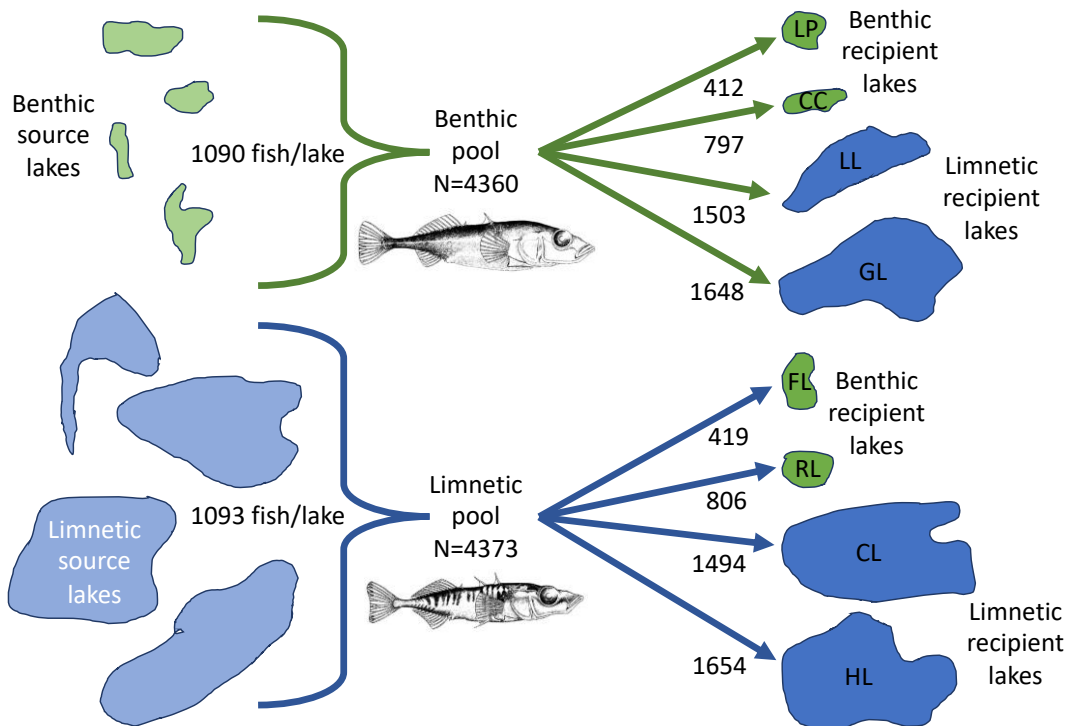# Experimental evolution to the rescue?

Blount et al (2018) *Scienc...*
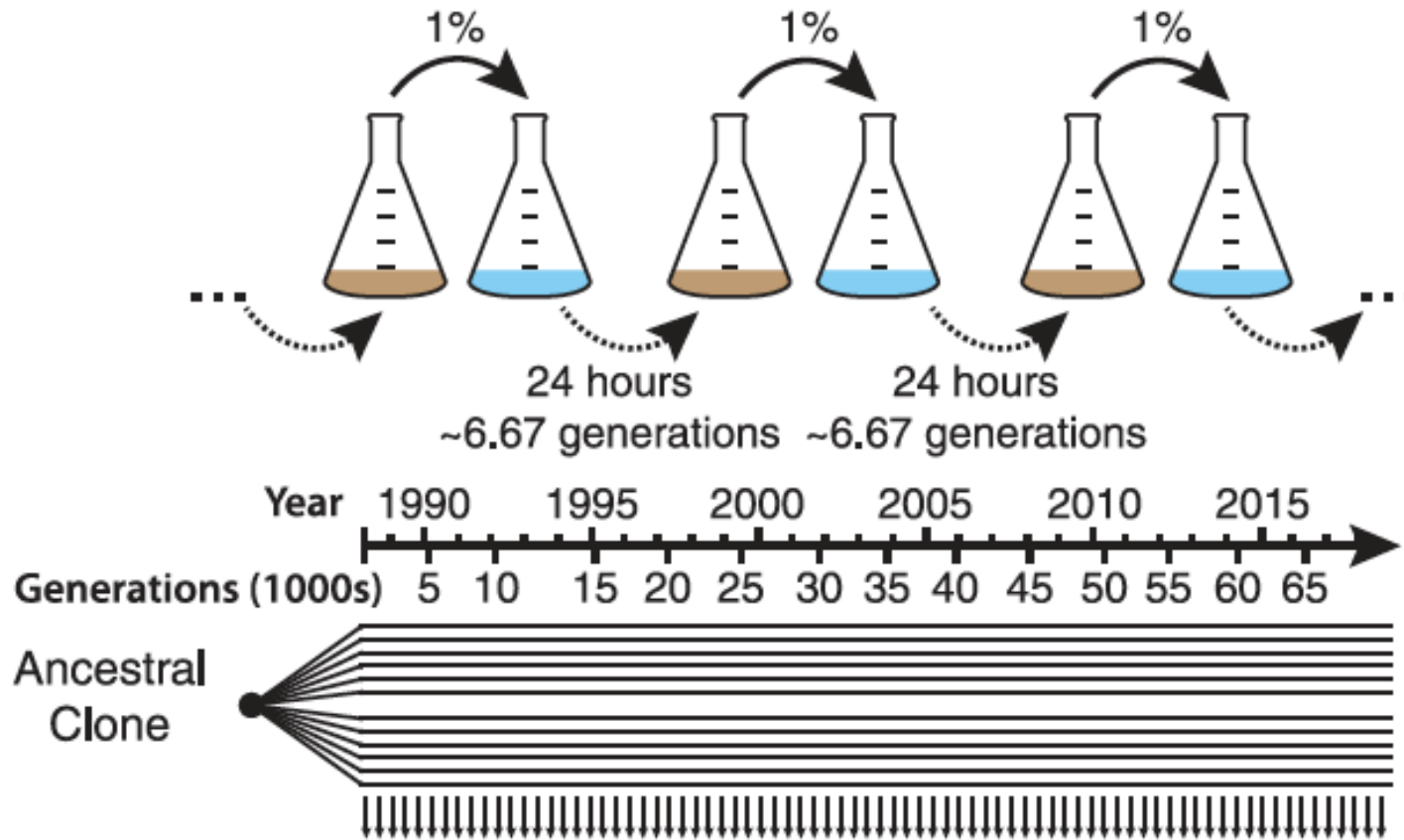
Can we do experimental evolution in **natural ecosystems**?

Hope Lake, Alaska

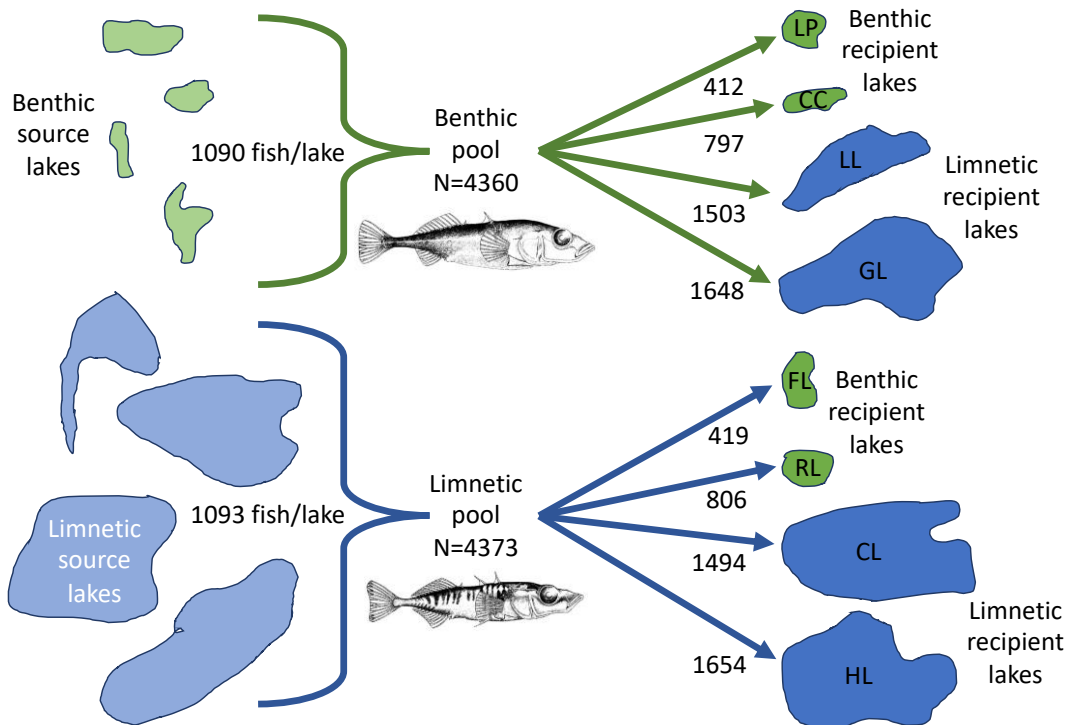# FITNESS: Forward-In-Time Natural Experimental Study of Selection

# FITNESS: Forward-In-Time Natural Experimental Study of Selection

- Whole genome sequences and phenotypes from 8733 founding fish

- Follow evolutionary trajectories of genotypes and phenotypes in recipient lakes for 8 generations (9216 fish)

FITNESS: Forward-In-Time Natural Experimental Study of Selection

- Starting **genotypic**, phenotypic, and ecological conditions are known

- Evolutionary trajectories will be followed over time in **natural ecosystems**

FITNESS: Forward-In-Time Natural Experimental Study of Selection
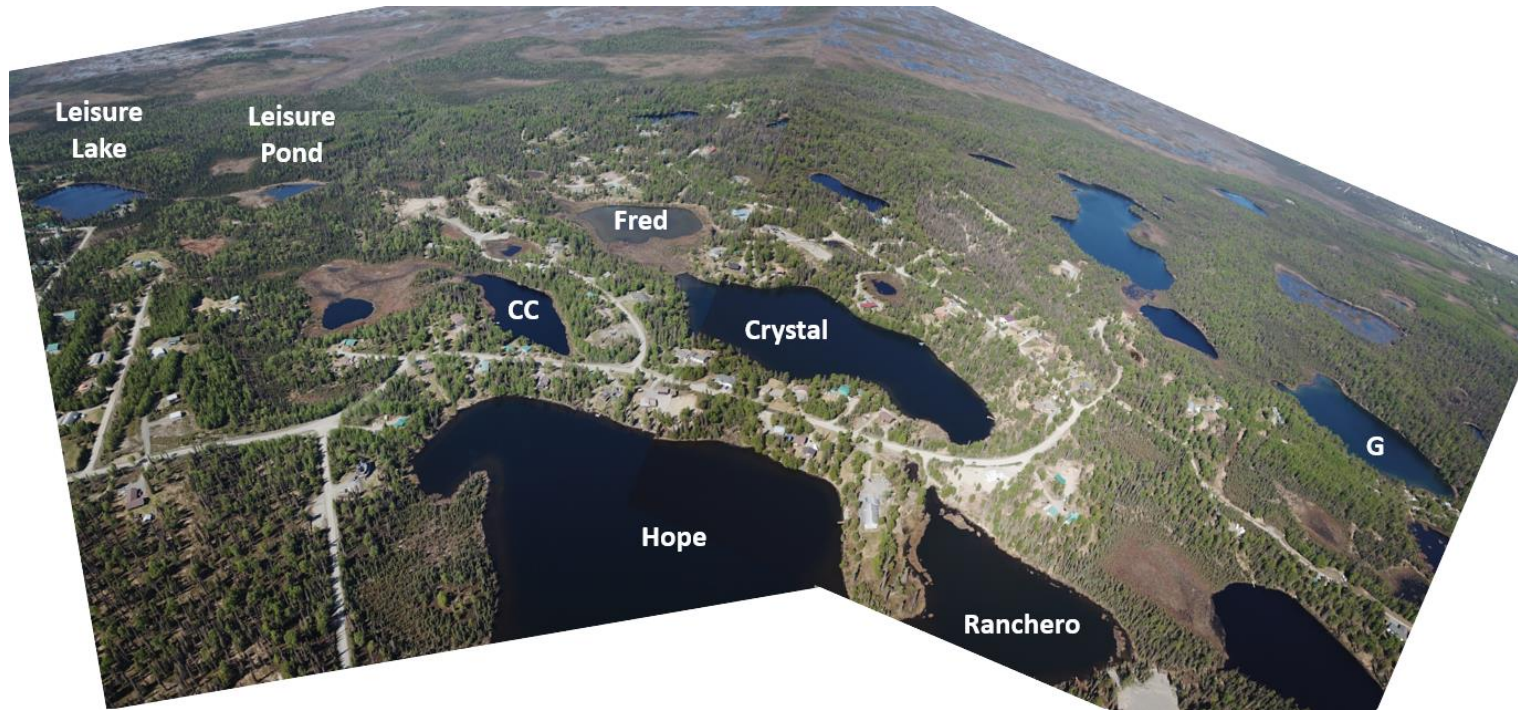
How repeatable are genotypic and phenotypic trajectories?
Can we predict the evolutionary trajectories we see?

Collaborators

Benthic-limnetic pairs
Dolph Schluter (U British Columbia)
Gina Conte (U British Columbia)
Matt Arnegard (Fred Hutch)
Diana Rennison (UC San Diego)

Lake-stream pairs
Dan Bolnick (U Connecticut)
Andrew Hendry (McGill)
Rowan Barrett (McGill)
Yoel Stuart (Loyola University)
Diana Rennison (UC San Diego)
Hilary Poore (U Bern)

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US NIH
US NSF
EU Marie Curie
Swiss NSF

FITNESS project
Andrew Hendry (McGill)
Dan Bolnick (U Connecticut)
Rowan Barrett (McGill)
Blake Matthews (EAWAG)
Alison Derry (U Quebec)
Matt Walsh (U Texas)
Kat Milligan-Myhre (U Connecticut)
Jesse Weber (U Wisconsin)
Natalie Steinel (U Massachusetts)
Hilary Poore (McGill)
Mike Bell (retired)