



Genomics of local adaptation from the Arctic to the desert

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Imperial College – November 12, 2020





Charlie Harper

A central goal in evolutionary biology is to understand how local adaptations evolve and are maintained



Local adaptation is the result of the interplay of

Mutation

Selection

Gene flow

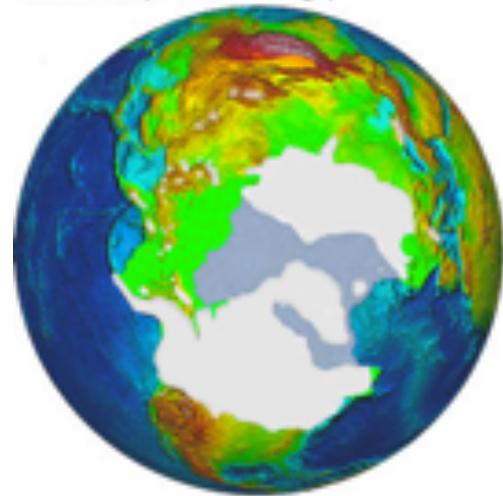
Genetic drift

Understanding their relative roles is important

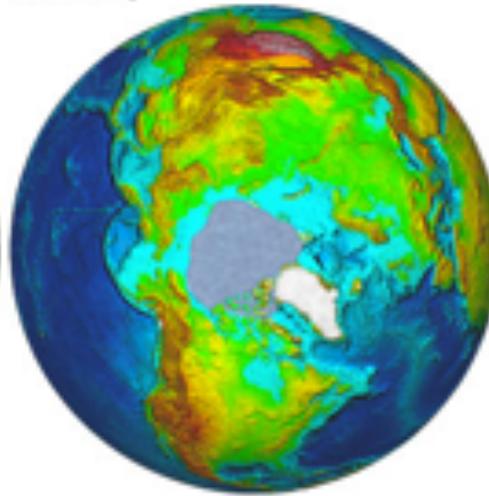
1. To understand the processes underlying adaptation and speciation
2. To help predict the potential of species to adapt to future changes

Adapting to a changing world

Pleistocene (18,000 Years Ago)



Modern Day



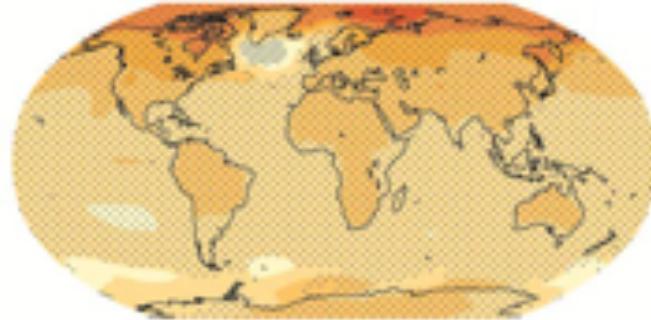
□ Glacial Ice ■ Sea Ice

Looking
backward...

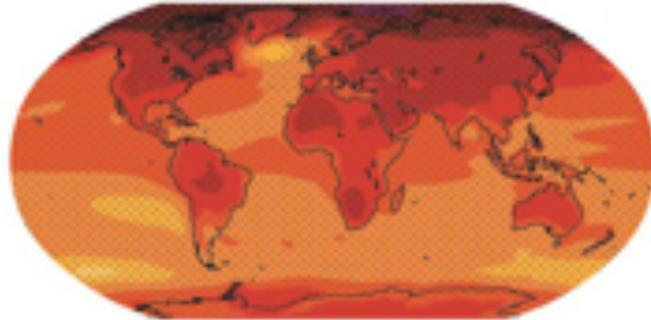
...to look
forward

RCP 2.6

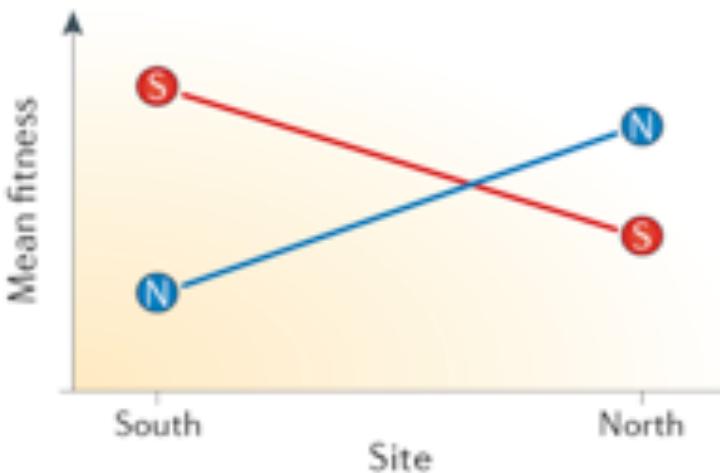
Change in average surface temperature (1986–2005 to 2081–2100)



RCP 8.5

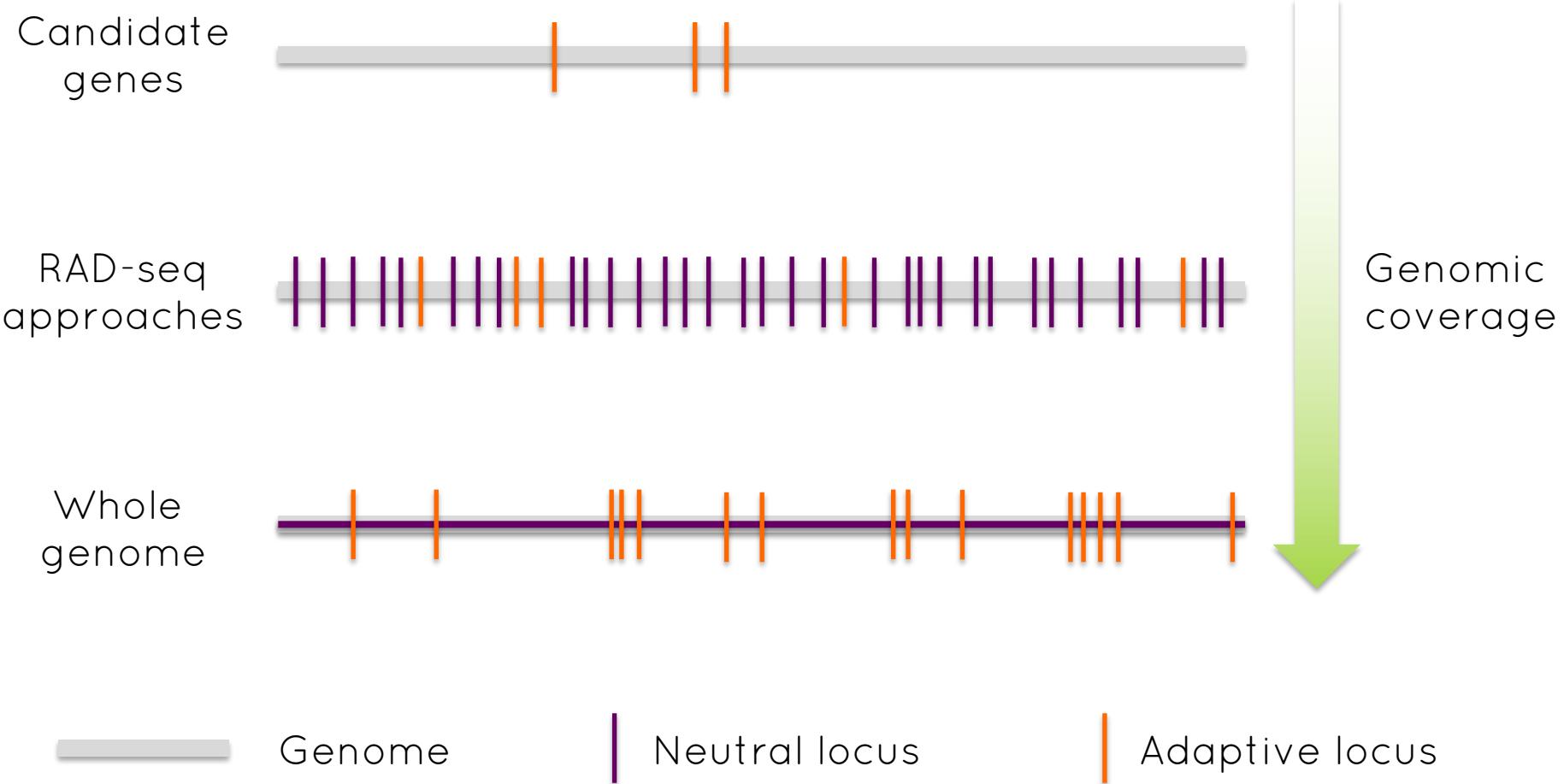


-2 -1.5 -1 -0.5 0 0.5 1 1.5 2 3 4 5 7 9 11 (°C)



Despite widespread evidence for local adaptation, the genomic basis of many adaptive traits is still poorly understood.

The ‘next-generation sequencing’ revolution



Evolutionary genomics & climate change



CORAL GENOMICS

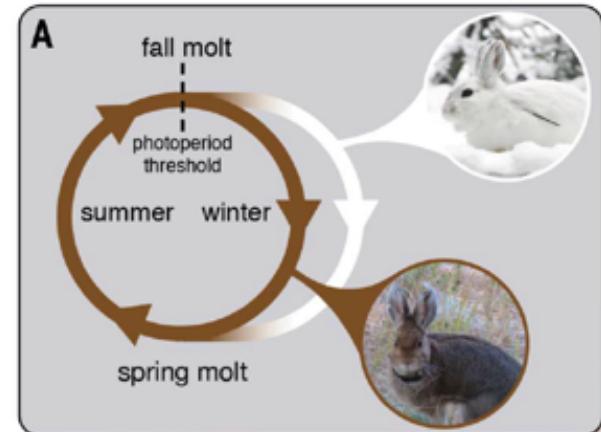
Population genetics of the coral *Acropora millepora*: Toward genomic prediction of bleaching

Zachary L. Fuller^{1*}, Veronique J. L. Mocellin², Luke A. Morris^{2,3,4}, Neal Cantin², Jianne Shepherd¹, Luke Sarre¹, Julie Peng⁵, Yi Liao^{6,7}, Joseph Pickrell⁸, Peter Andolfatto¹, Mikhail Matz^{6†}, Line K. Bay^{2*†}, Molly Przeworski^{1,9,10*†}

EVOLUTIONARY BIOLOGY

Adaptive introgression underlies polymorphic seasonal camouflage in snowshoe hares

Matthew R. Jones^{1*}, L. Scott Mills^{2,3,4}, Paulo Célio Alves^{2,5,6}, Colin M. Callahan¹, Joel M. Alves^{5,7}, Diana J. R. Lafferty^{2,4,8}, Francis M. Jiggins⁷, Jeffrey D. Jensen^{9,10}, José Melo-Ferreira^{5,6*}, Jeffrey M. Good^{1,2*}



From the Arctic to the desert



From the Arctic to the desert

Random mating



No gene flow



Varying gene flow
across the range

Mutation

Selection

Gene flow

Genetic drift

Different systems with different levels of gene flow

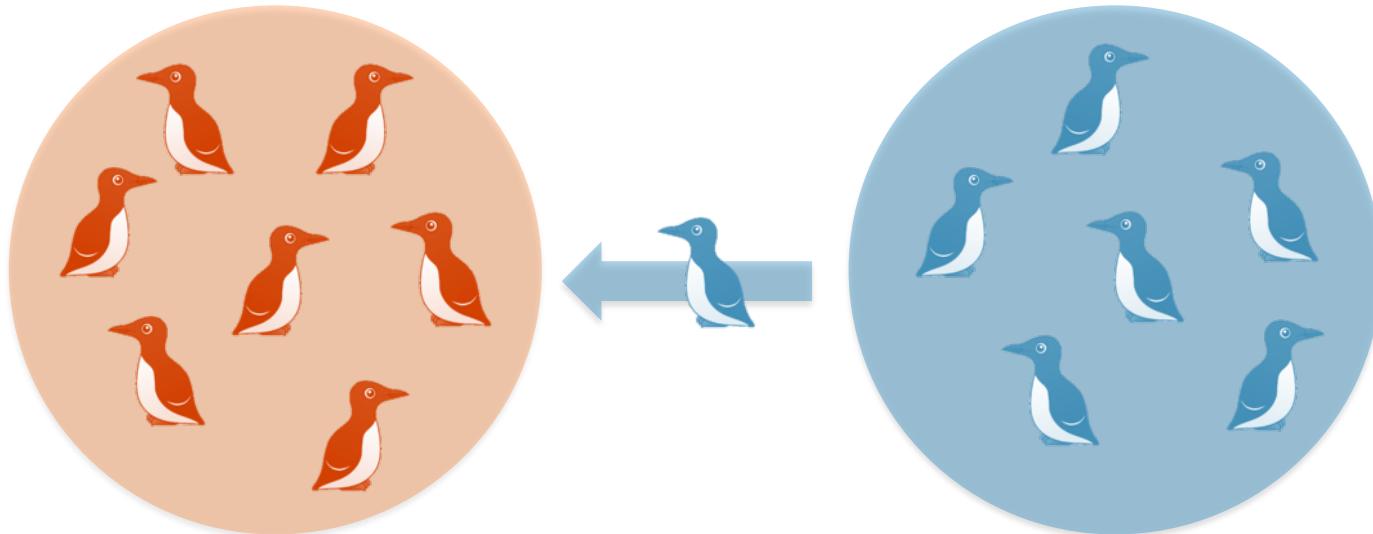
- help understand role of gene flow in adaptation
- show us that we need different approaches to study adaptation

INVITED REVIEWS AND SYNTHESES

Genomics of local adaptation with gene flow

ANNA TIGANO and VICKI L. FRIESEN

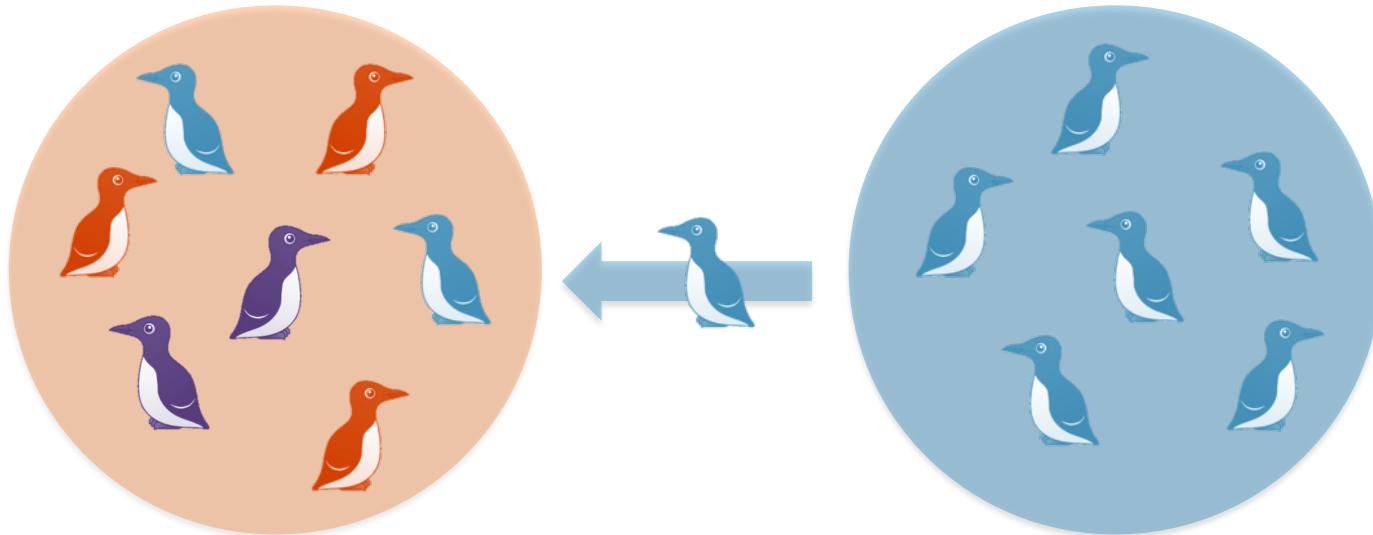
Department of Biology, Queen's University, Kingston, ON K7L 3N6, Canada



Genomics of local adaptation with gene flow

ANNA TIGANO and VICKI L. FRIESEN

Department of Biology, Queen's University, Kingston, ON K7L 3N6, Canada



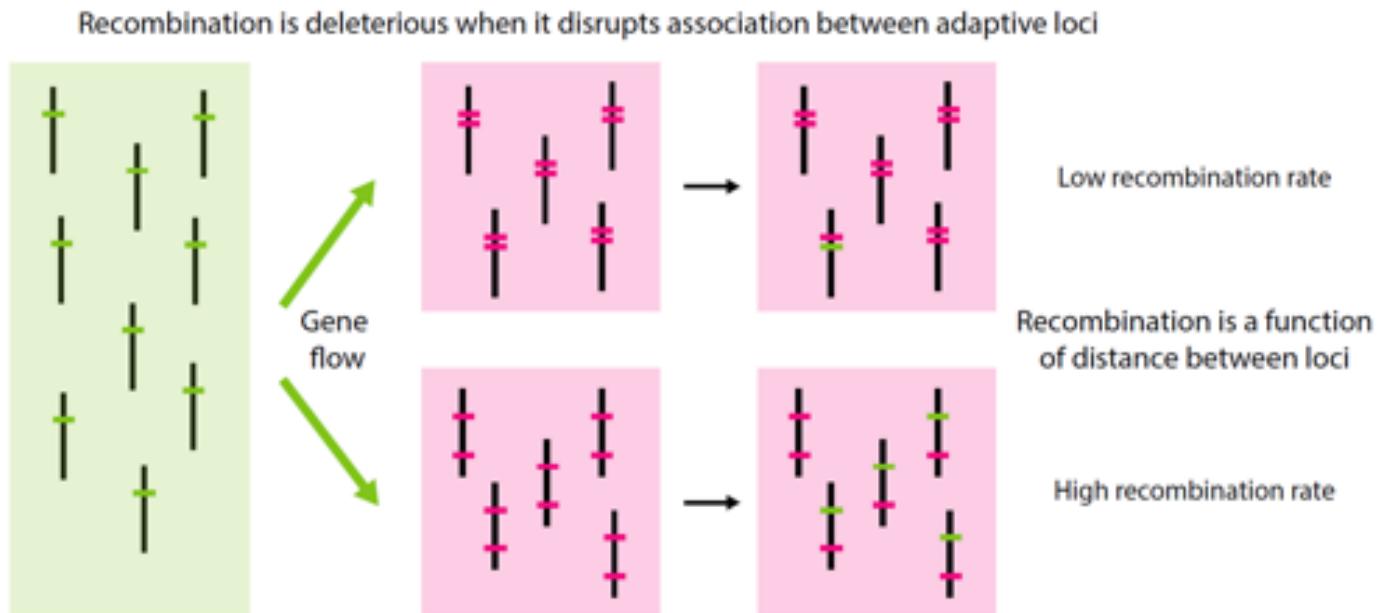
Single-locus theory (Haldane 1930)

If gene flow > selection
locally adapted alleles are swamped

How is adaptation maintained despite gene flow?

Maintenance of local adaptation with gene flow

Most adaptations are polygenic.

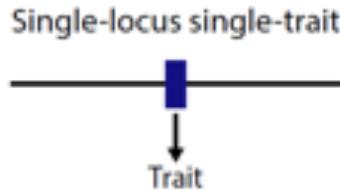


Local adaptation with gene flow favours tight genetic architectures.

Genetic architecture is important for maintaining local adaptation with gene flow.

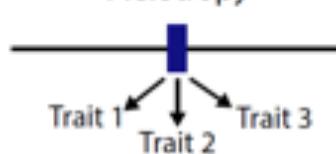
1 locus

No recombination



Single-locus single-trait

Pleiotropy



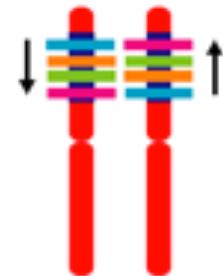
Recombination cannot disrupt association if the causal variant is a single mutation

2+ loci

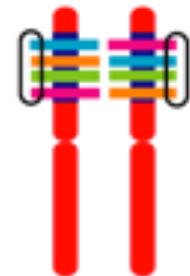


Cold spots
e.g. centromere

Chromosomal rearrangement
e.g. inversion



Epigenetic
modifications



Despite predictions about the disruptive effect of gene flow in adaptation

- local adaptations **can be maintained** despite high gene flow
- **genetic architecture** plays a fundamental role in the origin and maintenance of local adaptation with gene flow

From the Arctic to the desert

Random mating



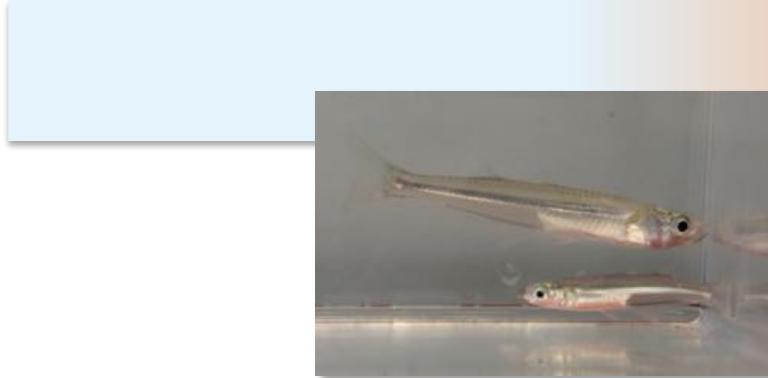
No gene flow



Varying gene flow
across the range

From cold to hot

No gene flow

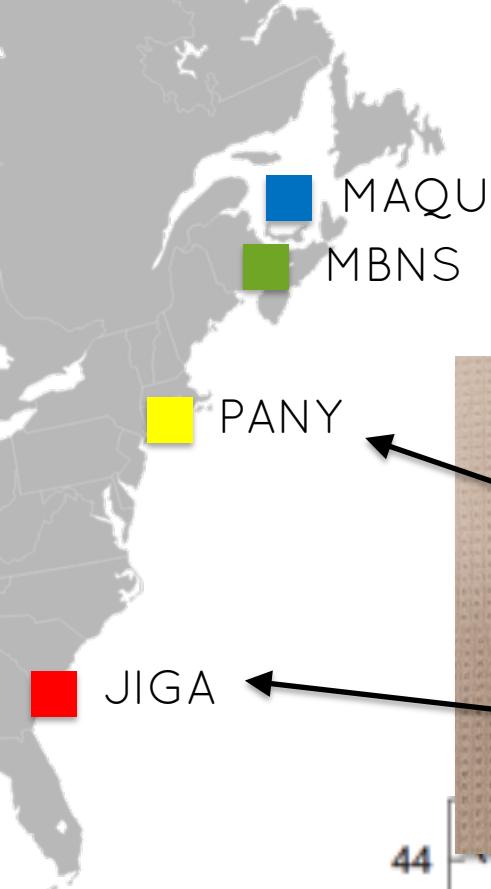


Varying gene flow
across the range

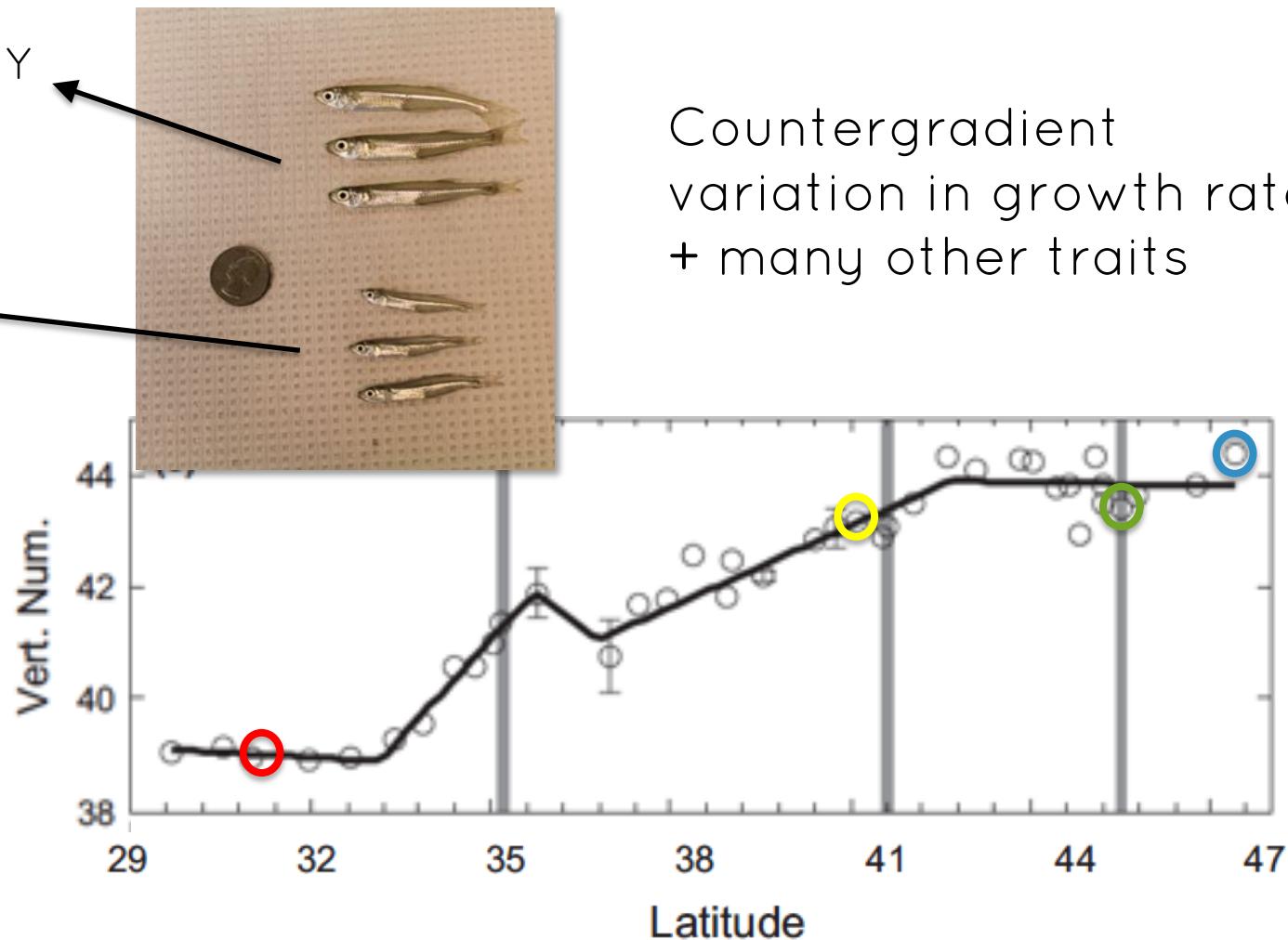
Atlantic silverside
(*Menidia menidia*)



Varying gene flow
across the range



Variation in local adaptation

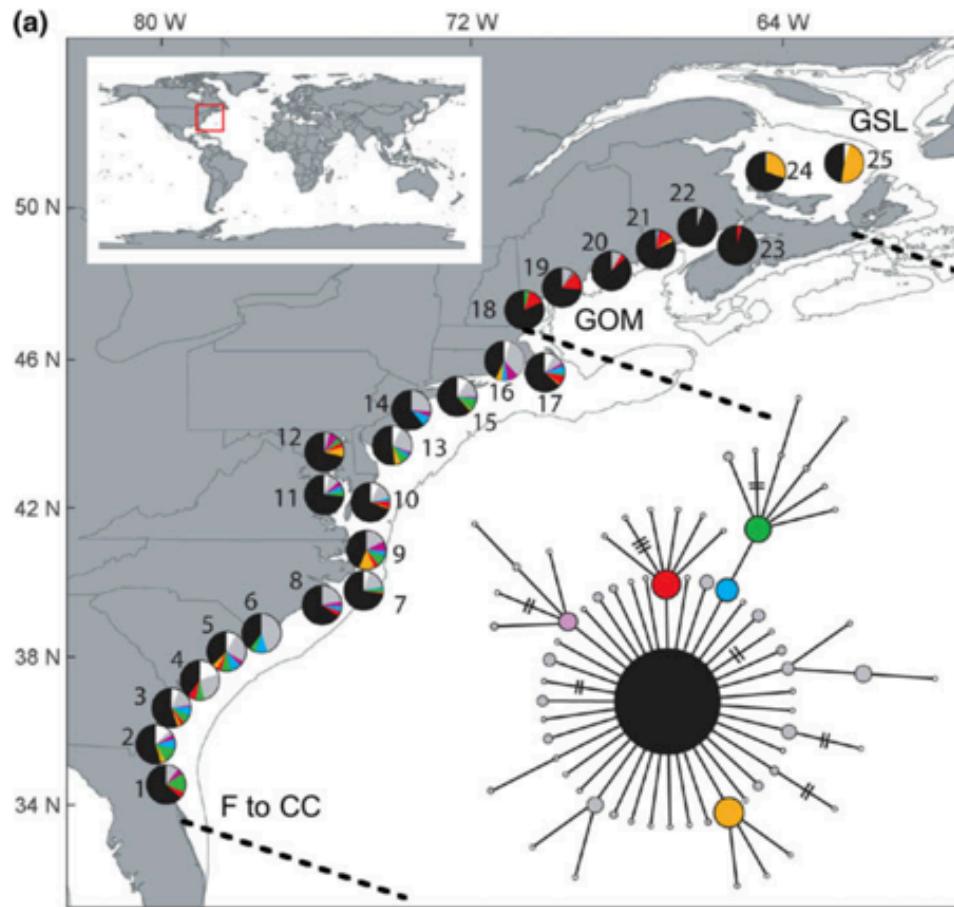


Latitude

Hice et al. 2012

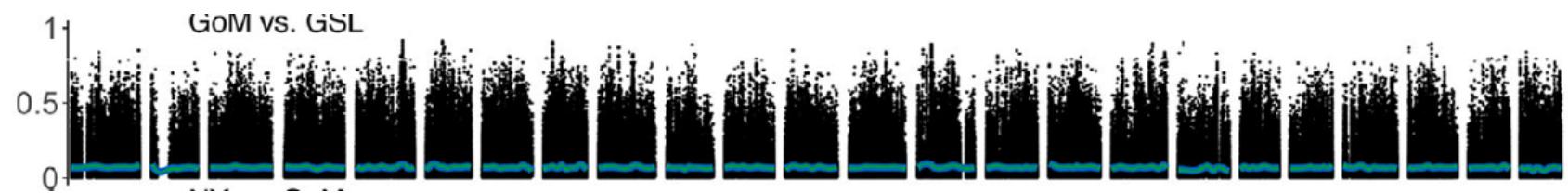
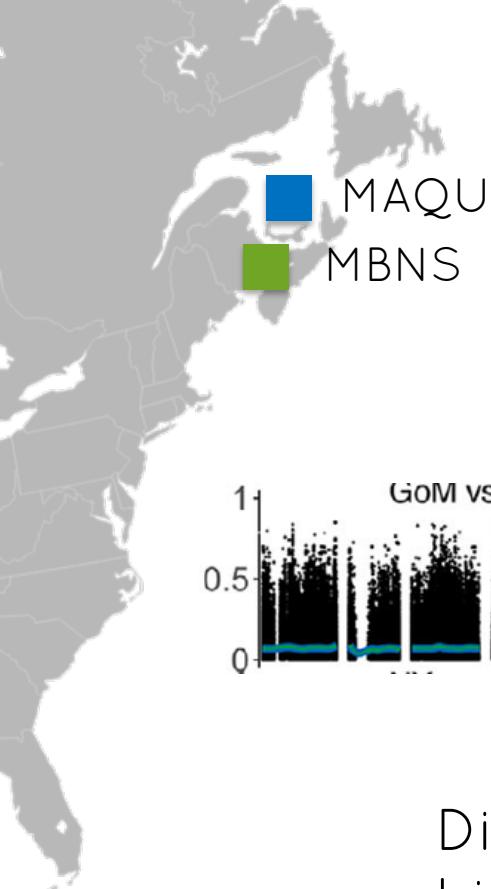
Countergradient
variation in growth rate
+ many other traits

Varying levels of connectivity



Based on one mtDNA locus

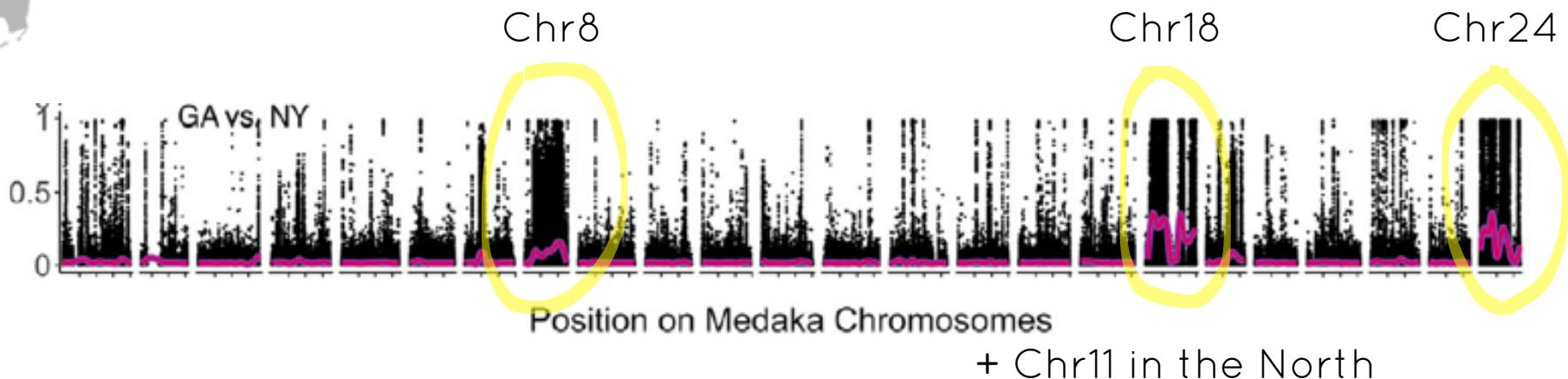
**How are these adaptation maintained with and
without gene flow along the cline?**



Differentiation in the north is very high, so it's hard to identify 'outlier' loci that could be under divergent selection.



Based on transcripts mapped to the medaka genome, four chromosomes show large blocks of differentiation associated with different local adaptations in the south.



Enriched GO terms associated with blocks of differentiation

Metabolism Cartilage morphogenesis
Spermatogenesis Lipid storage Heart formation
Immunity Kidney morphogenesis



Salinity
Length of spawning season
Temperature

How large are those blocks of differentiation?

What mechanisms maintains them in the face of gene flow?

Chromosome-level assembly of the Atlantic silverside genome

+ 210 whole genome resequenced



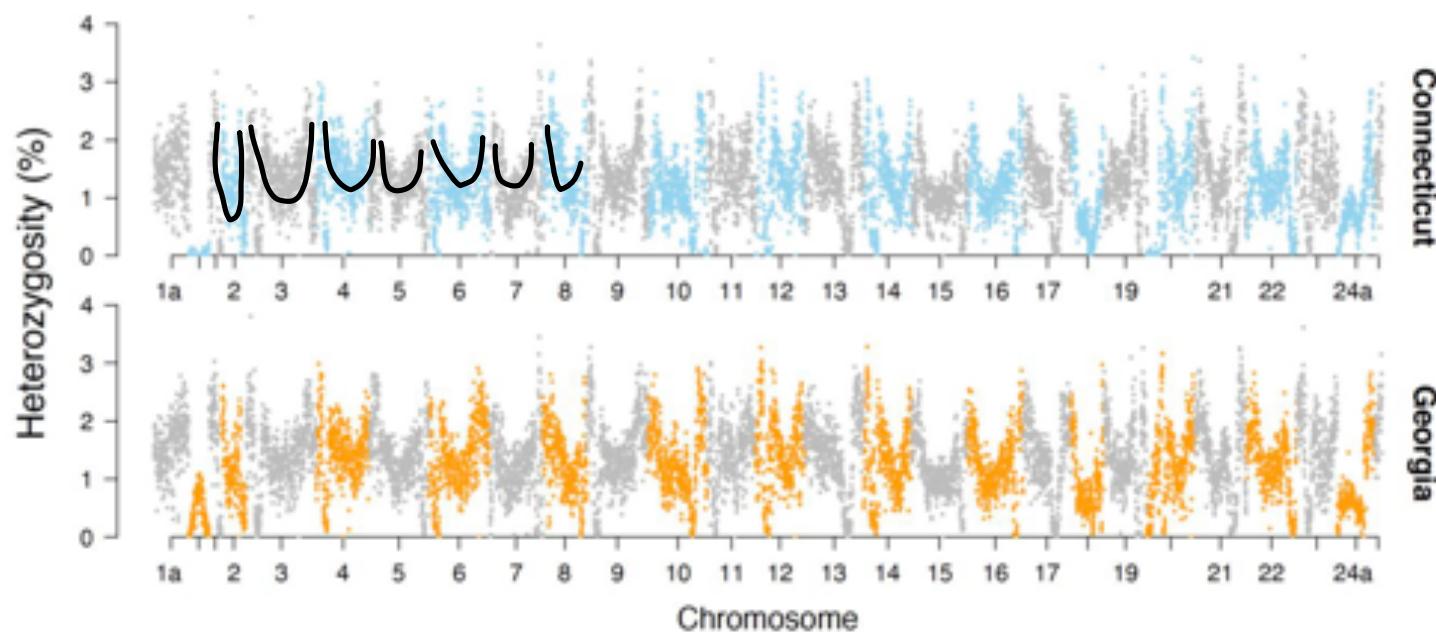
Characterization of the genomic landscape
of diversity and differentiation



Standing genetic variation

Sequence variation

- U-shape pattern

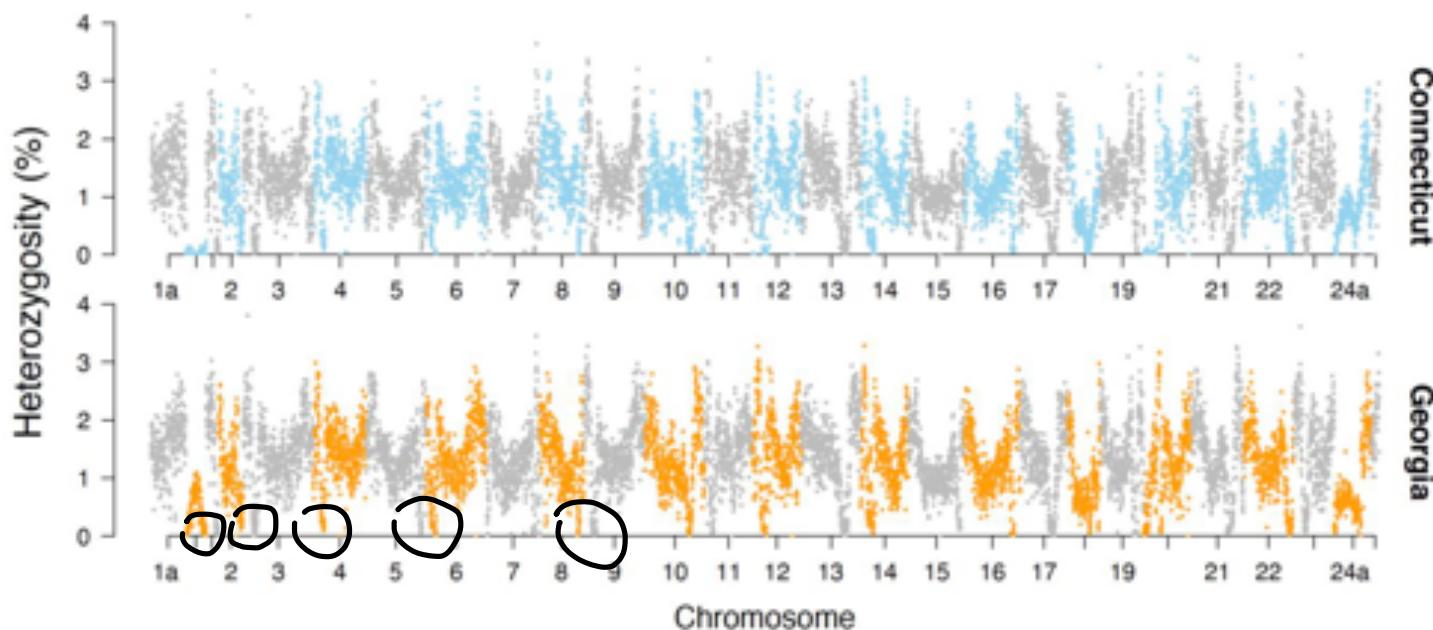




Standing genetic variation

Sequence variation

- U-shape pattern
- One major dip per chromosome

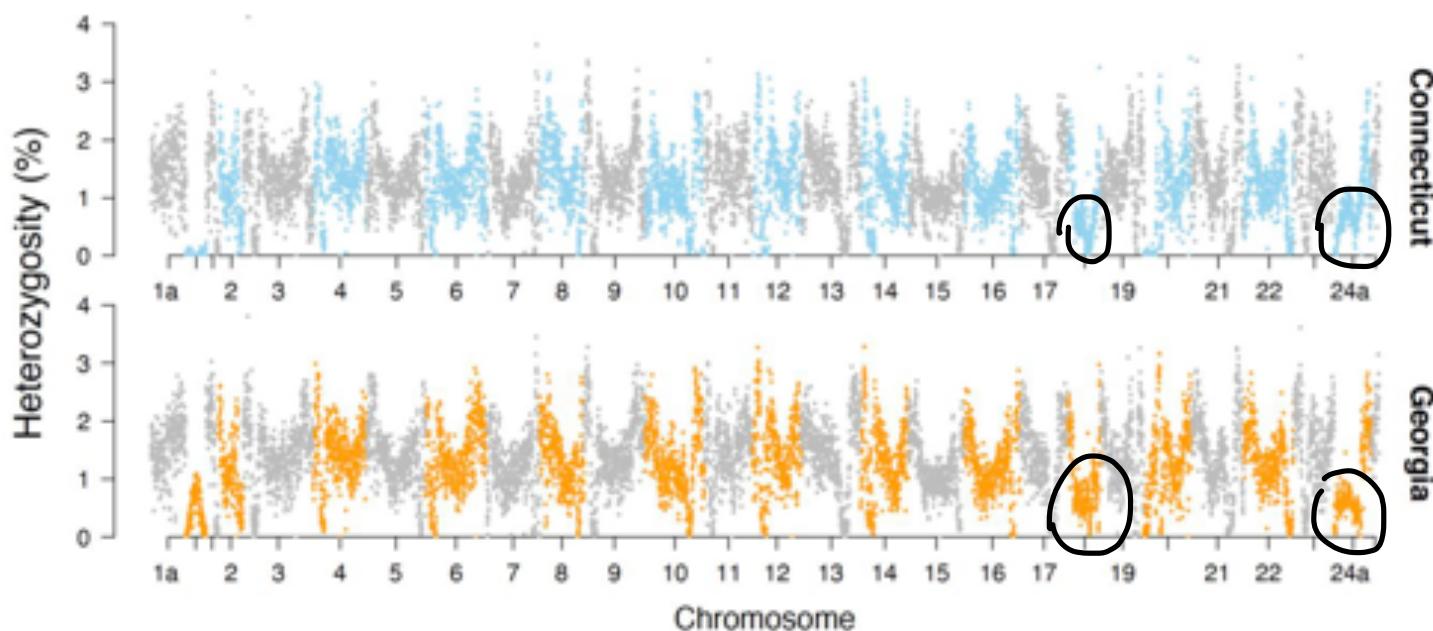




Standing genetic variation

Sequence variation

- U-shape pattern
- One major dip per chromosome
- Swaths of low diversity

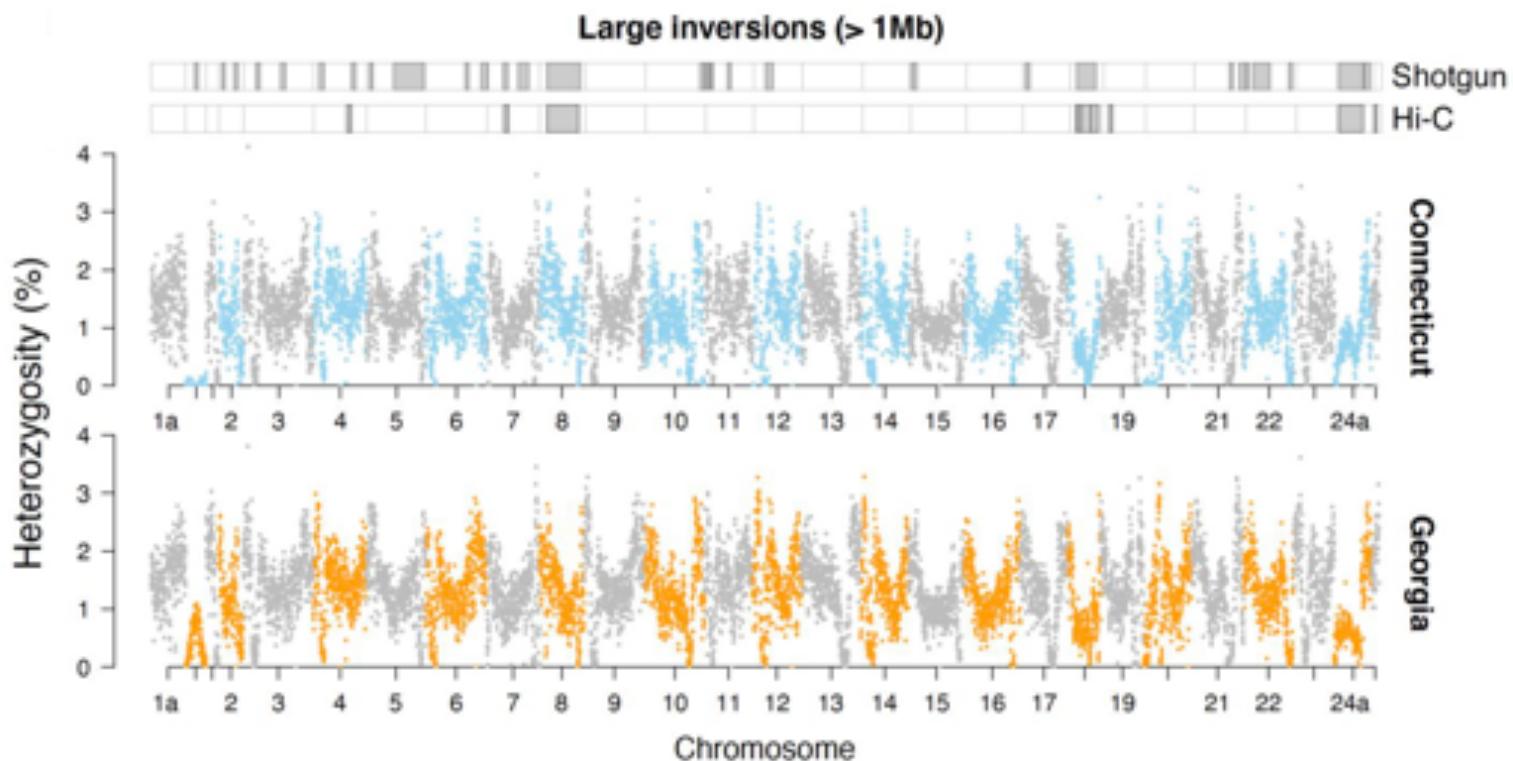




Standing genetic variation

Structural variation

- 4900 structural variants

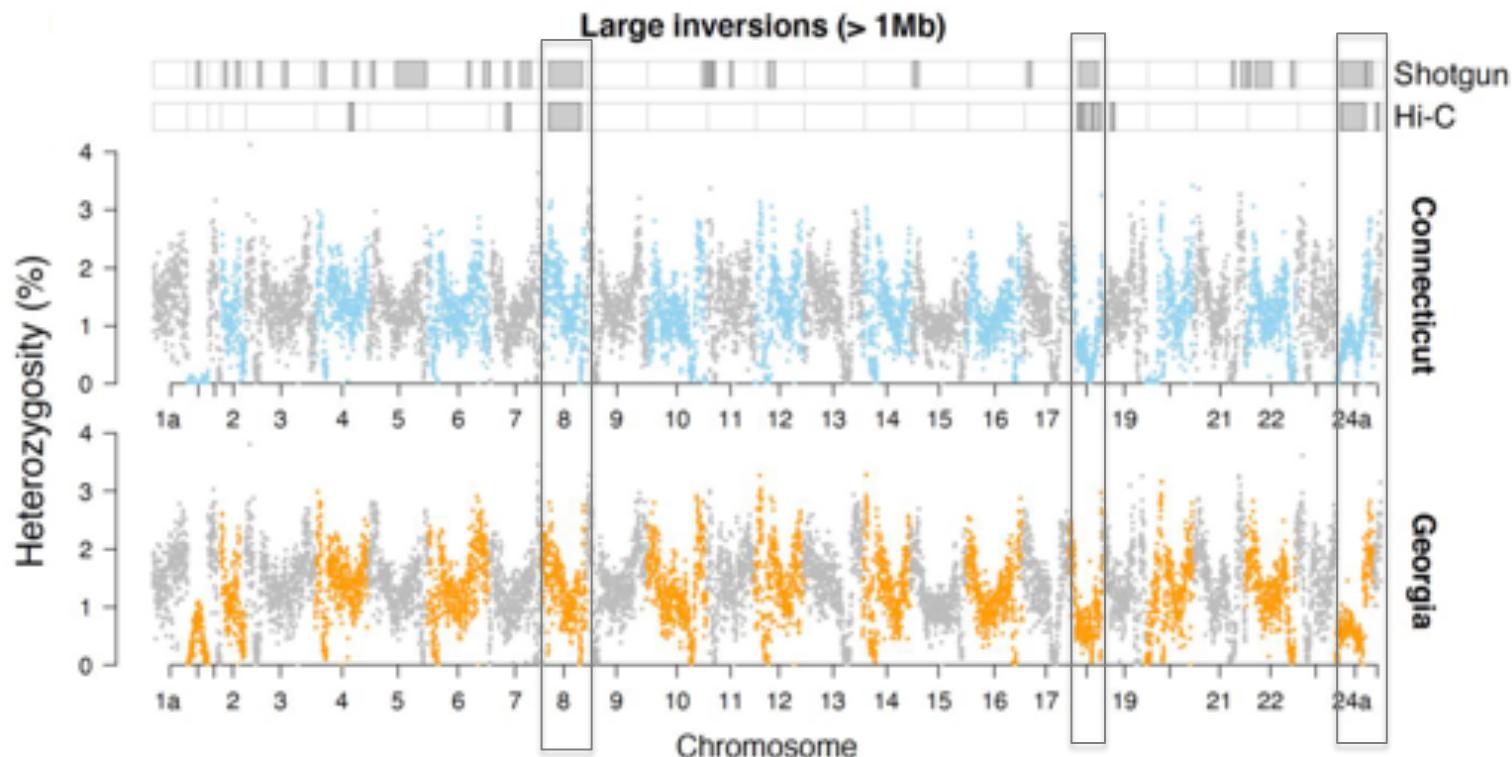




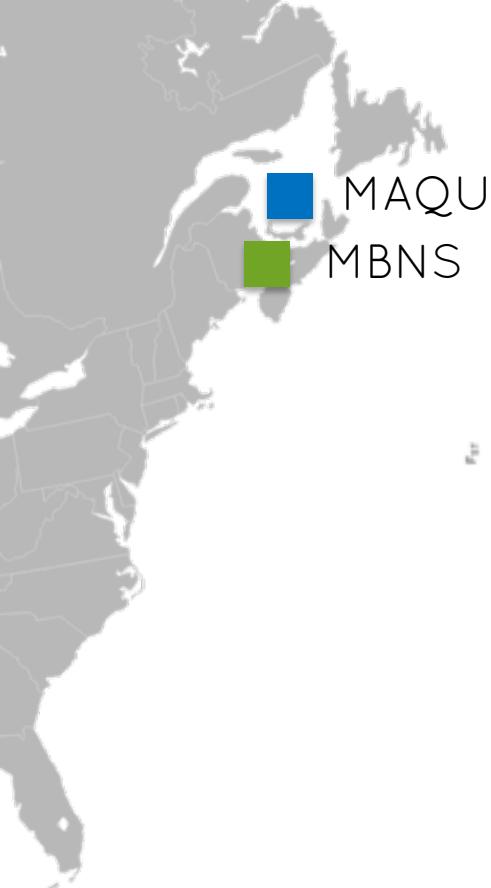
Standing genetic variation

Structural variation

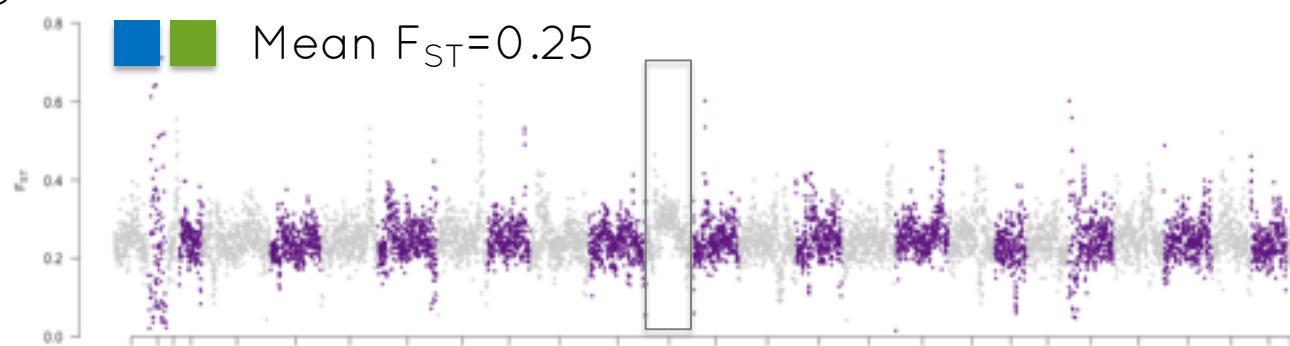
- 4900 structural variants
- Many large inversions

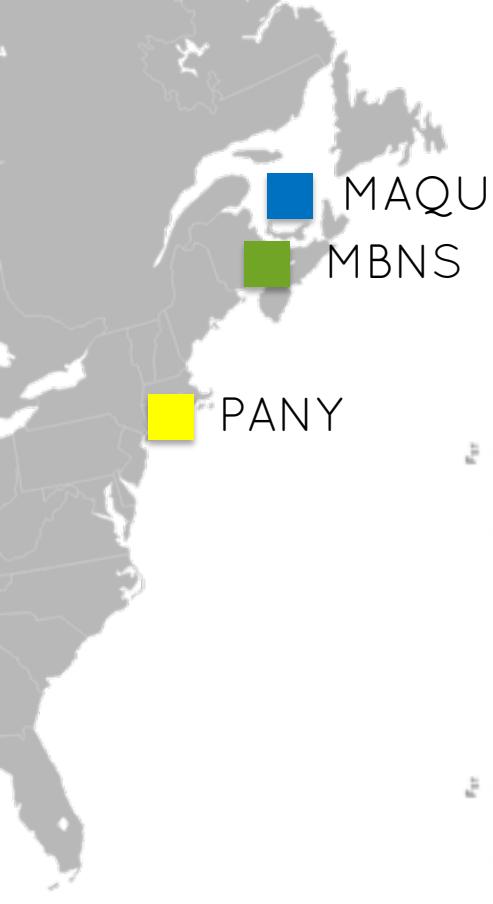


What are the real patterns of differentiation then?

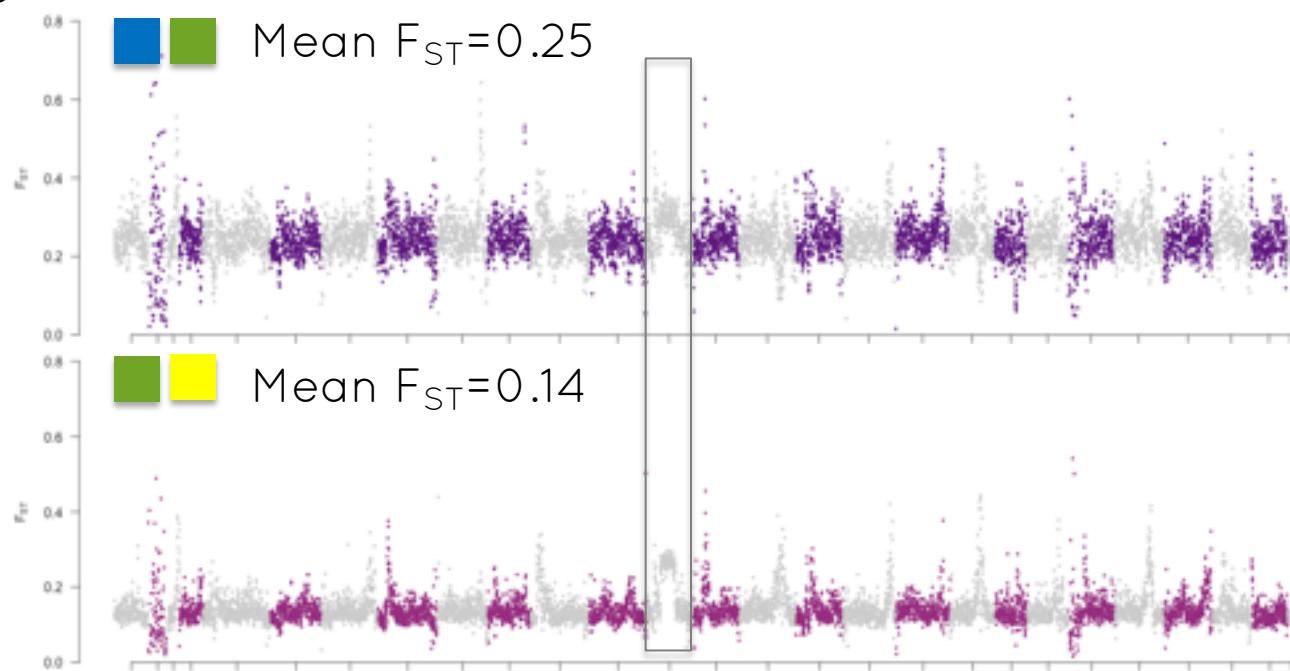


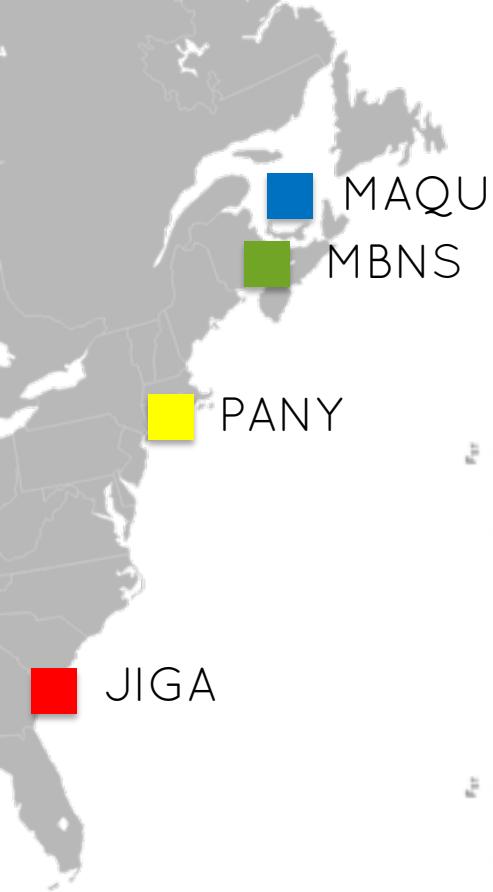
Pairwise differentiation along the cline - F_{ST}



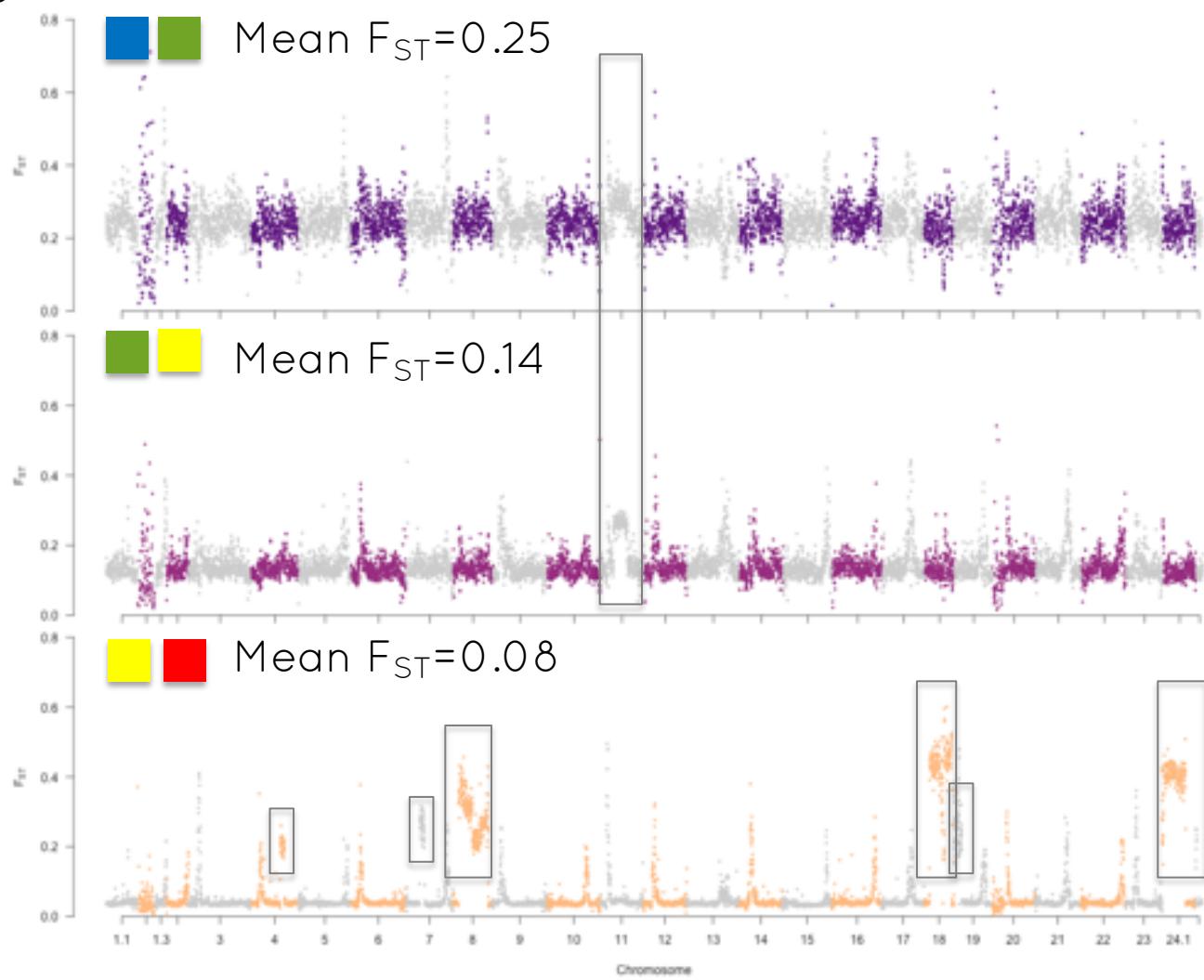


Pairwise differentiation along the cline - F_{ST}

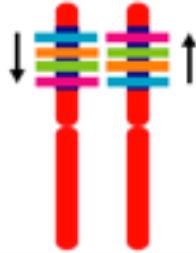




Pairwise differentiation along the cline - F_{ST}



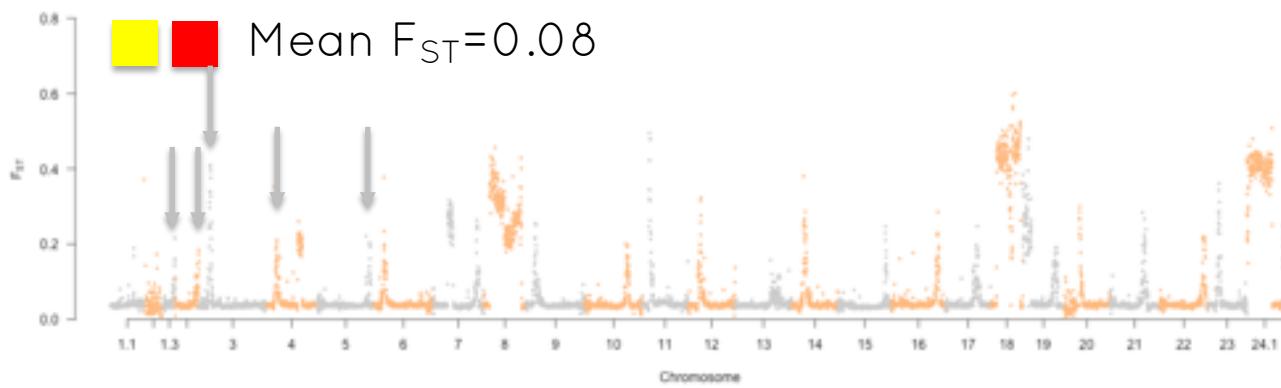
Chromosomal rearrangement
e.g. inversion

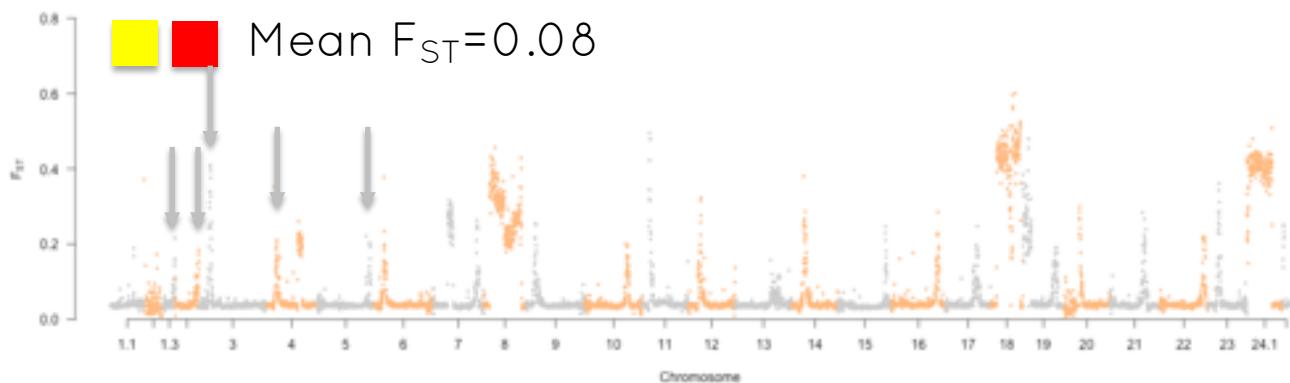


Chromosome	Inversion?	Inversion size
8	Yes	12 Mb
11	Yes	0.3 Mb
		0.9 Mb
18	Yes	5.7 Mb
		3.4 Mb
24	Yes	9.4 Mb
4	Gap in assembly?	-
7	Yes	1.7 Mb
19	Yes	1.2 Mb

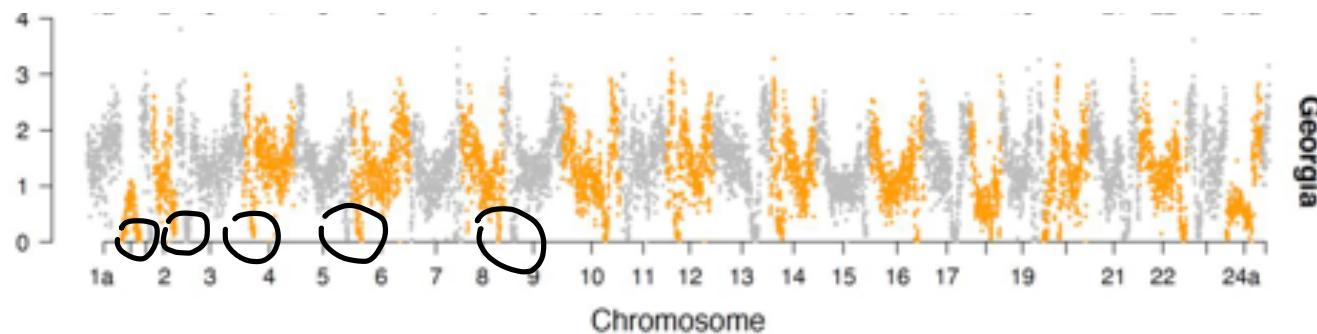
All blocks are associated with inversions.

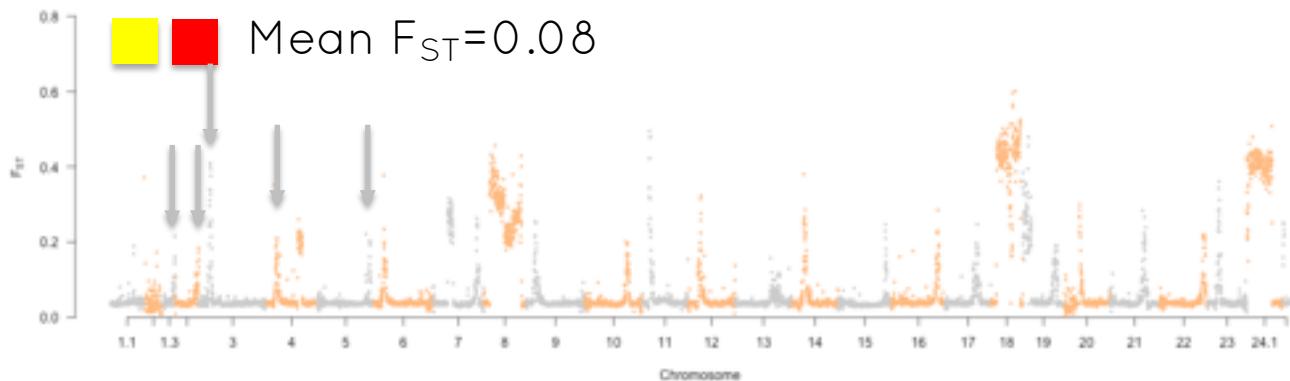
Inversions suppress recombination between rearranged haplotypes thus promoting evolution and maintenance of adaptive divergence



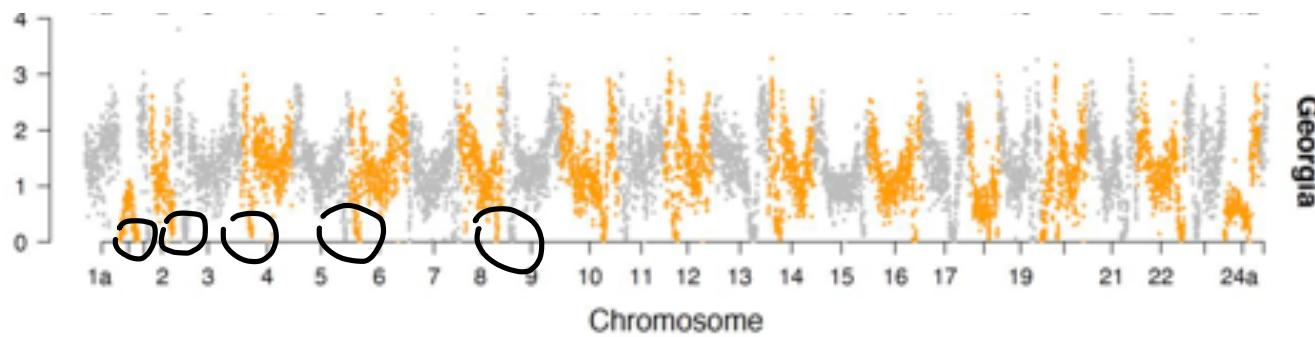


Mountain-like peaks present in each chromosome
are associated with centromeres, not adaptation





Mountain-like peaks present in each chromosome are associated with centromeres, not adaptation



Recombination plays a fundamental role in shaping the patterns of diversity and differentiation in the genome



Adaptation with **gene flow** is common in **marine environments** due to facilitated dispersal and offers the opportunity

- To study the interaction between selection and gene flow

- To identify the genomic basis of adaptive traits using a population genomics approach

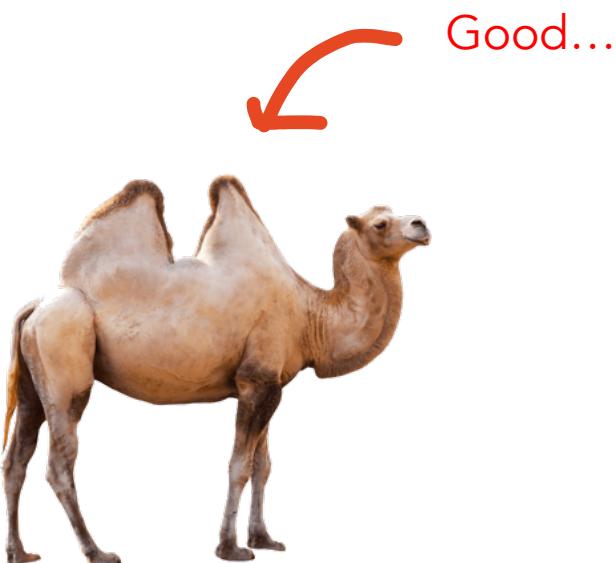
No gene flow



Cactus mouse
(*Peromyscus eremicus*)



Model species to study the
genomic and physiological basis
of desert adaptations



Good...

Model species to study the genomic and physiological basis of desert adaptations



Good...

Perfect!



Cactus mouse
Peromyscus eremicus

- Extreme adaptations to the desert
- Small
- Amenable to life in captivity and experimental manipulation

Experimental manipulation of dehydration

Severe acute **dehydration** treatment in the cactus mouse

- Behaviorally intact: active and healthy
- Physiological stress but limited tissue injury and apoptosis in the kidneys
- Loss of weight → metabolic water?

A human would have most likely died.

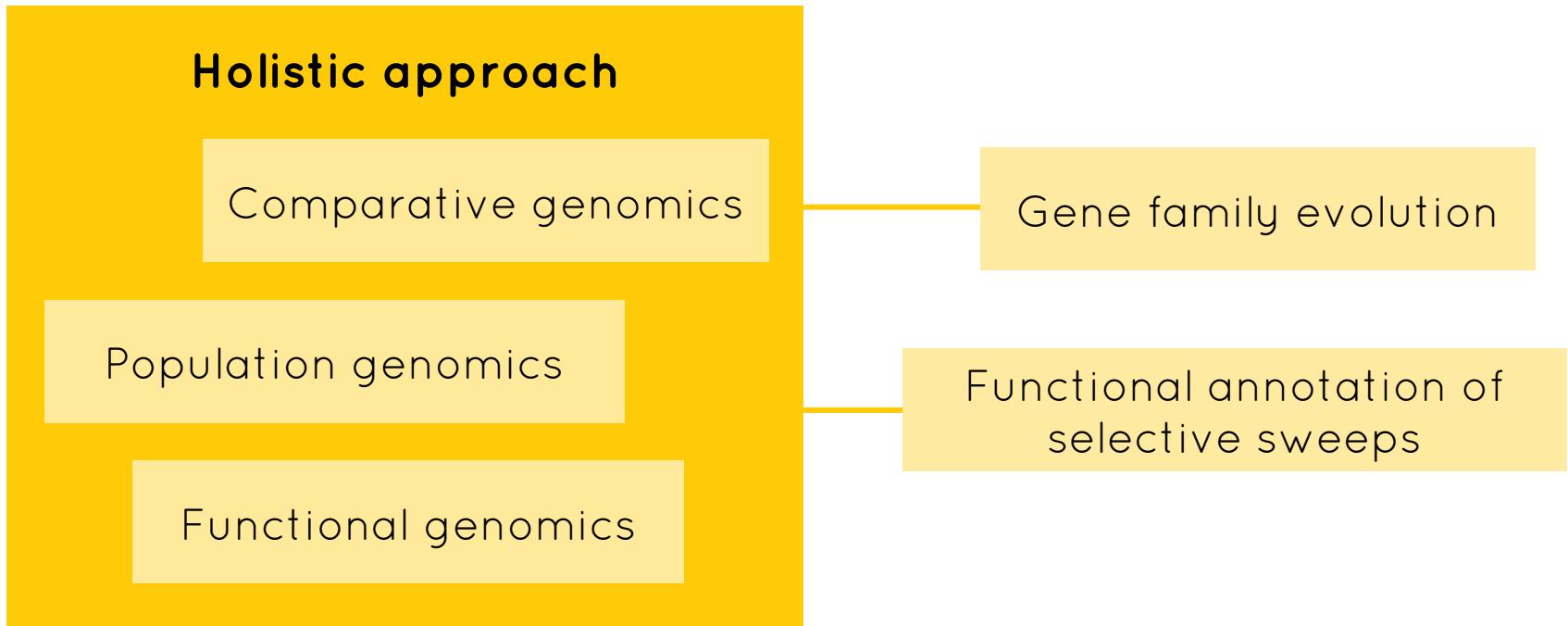


What is the genomic basis of
desert adaptations in the Cactus mouse?

(which are expected to be highly polygenic)

Chromosome-level assembly of the Cactus mouse genome

+ 26 whole genome resequenced

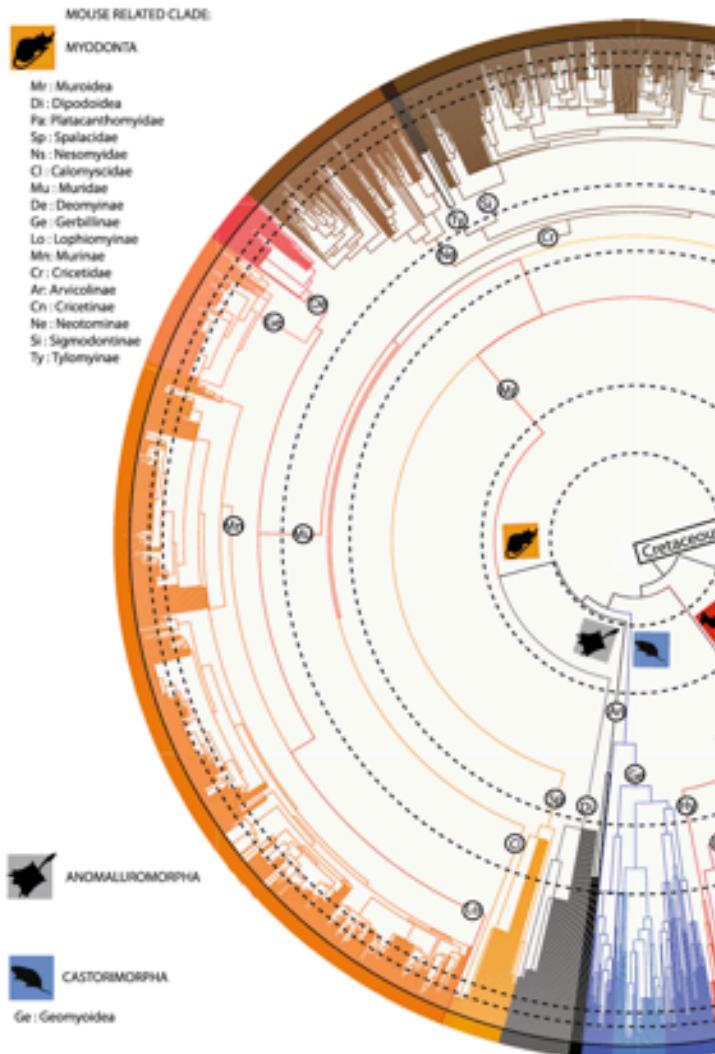


Gene family evolution

Focus on 18 species in Myodonta,
Rodents

Clustering on genes in
orthogroups and species tree in
Orthofinder2

Analysis of gene family
(orthogroups) in CAFE



Gene family evolution

Gene families

Sperm motility (4)

Ribosomal proteins (4)

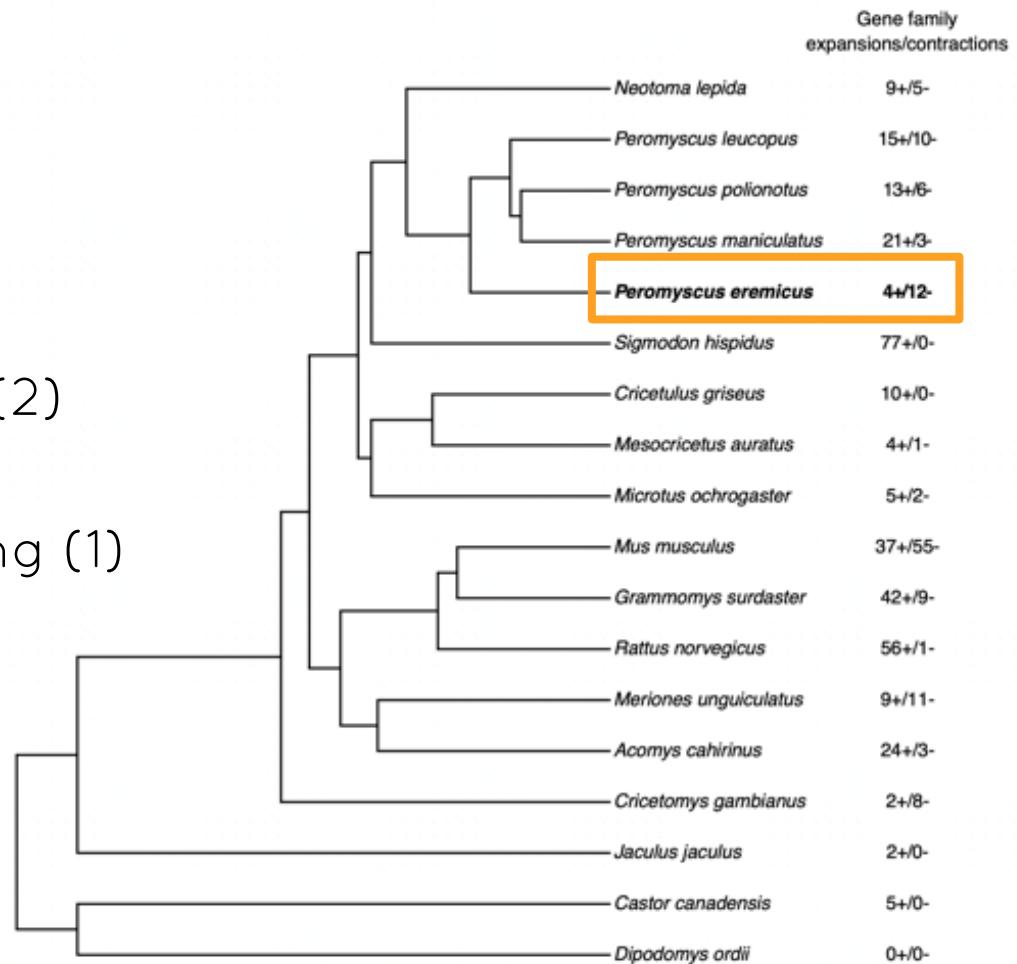
Immune response (3)

Ubl conjugation pathway (2)

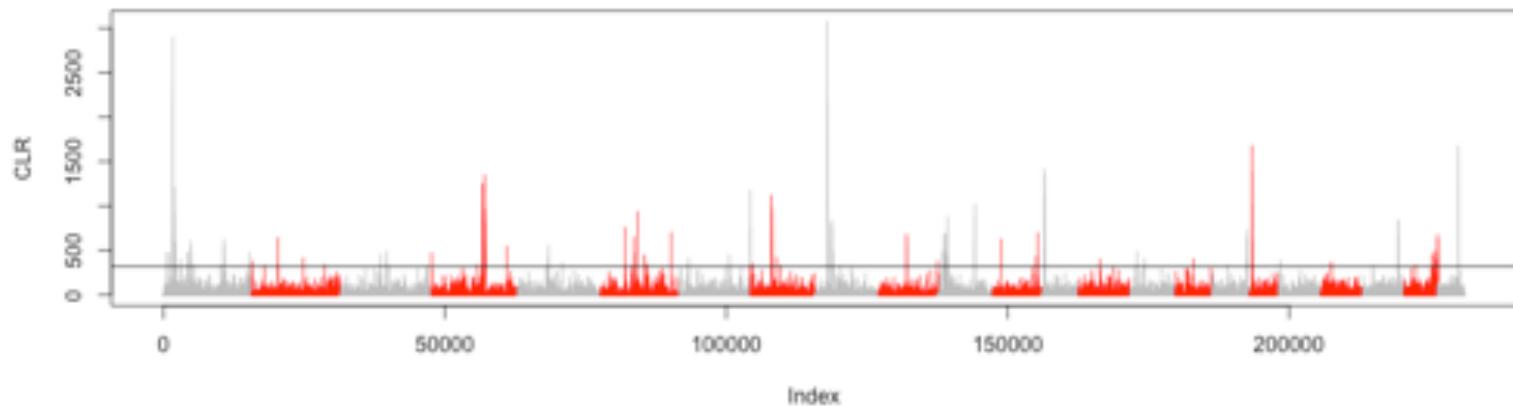
pheromone reception (1)

cytoskeletal protein binding (1)

prohibitin (1)



Selective sweeps – 25 Cactus mouse genomes

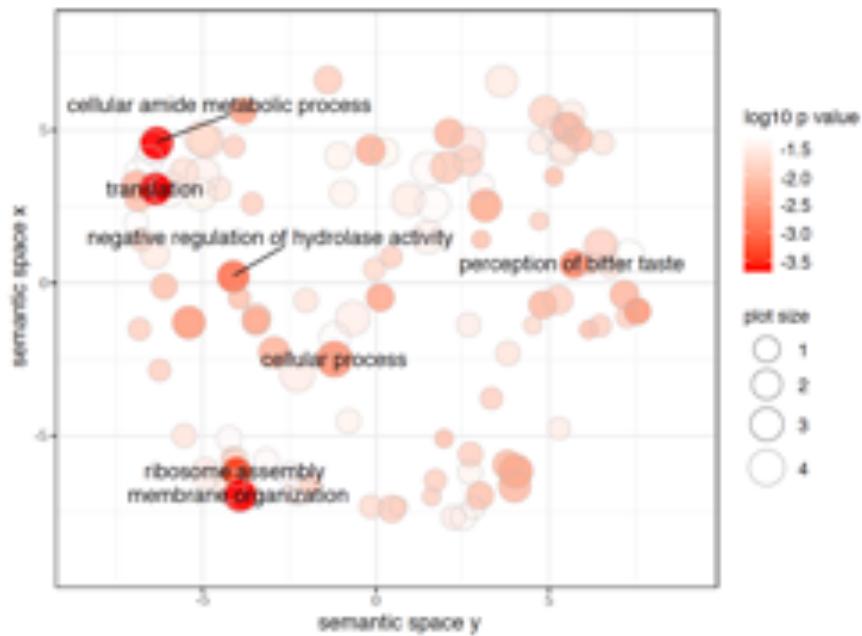


Detection of 232 putative selective sweeps
(above 99.9th percentile line)

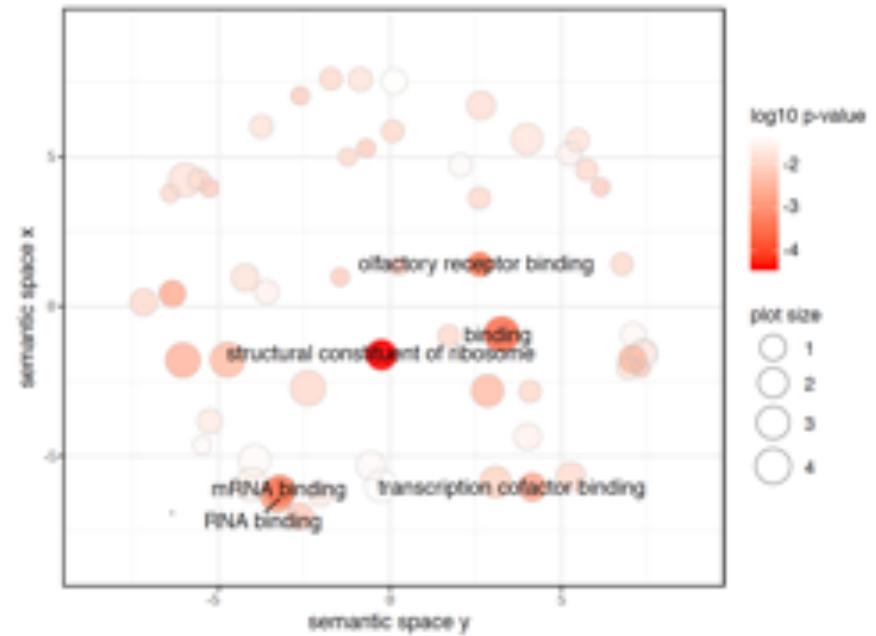
- Peaks in all chromosomes
- 119 of these cluster in 44 wider peaks
- List of 186 ‘closest genes’

Gene Ontology enrichment analysis + clustering algorithm to condense redundant terms

Biological process GO terms



Molecular function GO terms



+ GO cellular component = ribosome (highly significant)

Gene family evolution + Sweepfinder

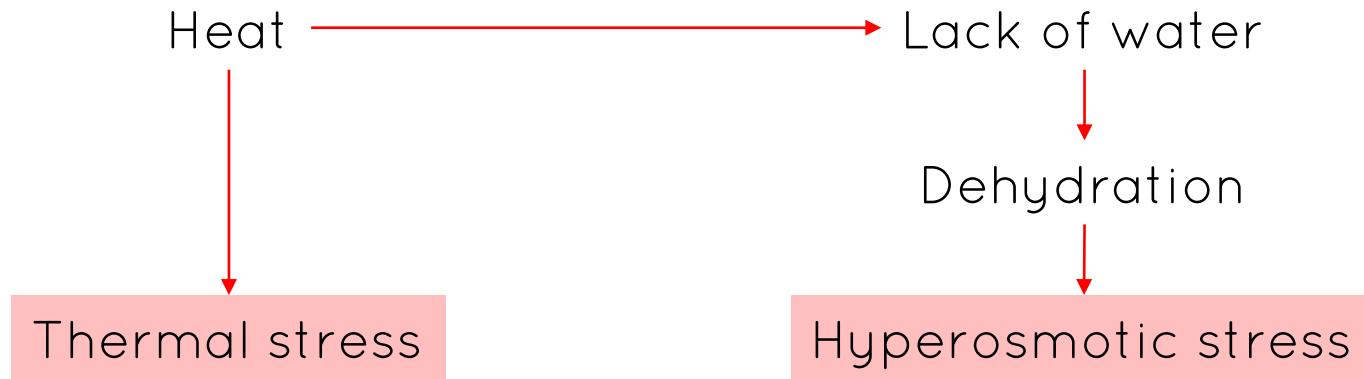
Ubl conjugation pathway mRNA binding binding

translation cytoskeletal protein binding

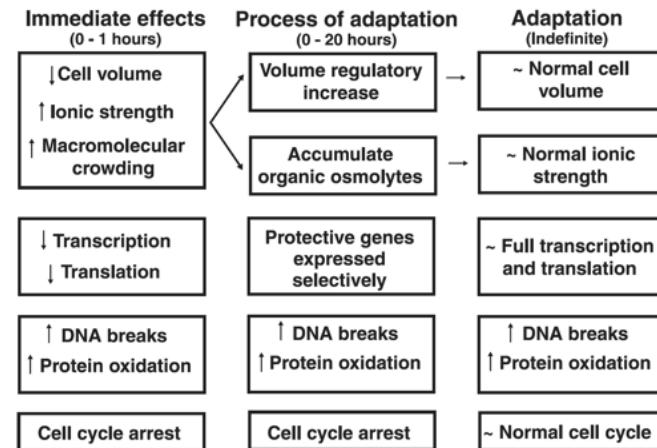
Ribosomal proteins Structural constituents of ribosome

Cellular amide metabolic process Ribosome assembly

Synthesis and degradation of proteins



GO category	Recurrent GO terms (%)
Biological processes	Translation (35.7%); metabolic processes (28.5%)
Molecular function	ATP binding (37%); structural constituent of ribosome (37%)
Cellular component	Ribosome (50%); membrane (37%)



Negative regulation of cell death

A mechanism helping preventing apoptosis could be advantageous in the kidney, but potentially in other tissues as well.

Perception of bitter taste

Bitterness (toxicity) is a defence mechanism against herbivory in the desert.

Lots of bitter plants and insects in the desert...

**Many traits, many genes, selective sweeps and
gene family expansions/contractions associated
with recent adaptations to desert.**

**One single area
of differentiation**

Random mating



**Widespread signature of
selection across the genome**

No gene flow



Varying gene flow
across the range

**A handful of areas
maintained by inversions**

Candidate genes for adaptation

- Point to the **physiological basis** of adaptive traits
- Help identify **what** species/populations/morphs are adapted to (reverse ecology)
- Help understand the **process** of adaptation and the **genomic architecture** of adaptive traits
- Are useful to understanding and predicting **evolutionary response** to environmental change



Cornell University



Acknowledgements



Environment and
Climate Change Canada



University of
New Hampshire

