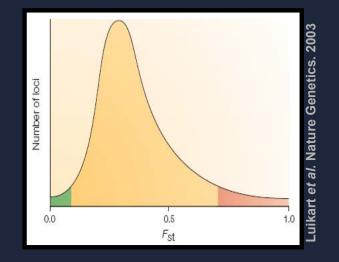


#### **Research in the Lawton-Rauh Lab: Plant Population Genetics** *Arabidopsis* sister species and the Hawaiian silversword alliance

#### **Brief overview:**

**'Comparative Molecular Population Genetics & Molecular Evolution'** *...tools to investigate population dynamics and gene-specific processes* 

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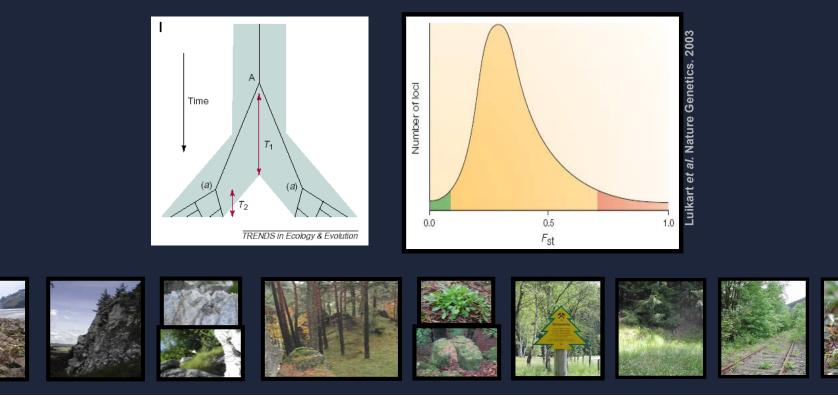






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### European Arabidopsis lyrata (A. petraea):

## glacial tills in Scandinavia

Vatnajokull glacier Iceland (photo by Ramos-Onsins)

#### on isolated Dolomitic rock outcrops in Central Europe

Harz Mtns, Germany







Franconian Alps, Germany





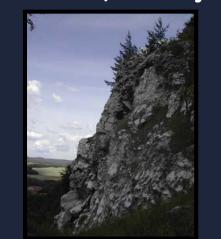


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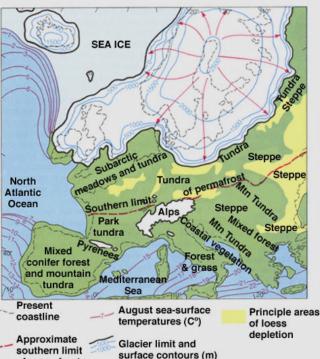
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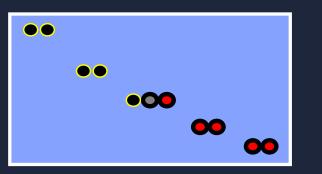


of permafrost



### **Relative Roles of Demography vs. Selection** *in shaping genetic variation: Arabidopsis petraea & Arabidopsis arenosa*

- 1). Frequency of unique polymorphisms increases with geographical distance between populations
- 2.) Frequency distributions of sequence diversity differ between populations with different postglacial histories
- 3.) Between-species isolation by distance is correlated with higher levels of linkage disequilibrium than within-species isolation by distance



- Population, A. petraea
- Population, A. arenosa
- Population, hybrids of A. petraea & A. arenosa



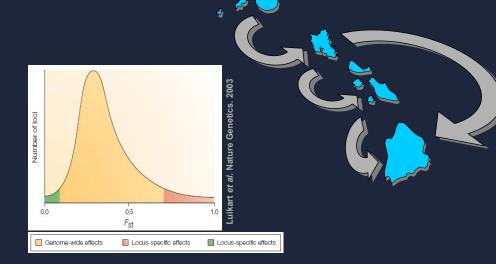
### Hawaiian Silversword Alliance: origin & radiation





What are the relative roles of genome dynamics (heredity) morphology (form) physiology (function) in shaping the adaptive evolution in the Hawaiian silversword alliance?

What are the relative contributions of: demography & selection in shaping genetic and genomic variation?





How many genes are involved in the morphological and physiological differentiation among Hawaiian silversword alliance species?















### Mahalo nui loa (und Danke schön)!

#### Lawton-Rauh Lab Folks:

Cindy Climer, Ph.D. Student HSA, Arabidopsis petraea-A.arenosa Brad Rauh, Research Associate, Manager HSA, Madia, Arabidopsis petraea

Margaret Beaudrot Clemson, HHMI/SCLife Honors Intern Katie Horton Clemson, Research Assistant

Chelsea Reighard Clemson, HHMI/SCLife & Calhoun Intern Jennifer Smith

Francis Marion Uni, NSF-REU Intern, S '07 Sarah Walker

Clemson, GEN-491 Intern 2006-07

**Collaborators, HSA:** 

Bruce Baldwin UC-Berkeley Elizabeth Friar Rancho Santa Ana Botanic Garden David Remington UNC-Greensboro Rob Robichaux University of Arizona

Collaborators, Arabidopsis sp:. Juliette deMeaux Max Planck Institute, Cologne Marcus Koch Universität Heidelberg

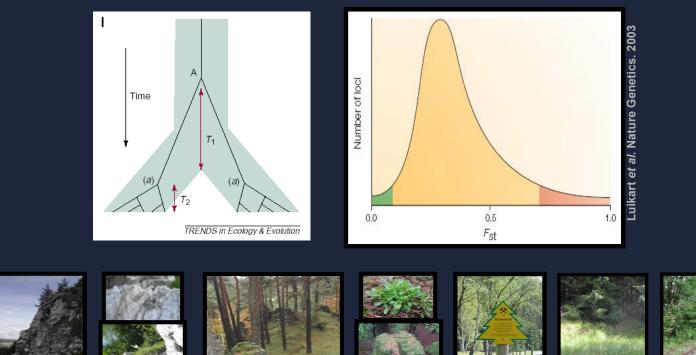




#### **Research in the Lawton-Rauh Lab: Plant Population Genetics**

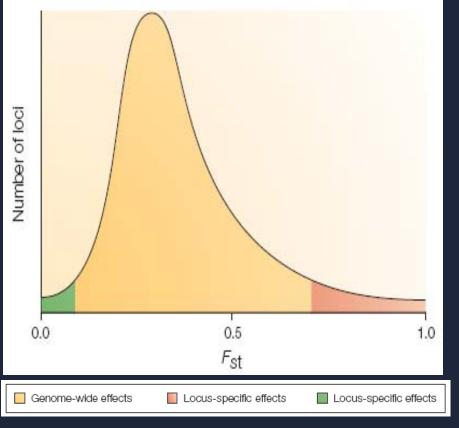
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### **'Population Genomics': Brief Introduction**



#### relevant to:

association genetics functional genomics

(human) disease mapping crop science, breeding conservation genetics

Luikart et al. Nature Genetics. 2003

### **Nucleotide Sequence Diversity**

ATCGTGTGCCCATAAACCCTTTACTCCTTACGGGC •••C••••••••••••••••••••••••••••••• ••••••G••••G••••G••••• 

Expect some rare polymorphisms, some singletons "singleton":

single nucleotide polymorphisms found in 1 individual

# Allele/haplotype segregation patterns differ among populations

•																 		** **
<b>.</b>	760		770	780	-	790	800	 10	820	-	30	840		850	86	 870		880
standard							CTTGACTCC											
consensus	T	c															• • • • • •	
56-5 i1	т																•••••	
d3																		
s5	T	. č.						 								 		
c4	т							 		c		<b>F</b>	c			 		
r3	т	c		c				 		c				. <b>.</b>		 d		
80-4	<b>.</b> T	c						 								 		
91-3	<b>.</b> T							 								 		
71-1	T		• • • • • • •															
e2	<b>.</b> T																	
73-2	•••••		•••••															
83-2 50-3	••••• <u></u>							 								 		• • • • •
50-3 e4																		
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r2	<b>.</b> T																	
50-1	T																	
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83-3	•••• <u></u>																	
j1 61-4	·····																	
61-4 70-1		· · · · ·																
73-5	· · · · · · · · · ·	. č.																
f2	т	. č.																
spit 11n3	T	. C																
spit 38n23	<b>.</b> T	c		c				 		c		<b>F</b>	c			 		
spit 17	<b>.</b> T	c		c														
spit 8	T	c		c				 					•••••	<b>.</b>		 		
spit 16	<b>.</b> T	c												• • • • • •				
spit 59n25	•••• <u>T</u>	c	• • • • • • •											•••••		 		
spit 18n21	•••••	c						 						•••••		 		
spit 40n29	· · · · · ·							 								 		
spit 14n11 spit 26n13		· · · · ·																
spit 26n13	· · · · · · · · ·	·																
spit 19		. č.						 										
spit 15	т	. č						 								 		
spit 9	т	.c.																
spit 12	T	c																
spit 33n16	T	c		c				 		c		<b>F</b>	c			 		
opro_comro								 								 		

### Demography

- Population expansion
- Population contraction (bottleneck)
- Population subdivision
- Gene flow, migration

...affects the entire genome (all loci at the same time)

### **Genetic variation is shaped by many factors** Nucleotide Diversity ( $\theta$ ) = 4 N<sub>e</sub> $\mu$

**Genetic Drift** 

**Gene Flow** 

**Inbreeding Depression** 

Population Subdivision Population Bottleneck Population Expansion Habitat Fragmentation
Interspecific Competition

Large-scale Events

Effective Population Size (N<sub>e</sub>)

### Non random association of alleles...

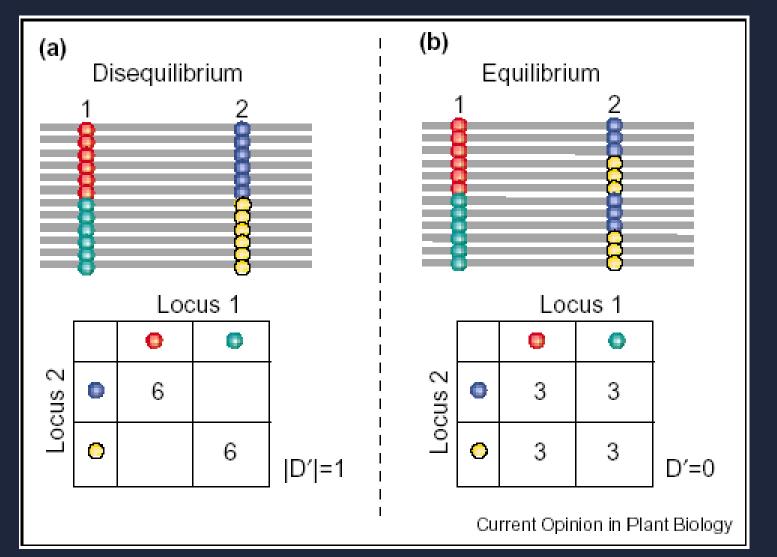
Linkage disequilibrium and selection for linked loci?



Migration of unique alleles that appear to be linked (via gene flow)?



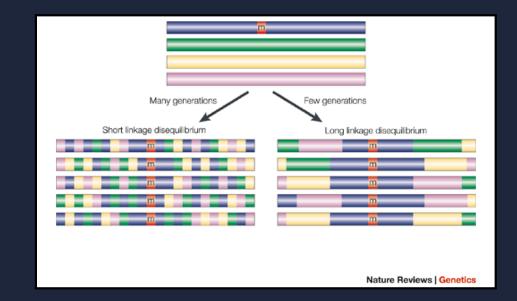
### Linkage Disequilibrium (LD): Non-random association among alleles



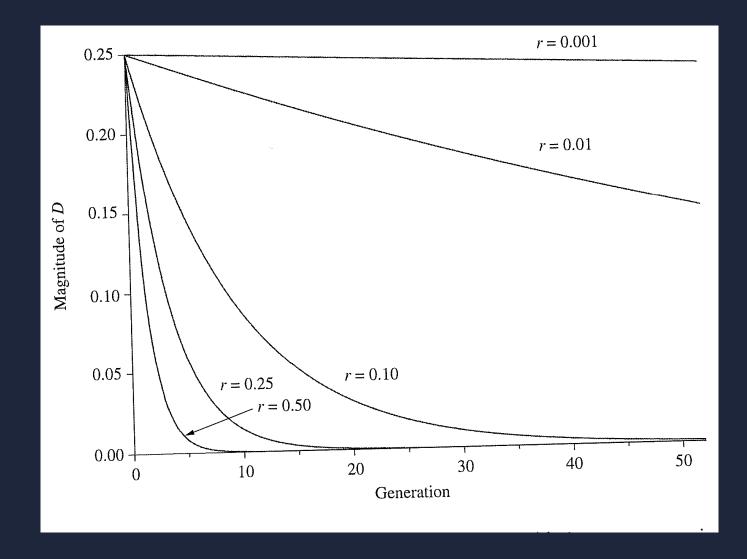
Rafalski A. (2002) Applications of single nucleotide polymorphisms in crop genetics. Curr. Opin. Plant Bio. 5:94-100.

### Linkage Disequilibrium Influences Nucleotide Sequence Diversity

Linkage Disequilibrium: Non-random association among loci/sites Depends on recombination rates & distance between loci/sites



### Linkage disequilibrium breaks down at different rates, correlated with recombination rate (c)



### Recombination Rate Impacts LD and *Nucleotide Sequence Diversity*

When a mutation arises, it is in complete linkage (LD) with entire genome.

Over generations, recombination breaks up LD in proportion to genetic distance between loci.

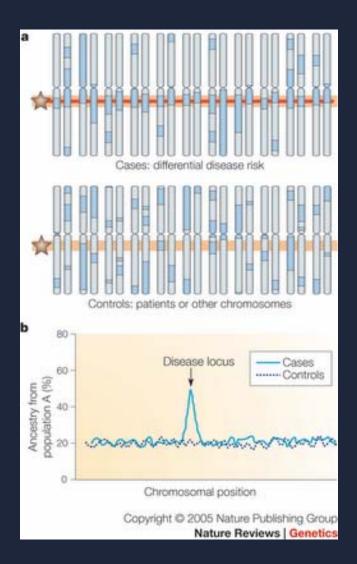
### Recombination Rate Impacts LD and *Nucleotide Sequence Diversity*

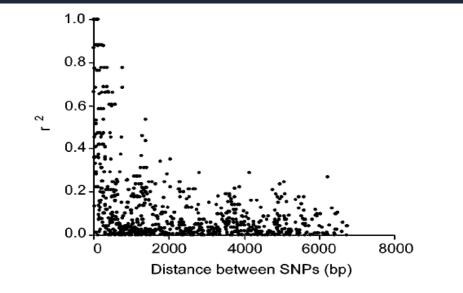
When a mutation arises, it is in complete linkage (LD) with entire genome.

Over generations, recombination breaks up LD in proportion to genetic distance between loci.

Other factors 'interrupt' linkage breakdown Selection Demography

### Linkage Disequilibrium (LD) breaks down in regions outside selection





**Figure 2** Linkage disequilibrium (LD) decay plot of *shrunken 1* (*sh1*) in maize. LD, measured as  $r^2$ , between pairs of polymorphic sites is plotted against the distance between the sites. For this particular gene, LD decayed within 1500 bp. Data from Reference 48. *Flint-Garcia & Buckler 2003.* 

### **Molecular population genetics**

#### **Genetic scales to consider:**

Single gene: selection or neutrality at 'favorite/candidate' locus (YFG)

<u>Genomic neighborhood</u>: selection or neutrality in region containing YFG,

related to effective recombination rate (linkage disequilibrium)

**Entire genome:** changes in population size, gene flow, population structure

...all relate to timeframe and organismal life history; scope of hypothesis determines analysis tools used Molecular evolution, phylogenetics, population genetics

### **Molecular population genetics:**

Different ways to estimate and compare levels of genetic variation

#### **Different genetic markers:**

AFLP Microsatellites Organelle Loci (cpDNA, mtDNA) Nuclear Loci (Coding regions, Introns) General loci Candidate genes and 'your favorite gene (YFG)'

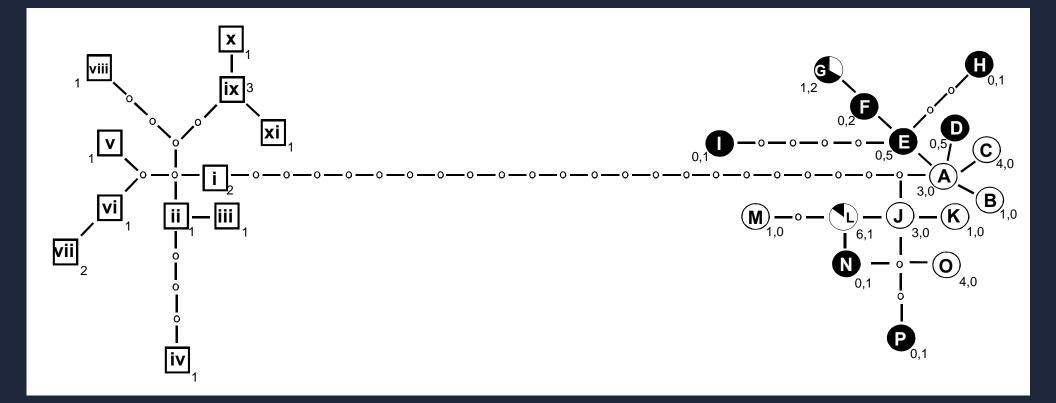
#### **Must consider:**

Levels of genetic variation/polymorphism Mutational model limitations Analytical power for detecting specific processes Allele/haplotype age Gene divergence times (for molecular evolution studies)

### Phylogenetic trees for the population level: haplotype network analysis

Statistical parsimony-based haplotype network: ASAPETALA1-B





#### **Shared alleles between taxon 1 and taxon 2:** Testing *Isolation* vs. *Migration* using the coalescent & Bayesian statistics

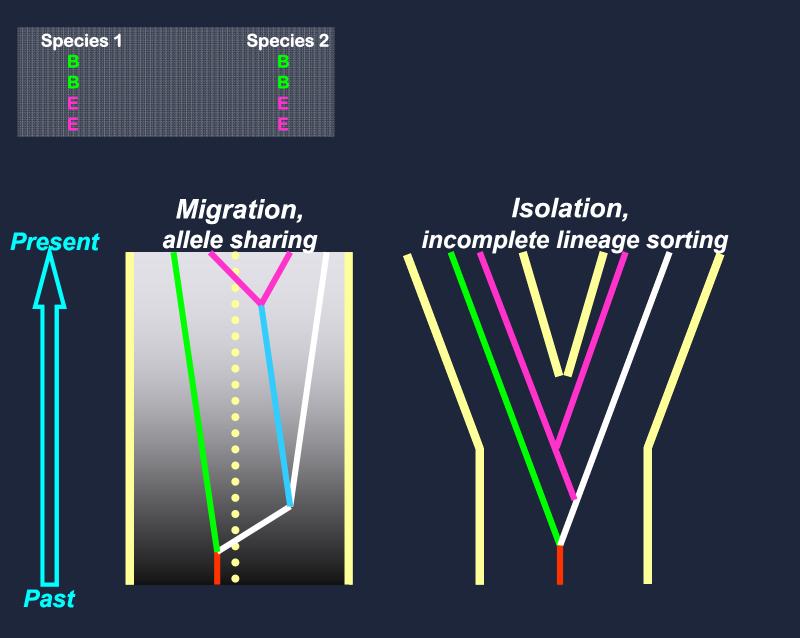


figure modified from: Wakeley J (1996) Theoretical Population Biology 49, 369-386

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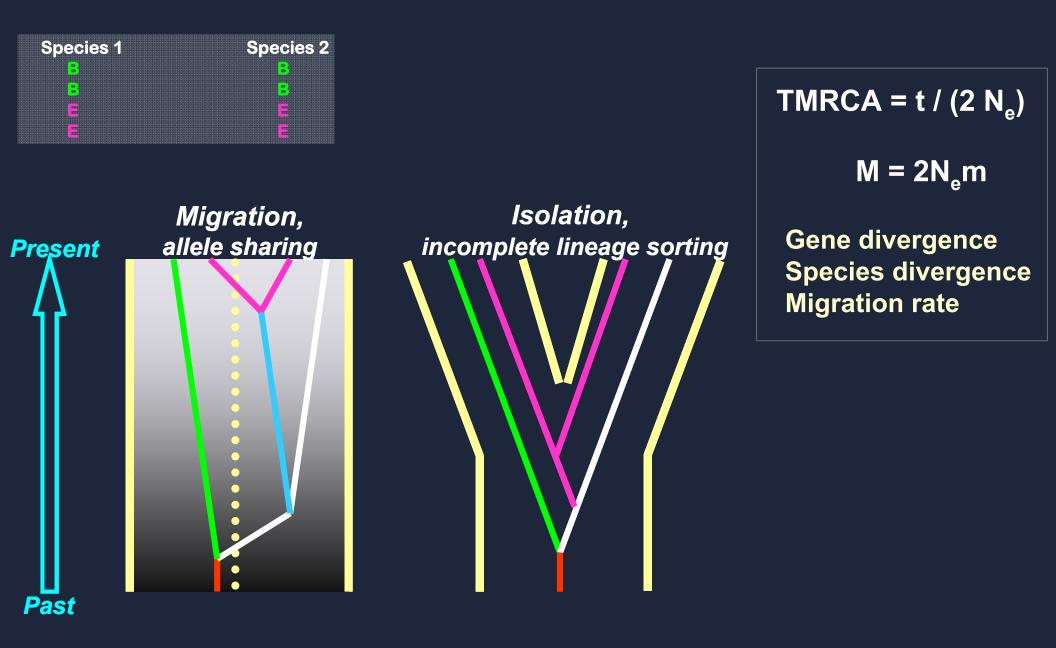


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# Testing "Isolation" vs. "Migration" using the coalescent & Bayesian statistics

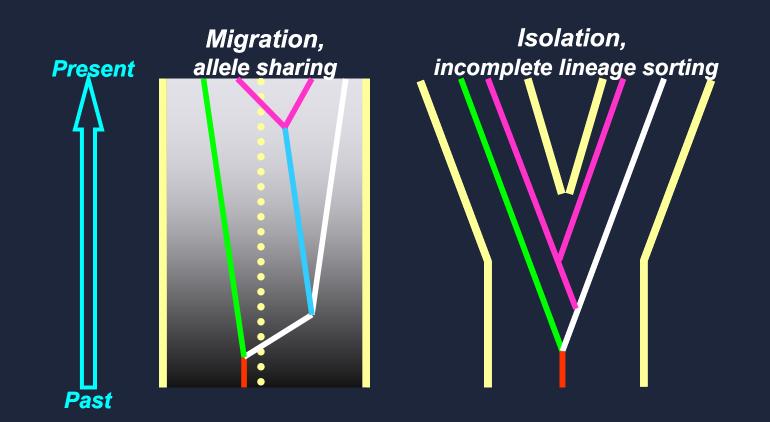
Nielsen R and Wakeley J (2001) Genetics 158, 885-896

### $TMRCA = t / (2 N_e)$

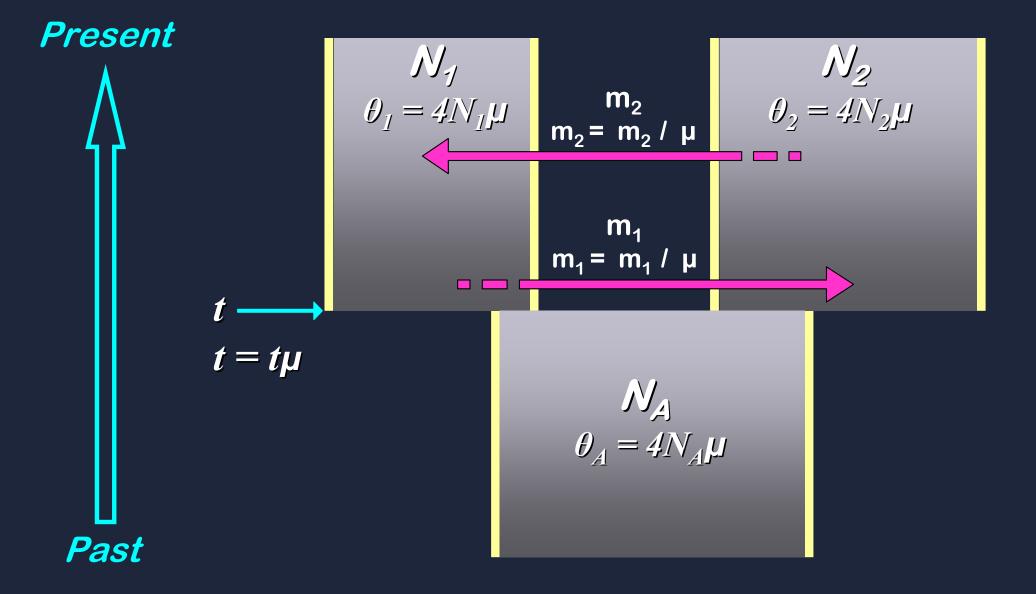
### $M = 2N_em$

Gene divergence Species divergence

Species divergence (credibility interval) Migration rate (credibility interval)



#### Isolation with migration model: gene flow directionality (Hey et al. 2004)



### **Distribution of estimates across loci:**

coordinates of 'favorite/candidate' genes?

#### **Population expansion** Demography-equilibrium

**YFG** 

0

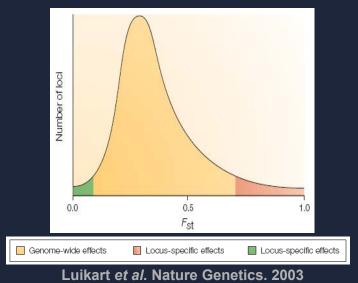
2

3

-2

-1

-3



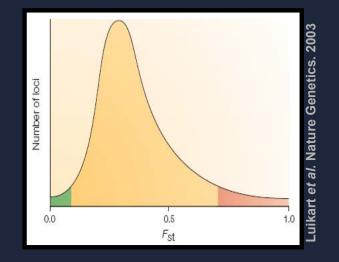


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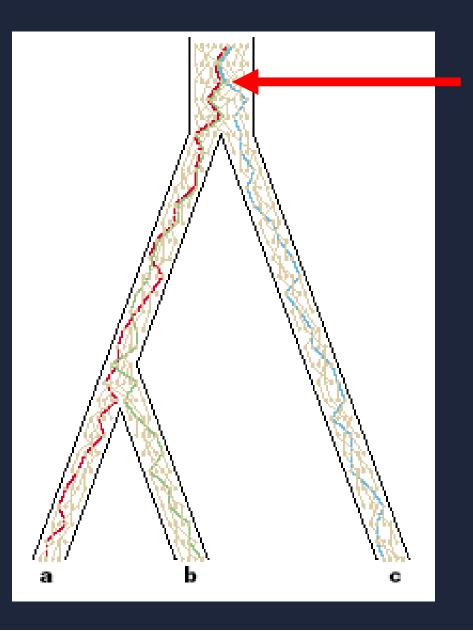
> Amy Lawton-Rauh Dept. Genetics & Biochemistry Clemson University AmyLR@clemson.edu www.clemson.edu/~amylr





### **Estimating Migration depends upon T**

(T = time to most recent common ancestor)



coalescence @ common ancestor species (entire genome) alleles @ specific locus

