

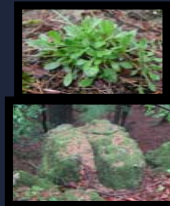
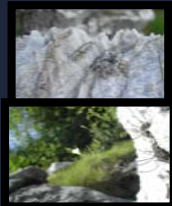
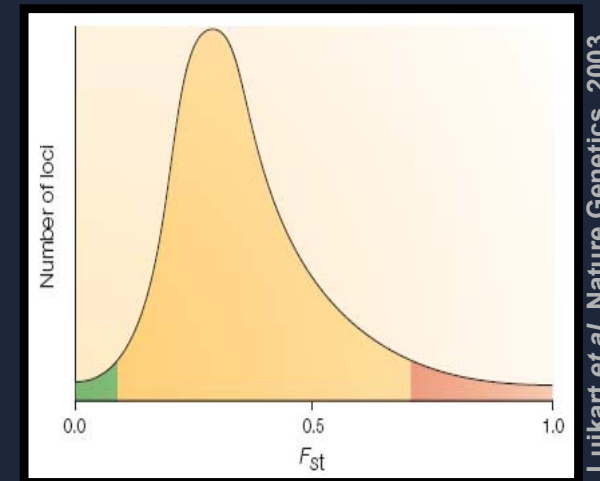
Research in the Lawton-Rauh Lab: Plant Population Genetics

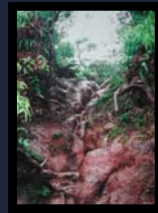
Arabidopsis sister species and the Hawaiian silversword alliance

Brief overview:

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

Amy Lawton-Rauh
Dept. Genetics & Biochemistry
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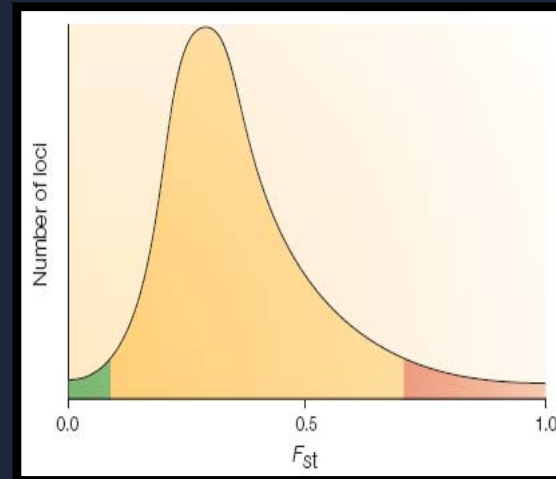
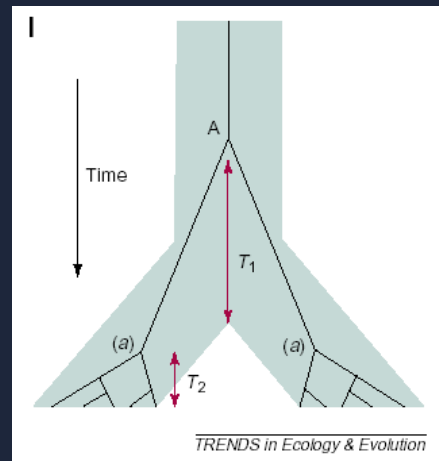
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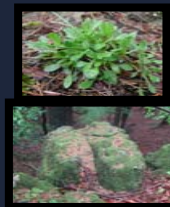
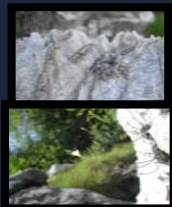
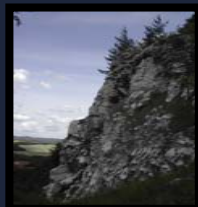
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Luikart et al. Nature Genetics. 2003



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

glacial tills
in Scandinavia

Vatnajökull glacier
Iceland
(photo by Ramos-Onsins)



on isolated Dolomitic rock outcrops
in Central Europe

Harz Mtns, Germany



Franconian Alps, Germany



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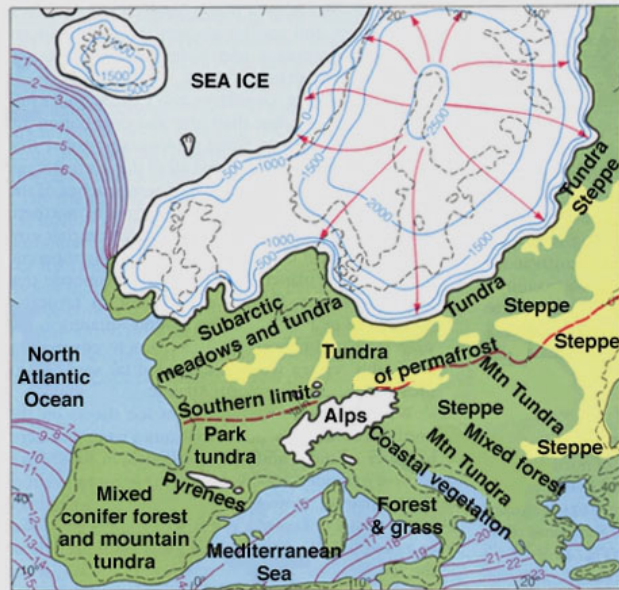


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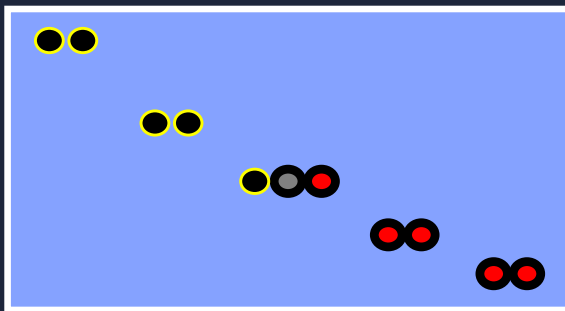
Franconian Alps, Germany



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



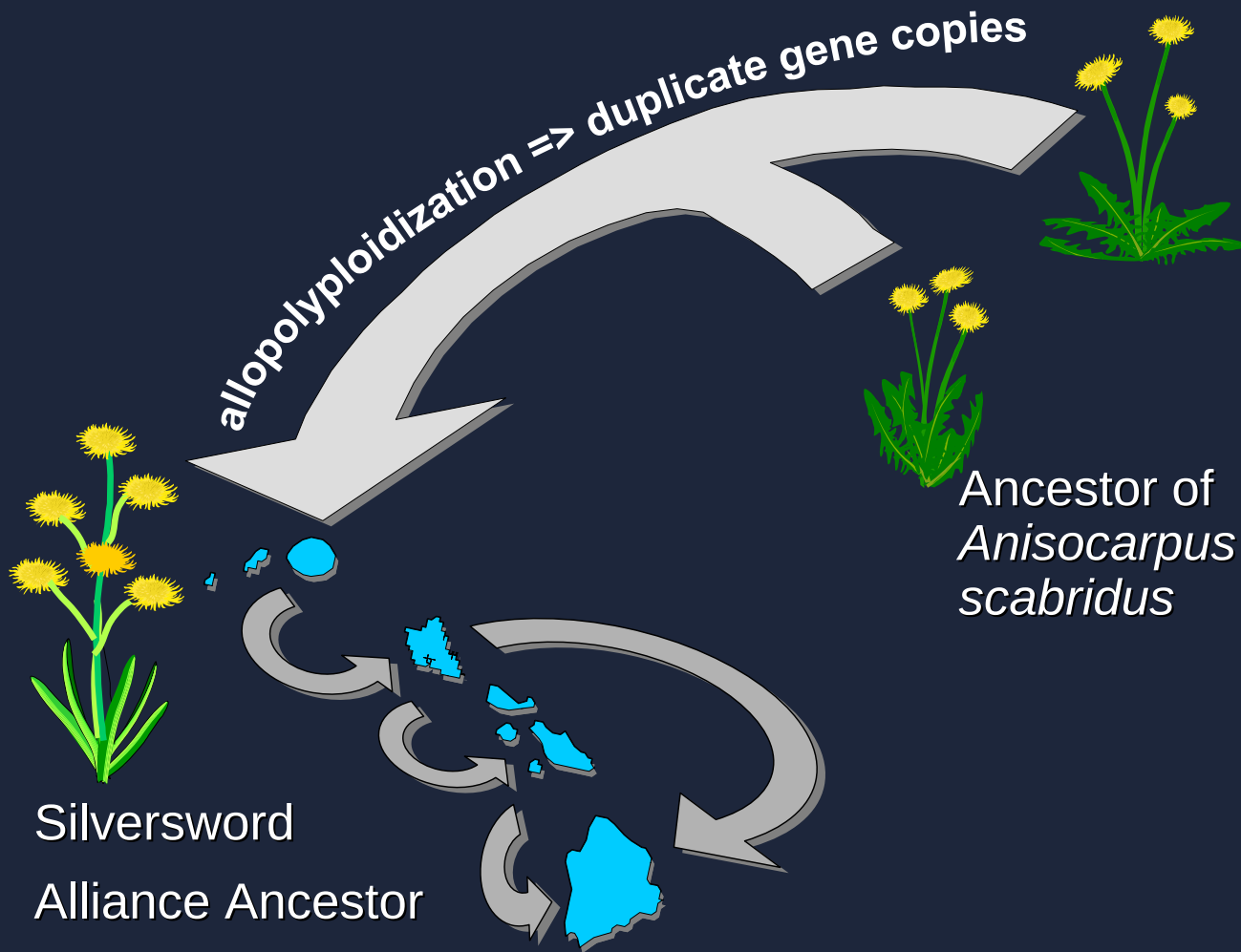
○ Population, *A. petraea*

● Population, *A. arenosa*

● Population, hybrids of *A. petraea* & *A. arenosa*



Hawaiian Silversword Alliance: origin & radiation



Ancestor of
Carlquistia muirii,
Madia nutans



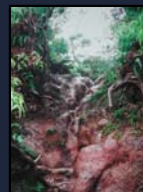
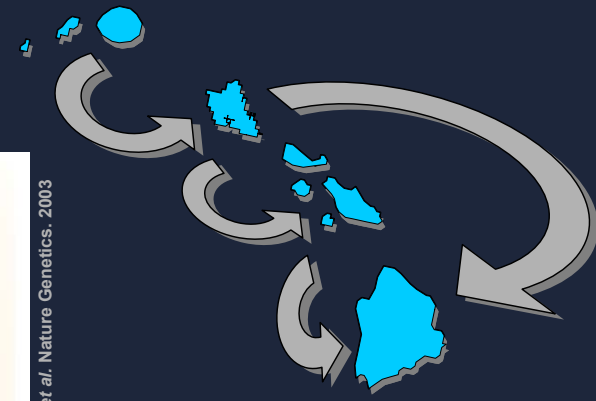
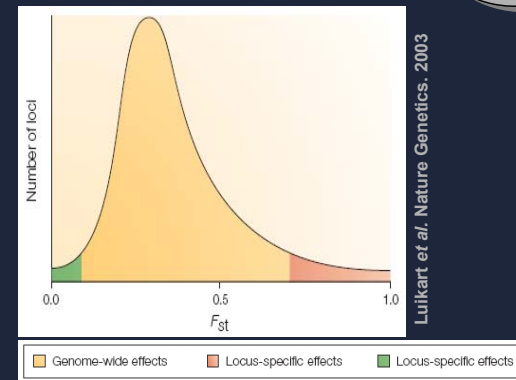
Ancestor of
Anisocarpus
scabridus



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.:

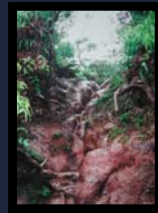
Juliette deMeaux

Max Planck Institute, Cologne

Marcus Koch

Universität Heidelberg



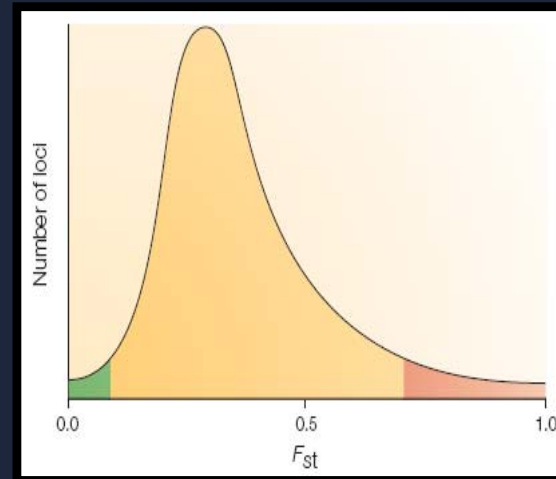
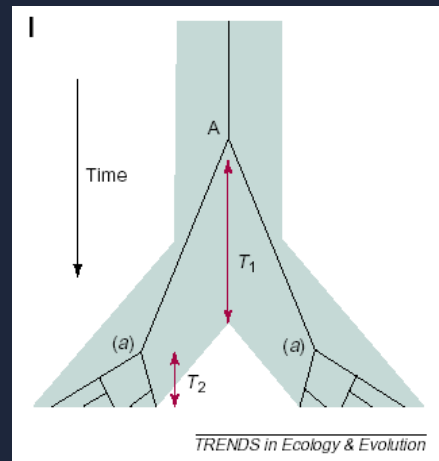


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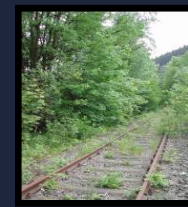
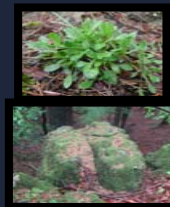
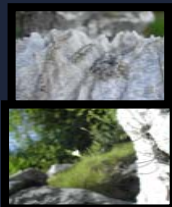
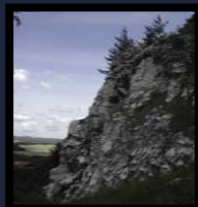
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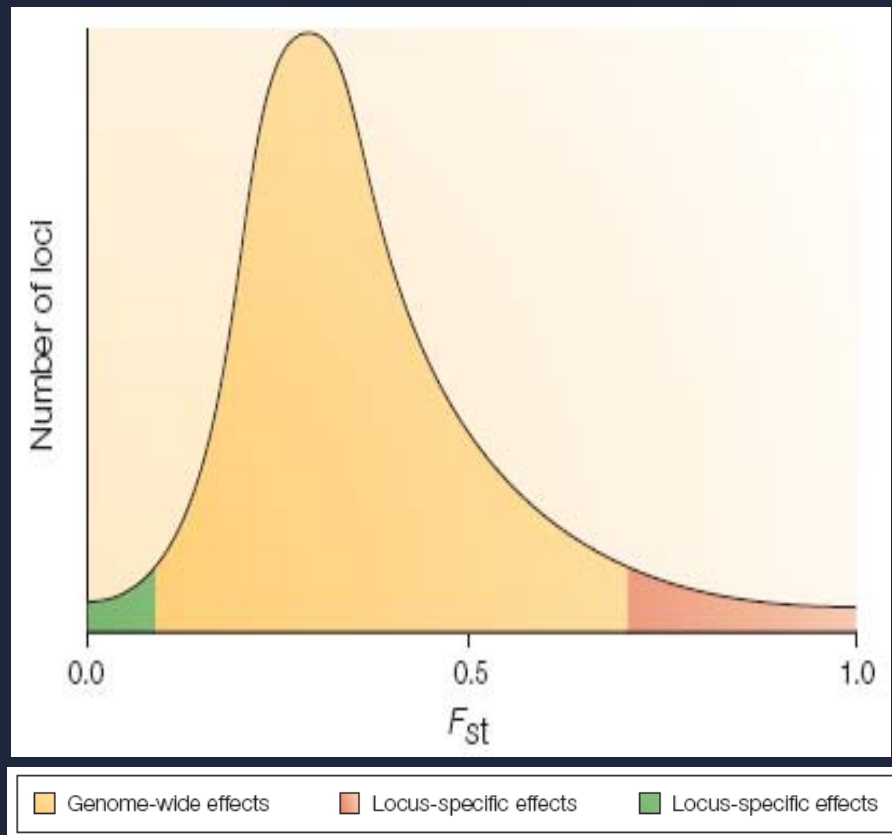
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Luikart et al. Nature Genetics. 2003



'Population Genomics': Brief Introduction



Luikart *et al.* Nature Genetics. 2003

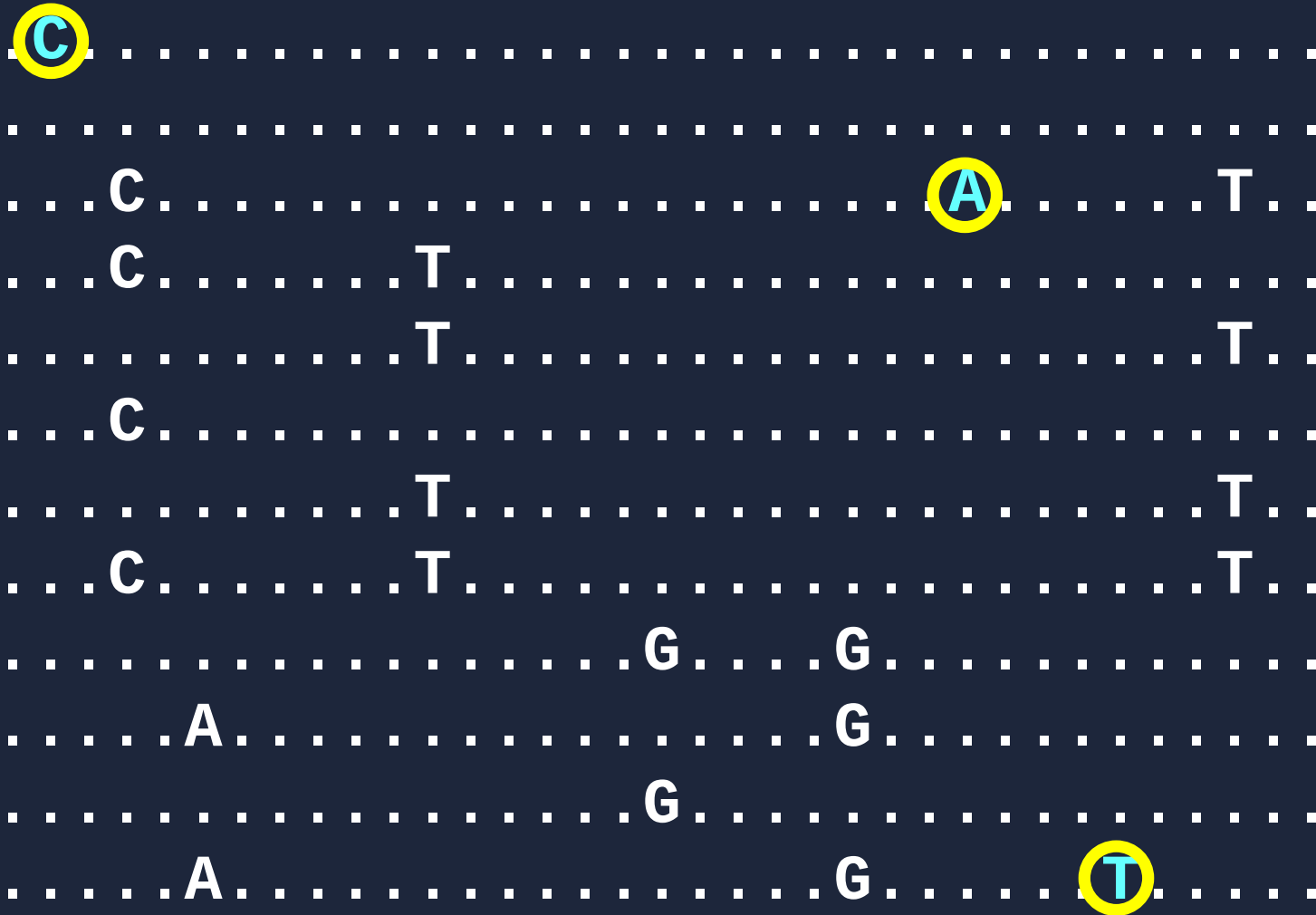
relevant to:

association genetics
functional genomics

(human) disease mapping
crop science, breeding
conservation genetics

Nucleotide Sequence Diversity

ATCGTGTGCCCATAAACCCTTTACTCCTTACGGGC

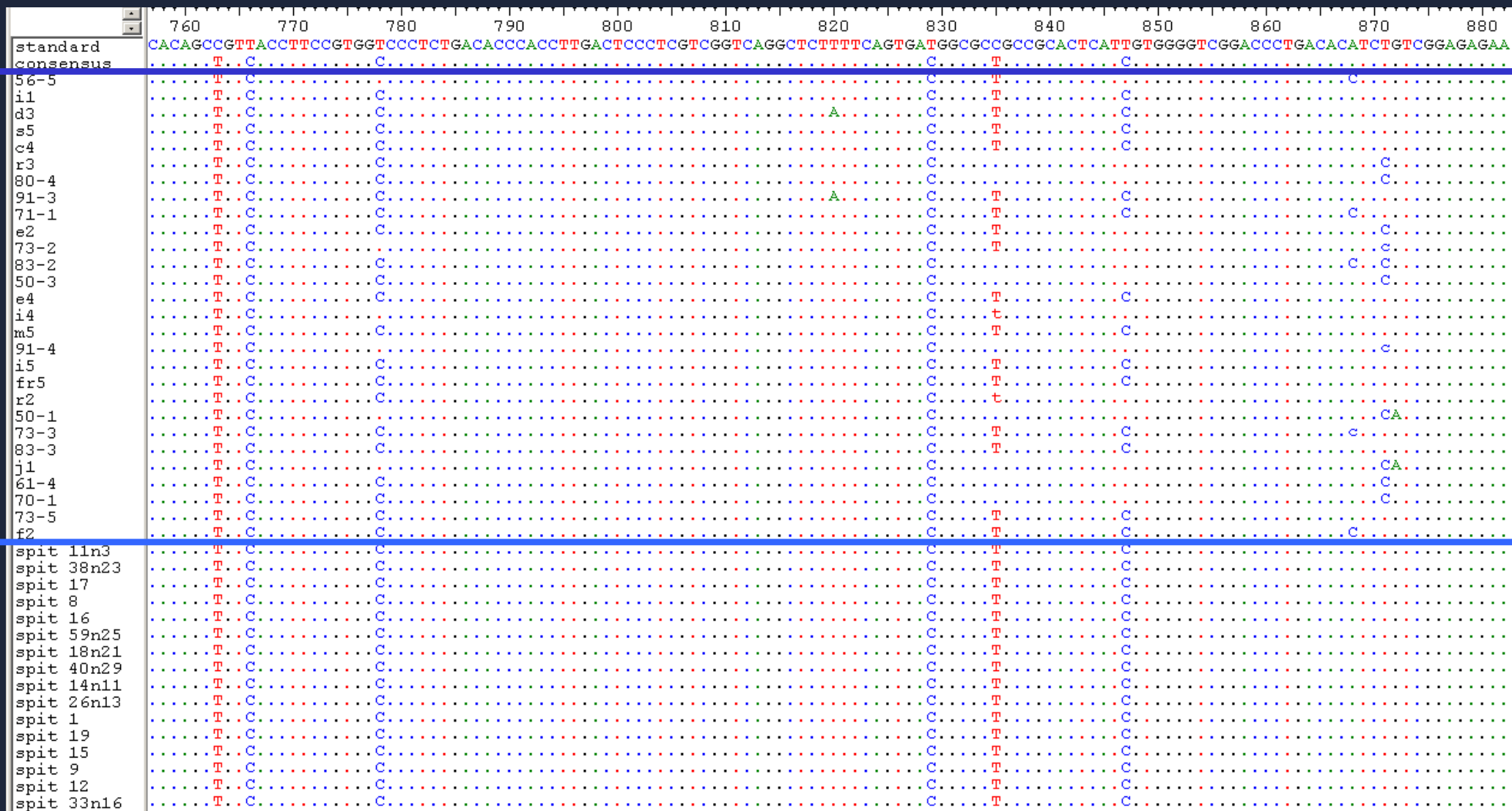


Expect some rare polymorphisms, some singletons

“singleton”:

 *single nucleotide polymorphisms found in 1 individual*

Allele/haplotype segregation patterns differ among populations



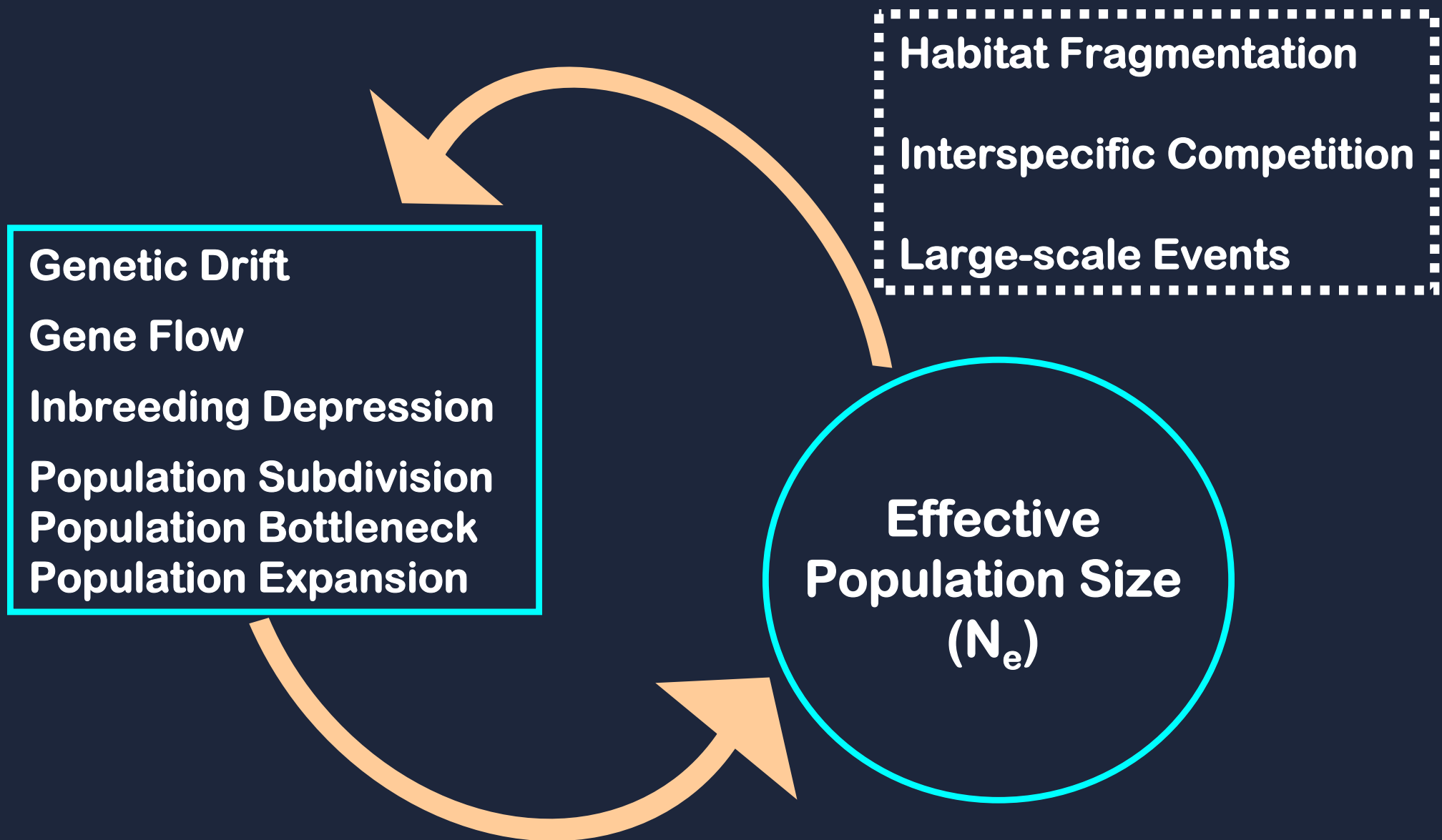
Demography

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

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

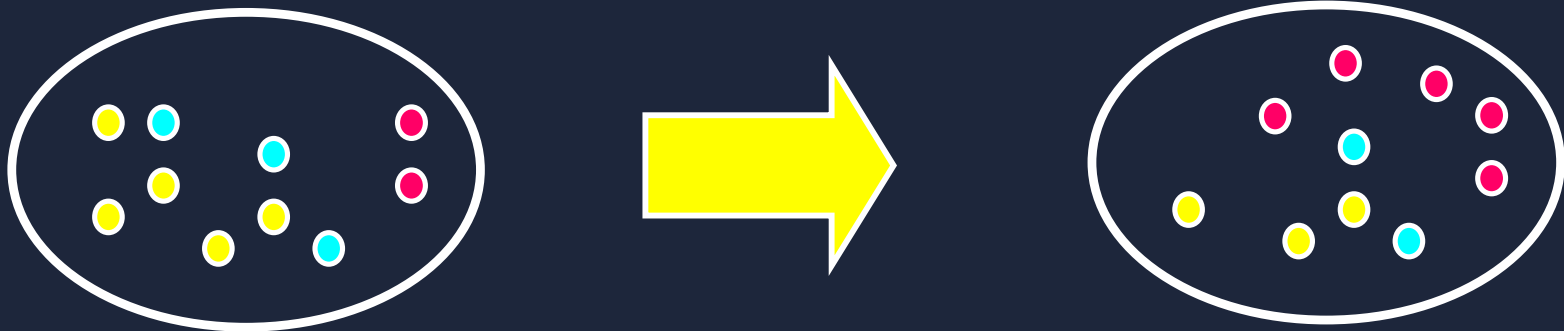
Genetic variation is shaped by many factors

$$\text{Nucleotide Diversity } (\theta) = 4 N_e \mu$$

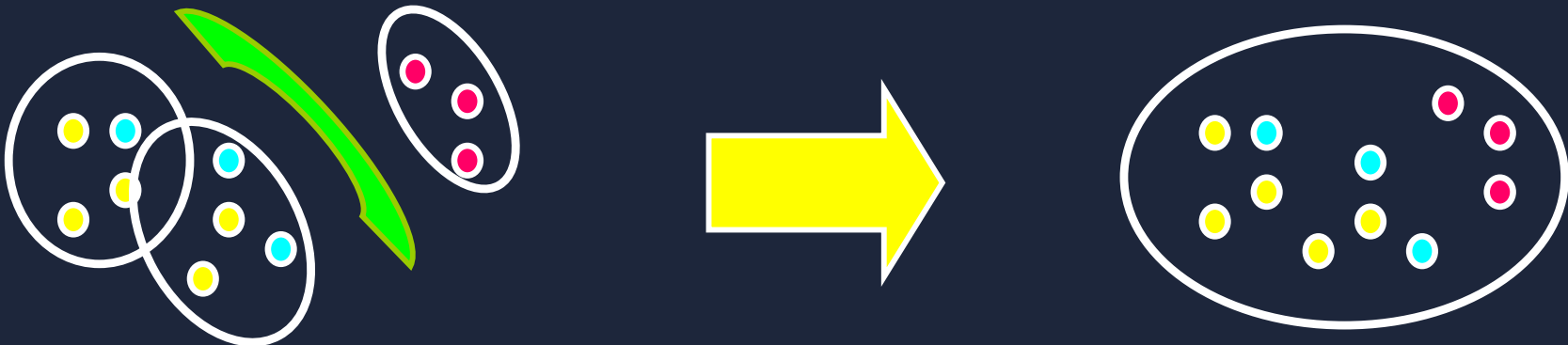


Non random association of alleles...

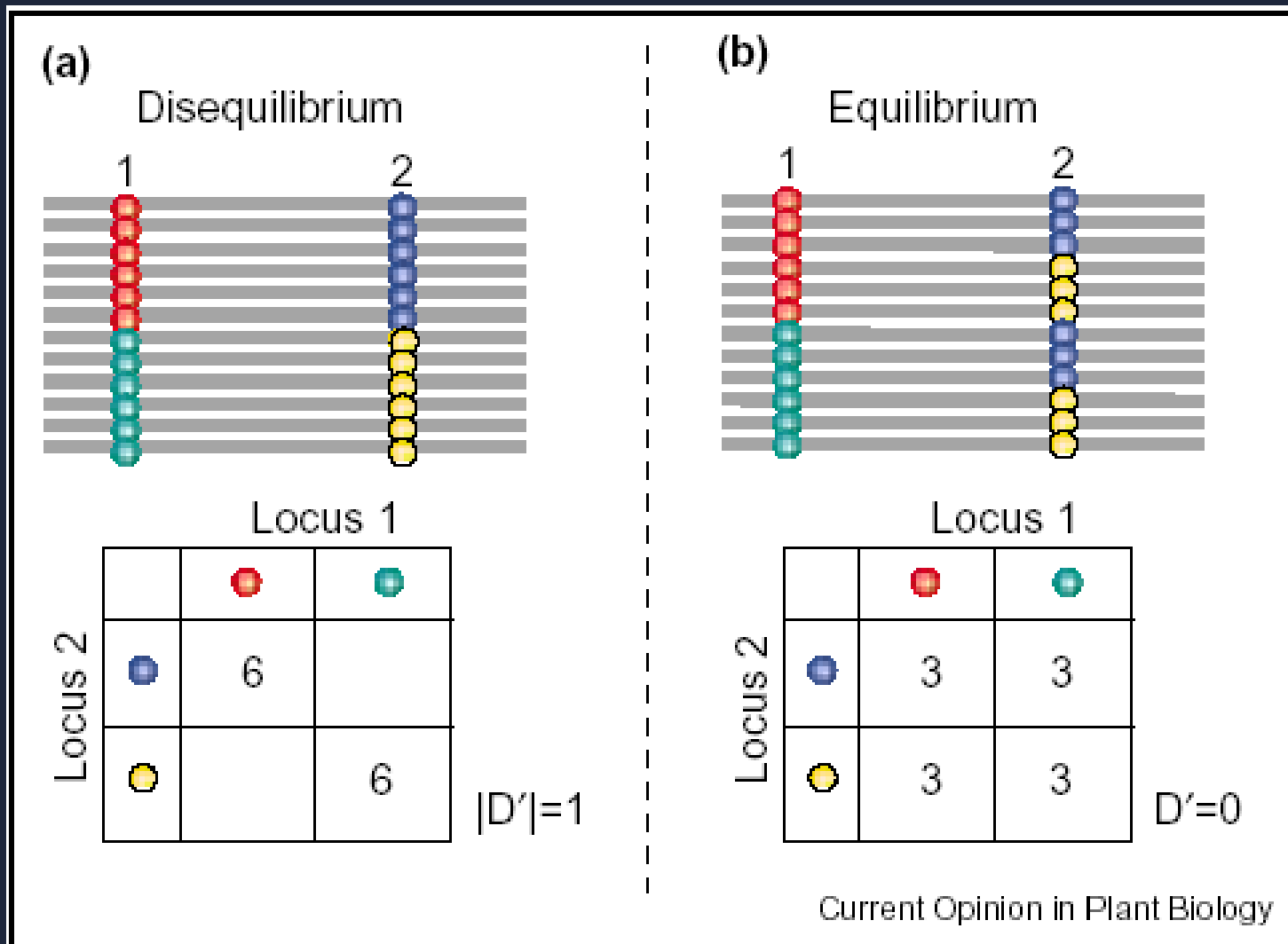
Linkage disequilibrium and selection for linked loci?



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



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



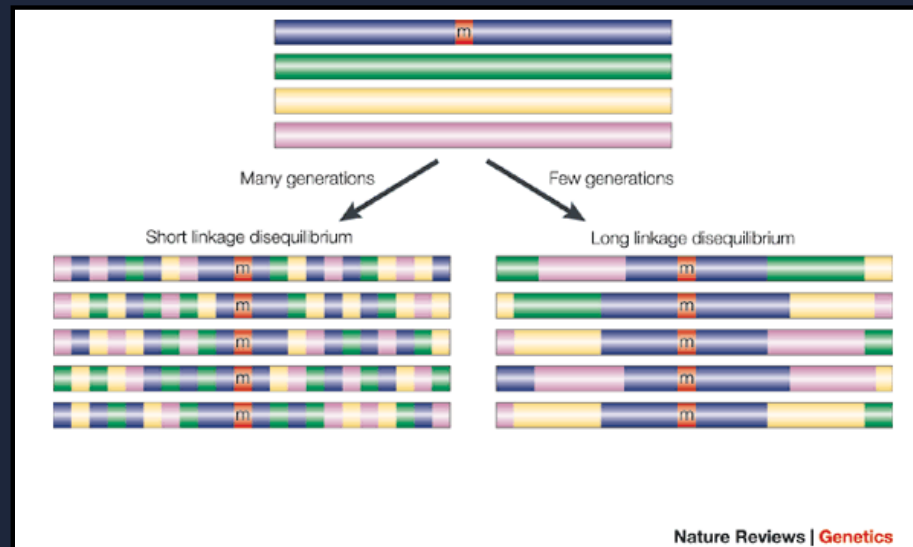
Rafalski A. (2002) Applications of single nucleotide polymorphisms in crop genetics.
Curr. Opin. Plant Bio. 5:94-100.

Linkage Disequilibrium Influences Nucleotide Sequence Diversity

Linkage Disequilibrium:

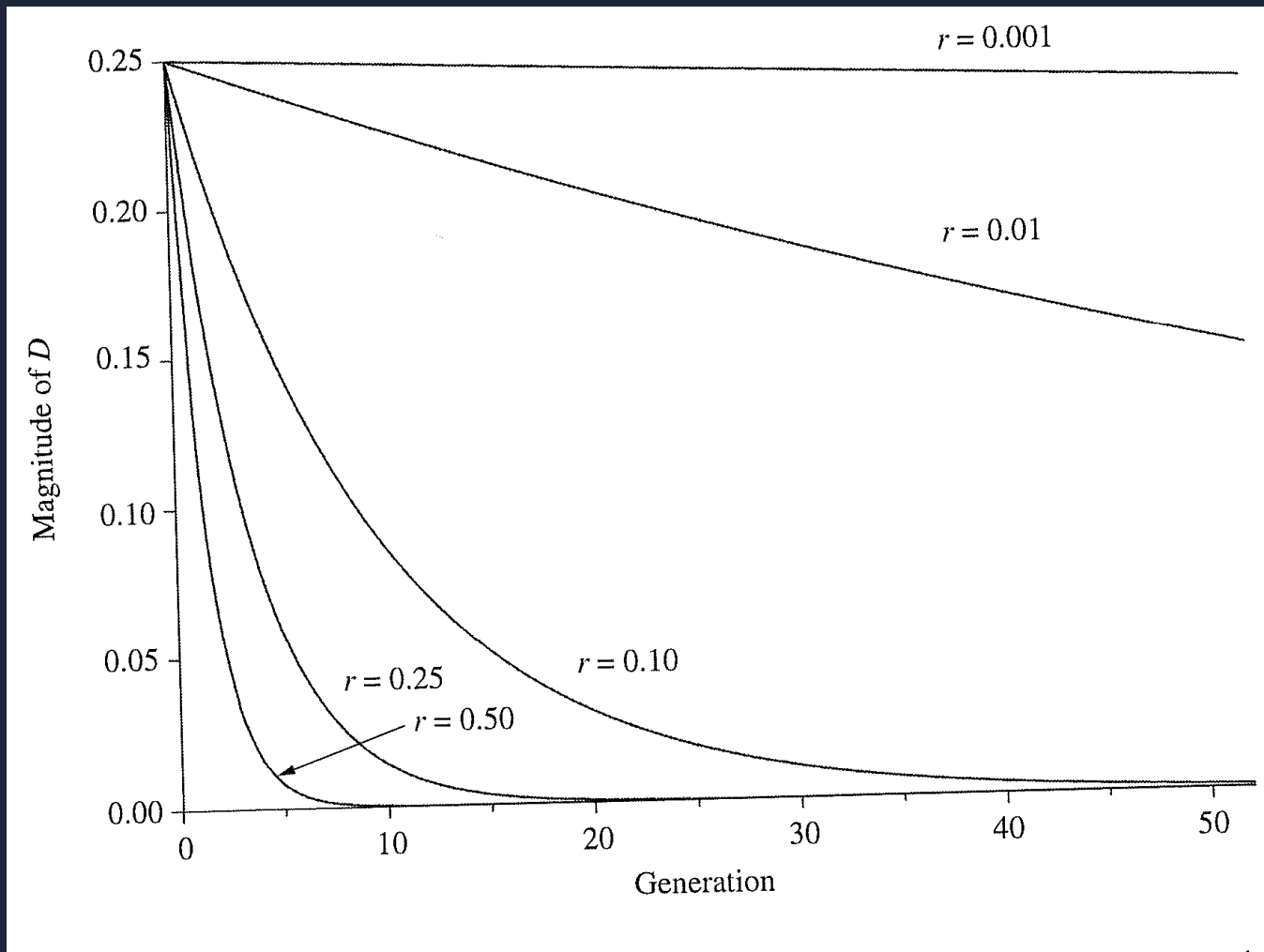
Non-random association among loci/sites

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

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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

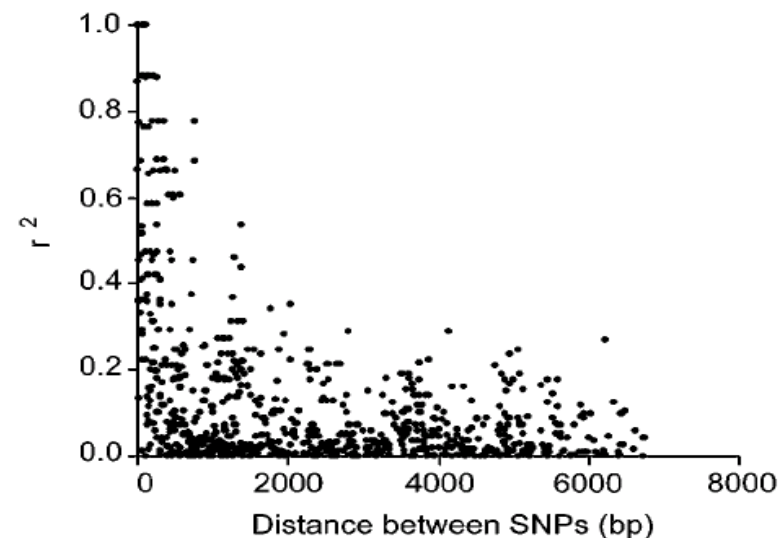
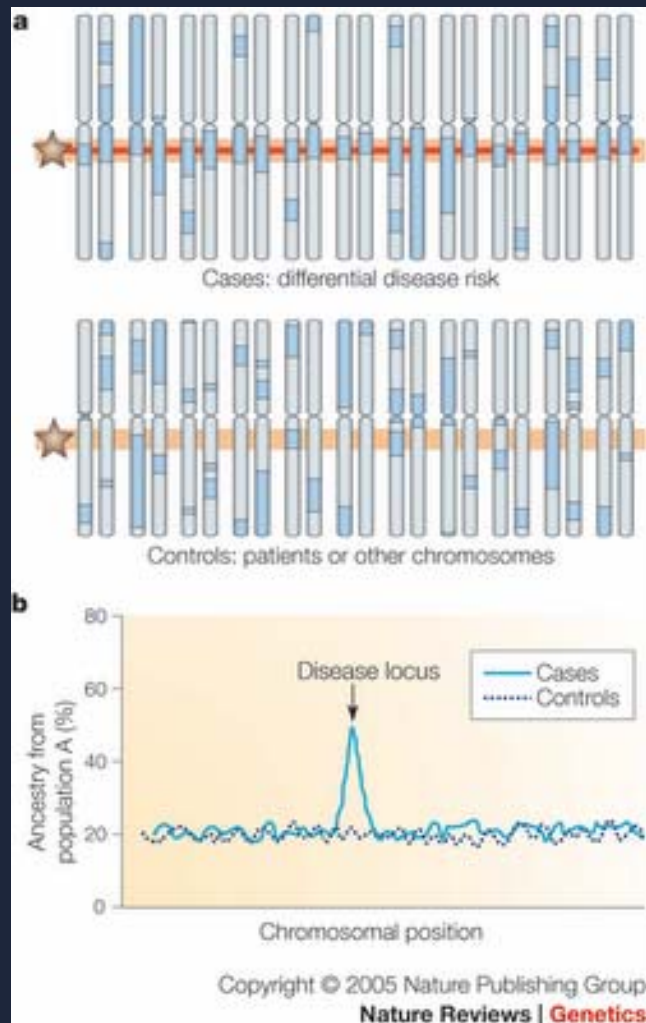


Figure 2 Linkage disequilibrium (LD) decay plot of *shrunken 1* (*sh1*) in maize. LD, measured as r^2 , between pairs of polymorphic sites is plotted against the distance between the sites. For this particular gene, LD decayed within 1500 bp. Data from Reference 48.
Flint-Garcia & Buckler 2003.

Molecular population genetics

Genetic scales to consider:

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

Genomic neighborhood: 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

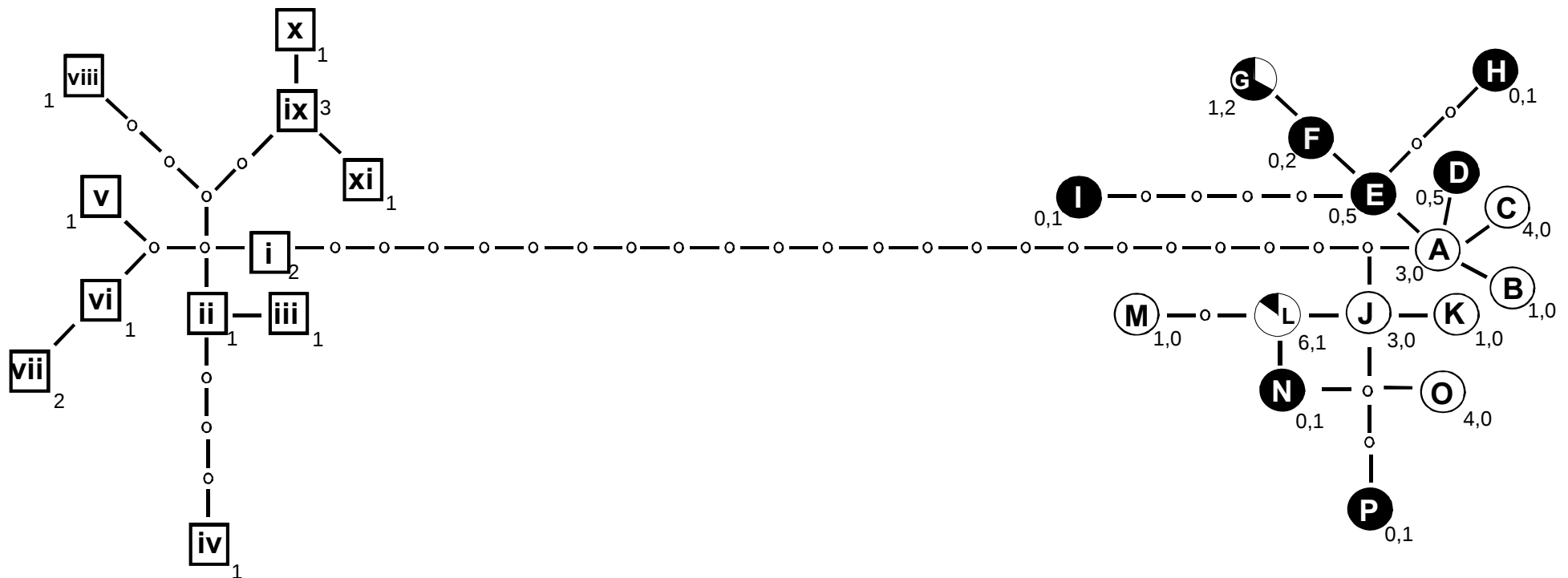
Allele/haplotype age

Gene divergence times (for molecular evolution studies)

Phylogenetic trees for the population level: haplotype network analysis

Statistical parsimony-based haplotype network: *ASAPETALA1-B*

□ *A. sandwicense* ● *D. arborea* ○ *D. ciliolata*



Shared alleles between taxon 1 and taxon 2:

Testing *Isolation vs. Migration* using the coalescent & Bayesian statistics

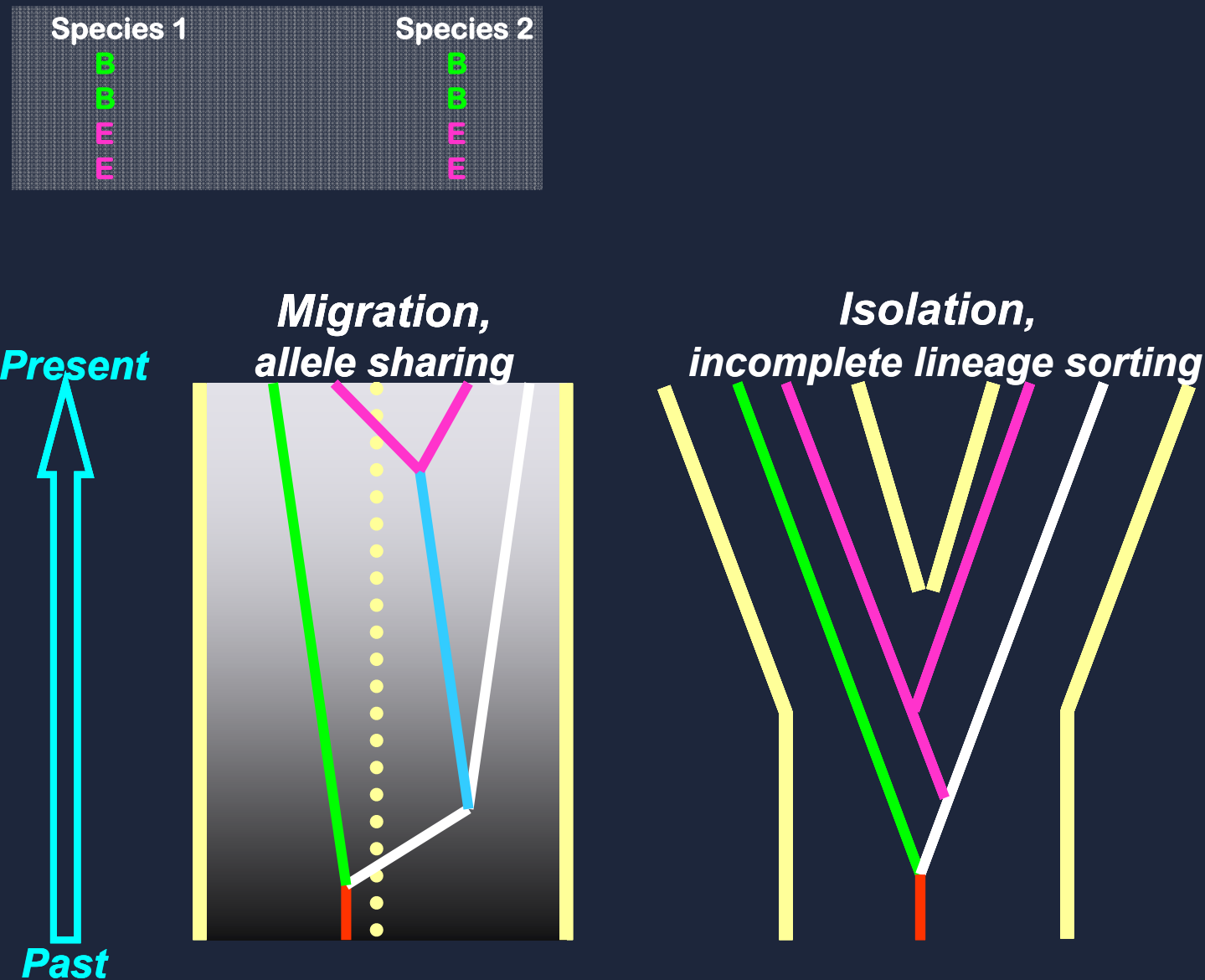


figure modified from: Wakeley J (1996) *Theoretical Population Biology* 49, 369-386

Shared alleles between taxon 1 and taxon 2:

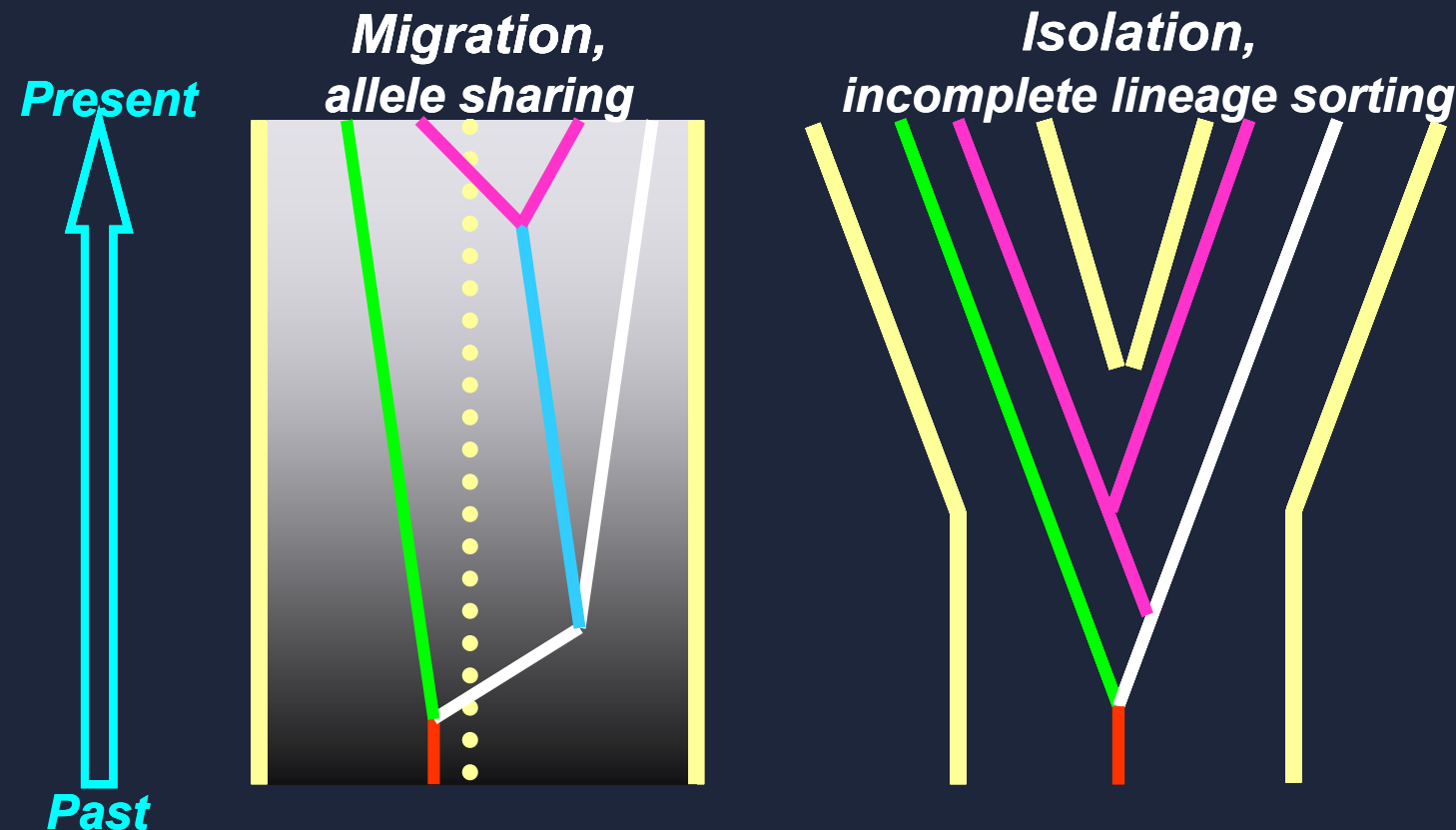
Testing *Isolation* vs. *Migration* using the coalescent & Bayesian statistics



$$\text{TMRCA} = t / (2 N_e)$$

$$M = 2N_e m$$

Gene divergence
Species divergence
Migration rate



Testing “Isolation” vs. “Migration” using the coalescent & Bayesian statistics

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

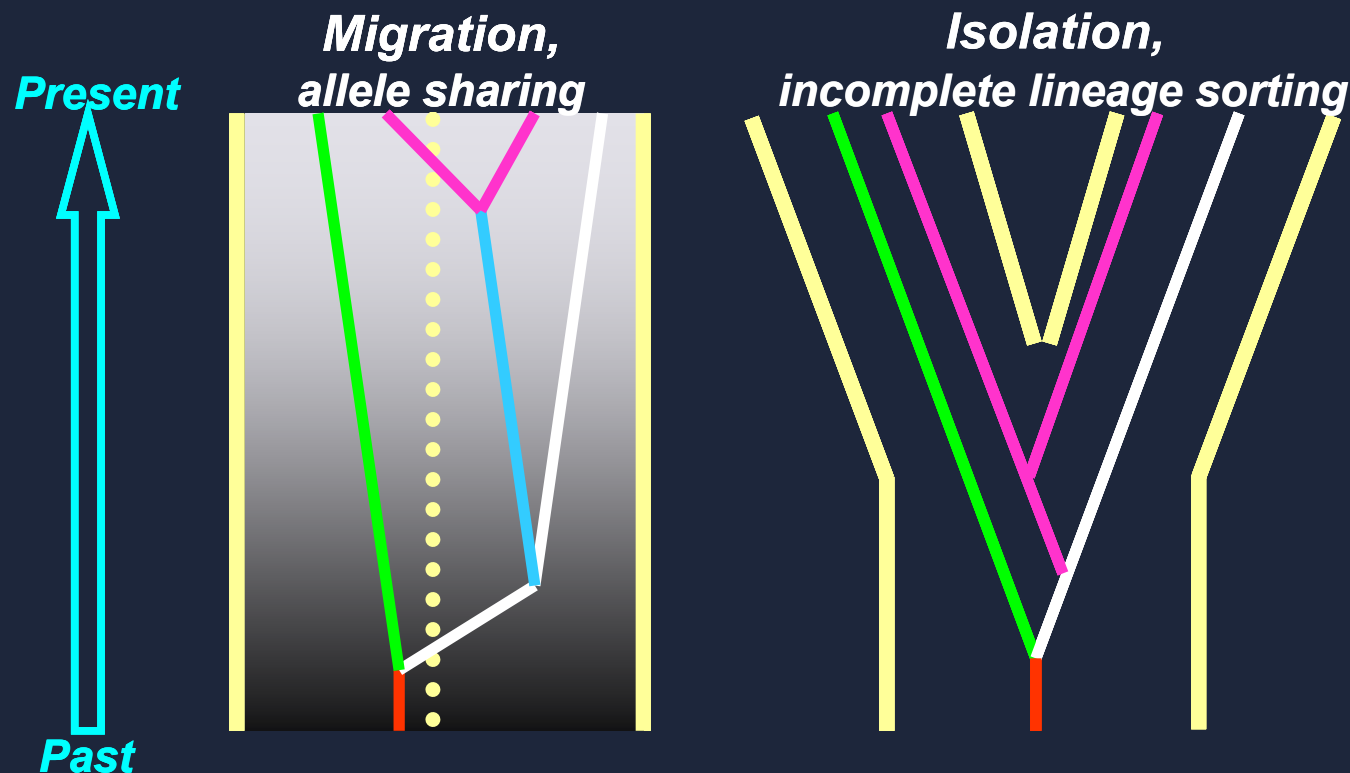
$$\text{TMRCA} = t / (2 N_e)$$

Gene divergence

Species divergence
(credibility interval)

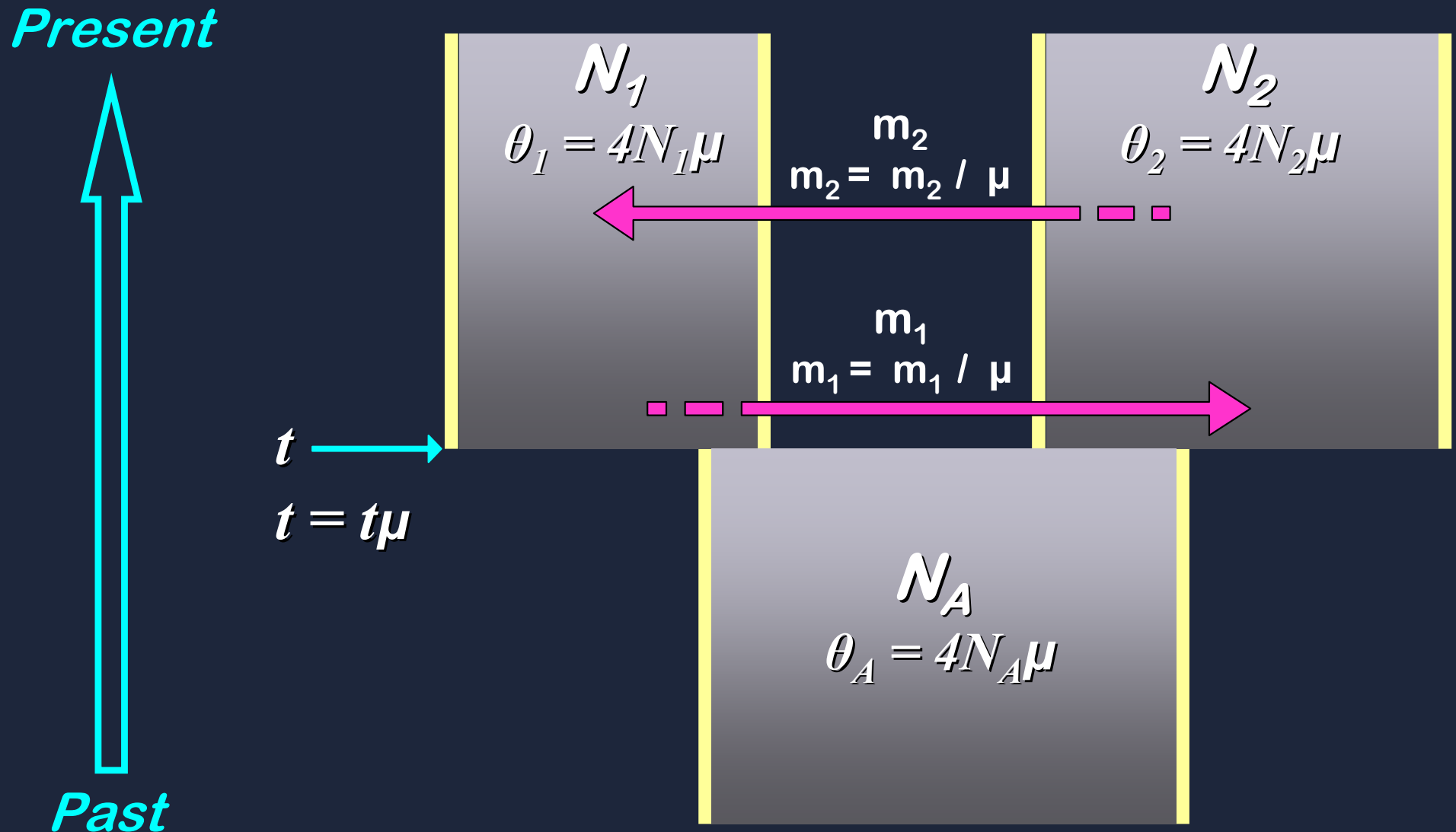
$$M = 2N_e m$$

Migration rate
(credibility interval)



Isolation with migration model: gene flow directionality

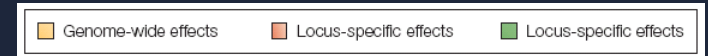
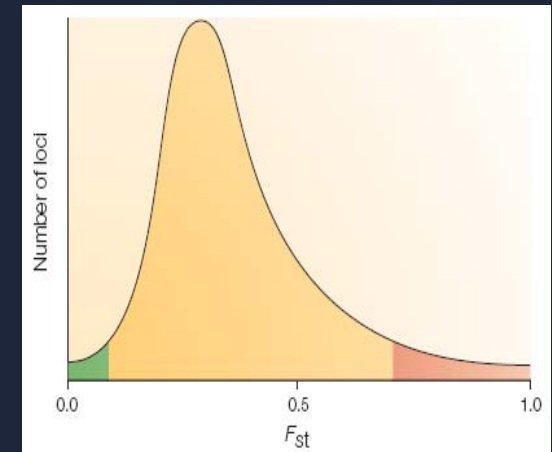
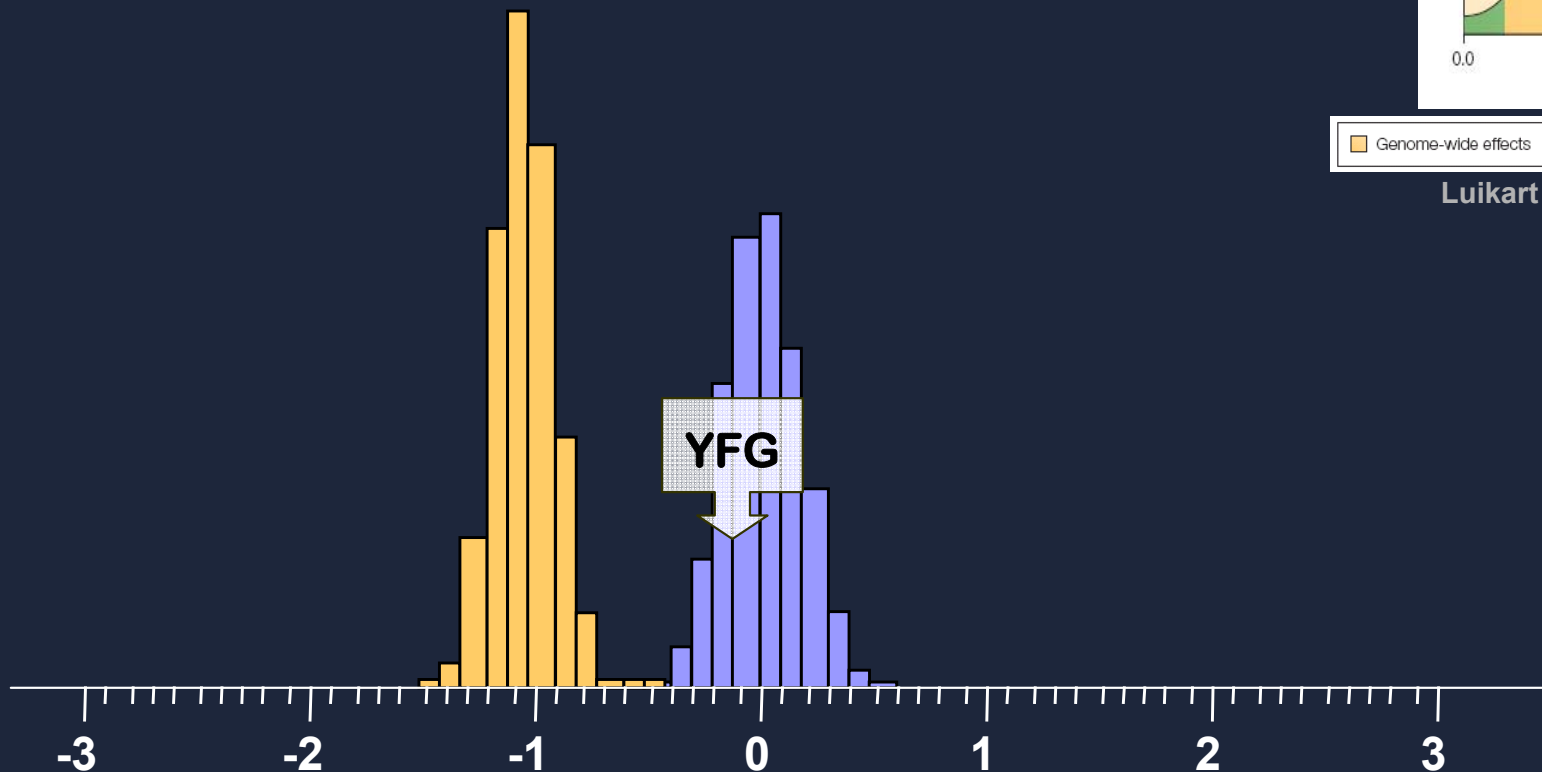
(Hey *et al.* 2004)



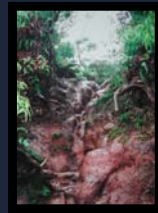
Distribution of estimates across loci:

coordinates of 'favorite/candidate' genes?

Population expansion Demography-equilibrium



Luikart *et al.* Nature Genetics. 2003



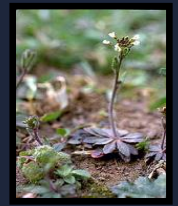
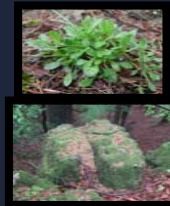
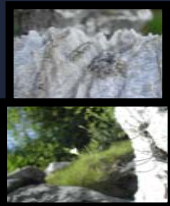
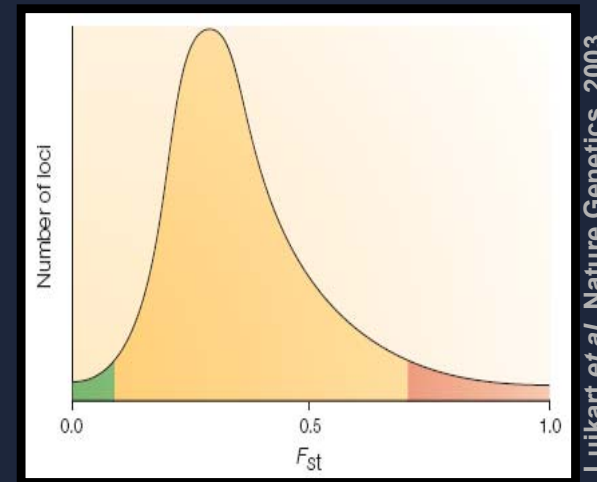
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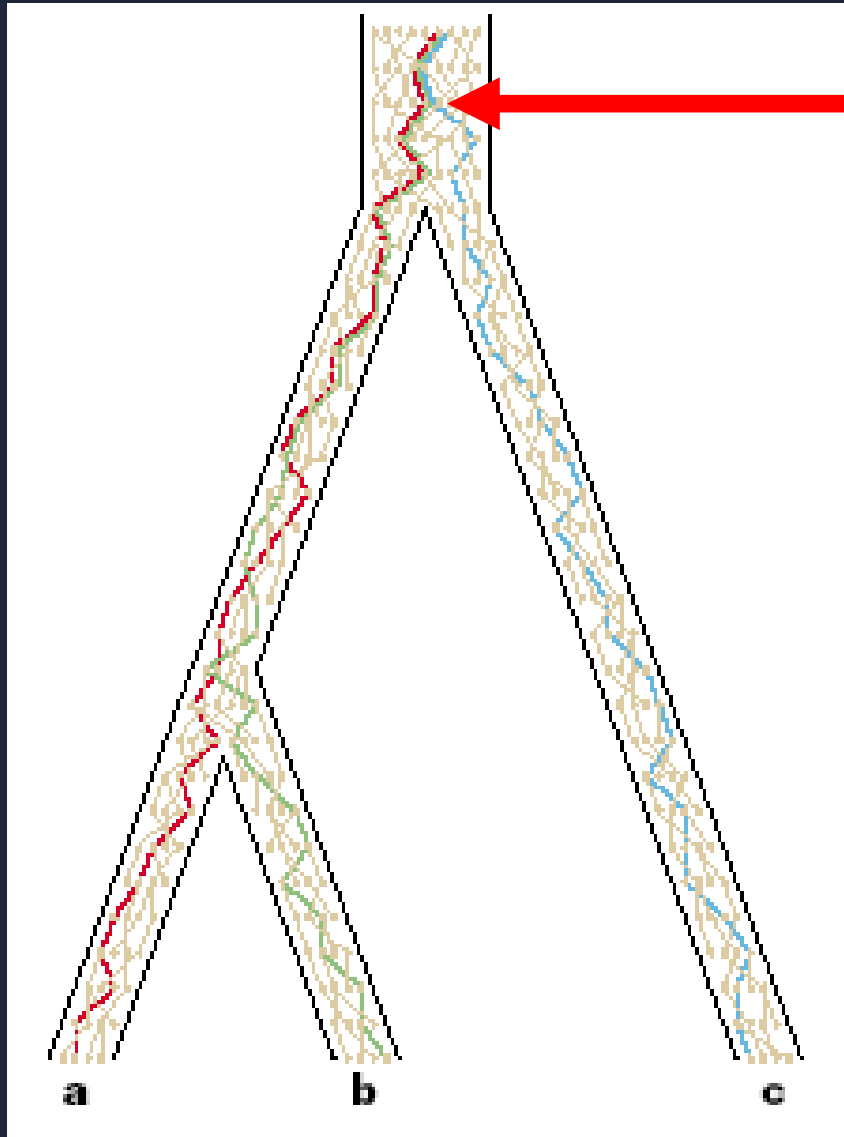
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Estimating Migration depends upon T

(T = time to most recent common ancestor)



coalescence @ common ancestor

species (entire genome)

alleles @ specific locus

