Goal I: Conserving species What is needed for long-term persistence of populations?

• Why is genetic diversity important for population persistence?

• Why are some populations more vulnerable to extinction?

· What processes affect population genetic diversity?

• How and why do populations change in size?

Populations/	How do populations work?	
species	· · · ↓	
	Population genetics	Popul

Population genetics

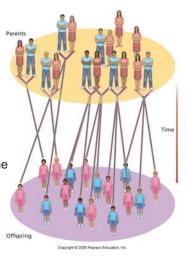
Population dynamics

Population dynamics

Population - a group of individuals of one species in a specified area - continuous through time

Population genetics - change in allele frequencies through time

Population dynamics - change in number of individuals through time

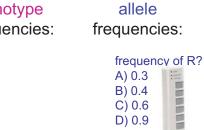


Q: What exactly is continuous through time?

Basics: calculating genotype frequencies and allele frequencies in a population



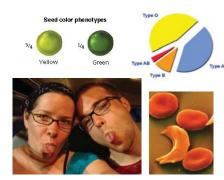
total = 1000 individuals



E) 1.8

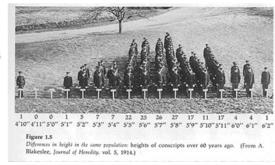
Types of phenotypic variation

Discrete (= qualitative)



Often based on possession of allele at a single locus (gene)

Continuous (= quantitative)

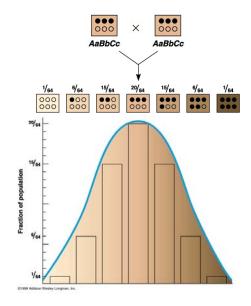


Based on multiple loci

…but continuous variation still results from discrete alleles…

Q: How does quantitative variation arise from discrete alleles?

Ex. Human skin color - at least <u>3 loci</u> affect melanin production



"polygenic (=multilocus) trait"
→ depends on more than one locus

"additive effect"
→ each ● allele "adds" pigment

plus...an environmental contribution – diet, allergens, disease, etc. – sun exposure

"phenotypic plasticity"



Genetic variation is essential for evolution

1. variation: flower size varies in a natural population





Darwin's simple insight: Only 3 conditions needed for evolution to occur in response to natural selection

IF: 1) a trait ("X") *varies* among individuals

2) some of the variation in X is *heritable*

Q: Why is genetic variation important?

- → phenotypic variation
- → genetic variation
- 3) survival/reproduction wrt X is **non-random** \rightarrow **natural selection**

natural selection

phenotypic change ns <u>within</u> a generation

"individuals that survive & reproduce better will leave more offspring, and more of their alleles, and more of their heritable phenotypic traits, in the next generation, and so on..."

A: Genetic variation is <u>essential</u> for adaptive evolution!

offspring's flower size

Q: What's wrong with Darwin's photo?

Genetic variation is essential for evolution

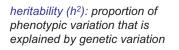
variation: flower size varies in a natural population
 variation is heritable: mother's flower size predicts



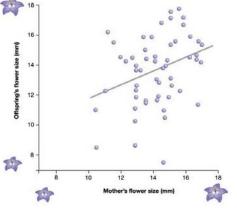
Ex: flower size in Alpine Sky Pilots

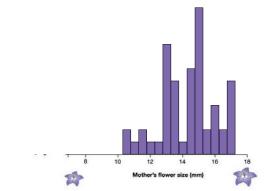


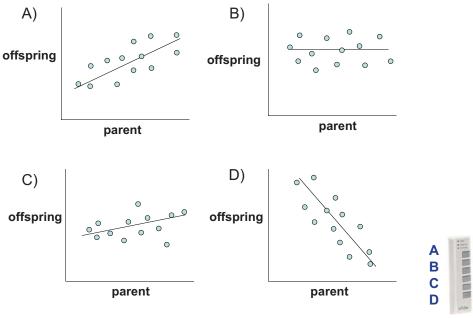
Ex: flower size in Alpine Sky Pilots



Q: How would you measure it?







Q: Which trait shows the greatest heritability? Q: What do each of these relationships mean?

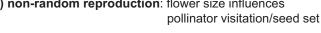
Genetic variation is essential for evolution

1) variation: flower size varies in a natural population

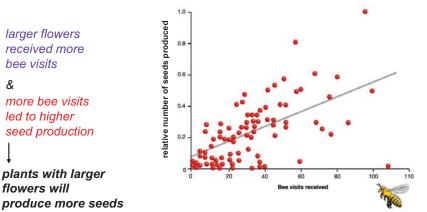
2) variation is heritable: mother's flower size predicts offspring's flower size

3) non-random reproduction: flower size influences









Q: Has natural selection happened? A)YES B)NO C) Cannot tell Q: Will evolution happen ? A)YES B)NO C) Cannot tell

Genetic variation is essential for evolution

1) variation: flower size varies in a natural population

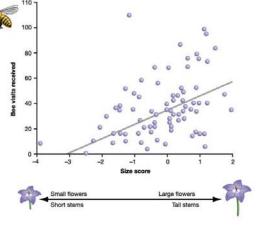
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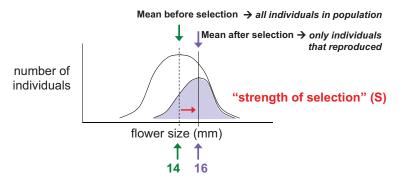
Ex: flower size in **Alpine Sky Pilots**





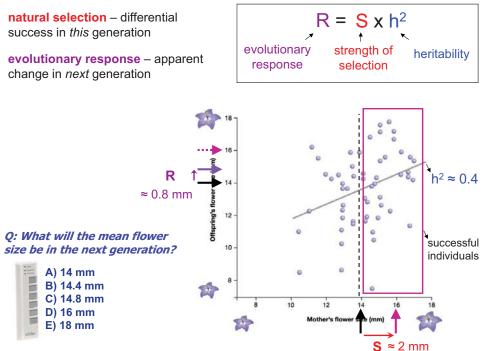
pollinator visitation/seed set

How "strong" is natural selection?



S = mean after selection – mean before selection

Q: Does selection happen within or between generations?

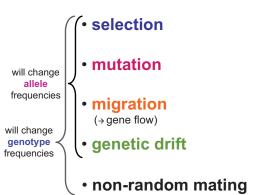


Q: So, why is heritable genetic variation critical for populations?

What can change population genetic structure?

(allele and genotype frequencies)

Aa AAAaaa



In the absence of these 4 processes

 <u>no change</u> in <u>allele</u> frequencies between generations (no evolution)

In the absence of these 5 processes

• genotype frequencies <u>as predicted</u> <u>from random pairing</u> of <u>alleles</u>

Hardy-Weinberg Equilibrium (HWE)

- **null mode** of what is expected in the absence of the 5 processes
- the presence of any of the 5 will cause deviation from HWE:

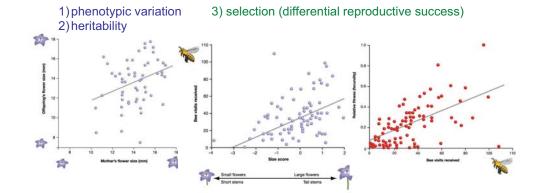
genotype frequencies <u>not</u> as predicted from allele frequencies



Ex. Alpine Sky Pilot

- evidence of *microevolution* = change in allele frequencies
- mechanism: *natural selection*

Q: What other mechanisms can cause changes in allele frequencies?



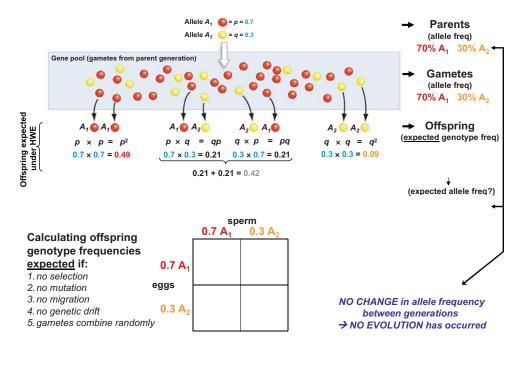
aside: calculating combined probabilities

 Probability of having a girl <u>and</u> then a boy? (assume probability of X from dad is 50%)



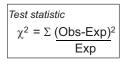
- Probability that a random date will be rainy <u>and</u> a Tuesday? (assume probability of rain is 70%)
 - The combined probability of two independent events happening together ("and") is their <u>product</u>.
- What is probability that a random date will be Tues or Fri?
 - The probability of either of two independent events happening ("or") is their <u>sum</u>.

A NUMERICAL EXAMPLE OF THE HARDY-WEINBERG PRINCIPLE



		A 1 A 1	A_1A_2	A_2A_2
Imagine that allele A ₁ = 0.7	Q2: Are these genotype	0.5	0.4	0.1 ?
frequencies are $A_2 = 0.3$	frequencies in HWE? (null hypothesis)	0.6	0.2	0.2 ?

A: Use χ^2 -test to test for deviations from the expected genotype frequencies

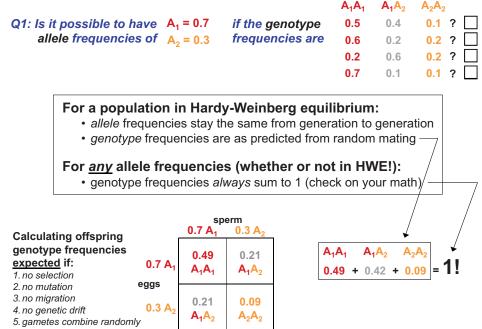


Expected frequencies 0.49 0.42 0.09 (assuming gametes combine randomly)

Exp and Obs are numbers, not frequencies

Imagine that you have measured genotypes for 100 individuals





Q3: Suppose you measure genotype frequencies in a population of plants. Are these genotypes in HWE?

$\begin{array}{ccc} & \mathbf{A}_1 \mathbf{A}_1 & \mathbf{A}_1 \\ \mathbf{A}_1 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_1 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_1 \\ \mathbf{A}_1 & \mathbf{A}_2 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_1 & \mathbf{A}_1 & \mathbf{A}_1 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_1 & \mathbf{A}_1 & \mathbf{A}_1 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_1 & \mathbf{A}_1 & \mathbf{A}_1 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_1 & \mathbf{A}_1 & \mathbf{A}_1 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_1 & \mathbf{A}_1 & \mathbf{A}_1 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 & \mathbf{A}_2 \\ \mathbf{A}_2 & A$	$A_2 A_2 A_2$

- Step 2: Calculate observed allele frequencies from observed genotype frequencies
- Step 3: Calculate genotype frequencies expected under HWE from the observed allele frequencies

Step 4: Compare <u>observed</u> to <u>expected</u> genotype numbers using χ^2 -test.

- Is test statistic less than threshold? \checkmark \rightarrow population is in HWE
- p^2 = frequency of $A_1A_1 =$ 2pq = frequency of $A_1A_2 =$ q^2 = frequency of $A_2A_2 =$ $p^2 + 2pq + q^2 = ?$

p + q = ?

p = frequency of $A_1 =$ q = frequency of $A_2 =$

- χ² = ?
- $\checkmark \rightarrow$ population is in HWE $\bigstar \rightarrow$ some assumption of HW is violated

o.5 A (allele and genotype frequencies) 0.5 a selection 0.25 0.25 0.5 A Parental allele freq: AA Aa 0.5 A 0.5 a eggs mutation 0.25 0.25 0.5 a will change Aa aa allele frequencies (**p** & **q**) 10000 zygotes: Survival to repro: Survivors: migration 250 AA 1.0 for AA 250 AA $(\rightarrow \text{gene flow})$ 425 Aa 500 **A**a 0.85 for Aa will cause 250 aa 0.5 for aa 125 aa deviations 800 indy's from "HWE" genetic drift (genotype freg not Offspring allele freq: p², 2pq, q²) 250 x 2 + 425 = 925 / 1600 = 0.578 A Is this population in HWE? $125 \times 2 + 425 = 675 / 1600 = 0.422 a$ non-random mating $\chi^2 =$

How does **selection** cause change in allele frequencies?

sperm 0.5 A 0.5 a 0.25 0.25 0.5 A Parental allele freq: AA Aa 0.5 A 0.5 a eggs 0.25 0.25 0.5 a Aa aa 10000 zygotes: Survival to repro: Survivors: 2500 AA 1.0 for AA 2500 AA 5000 **A**a 0.75 for Aa 3750 Aa 2500 aa 0.5 for aa 1250 aa 7500 indv's Offspring allele freq: 2500 x 2 + 3750 = 8750 / 15000 = 0.583 A Is this population in HWE? $1250 \times 2 + 3750 = 6250 / 15000 = 0.417 a$ $\gamma^2 = 6.12 > 3.84$ X

What are the consequences for genetic diversity? Within vs. between populations?

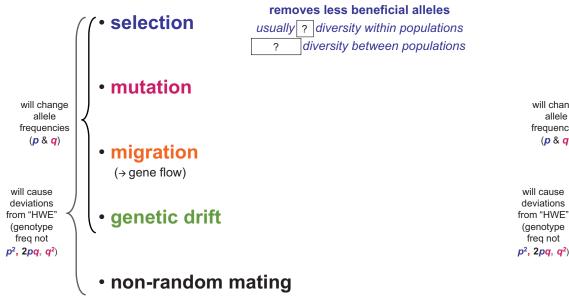




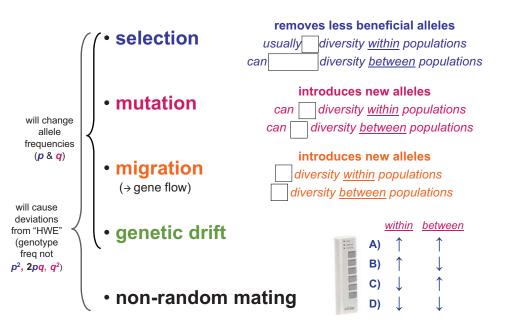
What can change population genetic structure?

How does selection cause change in allele frequencies?

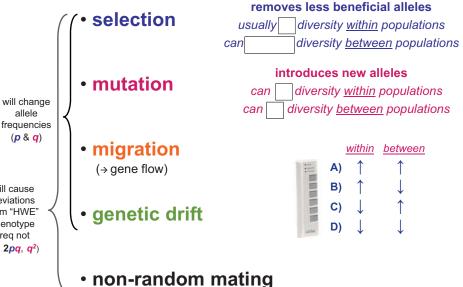
What are the consequences for genetic diversity?



What are the consequences for genetic diversity?



What are the consequences for genetic diversity?



What is genetic drift??

random change in allele frequencies due to sampling error
 random difference between expected and observed results
 all alleles in gene pool will be included
 by chance, only some alleles are included in next generation

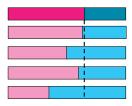
Example 1: Toss a coin 10 times--how many heads? how about 1000 times?

> Would you be more suspicious to get 7/10 heads or 700/1000 heads?

Example 2: CofC student body: 65% f, 35% m

- > 2011: 78 students: 63% female, 37% male
- > 2010: 40 students: 50% female, 50% male
- > 2009: 30 students: 60% female, 40% male

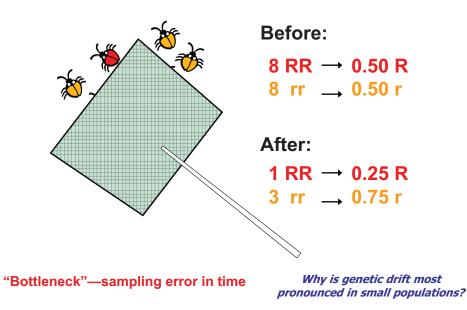
> 2008: 17 students: 35% female, 65% male



→ smaller samples have on average a greater sampling error

Genetic drift - survival is unrelated to trait of interest

alleles that increase in next generation based purely on chance



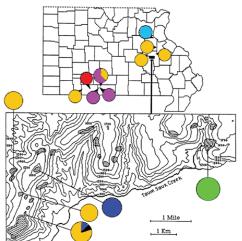
Effects of Genetic Drift in Small Populations

- 1. Each population will have a *unique, unpredictable trajectory*
- 2. Will more strongly influence small populations
- 3. Causes changes in allele frequencies or loss of alleles over time

Ex 2: Collared lizard, Crotaphytus collaris



different alleles are fixed even in very close populations

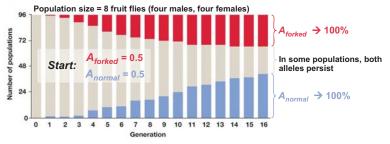


Effects of Genetic Drift in Small Populations

Ex 1: bristle shape in laboratory flies



In most experimental populations, one or the other bristle type became fixed



Effects of Genetic Drift in Small Populations

→ Unlike selection, does <u>not</u> lead to adaptation

Ex 3: Porphyria

- rare autosomal dominant disorder
- extreme sensitivity to light, skin lesions neurological complications, excess hair

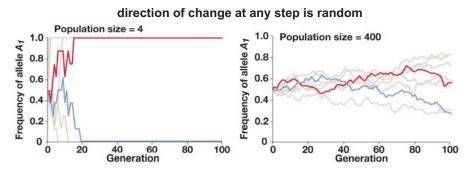


- in the 1600's, 80 Dutch immigrants settled in South Africa
 → have left over 1 million descendents
- one immigrant couple had the gene for porphyria
- now 3 out of every 1,000 Dutch S. Africans have the gene for porphyria (compare to general population: 3 : 1,000,000)

Founder effect = sampling error "in space"

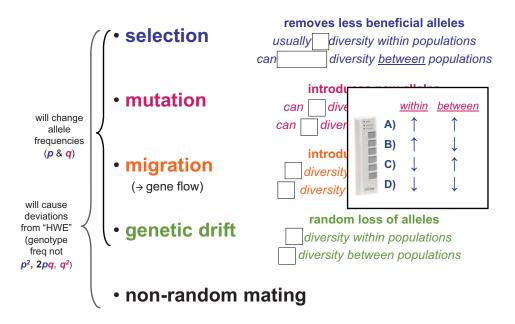
What are the consequences for genetic diversity?

Genetic Drift Is More Pronounced in Small Populations

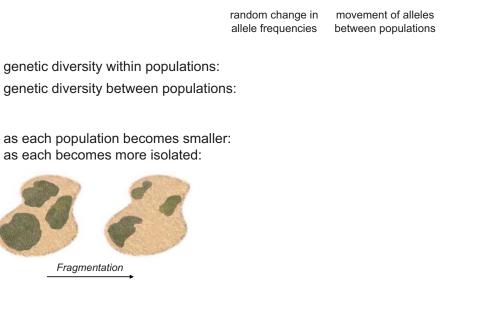


Q: In what two ways do the lines for large and small populations differ?

- smaller populations have bigger jumps at each step
- smaller populations go to fixation at a faster rate (slope of line)
- Q: Can an allele "come back" once lost?
- Q: Are any populations immune from genetic drift?
- Q: What are the consequences for genetic diversity?

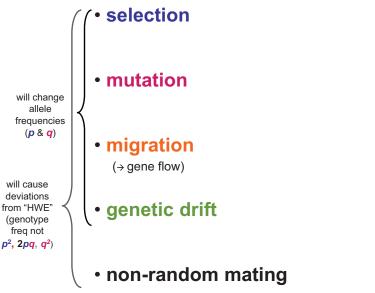


Expected consequences of genetic drift vs. gene flow

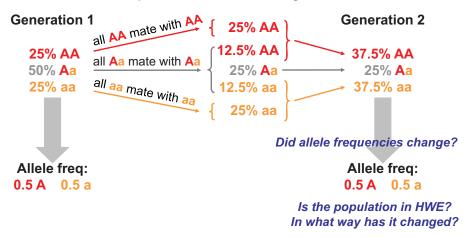


What can change population genetic structure?

(allele and genotype frequencies)



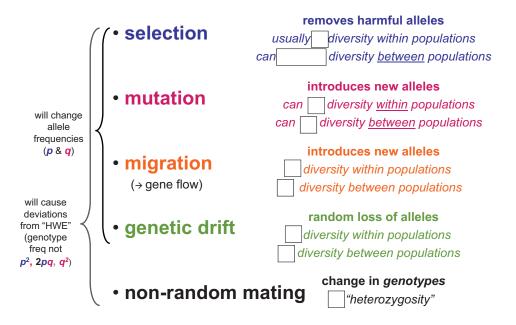
How does non-random mating affect pop'n genetic structure?



Example of assortative mating

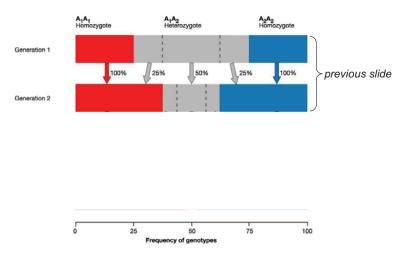
Q: How does assortative (non-random) mating differ from sexual selection?

What are the consequences for genetic diversity?



How does non-random mating affect pop'n genetic structure?

Over many generations...

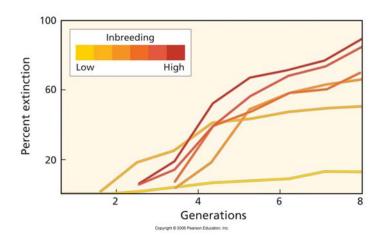


> Assortative mating <u>always</u> leads to excess homozygotes

Inbreeding depression

Ex. Drosophila with different degrees of inbreeding enforced

- inbreeding leads to greater homozygosity
- greater homozygosity leads to <u>decreased fitness</u>
 unmasking of deleterious recessive alleles in homozygotes
- more likely in small, isolated populations



Inbreeding depression

• more likely in small, isolated populations



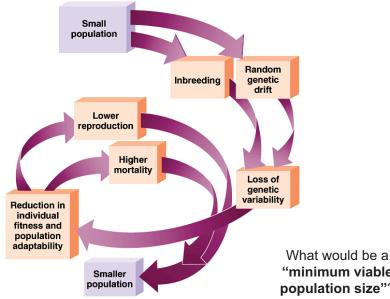


- Ex. Florida panther
 - Puma concolor coryii
- subspecies (mountain lion, cougar)
- fragments of south Florida
- federally endangered species
- 80-100 remain

Major threats

- habitat loss & fragmentation
- inbreeding depression
 - → heart defects
 - \rightarrow undescended testes
 - → defective sperm
- recent introduction of 8 females from Texas subpopulation



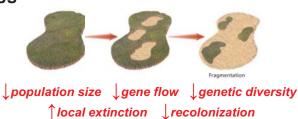


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"minimum viable population size"?

d. What are the biggest threats?

habitat fragmentation



· conservation approach: reduce fragmentation

↑ connectivity **↑**gene flow





Panther corridor