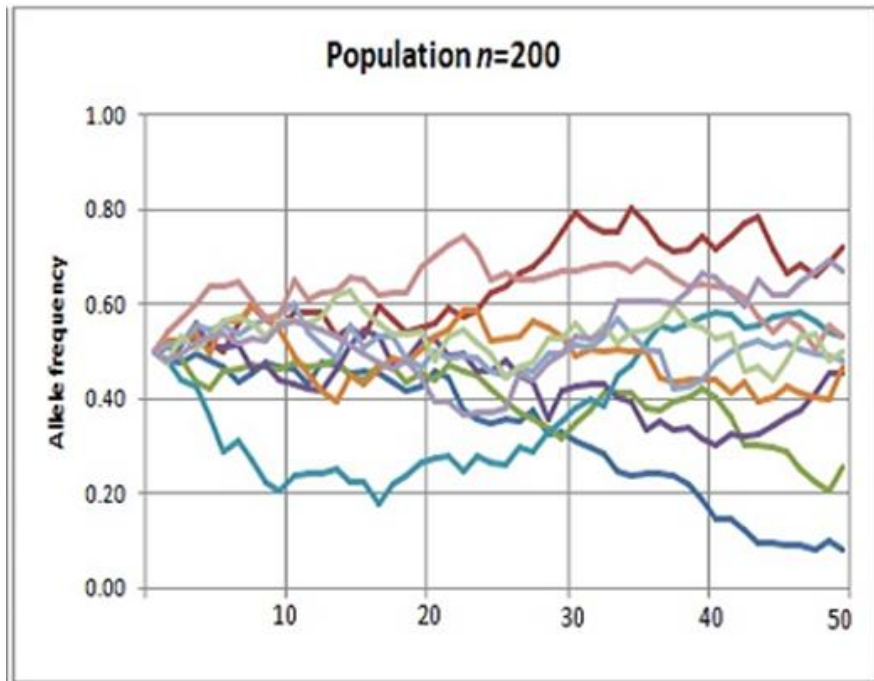


Today's class: Genetic Drift (#4) & HWE (start)



Expected genotype
frequency, heterozygotes
(Aa)

$$p^2 + 2pq + q^2 = 1$$

Expected genotype
frequency, homozygous
dominant (AA)

Expected genotype
frequency, homozygous
recessive (aa)

Housekeeping

Please pick up midterm during break. Thank you 😊

- I need to be in another class at 12:30 pm – so I must leave as soon as the class ends

Location of:

- Learning objectives (Canvas homepage)
- Answer keys for worksheets (e.g. Canvas > Genetics Worksheets & Quizzes > Worksheet page)
- Practice questions for the exam (e.g. Canvas > Midterm #1).
 - I will post Midterm #2 module tomorrow (Friday)

Organism of the Day – Midshipman Fish, *Porichthys notatus*

A local species of fish

- common name comes from photophores that look like the button on a naval uniform
- Photophores produce light that attract prey



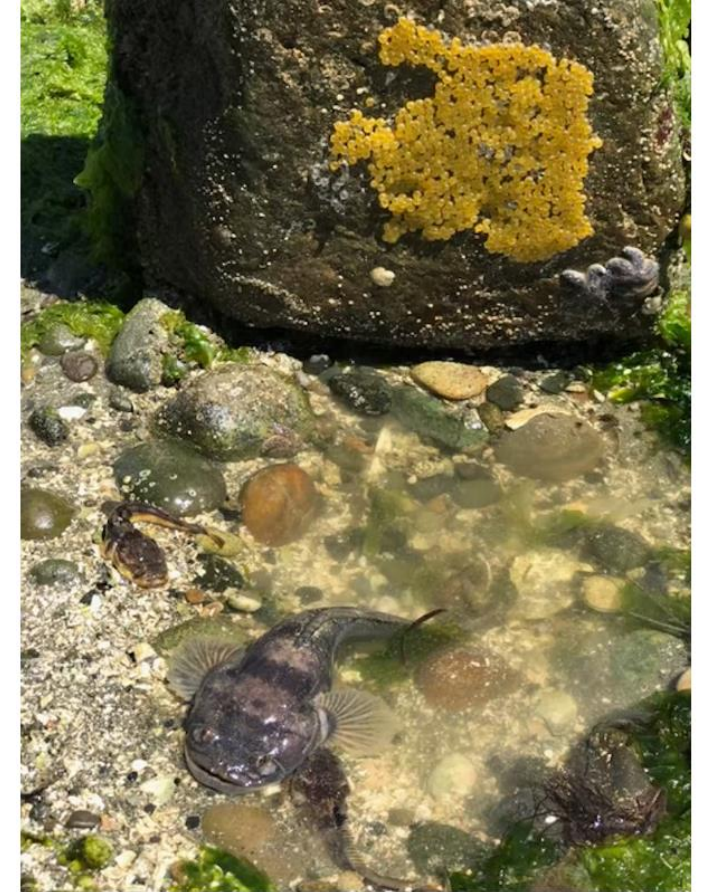
<https://www.fishbase.se/summary/3071>



<https://brighterworld.mcmaster.ca/articles/being-a-dad-is-hard-when-youre-a-plainfin-midshipman-fish/>

Midshipman Fish

- Outside of the breeding season, midshipman fish inhabit deep waters
- But, during the breeding season the males and female move into the intertidal zone to breed
- The males excavate a nest in the sand, usually under large rocks.
- Once finished, the males will sing (hum) to attract mates.
- A successful male may have hundreds of yellow eggs deposited on the roof of his nest.
- The females immediately return to deep waters.
- The male will then spend the next three months caring for the eggs (cleaning them, protecting, and caring for them).
- Each day, when the tide retreats, the male midshipman fish will be exposed to the air for potentially hours.
- The water and oxygen levels drop, the temperatures will increase, the concentration of wastes increases, and the fish are more vulnerable to bird predation.



<https://brighterworld.mcmaster.ca/articles/being-a-dad-is-hard-when-youre-a-plainfin-midshipman-fish/>

Two reproductive morphs of male midshipman fish – Type I and Type II

Type I males (guarders) –can be:

- 8X larger than Type II males
- 2X as long
- have larger vocal organs
- these males acoustically court females (hum). Hums draw females to the nest where they will lay eggs and depart sooner after.
- they are also territorial, defending nests under rocks, and emitting aggressive vocalizations to other males.
- they also have wider heads and mouth that are used in combat (mouth locking)



Two reproductive morphs of male midshipman fish – Type I and Type II

Type II males (sneakers)

- are smaller, but their reproductive organs are 7X larger than the Type I's
- heads are narrower
- they do not hum
- they do not hold/defend territories.
- they reach sexual maturity earlier
- they sneak into the nests of Type I guarder males and fertilize the eggs

Note – Young Type I males that do not have a nest will also try to sneak into the nests of the guarders and fertilize the eggs.



https://abel.mcmaster.ca/publications/pdfs/JFB_2019Miller%20et%20al.pdf

Research by Dr. Sigal Balshine and colleagues from McMaster University on the relationship between guarder male (Type I) body size and female mate preference.

Number of females and number of eggs used as a proxy for female preference (more females, more eggs = greater preference)

What do these data suggest about female midshipmen preference for male body size?

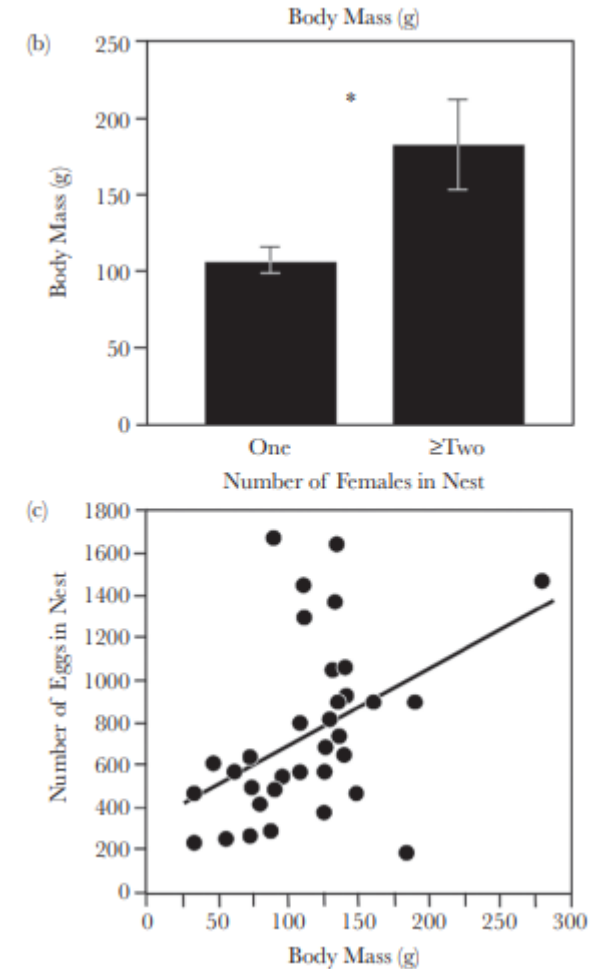
Female midshipman fish preferentially visit the nests of larger guarder males ($p=0.025$) (so selection for larger body size in males).

Not surprising, Dr. Balshine found that larger guard males experienced higher rates of attempted cuckoldry by sneaker males (no figure or p-value)



Members of the Aquatic Behavioural Ecology Lab (McMaster University) investigate midshipman fish nests on Crescent Beach, B.C. Aquatic Behavioural Ecology Lab (McMaster University)

* = $p < 0.05$



Original Article

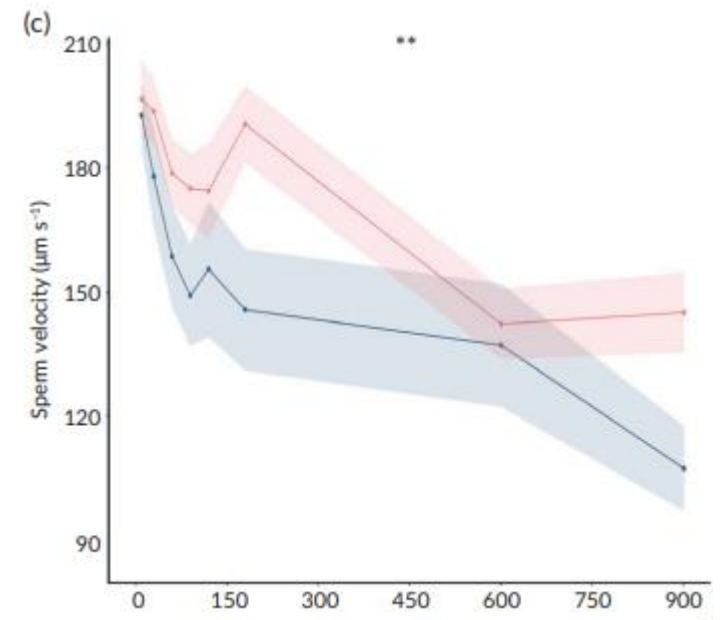
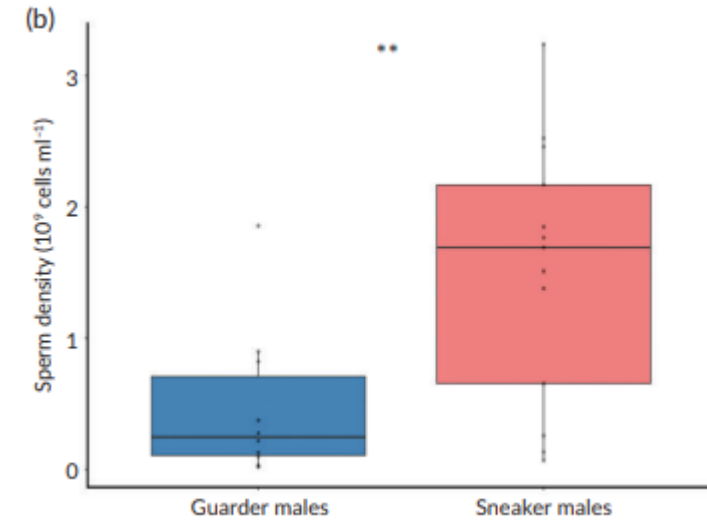
Postcopulatory consequences of female mate choice in a fish with alternative reproductive tactics

John L. Fitzpatrick,^{a,b,c} David J.D. Earn,^d Carol Bucking,^{b,c} Paul M. Craig,^{b,c} Sunita Nadella,^{b,c} Chris M. Wood,^{b,c} and Sigal Balshine^e

Dr. Sigal Balshine also found that:

- Sneaker males released 3X as much sperm as guarder males.
- Sneaker males produced significantly faster sperm than guarder males.

** = $p < 0.01$



REGULAR PAPER

JOURNAL OF FISH BIOLOGY

Sperm maturation and male tactic-specific differences in ejaculates in the plainfin midshipman fish *Porichthys notatus*

Jessica S. Miller¹ | Aneesh P.H. Bose^{1,2} | John L. Fitzpatrick³ | Sigal Balshine¹

Reproductive success

- Guarder males sired 52% - 58% of the young, on average
- Therefore, sneakers must have sired 42%-48% of the young, on average.

Comparing population level sexual selection in a species with alternative reproductive tactics

Karen M. Cogliati, Allison F. Mistakidis, Julie R. Marentette, Adrienne Lau,
Benjamin M. Bolker, Bryan D. Neff, Sigal Balshine [Author Notes](#)

Behavioral Ecology, Volume 25, Issue 6, November-December 2014, Pages 1524–1533,
<https://doi.org/10.1093/beheco/aru147>

Take home point – dimorphism in midshipmen fish (Type I guarders and Type II sneakers) can be explained by sexual selection (female mate choice, and male-male interactions)

Sexual selection will occur if three conditions are met.

The same as the three conditions for natural selection (environmental).

1. There is variation in a trait (e.g. differences in body size, or colour)
2. Variation (or trait) is heritable
3. Variants have differences in fitness due to differences in mating success or fertilization, resulting in different numbers of offspring.

Evolutionary Mechanism #4 – Genetic Drift

Genetic drift describes RANDOM (unpredictable) fluctuations in allele frequencies in a population over time/generations due to chance (sampling error).

“In each generation, **by CHANCE**, some “lucky” individuals in a population may, by chance, leave more descendants (and genes) than other “unlucky” individuals. It is NOT because the lucky individuals have better genes that give them a survival or reproductive advantage.” This can affect allele frequencies in the next generation. (*Berkeley Evolution webpage*)

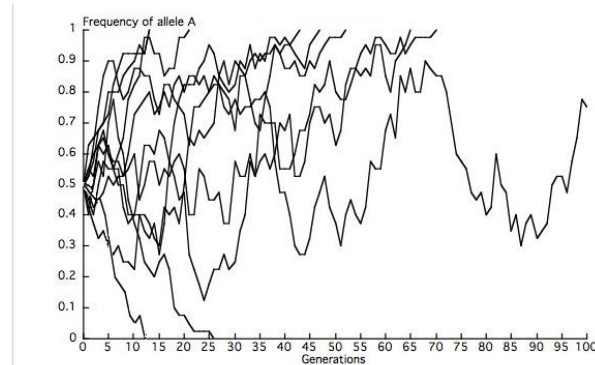


Figure 2: Simulations of allele-frequency change in 10 replicate populations ($N = 20$)
Since the initial frequency of the A allele = 0.5, we expect A to be fixed in 5 populations and lost in 5 populations, but our observations deviate from expectations because of the finite number of populations. In this run of simulations, we see 7 instances of fixation ($p = 1$), 2 instances of loss ($p = 0$), and one instance in which there are still two alleles after 100 generations. In this last population, A would eventually reach fixation or loss.
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<https://www.nature.com/scitable/knowledge/library/natural-selection-genetic-drift-and-gene-flow-15186648/>

Chance and survival – a personal story

Dan Tenaglia, incredible botanist and cyclist



http://www.missouriplants.com/Dans_page.html

Karen Luepke/Tenaglia/Hoksbergen, my field partner on Missouri Herp Team in 2001

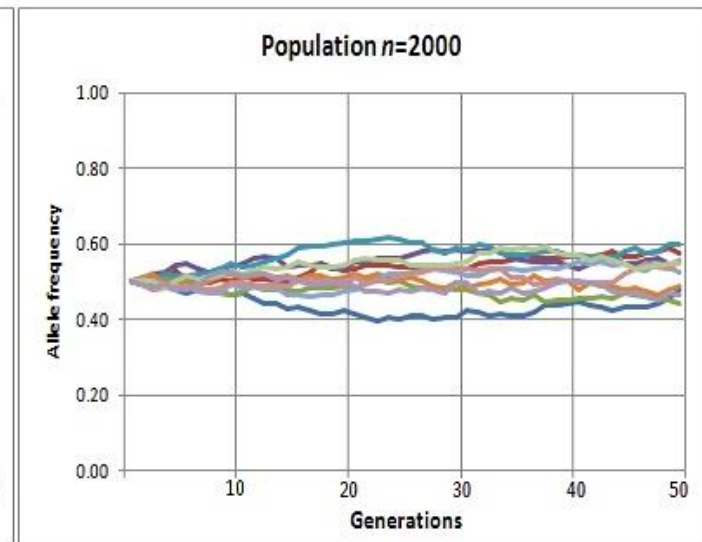
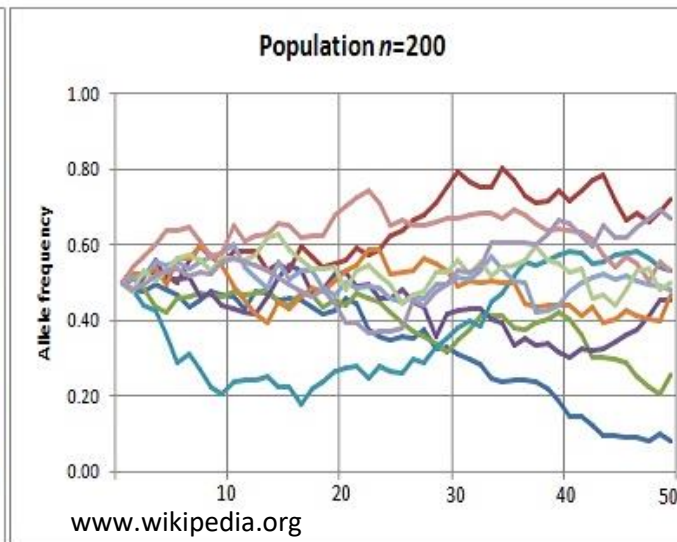
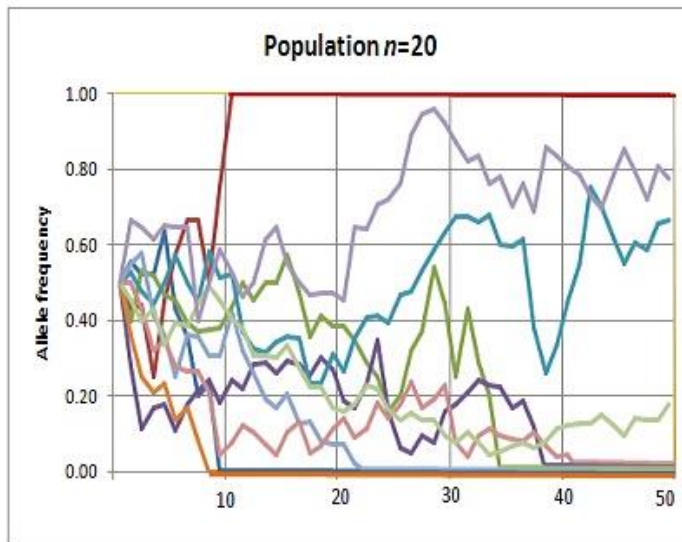


http://webhome.auburn.edu/~santosr/karen_tenaglia.htm

Genetic Drift

Important to understand:

- Genetic drift (i.e. random fluctuations in allele frequencies) occurs in ALL populations (including the human population) that are not infinite in size.
- Random fluctuations in allele frequencies tend to be more pronounced in smaller populations than larger populations.
- Small populations tend to lose genetic diversity (i.e. alleles) more quickly than larger populations (e.g. in first figure with a small population, an allele was lost within 10 generations).



Genetic Drift – effect of population size

<https://evound.shinyapps.io/AlleleA1Shiny/>

Alleleone website by Jon C. Herron, University
of Washington

<https://www.youtube.com/watch?v=Qt2qE0ceM9c>

Perhaps it is easiest to image genetic drift occurring with a decrease in population size



<https://evolution.berkeley.edu/evolution-101/mechanisms-the-processes-of-evolution/genetic-drift/>

On campus there is a very small population of beetles ($n=16$) with green ($n=7$) and brown ($n=9$) phenotypes. Imagine, brown colour is coded for by a C1 allele and green colour is coded for by a C2 allele.

A student walking between classes, accidentally steps on 2 of the green individuals, that did not survive. Would this affect the frequencies of the C1 and C2 allele in the population?

Originally a total of 32 C alleles in the population (16×2)

- Assuming beetles are homozygous

- Original frequency of C1 (brown allele) = $(9 \times 2) / 32 = 0.56$
- Original frequency of C2 (green allele) = $(7 \times 2) / 32 = 0.44$

After accident – 14 beetles (=28 C alleles):

- New frequency of C1 (brown allele) = $(9 \times 2) / 28 = 0.64$
- New frequency of C2 (green allele) = $(5 \times 2) / 28 = 0.36$



If the population size of beetles was larger (e.g. 10,000) would the death of those two beetles have had such a strong affect on allele frequencies?

Two opportunities for genetic drift – dramatic change in population size

1. (Population) Bottleneck Event
2. Founder Event

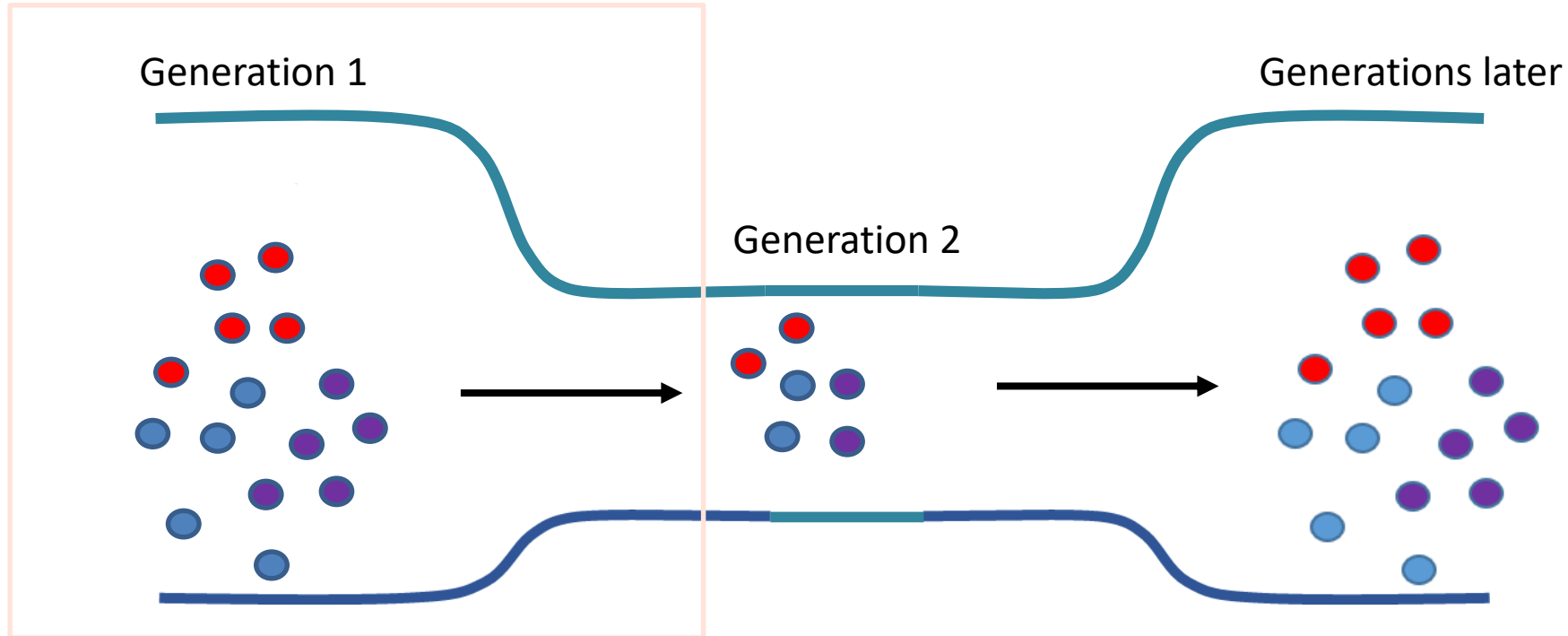
Both events involve a change in population size.

The new population is substantially smaller than original population
Allele frequencies in new population will likely differ from original population.

Important - please know that allele frequencies in a population can change by due to chance events with no change in population size.

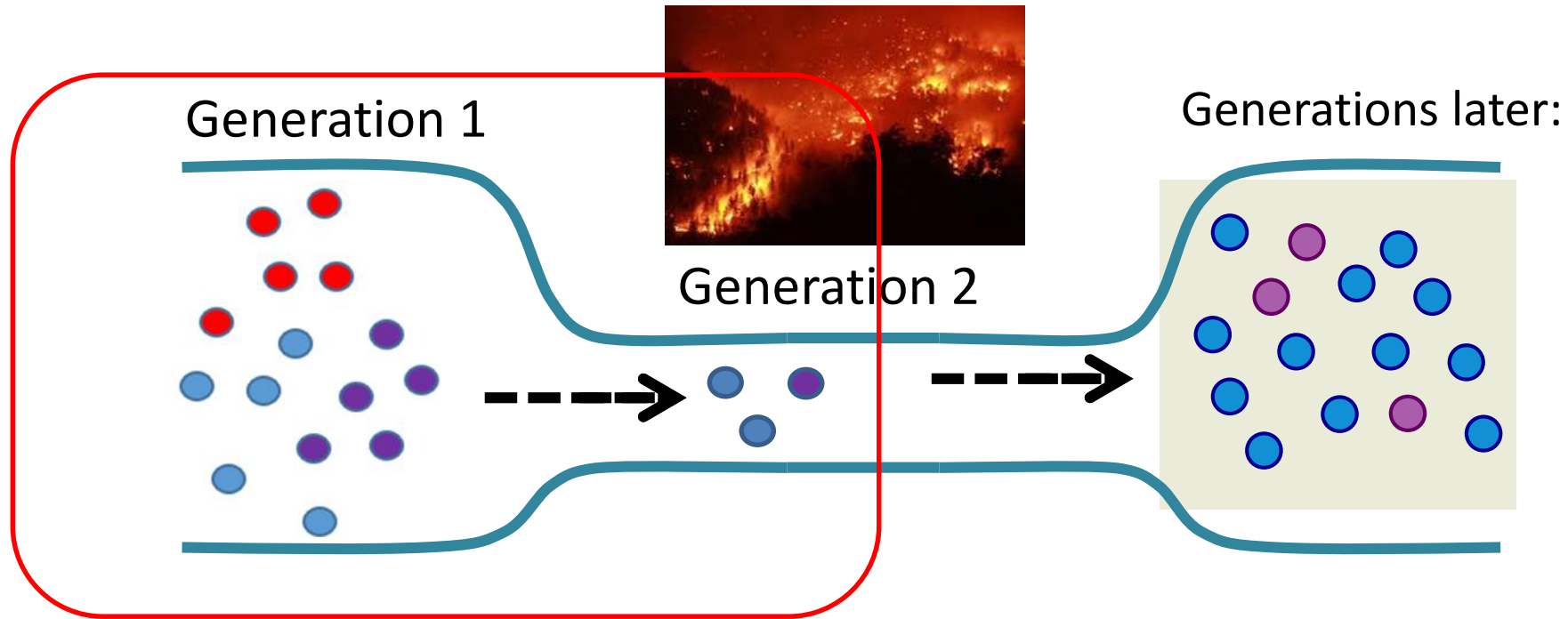
Population Bottleneck Event

Where the name bottleneck event comes from



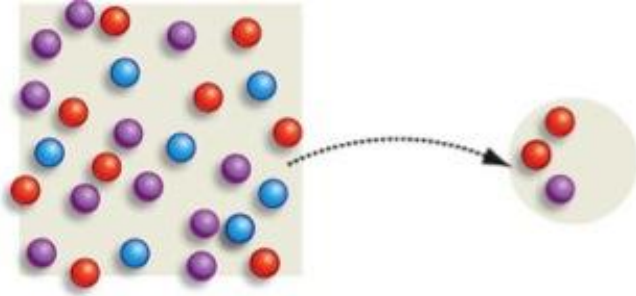
- Population size is greatly reduced (often over a short time period) for at least one generation (usually due to a catastrophe such as a wildfire, flood, etc.).

Example: Bottleneck event - with a change in allele frequencies



- Generation 1: 3 alleles (red, blue, purple) with equal frequencies (0.33 or 5/15)
- By chance, individuals blue and purple phenotypes survived the fire (lucky individuals).
- All individuals with a red phenotype perish
- Allele frequencies in generation 2 are very different, blue = 0.66, purple = 0.33, red=0.00;
- Red alleles missing = reduction in genetic variation
- Genetic drift has occurred

Founder Event – another opportunity for genetic drift



- A founder effect occurs when a new population is founded (started) by a few individuals.
 - Original parental population still exists.
 - Founding members isolated from the original parental population (so no gene flow)
 - Allele frequencies in the newly founded population will likely be very different from the original population (by chance)
 - Some alleles may be lost (reduced genetic variation, e.g. genetic bottleneck).
- | | |
|-----------------------------------|-----------------------------------|
| - In <u>parental population</u> : | In <u>founding population</u> : |
| purple - $11/27 = 0.41$; | purple – $1/3 = 0.33$ (decreased) |
| red - $9/27 = 0.33$ | red – $2/3 = 0.67$ (increased) |
| blue - $7/27 = 0.39$ | blue – $0/3 = 0.0$ (lost) |
- Allele frequencies have “drifted” due to random/chance events = genetic drift

Founder event - Invasive species

Can be significant losses in allelic diversity (i.e. number of different alleles) in the new population.



Saint John's wort

Molecular Ecology (2008) 17, 431–449

doi: 10.1111/j.1365-294X.2007.03538.x

Founding events in species invasions: genetic variation, adaptive evolution, and the role of multiple introductions

K. M. DLUGOSCH and I. M. PARKER

Ecology and Evolutionary Biology, University of California Santa Cruz, Santa Cruz, California 95064, USA



Journal List > Ecol Evol > v.6(15); 2016 Aug > PMC4984499

Ecology and Evolution

Open Access

Ecol Evol. 2016 Aug; 6(15): 5221–5233.

Published online 2016 Jun 28. doi: [10.1002/ece3.2278](https://doi.org/10.1002/ece3.2278)

PMCID: PMC4984499

PMID: [27551378](https://pubmed.ncbi.nlm.nih.gov/27551378/)

Genetic reconstruction of a bullfrog invasion to elucidate vectors of introduction and secondary spread

Pauline L. Kamath¹, Adam J. Sepulveda¹ and Megan Layhee¹

► Author information ► Article notes ► Copyright and License information [Disclaimer](#)

Rare alleles can increase in frequency in a population due to a founder event

The Pennsylvania Amish (Lancaster County) have a higher incidence* of polydactyly (extra fingers and/or toes), which can be traced back to Samuel King and his wife (who immigrated to Pennsylvania from what is now Germany in 1744)

There were part of the “founding 200 members” of this population.

It is assumed that Samuel King and/or his wife were carriers for the mutated recessive rare allele (EvC gene).

The frequency of this rare allele would be higher in the founding community (of approximately 200 individuals) than the original population

The Amish also observes strict endogamy (i.e. individuals marry within community); so, no introduction of new alleles (except through mutations). Not testable – but inbreeding can also result in higher homozygosity – and polydactyly is a recessive phenotype



<https://en.wikipedia.org/wiki/Polydactyly>

Genetic drift can happen **WITHOUT** a reduction in population size! (Testable)

Due to random events related to reproduction (rather than survival).

(Lucky) individuals being in the right place at the right time for reproduction resulting in more mating opportunities.

Example: Genetic drift with no change in population size

- We will look at 3 generations of rabbits, $n=10$ rabbits per generation (so no change in population size)
- 2 fur colours (brown and white) determined by B/b gene
- 1st generation: allele frequencies are $B=0.5$, $b=0.5$
- By chance, only five rabbits reproduce (circled)
 - 2 BB, 3 Bb, 0 bb

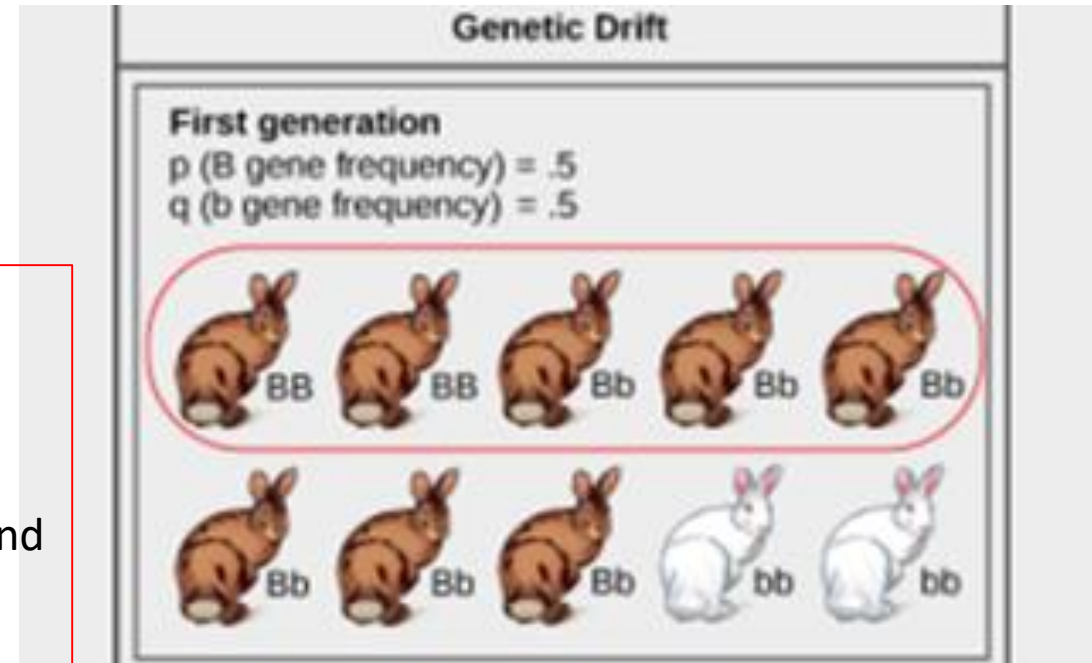
How allele frequencies were calculated:

Generation 1 – 10 rabbits; so 20 alleles in gene pool (10×2)

2 BB rabbits = 4 B alleles; 6 Bb rabbits = 6 B alleles and 6 b alleles; and
2 bb rabbits = 4 b alleles

- Total number of B alleles = $6 + 4 = 10$ $10/20 = f(B) = 0.5$

- Total number of b alleles = $6 + 4 = 10$ $10/20 = f(b) = 0.5$



- 2nd generation: the allele frequencies have changed to $B=0.7$ and $b=0.3$; so, genetic drift has occurred.

Calculating those generation 2 frequencies: 10 diploid rabbits = 20 alleles in population.

5 BB rabbits = 10 B alleles

4 Bb rabbits = 4 B alleles and 4 b alleles

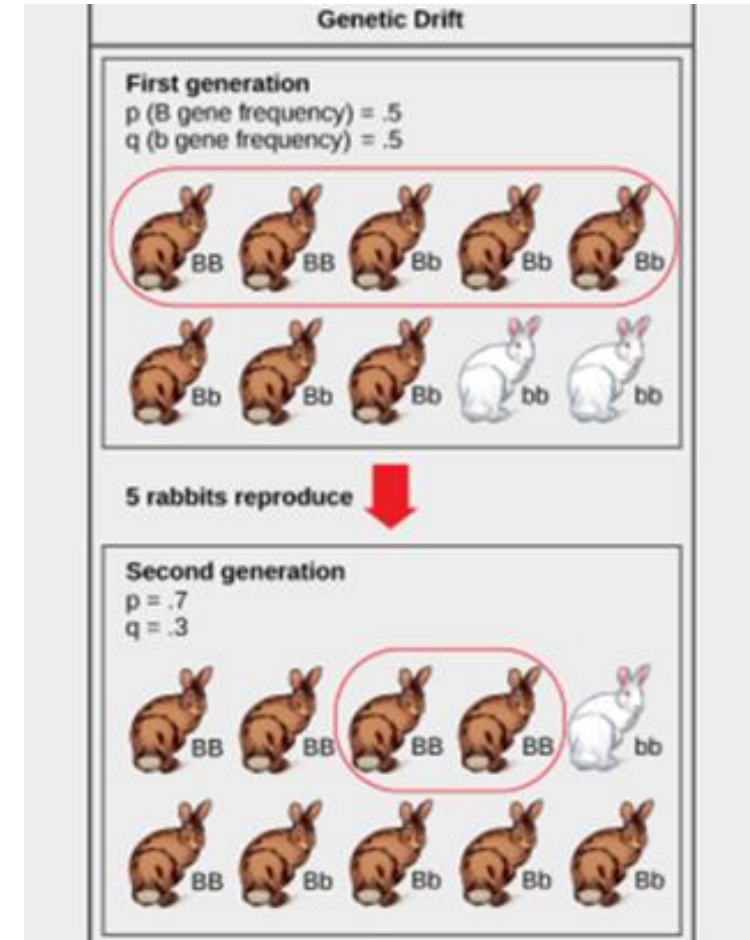
1 bb rabbit = 2 b alleles

Frequency of B allele in gen 2 = $(10 + 4) / 20 = 0.7$

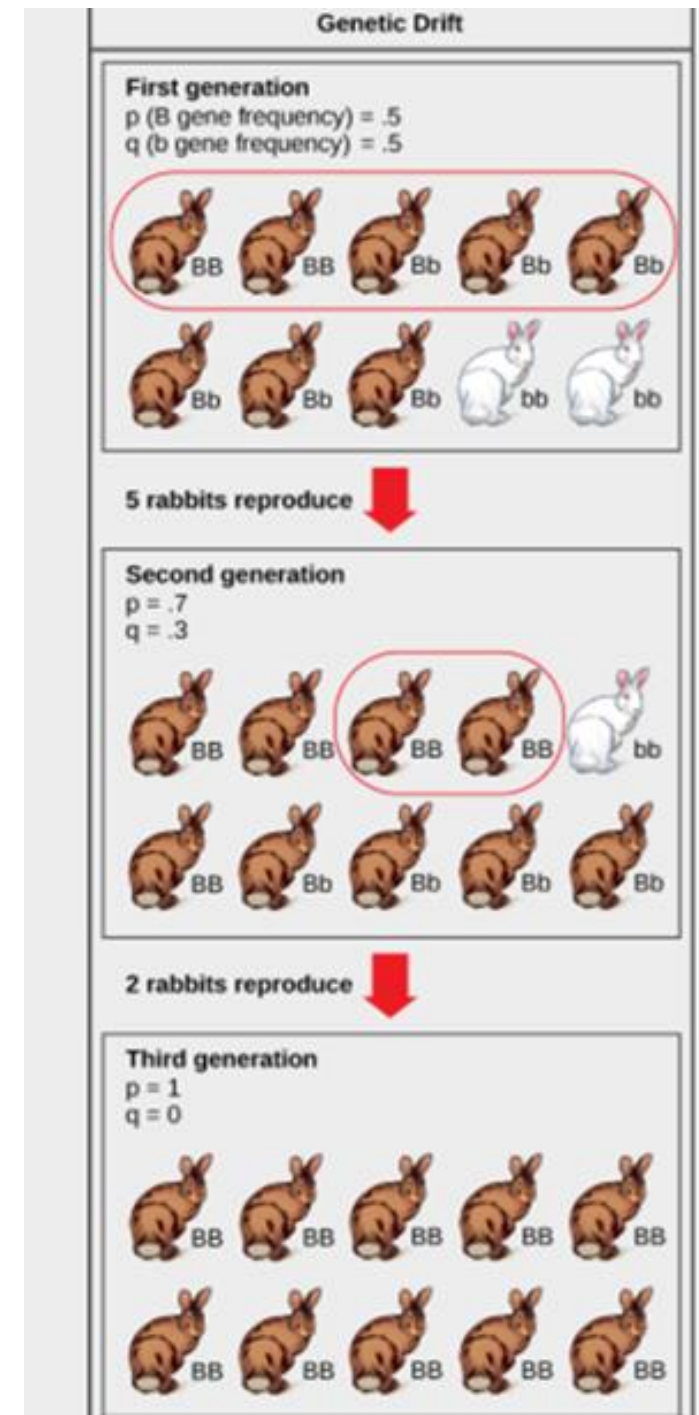
Frequency of b allele in gen 2 = $(4 + 2) / 20 = 0.3$

So both allele still present, but there is an increase in frequency of the B allele and a decrease in frequency of the b allele (drift has occurred)

- Only two (lucky) rabbits in this generation (2nd generation) reproduce. Both individuals have the genotype BB (circled).



- 3rd generation, $p(B)=1$ (fixed), $q(b)=0$ (lost)
- No white phenotype.
- Genetic drift (change in allele frequencies due to random events); but, no change in population size.

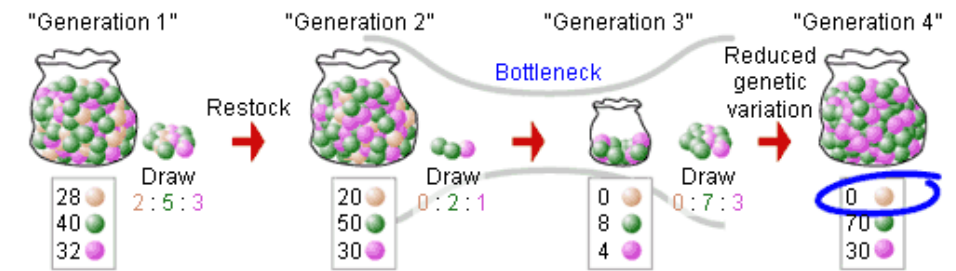


Genetic drift is the evolutionary equivalent of sampling error

Sampling error – a sample does not represent a population.

Imagine a starting population (generation 1) of 100 alleles.

- A sample of the population (gen 1) is taken
 - sample is not representative of population – so sampling error
 - 3 alleles present, but frequency has changed
 - population increases back to 100 individuals
- A sample of the population (gen 2) is taken
 - sample is not representative of the population
 - cream allele is lost, i.e. has drifted out of population
 - population increases back to 100 individuals
- A sample of the population is taken
 - sample is not representative of the population
 - Population increase back to 100 individuals; allele frequencies have changed
- With less genetic variation there is less for natural selection to act on.



<https://evolution.berkeley.edu/bottlenecks-and-founder-effects/>

Implications for a reduction in genetic variation

Northern elephant seals

- Population size reduced from >100,000 individuals to one population of 10-30 individuals by the end of the 19th century due to human hunting.
- Population has recovered to >80,000 individuals today.
- Reduced genetic variation.



www.wikipedia.org



<https://i.honesttopaws.com/animals-wild-facts>

Impact of a population bottleneck on symmetry and genetic diversity in the northern elephant seal (*Holzel et al.* 2002)

Findings:

- reduced genetic diversity
- entire genotypes lost
- increased asymmetry
- likely a function of developmental instability due to loss of alleles

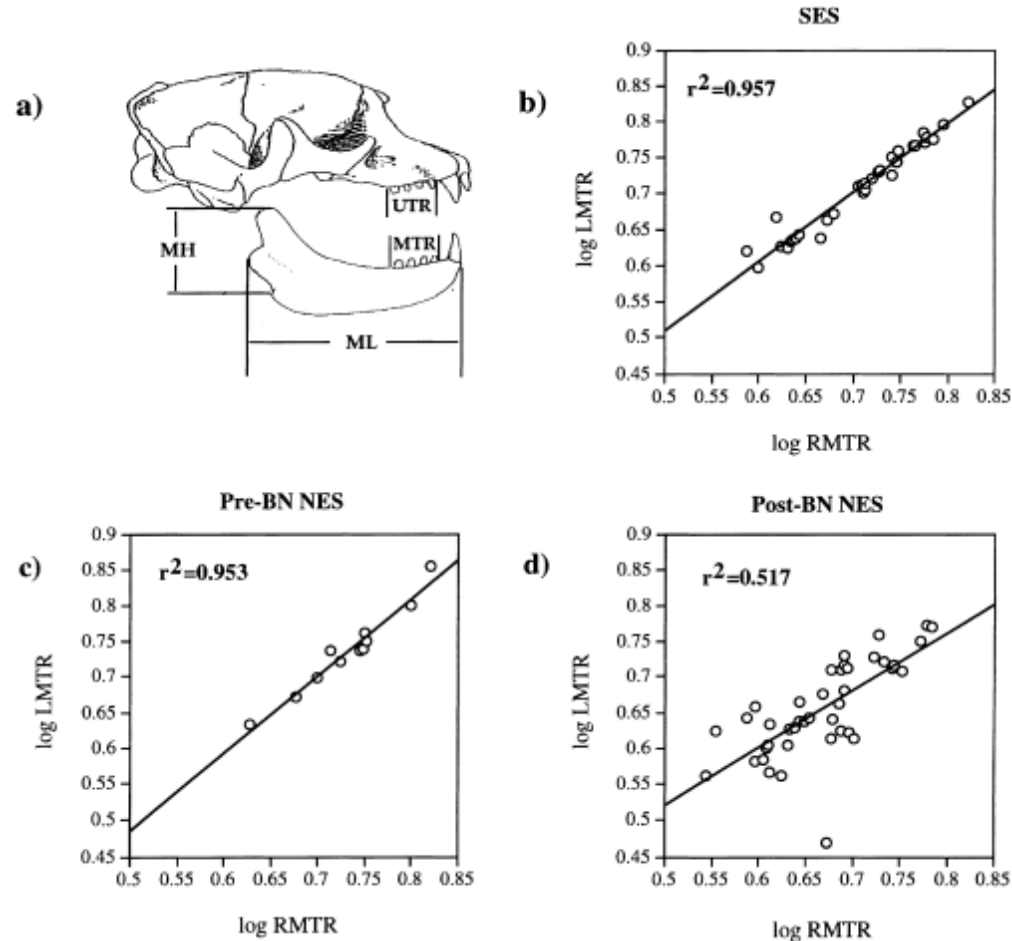
NES = Northern Elephant Seal

SES = Southern Elephant Seal

BN = Bottleneck

RMTR = Mandibular Tooth Row

LMTR = Upper Tooth Row



iClicker Question

What are the possible consequences of genetic drift?

- A. Reduction in the frequency of an allele
- B. Increase in the frequency of an allele
- C. Introduction of new alleles
- D. A & B
- E. All of the above

Answer

What are the possible consequences of genetic drift?

- A. Reduction in allele frequency
- B. Increase in allele frequency
- C. Introduction of new alleles
- D. A & B
- E. All of the above

iClicker Question

If genetic drift tends to reduce genetic variation in a population, what evolutionary mechanism(s) can increase genetic variation in a population?

- A. Mutation
- B. Gene flow
- C. Natural selection
- D. A & B
- E. A & C

Answer

If genetic drift tends to reduce genetic variation in a population, what evolutionary mechanism(s) can increase genetic variation in a population?

- A. Mutation
- B. Gene flow
- C. Natural selection
- D. A & B
- E. A & C

Activity #1 – 3 minutes: Answer the following question

The B gene in fish codes for body size.

There are two possible alleles:

- the B1 allele codes for large body size
- the B2 allele codes for small body size.

In 2000, a small population ($N=30$) of isolated fish in Spectacle Lake had equal frequencies of each allele (0.5:0.5).

In 2010, the allele frequencies had changed to 0.7 B1: 0.3 B2

There had been no change in the population size.

Explain how genetic drift could be the evolutionary mechanism that explains the change in allele frequencies in this fish population.

A complete answer should include:

- Definition of genetic drift should be clear.
- Clear explanation of how allele frequencies could change that is specific to the scenario BY CHANCE, i.e.
 - explains how there was an increase in the frequency of the B1 allele from 0.5 to 0.7
 - and a decrease in the frequency of the B2 allele from 0.5 to 0.3
 - in the Spectacle Lake fish population
 - over a decade (2000-2010)
 - With no change in population size ($N=30$)

One possible answer

Genetic drift occurs in a population when, by chance, some individuals in a population leave more descendants (and their alleles) than other individuals in the population, resulting in a change in allele frequencies in the next generation.

It is possible that, by chance, fish that were carrying the B1 allele had more opportunities to mate than the fish carrying the B2 allele. As a result, the fish carrying the B1 alleles would potentially leave more descendants than the fish carrying the B2 allele. This would result in an increase in the B1 allele frequency from 0.5 to 0.7 and a decrease in the B2 allele frequency from 0.5 to 0.3. between 2000 and 2010.

Given that the population size of the fish in Spectacle Lake is small ($N=30$), genetic drift could have a relatively large impact on the population, and the allele frequencies could have changed by 0.2 in just a decade.

Activity #2 – 3 minutes: Answer the following questions

The B gene in fish codes for body size. There are two possible alleles:
the B1 allele codes for large body size
the B2 allele codes for small body size.

In 2000, a small population ($N=30$) of isolated fish in Spectacle Lake had equal frequencies of each allele (0.5:0.5).

In 2008, the fish population declined from 30 fish to 5 fish due to drought.

In 2010, the fish population size had increased back to its original size of 30 fish, but the allele frequencies had changed from 0.7 B1 to 0.3 B2.

Explain how genetic drift could be the evolutionary mechanism that explains the change in allele frequencies in this fish population.

One possible answer

In 2008, the small fish population at Spectacle Lake went through a population bottleneck when the population decreased from 30 fish to 5 fish due to a drought. It is possible that, by chance, most of fish that survived the drought were carrying at least one B1 allele, whereas many of the fish that perished were carrying the B2 alleles. This would have resulted in an increase in the frequency of the B1 allele and a decrease in the frequency of the B2 allele in the surviving population.

The surviving fish would be the ones to pass their alleles to the next generation. This could explain why in 2010, even though the population had recovered, the allele frequencies in the Spectacle Lake fish population had a higher frequency of the B1 allele (0.7) and a lower frequency of the B2 allele (0.3) compared to the original 2000 population (0.5:0.5)

Just a fun biology fact – genetic bottleneck events & human genetic diversity



- One genetic bottleneck event when humans first left Africa to move to the Middle East (50,000-60,000 years ago).
- A second genetic bottleneck event occurred 19,000 miles away when humans migrated from Asia over the Bering Land Bridge.
- Genetic bottlenecks – likely due to founder events. Barriers like the Himalaya Mountains and Bering Sea acted as a barrier that held many people back; much smaller groups moved on.

Learning objectives for evolutionary mechanisms

Given a scenario:

C2. Be able to identify, describe, predict and/or explain how mutations, gene flow, selection and/or genetic drift may affect populations in terms of their genotype and phenotype frequencies through time.

C4. Be able to explain the steps (or conditions) necessary for selection and how they can lead to a population becoming more well adapted.

Be able to identify traits that are likely adaptations.

Be able to provide a logical justification for your conclusions supported by evidence from a scenario and reasons.

If requested, be able to describe the conditions required for natural selection/sexual selection, and/or the requirements for a trait to be considered an adaptation.

Evolutionary Mechanisms – Learning Objectives

C2 Predict and explain how the main mechanisms of evolution may affect populations in terms of their genotype and phenotype frequencies through time.

- Recognize situations where selection, drift, mutation or gene flow have likely impacted genotype or allele frequencies in a population
- Describe how evolutionary mechanisms can affect genotype and allele frequencies in a population.
- Given a description of a population or scenario, **identify** evolutionary mechanisms likely affecting allele or phenotypic frequencies and **describe** or **explain** how evolutionary mechanisms could affect allele or phenotypic frequencies. **Logically justify** your explanation with specific evidence from the description or scenario.
- **Describe and evaluate** the likely evolutionary mechanisms responsible for observed changes in genotype or phenotype within a population

- Terms and concepts related to evolutionary mechanisms (genetic drift, natural selection, artificial selection, sexual selection, gene flow, random mating, non-random mating, population size, finite or infinite population, bottleneck, founder effect, mutation, genotype, phenotype, fitness, random sampling)
- Requirements for evolution by natural selection.

Please see learning objectives file on Canvas homepage.

Before moving onto HWE

Please know that the four evolutionary mechanisms do not act in isolation of each other.

All four evolutionary mechanisms can be acting on a population at the same time.

It is possible for one mechanism (e.g. genetic drift) to mask, or counter, or amplify the effect of other mechanisms (e.g. natural selection, or gene flow).

For example, you might expect natural selection to eventually fix a beneficial allele in a population (i.e. beneficial allele will ultimately have a frequency of 1.0), but chance events (genetic drift) can overcome the effects of selection, especially if selection is weak.

Summary

Mechanism	Description	Effect on Genetic Variation	Effect on average fitness
Mutation	<ul style="list-style-type: none"> Refers to changes in an individual's DNA (sequences of nucleotides). Must occur in the gametes of an organism to be a mechanism for evolution (i.e., the change must be heritable). Random process Weak evolutionary force, but essential for evolution. 	<ul style="list-style-type: none"> Increased genetic variation by producing new alleles 	<ul style="list-style-type: none"> Not all mutations have fitness consequences (e.g. neutral mutations). Those that do have fitness consequences can have harmful or beneficial consequences. Fitness consequences may depend upon environment.
Gene flow	<p>Refers to the movement of genes into or out of populations due to individuals dispersing* and/or due to the movement of gametes.</p> <p>*note – individuals must mate with individuals in new population for gene flow to be complete.</p>	<p>May increase genetic variation (receiving population).</p> <p>May decrease genetic variation (by removing alleles)</p> <p>Can homogenize populations over time</p>	<p>Gene flow may increase, decrease or have no effect on average fitness depending upon the allele</p>

Summary

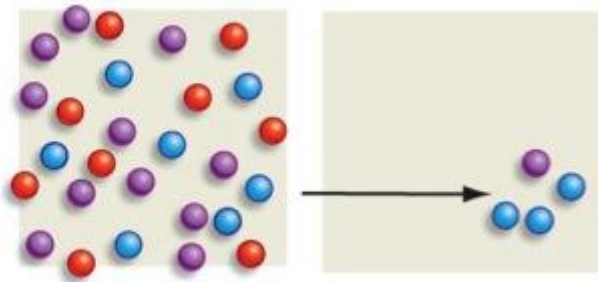
Mechanism	Description	Effect on Genetic Variation	Effect on average fitness
Natural Selection (ecological and sexual)	<ul style="list-style-type: none">Occurs when individuals with a heritable trait have higher fitness (via increased survival and offspring number) than individuals without the trait.Individuals with the trait pass their alleles to their offspring.This is a <u>non-random</u> process.Requires that individuals in the population vary in the trait; that the trait is heritable; and variation in the trait is linked to the individual's fitness.	<ul style="list-style-type: none">Natural selection tends to decrease genetic variation.Alleles that aid in survival and reproduction are much more likely to become common in the population than alleles that do not contribute to an organism's fitness.	<ul style="list-style-type: none">Increases fitnessOver generational time, natural selection can produce populations that have adapted to succeed in specific environments (e.g. mice.)
Genetic Drift	Changes in allele frequencies in a population over time due to chance/random events (or events that are random with respect to the allele of interest). - effect stronger in small populations.	Tends to reduce genetic variation (alleles are lost)	Drift is random with respect to fitness; usually reduces average fitness (elephant seals)

(Population) Bottleneck Event vs. Founder Event

Not shown in class

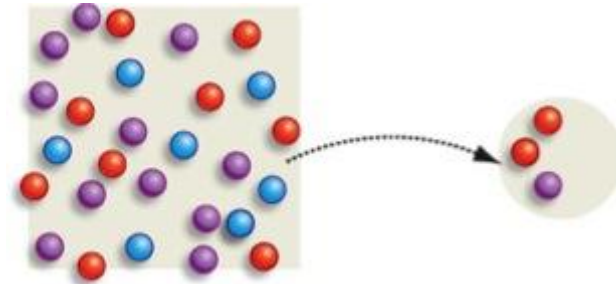
Bottleneck event:

- Existing population decreases in size by a large amount.
- Allele frequencies in the remaining population will likely differ from those in the earlier generation.



Founder effect:

- New population is founded
- Allele frequencies in new population differ from those in the parent population



Both types of events may result in a genetic bottleneck, i.e. reduction in allele diversity

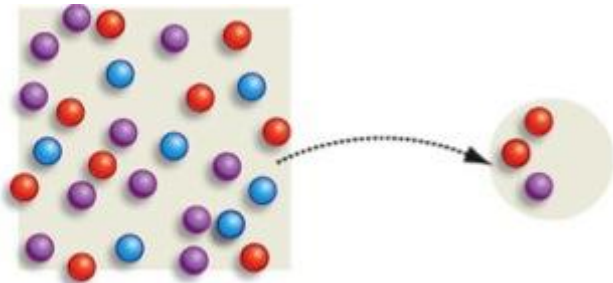
Founder effect vs. Gene flow

Not shown in class

Founder effect:

(can result in genetic drift)

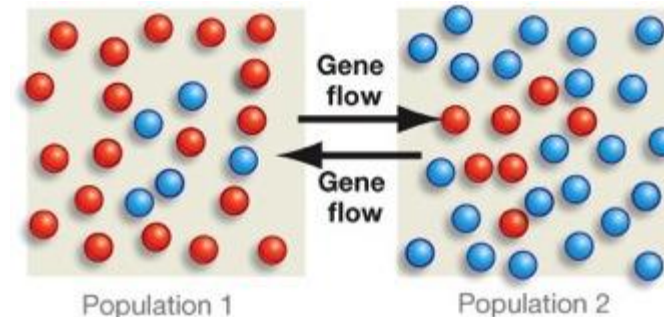
- Brand new population is founded from a random sample of individuals
- Can decrease genetic variation.
- Allele frequencies in new population may differ from those in the parent population



Gene flow:

(= movement of individual/alleles)

- Alleles entering or leaving an already established population.
- May increase genetic variation in receiving population.
- Allele frequencies in two populations can become more similar to one another



Genetic drift vs. Natural selection

Not shown in class

Genetic drift:

Change in allele frequency due to chance/random sampling(with respect to the gene of interest)

- Random sample of individuals survive and/or reproduce to pass alleles to the next generation
- Unpredictable
- Stronger effect on smaller populations

Natural selection:

Change in allele frequency due to specific sampling

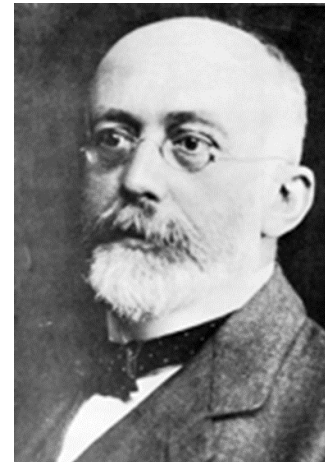
- Non-random sample of individuals survive/reproduce to pass alleles to the next generation.
- Predictable

Population genetics

How do we know if evolution is acting on a specific gene or not in a population?

- Use the Hardy-Weinberg Equation - developed independently by George Hardy and Wilhelm Weinberg in the early 1900s

$$p^2 + 2pq + q^2 = 1$$



Using the Hardy-Weinberg Equation

$$\begin{array}{ccc} \text{AA} & \text{Aa} & \text{aa} \\ \downarrow & \downarrow & \downarrow \\ p^2 & + & 2pq & + & q^2 = 1 \end{array}$$

This equation is used to calculate **EXPECTED genotype frequencies** in a population – if the gene is in HWE (meaning evolution is not acting on the gene)

HWE - null hypothesis

The expected (genotype) values calculated using the Hardy-Weinberg equation act as a null hypothesis:

- that evolution is NOT acting on the gene –

You compare these HWE expected genotype frequencies to the “OBSERVED” genotype frequencies in the population to determine if gene frequencies are changing, and evolution is occurring.

Step 1: Calculate the observed (real) genotype frequencies

For example: If there were 10 diploid individuals in a population



What is the frequency of AA genotype/individuals?

What is the frequency of Aa genotype/individual?

What is the frequency of aa genotype/individuals?

Step 1: Calculate observed (real) genotype frequencies

For example: If there were 10 diploid individuals in a population



What is the frequency of AA individuals? $6/10 = 0.6$

What is the frequency of Aa individual? $1/10 = 0.1$

What is the frequency of aa individuals? $3/10 = 0.3$

check arithmetic ($0.6 + 0.1 + 0.3 = 1$)

We made it this far in Thursday's lecture

We will pick up from this point in Tuesday's class.

Remainder of slides for this example included for students who would like to read ahead.

Step 2: Calculate the allele frequencies

p = freq. of A allele

q = frequency of a allele

$p+q=1$



Total number of alleles in population?

Number of A alleles in population?

Number of a alleles in population?

Frequency of p (A)?

Frequency of q (a)?

Step 2 – calculating allele frequencies

Step 2: Calculate the allele frequencies in the population



Total number of alleles = $10 \times 2 = 20$

Number of A alleles in population = 13

Frequency of A allele (p) = $(13) / 20 = 0.65$

Number of a alleles in population = 7

Frequency of a allele (q) = $(7) / 20 = 0.35$

- 6 individuals = AA $\longrightarrow 6 \times 2 = 12$ A alleles
- 1 individual = Aa $\longrightarrow 1$ A allele and 1 a allele
- 3 individuals = aa $\longrightarrow 3 \times 2 = 6$ a alleles

$$p + q = 1$$

Step 3: Calculate the EXPECTED genotype frequencies – if the population is in HWE - using observed allele frequencies

Genotype: AA Aa aa

 ↓ ↓ ↓

Expected p² + 2pq + q² = 1

Frequency:

p (A) = 0.65 q (a) = 0.35

	Expected Frequencies
AA – p ²	
Aa – 2pq	
aa – q ²	

Step 3: Calculating expected genotype frequencies (if gene is in HWE)

	Expected Frequencies
$AA - p^2$	$0.65^2 = 0.42$
$Aa - 2pq$	$2(0.65)(0.35) = 0.46$
$aa - q^2$	$0.35^2 = 0.12$



$$p(A) = 0.65 \quad q(a) = 0.35$$

Step 4: Compare observed and expected values

	Expected Frequencies	Observed frequencies
$AA - p^2$	$0.65^2 = 0.42$	$6/10 = 0.60$
$Aa - 2pq$	$2(0.65)(0.35) = 0.46$	$1/10 = 0.10$
$aa - q^2$	$0.35^2 = 0.12$	$3/10 = 0.30$

Are the expected and observed genotype frequencies close to each other (i.e. within 0.10)? If they are all close this gene is in HWE. Evolution is not acting on this gene.

If at least one of the expected and observed genotype frequencies are not close, which genotype frequencies were observed more frequently than expected? Less frequently than expected?

iClicker Question – Is this population in HWE for the A gene?

	Expected Frequencies	Observed frequencies
$AA - p^2$	$0.65^2 = 0.42$	$6/10 = 0.60$
$Aa - 2pq$	$2(0.65)(0.35) = 0.46$	$1/10 = 0.10$
$aa - q^2$	$0.35^2 = 0.12$	$3/10 = 0.30$

A. Yes

B. No

C. Not sure

iClicker Question – Is this population in HWE for the A gene?

	Expected Frequencies	Observed frequencies
AA – p^2	$0.65^2 = 0.42$	$6/10 = 0.60$
Aa – $2pq$	$2(0.65)(0.35) = 0.46$	$1/10 = 0.10$
aa – q^2	$0.35^2 = 0.12$	$3/10 = 0.30$

- A. Yes
- Observed more homozygous individuals (AA and aa) than expected if evolution not acting on gene and
and observed fewer heterozygotes (Aa) than expected.

B. No

- C. Not sure

For BIOL121 – if the difference between an observed and expected frequency is greater than 0.10, it okay to assume the difference is big enough to conclude that population is not in HWE for that gene.

Scientists would do a statistical analysis (e.g. Chi-Square test)

Hardy-Weinberg Equilibrium

Like all models, Hardy-Weinberg equation makes some assumptions (n=5) about the population (all related to evolutionary mechanisms):

- Random fusion of gametes (random mating)
 - Organisms do not select mates with respect to this gene
- The population is infinitely large
 - So, no changes in allele frequencies due to sampling error (i.e., **no Genetic Drift**)
- No natural selection
- No gene flow
- No mutations

If a population is not in HWE for a gene – at least one of these assumptions has been violated

Hardy-Weinberg Principle – Learning Objectives

Understand/know:

- The 5 assumptions for the Hardy-Weinberg Equilibrium (HWE) Model
- How to calculate:
 - Observed genotype frequencies in a population.
 - Allele frequencies in a population from observed genotype frequencies
 - Expected (predicted) genotype frequencies, under HWE conditions (i.e. no evolution), using allele frequencies.
- How to use this information to test whether a population is in HWE for a particular gene.
- If not in HWE, be able to provide a reason why a population might not be at HWE equilibrium for a particular gene (what assumption has been violated). Be able to explain your answer using evidence and reasoning.

Next class

- Tuesday: Finish HWE; if needed;
Phylogenetic trees
- Reminder – this Sunday due dates for Quiz 6 (Evolutionary Mechanisms) and Worksheet 7 (Natural Selection and Drift)