Lecture II – Evolution in Populations – Dr. Kopeny

Delivered 1/21, 1/23 and 1/25/02

Three female African Swallowtail Butterflies (*Papillio dardanus*) from the same population.

Mechanisms of Evolution:
How evolution happens to populations

Lecture Outline

1. Introductory terms and concepts
2. Darwin, Mendel and the Modern Synthesis
3. Genetic Variation within Populations
4. Genetic composition of a population may change intergenerationally; this is evolution, and one or more processes may contribute to that change
5. The effect of natural selection on frequency distribution of phenotypes and genotypes within a population varies depending on the nature of the selective force or forces operating on individuals
6. We know that variability is often maintained (not lost) over time in populations [ie, recognize a pattern], and we know something about the processes by which variability is maintained.
1. Introductory terms and concepts

**Concepts and Terms**

*Species*: A group of populations whose individuals have the potential to interbreed and produce fertile offspring in nature

*Population*: Localized group of individuals belonging to the same species

*Gene Flow*: Consequence of migration between populations, followed by breeding.

*Deme*: Locally interbreeding group within a population
Meiosis: Diploid parent cell to haploid daughter cells (gametes)

- Chromosomes replicate
- Homologous Pair of replicated chromosomes
- Homologous chromosomes separate
- Sister chromatids separate
Meiosis I; homologs distributed to daughter cells

Meiosis II; chromatids distributed to daughter cells

Review terms:
- Sexual reproduction
- Meiosis, Mitosis
- Diploid organism
- Homologous chromosomes
- Sister chromatids
- Gene
- Locus
- Allele

Sexual Reproduction fosters genetic diversity
- Random selection of half a parent’s diploid chromosome set to make a haploid gamete
- Fusion of two such haploid gametes to produce a diploid organism

Products of Meiosis are genetically diverse for two reasons
- Crossing over results in recombinant chromatids that contain some genetic material from each chromosome
- It is a matter of chance as to which member of a homologous pair of chromosomes goes to which daughter cell

Overview of Meiosis

Genotype: The genetic constitution governing a heritable trait of an organism

Phenotype: Physical expression of an organism’s genotype

Gene Pool: Sum of all alleles in a population
How to quantify the **genetic structure** of a population, for a discrete trait controlled by a single gene locus with one recessive and one dominant allele:

- **Allele Frequency**
- **Genotype Frequency**

<table>
<thead>
<tr>
<th>Phenotypes</th>
<th>Genotypes</th>
</tr>
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<tbody>
<tr>
<td></td>
<td>BB</td>
</tr>
<tr>
<td>Genotypes</td>
<td>0.36</td>
</tr>
<tr>
<td>Genotype Frequencies</td>
<td><strong>0.6B</strong></td>
</tr>
<tr>
<td>Allele Frequencies</td>
<td><strong>0.36 + 0.24 = 0.6B</strong></td>
</tr>
</tbody>
</table>

**Consider a population with 500 diploid cats and the single gene locus that controls fur color.**

- **For individuals in this or any other diploid species, how many times is each locus represented?**
  - Two (each individual has two “copies” of the gene).
- **How many different allelic forms are there in the population?**
  - Two (B, b)
- **How many alleles in the population?**
  - 1000
- **What defines an individual “homozygous” at a particular locus?**
  - Same allele at both loci  
  - BB or bb
- **What defines an individual that is “heterozygous” at a particular locus?**
  - Different alleles at the two loci  
  - Bb
Genotype frequencies: BB 36%  Bb 48%  bb 16%

**In a population of 500 individuals, how many individuals have genotype:**

- BB  $500 \times 0.36 = 180$ individuals
- Bb  $500 \times 0.48 = 240$ individuals
- bb  $500 \times 0.16 = 80$ individuals

**In a population of 500 individuals, how many copies of the B allele are there?**

- 360 alleles from the BB individuals (all the alleles from BB cats)
- 240 alleles from the Bb individuals (1/2 the alleles from Bb cats)

$= 600$ copies of the B allele

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Model of polygenic inheritance based on three genes

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Connecting population genetics and evolutionary change

• Adaptive Evolution

-Of the agents of evolutionary change, only selection is likely to adapt a population to its environment

–Adaptive evolution involves random chance in the form of mutation and sexual recombination and probabilistic sorting in terms of the sifting action of natural selection

• Individual Fitness

–Relative contribution of individual to gene pool of the next generation – relative to contribution of other individuals in population. Alternatively – can measure "lifetime fitness" – lifetime contribution to the gene pool.

• Genotype Fitness

–Contribution of genotype at a given locus to the next generation relative to contribution of other genotypes in population

• Fitness Quantified

–Relative scale from 0.0 to 1.0, where individual with greatest contribution in population has fitness of 1.0

2. Darwin, Mendel and the Modern Synthesis
Architects of the modern synthesis -- theoretical population geneticists whose work reconciled Mendelian theory of heredity with Darwin’s theory of natural selection

Ernst Mayr, on the right, on an ornithological expedition in New Guinea in 1928, with his Malay assistant
Fundamental Perspectives of the Modern Synthesis

• **The units of Evolution**: Populations are the fundamental units of evolution

• **The mechanism of Evolution**: Natural Selection is the most important mechanism of evolution in that it alone results in adaptive evolutionary change

• **The tempo of evolution**: Gradualism -- large change can and does evolve as an accumulation of small changes over long periods of time.

![Diagram of the Breach between Darwinism and Mendelism](image-url)

**Figure 1.8** Early Mendelians and biometricians. (a) Early Mendelians studied large differences between organisms, and thought that evolution happened when a new species evolved from a “macromutation” in its ancestor. (b) Biometricians studied small interindividual differences, and explained evolutionary change by the transition of whole populations. Mendelians were less interested in the reasons for small interindividual variation. This figure is a simplification to historical debate between two groups of scientists lasting for three decades can be fully represented in a simple diagrammatic contrast.
3. Genetic variation within populations

Examine the “instantaneous” genetic structure of a sexually reproducing diploid population.

• Allele frequencies
• Genotype frequencies

Examine the “intergenerational behavior” of genetic structure in a sexually reproducing diploid population that is not evolving

• “Hardy-Weinberg Equilibrium”

Consider the causes of microevolution (“non-equilibrium”)

• Natural selection
• Mutation
• Genetic drift
• Gene flow
• Non-Random mating

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### Determining the Genetic Structure of a Population (at one locus)

- **Genotype Frequencies**
- **Allele Frequencies**

<table>
<thead>
<tr>
<th>Phenotypes</th>
<th>Genotypes</th>
<th>Number of plants (total = 500)</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>AA</td>
<td>320</td>
</tr>
<tr>
<td></td>
<td>Aa</td>
<td>160</td>
</tr>
<tr>
<td></td>
<td>aa</td>
<td>20</td>
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</table>

<table>
<thead>
<tr>
<th>Genotype frequencies</th>
<th>Number of alleles in gene pool (total = 1000)</th>
<th>Allele frequencies</th>
</tr>
</thead>
<tbody>
<tr>
<td>AA 320/500 = 0.64</td>
<td>640 A</td>
<td>800/1000 = 0.8 A</td>
</tr>
<tr>
<td>Aa 160/500 = 0.32</td>
<td>160 A</td>
<td>200/1000 = 0.2 A</td>
</tr>
<tr>
<td>aa 20/500 = 0.04</td>
<td>40 a</td>
<td></td>
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</tbody>
</table>

\[ p = \text{frequency of } A = 0.8 \quad q = \text{frequency of } a = 0.2 \]
The genetic (allelic) structure of a population may remain relatively constant over time, ie stay in “equilibrium”

Allele frequencies may change intergenerationally; this is evolution, and one or more processes may cause that change

“Microevolution” refers to evolutionary change within and among populations – ie, within species.

One or more of five processes may drive microevolution:

mutation, selection, drift (chance), nonrandom mating, migration

Hardy-Weinberg Theorem

Mathematical theorem that shows, based on probability theory, that frequencies of alleles and genotypes in a population's gene pool remain constant over generations unless acted on by agents other than sexual recombination:

•Recombination of alleles, via gametes, due to meiosis and random fertilization has no effect on overall genetic structure of a population
Assumptions of “Hardy-Weinberg” Equilibrium of Genetic Structure of Populations:

No Mutation  No Random changes in genetic material

No Migration [Gene Flow] No Movement of individuals among populations and subsequent breeding

No Genetic Drift No Changes in genetic structure due to chance (dumb luck)

• No Non-Random Mating Gametes (eggs and sperm) are not combined in a completely random fashion

• No Natural Selection Differential reproduction

Hardy-Weinberg Theorem

• Each gamete carries one allele for flower color

• Allele frequencies among gametes is same as among diploid cells in parent population

For A, p = .8
For a, q = .2
Note: p+q=1
Hardy-Weinberg Theorem

• One assumption of model (theorem) is that mating in population is completely random

• All sperm and egg unions (fertilizations) are completely random - analogous to putting all gametes in a bag and randomly drawing sperm and eggs, one pair at a time, for each zygote

Hardy-Weinberg Theorem

Use *multiplication rule* of probability to determine frequency of genotypes in the next generation:

Probability of two independent events “co-occurring” equals the product of the probabilities of each independent event occurring.

(“Co-occur” = be in the same zygote via fertilization of egg by sperm)
Hardy-Weinberg Theorem -- Its Importance in the Study of Evolution

H-W is a "model" that defines a pattern in nature; the line of reasoning is grounded in mathematics -- probability theory and algebra

• H-W has predictive value

• H-W tells us what genetic structure to expect if a population is not evolving or can tell us about the extent to which a population is evolving
Using the Hardy-Weinberg Equation

Frequency of allele for an inherited disease; the recessive allele coding for PKU in the U.S. population (A=normal, a=pku)

\[ p^2 + 2pq + q^2 = 1 \]

Freq. AA  Freq. Aa  Freq. aa  100%

• PKU genotype = aa  Carrier genotype = Aa
• Freq. of PKU genotype = \( q^2 \)  Freq. Carrier genotype = \( 2pq \)
• Occurrence of PKU = 1 in 10,000 births = .0001
• Freq. of \( q^2 \) = .0001  Freq. of \( q = (.0001)^{1/2} = 0.01 \)
• Freq. of \( p = 1 - q = 1 - .01 = .99 \)
• Freq. of carriers (heterozyotes) = \( 2pq = (2)(.99)(.01) = .0198 (=\sim 2\%) \)

Allele frequencies may change intergenerationally; this is evolution, and one or more processes may cause that change

mutation, selection, drift (chance), nonrandom mating, migration
Natural Selection Can Cause Evolution

• Inclusive Fitness

Cooperation among Florida Scrub Jays. Helpers, mostly offspring from previous breeding season, improve inclusive fitness by feeding nestlings, defending nest, etc. (p.1126)

Mutation Can Cause Evolution

• **Heritable change in DNA.** Various forms of mutation, but most common is point mutation’ substitution of one nucleotide for another “accidentally” during the synthesis of a new DNA strand

• **Rate at which mutations occur vary** among nucleotide sites in a gene, among genes in an organism

• Mutation frequency typically less than one mutation per $10^4$ genes per DNA duplication
Non-Random Mating Can Cause Evolution

For H-W equilibrium to hold, individuals must select mates completely at random from a population, but often not the case

- Assortative Mating (individuals select as mates individuals that look like them - e.g. humans)
- Selection of “nearby” mates (promotes inbreeding -- mating of closely related individuals)
- Self fertilization - common in plants (extreme case of inbreeding)

Assortative mating in toads (*Bufo bufo*); males and females tend to mate with individuals similar in size to themselves (in Campbell 1999).

Genetic Drift Can Cause Evolution

Genetic Drift:
Random change in genetic structure of a population

Thought Experiment:

- What is your expectation regarding the probability of getting heads when you flip a coin? Tails? Why?
- Flip a coin 4 times, record results.
- Flip a coin 4000 times, record results.
- What is your expectation for both sets of trials?
- Which set of trials would you expect to be closer to your expectation?
- Why?
Sampling Error and Genetic Drift

**Sampling Error.** Chance events (random departures from expectations based on underlying probabilities) are more likely to occur in small populations than in large populations...

• Chance events are **random deviations from expected outcomes**.

• Chance events are **more likely in small “samples”** than in large “samples”

• This phenomenon is know as “**sampling error**”

*Let’s go from dimes and nickels to gametes and alleles…*

Relevance of Sampling Error/Genetic Drift to Population-Level Evolutionary Processes

• Following **reproduction in small populations**, genetic structure may change, i.e., evolution may happen, strictly due to random deviations from expected outcomes (allele and genotype frequencies in the next generation -- in the Punnett Square)

• A **population that experiences a bottleneck** -- size reduced to a small number -- may well experience an evolutionary shift in allele and genotype frequencies due to “sampling error” (small sample size of individuals that survive)

• A **small founding population**, isolated from population at large either by dispersal or vicariance, may well give rise to a new species in time, but will have started that trajectory towards speciation with allele and genotype frequencies different from the larger population from which the founding population arose
**Bottleneck Effect**: The likely consequence of a decrease in population size to very small number of individuals is that genetic structure of the surviving population is not representative of the original population.

Original population has about equal frequencies of red and yellow alleles

A chance environmental event greatly reduces the populations size

The surviving individuals have different allele frequencies from the original population...

...which generates a new populations with more red than yellow alleles

...in extreme cases, alleles are lost…purged from the population...

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Northern Elephant seals. Reduced to ~20 individuals in 1890’s. Now populations are large and growing, but show extremely low genetic variation. Is low variation a problem, a potential problem? Why?
Migration can cause evolution

Populations Size
N = 10 individuals

Allele Frequencies
A = 13/20 = .65
a = 7/20 = .35
Allele frequencies in the “pool of gametes” is the same;

A = .65

a = .35
Of evolutionary importance are point mutations and other mutations in germ line cells.
No net mutational change
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<td>Aa</td>
<td></td>
<td>Aa</td>
<td>AA</td>
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</tr>
</tbody>
</table>

**Migration (Gene Flow) Can Cause Evolution**

Deer Mouse population on west side of river

Deer Mouse population on east side of river

Deer Mouse population on west side of river

Deer Mouse population on east side of river
Pollination and Gene Flow

Dr. Galloway’s grad student, Linda Johnson is studying whether or not the origin of the plants affected the distances that birds carried the pollen and fertilized “target” flowers.
The effect of natural selection on frequency distribution of phenotypes and genotypes within a population varies depending on the nature of the selective force or forces operating on individuals.

**Natural Selection Affects Frequency Distribution of Phenotypes and Underlying Genotypes**

- Organismal-level *process* generates population-level *pattern*

- The outcome (pattern) depends on which forms of a heritable trait are favored by natural selection. Three principal “modes” of selection are:

  - **Stabilizing**
  - **Directional**
  - **Diversifying**
**Stabilizing Selection**

- *Culls extreme phenotypes* at both ends of distribution; phenotypes near mean are favored (pass on more alleles)
- *Reduces variation* and preserves existing mean phenotype, which is also most common if distribution is normal (bell-shaped)
- *Counters evolutionary change* because it counters shift in genetic structure

**Example of Stabilizing Selection**

Human infants with extremely high and low both weights have higher death rates.
Directional Selection

- Individuals at one extreme of distribution are “favored”
- Sustained directional selection leads to evolutionary trend
- Such trends may be reversed when environment changes

Example of Directional Selection
Pink Salmon and Artificial Selection
Example of Directional Selection

Example of Directional Selection in Galapagos Finches

(a) Beaks favored during drought years

(b) Beaks favored after the rains return
Disruptive Selection

- Environment favors phenotypic extremes; selection against intermediate phenotypes
- Sustained selection can leads to bimodal frequency distribution -- *balanced polymorphism*
- Occurrence rarely documented in nature

Example of Disruptive Selection

- Black billed Seed-crackers of West Africa
- Two main types of food available; large, hard seed, and small soft seeds
- Two bill polymorphisms are specialized to seed types - intermediates have low survival
We know that variability is maintained (not lost) over time in populations [ie, recognize a pattern], and we know something about the processes by which variability is maintained.

Genetic variation is the substrate of evolution.
Genetic variation is distributed in time and space.
The extent of genetic variation represents an equilibrium between agents that tend to reduce it, and agents that tend to create or maintain it.
Agents such as genetic drift, stabilizing selection, and directional selection tend to reduce genetic variation in a population.
What agents maintain genetic variation???

<table>
<thead>
<tr>
<th>Table 18-1</th>
<th>Genetic Polymorphism of Selected Enzymes within Plant and Animal Species</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Organism</strong></td>
<td><strong>Number of Species Examined</strong></td>
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<tr>
<td>Plants</td>
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<tr>
<td>Gymnosperms</td>
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<tr>
<td>Flowering plants (monocots)</td>
<td>111</td>
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<tr>
<td>Flowering plants (dikot)</td>
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<td>Invertebrates</td>
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<td>Land snails</td>
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<td>Vertebrates</td>
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<td>Fishes</td>
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<td>Reptiles</td>
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<tr>
<td>Birds</td>
<td>7</td>
</tr>
<tr>
<td>Mammals</td>
<td>46</td>
</tr>
</tbody>
</table>


Maintaining Genetic Variation

**Genetic polymorphism:**
- Two or more alleles at a locus in a population
- Polymorphism exists in genes… and gene products
- Table shows degree of genetic polymorphism in populations
- Genetic polymorphism is extensive in populations, although many of the alleles are present in low frequencies
Neutral Variation
Genetic variation that is invisible to natural selection; e.g. silent mutations

Example:
Variable nucleotide sites (top line) in mitochondrial gene in Margined Madtom (*Noturus insignis*)

```
101                                                150
AGCAGTAGAA GCCGCCCGCG AATACTTCT GCCGCCCGCG GCCTCCCGCG
151                                                200
GCACCTCTCT TATCCCGCG AATACTTCT GCCGGCCCGCG TACAGAGCTG
201                                                250
AAACATCTCT TGTTAACCC ACCCCGGCG ACCCGCGCG ACCCGCGCG ACCGCGCGCG
251                                                300
CACTGCTCT GTAAGTCC CAATCTCTCT TAATCTCTCT TAATCTCTCT TAATCTCTCT
301                                                350
GGTATAAGAG GCTAATACG AATACGCGA TAACTTCTCT CGACTCTGCA
3' End of NADH2 gene; *N. insignis* (variable sites in blue) Compared to *N. gilberti*.
```

Maintenance of Genetic Variation through Geographic Variation in Environmental Conditions

- **Geographic variation** in environmental conditions drives variation among populations
- **Clines**: gradual geographic shifts in frequency of genotypes and phenotypes are called clines
  - Recurring clinal variation in body size with temperature
  - Recurring clinal variation in external color with humidity
Clinal Variation in the Song Sparrow (Melospiza melodia)

• The song sparrow is found throughout North America. 31 subspecies, or “races” have been described.
• Along the west coast, the subspecies form well known cline in body size, plumage coloration and song characteristics, from small pale *M. m. saltonis* of desert southwest, to large dark *M. m. maxima* of aleutian islands.

Peregrine falcons show clinal variation in

*Peale’s* Peregrine Falcon of the Pacific Northwest and Aleutian Islands

*Anatum* Peregrine Falcon of the continental United States

*Tundra* Peregrine Falcon of the arctic tundra
Maintainence of Genetic Variation through Balanced Polymorphism

- **Balanced Polymorphism**: Special case of genetic polymorphism in which two or more alleles persist in a population over many generations as a result of natural selection
- Example: **Black-bellied seed crackers**. In this case, an obvious phenotypic polymorphism is associated with the underlying genetic polymorphism

Maintainence of Genetic Variation through Heterozygote Advantage

**Heterozygote Advantage.** Natural selection favors individuals heterozygous at a particular gene locus over individuals homozygous at that locus

- Distribution of Sickle Cell Anemia (red bars) and distribution of Falciparum malaria
- Homozygous dominants are at disadvantage due to anemia, homozygous recessives due to malaria.
Maintaining Genetic Variation: Frequency Dependent Selection

• Frequency-dependent selection. Fitness of particular phenotype depends on how frequently it appears in the population.

• Often maintains genetic variation in populations of species that are preyed on by a particular predator species.

• Demonstrated in water boatmen, which have three color phenotypes.