BGEN 55 - Advanced Genetics II

From Rearranging to Creating Variation

Week 5: Evolutionary Force I - Mutation

📚 Learning Objectives

After studying these notes, you will be able to:

- Contrast systems of mating with evolutionary forces
- Explain why mutation is considered the ultimate source of genetic variation
- Describe the fundamental properties of mutation as an evolutionary force
- Connect Week 4 concepts with the new focus on allele frequency changes

🔗 Bridging Week 4 and Week 5

Key Transition: We are moving from studying how existing variation is *rearranged* to understanding how new variation is *created* and how populations *evolve*.

Week 4 Recap: Systems of Mating

Last week, we explored how different mating systems affect genetic variation:

- **Random Mating:** Hardy-Weinberg equilibrium maintains constant genotype frequencies
- **Inbreeding:** Increases homozygosity, decreases heterozygosity
- **Assortative Mating:** Like genotypes mate preferentially
- **Disassortative Mating:** Unlike genotypes mate preferentially

Critical Insight: Systems of mating change **genotype frequencies** but do NOT change **allele frequencies** in the population gene pool.

Aspect	Systems of Mating (Week 4)	Evolutionary Forces (Week 5+)
What changes?	Genotype frequencies	Allele frequencies
Effect on variation	Rearranges existing variation	Creates/removes variation
Time scale	Immediate (one generation)	Gradual (multiple generations)
Examples	Inbreeding, assortative mating	Mutation, selection, drift, migration

🧬 Introduction to Mutation

What is Mutation?

Mutation is a heritable change in the DNA sequence. It is the *ultimate source* of all genetic variation in populations.

> **Fundamental Principle:** Every genetic difference between individuals, every allele in every gene pool, originated as a mutation at some point in evolutionary history.

Properties of Mutation as an Evolutionary Force

1. Weak but Constant

Mutation rates are typically very low:

$$\mu \approx 10^{-8} \text{ to } 10^{-11} \text{ per base pair per generation}$$
$$\text{or } 10^{-5} \text{ to } 10^{-6} \text{ per locus per generation}$$

This means mutation alone changes allele frequencies very slowly, but it acts continuously in every generation.

2. Random Process

Mutations occur:

- Without regard to organismal needs
- Without direction toward adaptation
- Randomly in time and location in the genome

3. Mostly Irreversible

While reverse mutations can occur, they are rare. For modeling purposes, we often treat mutation as a one-way process.

4. Diverse Effects

Mutations can have different impacts on fitness:

- **Neutral mutations:** No effect on fitness (most common)
- **Deleterious mutations:** Reduce fitness
- **Advantageous mutations:** Increase fitness (rare)

🎯 Looking Ahead

This week, we will explore two key models:

1. Recurrent Mutation Model

How does mutation pressure alone change allele frequencies over time?

$$p_1 = p_0(1 - \mu)$$
$$\Delta p = -\mu p_0$$

2. Mutation-Selection Balance

What happens when mutation creates deleterious alleles that selection removes?

$$\hat{q} = \sqrt{(\mu/s)} \text{ [for recessive deleterious alleles]}$$

Central Question: If natural selection efficiently removes deleterious alleles, why do genetic disorders persist in populations? The answer lies in mutation-selection balance.

💡 Key Takeaways

- Mutation is the **ultimate source** of all genetic variation
- Unlike mating systems, evolutionary forces change **allele frequencies**
- Mutation is a **weak but constant** force
- Most mutations are **neutral or deleterious**
- This week bridges rearrangement of variation to creation of variation

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