Bio1B Evolution 3

Last lecture:
• Natural selection - principles, lines of evidence in the “Origin”
• Descent with modification
• Estimation & interpretation of phylogeny
• Some major insights about the “Tree of Life”
  – 3 kingdoms: Archaea, Bacteria, Eukarya
  – Metazoan origins & relationships (not covered)

Today
• More history - Darwin+Mendel => the Neodarwinian synthesis
• Mechanisms of evolution:
  – Evolution in populations - population genetics
  – Allele, genotype and phenotype frequencies
  – Predicting genotype freq’s: Hardy (Castle) Weinberg Equilibrium
    • Application: Null model for evolution
    • Application: Predicting heterozygote frequencies for recessive traits
Mendel’s principles of inheritance (1865) [see Ch 14]

• Alternative versions of genes (alleles) account for variation in inherited characters

• For each character, an organism inherits 2 alleles, one from each parent

• If the 2 alleles at a locus differ, then the dominant allele determines phenotype

• The 2 alleles for a heritable character segregate during gamete formation (Law of Segregation)

• Each pair of alleles segregates independently of others during gamete formation [for unlinked genes]
Dominance of purple (P) over white (p) flower color: Fig. 14-5

- **P Generation**
  - Appearance: Purple flowers
  - Genetic makeup: PP
  - Gametes: P

- **F₁ Generation**
  - Appearance: Purple flowers
  - Genetic makeup: Pp
  - Gametes: 1/2 P, 1/2 p

- **F₂ Generation**
  - F₁ eggs: PP, Pp, pp
  - 3:1Ratio

Co-dominance - heterozygote is intermediate (pink) in snapdragons: Fig. 14.10

- **P Generation**
  - Red: C²R²
  - White: C²W²
  - Gametes: C², C², R², R², C², W², C², W²

- **F₁ Generation**
  - Pink: C²R²
  - Gametes: 1/2 C², 1/2 C²

- **F₂ Generation**
  - Eggs: 1/2 C²R², 1/2 C²W²
  - Sperm: 1/2 C²R², 1/2 C²W²
Genotype and allele frequencies for a locus with two alleles

**Genotypes**

- $A_1A_1$
- $A_1A_2$
- $A_2A_2$

**Alleles**

- $A_1$
- $A_2$

**Genotypes, parental generation**

- Genotype frequencies
- Parental generation

**Allele frequencies among gametes**

- Allele frequencies

**Genotype frequencies, offspring generation**

- Genotype frequencies
- Offspring generation
Hardy-Weinberg Equilibrium

**general case**

**male gametes**

\[ f(A_1) = p \quad f(A_2) = q \]

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**female gametes**

\[ f(A_2) = q \]

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**Expected genotype frequencies**

\[ A_1A_1 = p^2 \]
\[ A_1A_2 = 2pq \]
\[ A_2A_2 = q^2 \]

Gametes for each generation are drawn at random from the gene pool of the previous generation:

- 80% \( C^R \) (\( p = 0.8 \))
- 20% \( C^W \) (\( q = 0.2 \))

![Fig 23.7](image)

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Hardy-Weinberg Equilibrium

• Predicts genotype (& phenotype) frequencies from allele frequencies
• Genotype frequencies in expected proportions in a single generation
• Allele (& genotype) frequencies constant across generations => inheritance alone does not cause evolution
• Assumptions
  – Random mating (for this gene/trait)
  – No mutation, selection, migration
  – Large population (no drift)
Applications of HWE

- A null model for evolution
  - Deviations from expected proportions indicate something interesting - but what?
- Predicting frequency of heterozygotes for recessive alleles, e.g. cystic fibrosis

**Cystic fibrosis:** Mapped to chloride transport gene on chromosome 7

Common mutation, ΔF508 is recessive and at \( p = 0.02 \) in caucasian population

\[
F(\text{het}) = 2pq = 0.04 \text{ (carriers)}
\]

\[
F(\text{hom}) = p^2 = 0.0004 \text{ (affected)}
\]
Hardy-Weinberg genotype frequencies as a function of allele frequencies at a locus with two alleles

\[ q^2(A_2A_2) \]
\[ p^2(A_1A_1) \]
\[ 2pq(A_1A_2) \]