

# Principles of Genetics

## Class Schedule

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### Introduction

#### 1 Introduction

- Genetics might be described as the study of biological information flow. Defend this viewpoint.
- Genetics can be defined as the study of inheritance and of variation. Compare and contrast these views.
- What conceptual unification can you offer that integrates all of genetics into one coherent viewpoint?

#### 2 No class: Wednesday, 20 January

#### 3 Phenotypes: observations and measurements of traits

- What two basic observations motivate the study of genetics and the need for quantifying phenotypes?
- Define the term *phenotype* in a way that is both generally applicable to the full range of possibilities and clearly distinct from the term *genotype*. Justify the conceptual foundation of your definition.
- What two broad types of phenotypes are there and why must they be distinguished?
- Compare and contrast the terms *observation* and *measurement*.

#### 4 Distributions of traits: quantifying variation

- How is the variation within groups of individuals quantified?
- What two components compose a distribution for discrete, categorical characteristics.
- Interpret graphical and tabular representations of variation.
- Compare and contrast distributions for single traits and joint distributions for a collection of traits. What information is being conveyed by each?
- How can one recognize whether one discrete distribution represents a group with a greater or lesser degree of variation?

## Transmission of genetic information

### Mendel's principles

#### 5 Analysis of traits

- What were Mendel's critical insights that enabled him to unravel the general rules of inheritance?
- Why were the insights so important?
- Are all individuals with the same phenotype equivalent with respect to transmission of genetic information?
- How can one differentiate between different types of individuals with respect to the genetic information they transmit?

#### 6 Mendel's principles

##### Chapter 2

##### Problems: Chapter 2 C1

- What are Mendel's main principles?
- What observations support those principles?
- What are the two fundamental processes that these principles describe?

## 7 Cell division: mitosis and meiosis

## Chapter 3

Problems: Chapter 3 C7, C19

- How do cells divide so as to maintain the integrity of the information contained within a genome?
- What are the different types of cell division and how do they differ?
- What is the relationship between the genetically important events of independent segregation (Mendel's laws about segregation) and recombination and the physical events occurring during meiosis?

## 8 Basics of probability

## Chapter 2

Problems: Chapter 2 C15

Problems: Chapter 3 C13

- Define *probability*.
- Compare and contrast the terms *alternative events* and *joint events*. Compare and contrast the terms *mutually exclusive* and *not mutually exclusive*. Compare and contrast the terms *independent* and *dependent*. How do these last four terms relate to the first two?
- How is the probability of a set of *alternative events* calculated given information on the probability of each of the component events? Be sure you understand the general answer to this and how the simpler special case relates to that.
- How is the probability of a *joint event* calculated given information on the probability of each of the component events? Be sure you understand the general answer to this and how the simpler special case relates to that.
- Be sure you can recognize these concepts in practical situations, that you can apply the correct set of expressions, *and that you can illustrate clearly that you understand how to use these*.

## 9 Segregation: one locus

## Chapter 2

Problems: Chapter 2 C5, C6, E4

- How can the transmission of genetic information be modeled quantitatively?
- What is the probability distribution for gametes produced by each of the parents of Mendel's experiment?
- What is the probability distribution for the gametes produced by the  $F_1$  individuals of Mendel's experiment?
- What is the probability distribution for offspring genotypes produced by mating the parents of Mendel's experiment?
- What is the probability distribution for offspring genotypes produced by *back-crossing*  $F_1$  individuals with each of the parents of Mendel's experiment?
- What is the probability distribution for offspring genotypes produced by mating two  $F_1$  individuals?
- What is the probability distribution for the phenotypes produced in each of these crosses?
- What is the probability that the first three  $F_2$  seeds examined by Mendel are all wrinkled?
- What is the probability that the first three  $F_2$  seeds examined by Mendel were wrinkled, round, and wrinkled, in that order?
- What is the probability that the first three  $F_2$  seeds examined include only two wrinkled ones?
- What is the probability that a pair of mice, both heterozygous for the meiotic drive ( $T$ ) gene, will produce two  $Tt$  offspring?

## 10 Segregation: two independent loci

## Chapter 2

Problems: Chapter 2 C12, E10, E13

## 11 Segregation: two linked loci

Chapter 5

Problems: Chapter 5 C2, E1, E3, E7, E16

- Under what conditions is Mendel's law of independent segregation violated?
- How can one determine whether or not Mendel's law of independent segregation is, in fact, violated?
- What is the physical basis for nonindependent segregation?

**Prior problems due today**

## 12 Segregation: three linked loci

Chapter 5

Problems: Chapter 5 C6, E5, E9, E15, E22

## 13 Genetic linkage maps

Chapter 5

- How can nonindependent segregation be used to map the location of genes?
- How can the distance between genes be determined and what is the unit of measure for genetic linkage maps?
- What information is depicted in a genetic linkage map?
- Construct a linkage map from the data collected, for example, by Alfred Sturtevant.
- Construct a linkage map from data obtained from a trihybrid cross.

## 14 Mapping genetic traits

## 15 Applications of genetic maps: genetic counseling

**Exam**

## 16 Friday, 19 February 2009 (Chapters 2, 3, 5)

**Prior problems due today**

## Molecular genetics

### Physical storage of genetic information: DNA

17 Discovery of DNA's importance in genetics: Hershey and Chase

Chapter 9

Problems: Chapter 9 E1, E4, E6

- What evidence convinced scientists that DNA was the physical carrier of genetic information?

18 Structure of DNA

Chapter 9

Problems: Chapter 9 C4, C8, C13, C28, C34

- What are the basic structural properties of DNA?
- How does they differ from RNA?
- How do the structural properties influence the physico-chemical properties of these molecules?

19 Enzymes the manipulate DNA

Problems: Chapter 11 C7, C8, E8, E9

20 Biotechnology: PCR, LASA and sequencing

21 Genome structure: viral, prokaryotic, and eukaryotic genomes

Chapter 10

Problems: Chapter 9 C31

Problems: Chapter 10 C5, C6, C9, C15, C25

- What is a genome?
- What are the genetic and physical properties of entire genomes with respect to the following characteristics: size, gene content, chromosome organization, and physical arrangement?
- What constraints exist with respect to fitting genomes into cells?
- How are genomes organized so that they fit into cells?

## **Molecular aspects of transmission**

### 22 Central Dogma of Molecular Genetics

#### **Prior problems due today**

### 23 Overview of replication: semiconservative replication

Chapter 11

Problems: Chapter 11 C1, C3, C4, E1

- What is semiconservative replication and what evidence did Meselson and Stahl obtain to verify that hypothesis?
- What are the large scale features of DNA replication?

### 24 Enzymology of DNA replication

Chapter 11

Problems: Chapter 11 C6, C11, C16, E7

- How is DNA replication carried out enzymatically?
- How do the characteristics of DNA and the enzymes involved constrain the process of replication?

## **Molecular aspects of expression**

### 25 Prokaryotic transcription

Chapter 12

Problems: Chapter 12 C1, C12, C17, C25, C31, E1

- What is gene transcription and how does it fit within the Central Dogma of Genetics?
- Does all genetic information follow the tenets of the Central Dogma?
- What conventions are used to describe the locations of genetic elements and the nature of genetic information involved in transcription?
- How does the transcription process proceed in prokaryotes?

#### **Prior problems due today**

## 26 Translation

## Chapter 13

Problems: Chapter 13 C1, C2, C7, C9, C10, C29

- What is the meaning of genetic information?
- How is that meaning encoded within the genome?
- Explain the one gene-one enzyme hypothesis.
- What are the characteristics of the genetic code?
- How does polypeptide synthesis relate to the Central Dogma of Molecular Genetics?

## 27 Discovering the (almost) universal genetic code

## Chapter 13

Problems: Chapter 13 E1, E2

## 28 Enzymology of translation

## Chapter 13

Problems: Chapter 13 C23, C39

- What is the process of translating an mRNA into a polypeptide?
- What is the process of translating an mRNA into a polypeptide?
- Given data from a cell-free translation system (e.g., from Experiment 13A, page 329), what can you ascertain about the genetic code?
- What is the purpose of the 3 main sites in a ribosome?
- How does translation differ between eukaryotes and prokaryotes?
- What is the role of (some important factor) in the translation process?
- Why is the specificity of aminoacyl-tRNA synthetase so important?

**Regulation of gene expression**

29 General mechanisms of gene regulation and operons

Chapter 14

Problems: Chapter 14 C1, C2, C3, C5, C8

- How is gene expression controlled?
- Differentiate between repressors/activators, inducers/corepressors/inhibitors, inducible/repressible.
- Explain the regulatory mechanisms for the lac and trp operons.
- Why do inducible operons often encode catabolic enzymes, whereas repressible operons often encode anabolic enzymes?

**Prior problems due today**

30 Eukaryotic transcription factors

Chapter 15

Problems: Chapter 15 C1, C2, C4, C5, C18, E4

- What mechanisms control eukaryotic gene expression?
- How do they differ from prokaryotic mechanisms?

## Exam

31 Monday, 5 April (Chapters 9–15)

**Prior problems due today**

## Quantitative and population genetics

### Complex genetic traits

## 32 Nature of quantitative traits

Chapter 25

Problems: Chapter 25 C1, C4, C5, E1, E2

- What is the difference between quantitative and qualitative traits?
- What characteristics are used to describe probability distributions of quantitative traits?
- What descriptive statistics are used to describe probability distributions of quantitative traits and how are they calculated?

## 33 Multifactor models of genetic traits

Chapter 25

Problems: Chapter 25 C11

- What is the genetic basis for quantitative traits?
- What might cause two individuals to exhibit different values of a quantitative trait?
  - If they have the same (multilocus) genotype?
  - If they have different genotypes?
- What is a *QTL*?
- How can a *QTL* be found in a genome?

## 34 Genetic influence, heritability, and the response to selection

Chapter 25

Problems: Chapter 25 C20, C21, C22, C24, E17, E18

- What does heritability measure?
- What is the difference between broad- and narrow-sense heritability?
- How can the two forms of heritability be estimated from phenotypic observations?
- Why is heritability specific to a particular population within a particular environment?

## Organization of genetic information in populations

### 35 Introduction to population genetics

Chapter 24

Problems: Chapter 24 C1

- In what ways is population genetics similar and different from Mendelian genetics?
- How are genotype/allele/gamete frequencies calculated for a population?
- How does random mating create a relationship between allele frequencies and genotype frequencies?
- What is that relationship?

### **Prior problems due today**

### 36 Discovering the Hardy-Weinberg principle

### 37 Importance of false theories: the Hardy-Weinberg principle is both wrong and useful

Chapter 24

Problems: Chapter 24 C10, E3

### 38 Mechanisms of genetic change in populations: natural selection

Chapter 24

Problems: Chapter 24 C24, C28, E9

### 39 Random genetic drift and inbreeding

Chapter 24

Problems: Chapter 24 C15, C18, C20, C26

## 40 Migration

## Chapter 24

Problems: Chapter 24 C27, E4

- What mechanisms cause the composition of a population to change from generation to generation?
- What conditions allow the Hardy-Weinberg Association to persist?
- What factors determine the overall effect of mutation, genetic drift, and migration on allele frequencies?
- Which of these is likely to have the weakest effect?
- What are the effects of natural selection on a population?
- Define Darwinian fitness.

## Evolutionary genetics

## 41 Patterns of molecular evolution

## Chapter 26

Problems: Chapter 26 C1, C13, C16, C17, C19, C20, E7

- How do genes diversify in number and function?
- Explain the meanings of homology, orthology, and paralogy.
- What role does exon shuffling play in the diversification of gene function?

## Overview

## 42 Genetic functions, mechanisms and challenges

- From a mechanistic standpoint, how can the field of genetics be unified?
- What are the functions carried out by the genetic machinery?
- What challenges are faced in carrying out those functions?
- How do the processes involved in genetics at different scales relate to each other?

43 Understanding the diversification of life

- How can genetic information be used to understand the diversification of life?
- How does genetic information support the notion of a common ancestor for life?
- How does genetic information help understand the origin and diversification of complex traits?

**Prior problems due today**

## **Final Exam**

44 Monday, 3 May, 1:00–3:00