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 constrast 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 backcrossing \( F_1 \) 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 do 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 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